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PRIMARY LITERATURE RELEVANT TO BARTH SYNDROME BY TOPIC

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**Publications that acknowledge financial support contributed by BSF and/or BSF Affiliates.*

▼*Publications that acknowledge biological samples (and/or information) from Barth families, the Barth Syndrome Registry and Repository (BRR), and/or BSF Affiliates.*

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ANIMAL MODELS IN BARTH SYNDROME RESEARCH

NEW Kim J, Lee K, Fujioka H, Tandler B, Hoppel CL. **Cardiac mitochondrial structure and function in tafazzin-knockdown mice.** Mitochondrion. 2018 Oct 30. pii: S1567-7249(18)30074-6. doi: 10.1016/j.mito.2018.10.005. [Epub ahead of print] *(PubMed Abstract)** ▼

NEW Chao H, Anthonymuthu TS, Kenny EM, Amoscato AA, Cole LK, Hatch GM, Ji J, Kagan VE, Bayır H. **Disentangling oxidation/hydrolysis reactions of brain mitochondrial cardiolipins in pathogenesis of traumatic injury.** JCI Insight. 2018 Nov 2;3(21). pii: 97677. doi: 10.1172/jci.insight.97677. [Epub ahead of print] *(PubMed – Open Access)**

NEW Chowdhury A, Aich A, Jain G, Wozny K, Lüchtenborg C, Hartmann M, Bernhard O, Balleiniger M, Alfar EA, Zieseniss A, Toischer K, Guan K, Rizzoli SO, Brügger B, Fischer A, Katschinski DM, Rehling P, Dudek J. **Defective mitochondrial cardiolipin remodeling dampens HIF-1α expression in hypoxia.** Cell Rep. 2018 Oct 16;25(3):561-570.e6. doi:10.1016/j.celrep.2018.09.057. *(PubMed – Open Access)* ▼

NEW Suzuki-Hatano S, Saha M, Rizzo SA, Witko RL, Gosiker BJ, Ramanathan M, Soustek MS, Jones MD, Kang PB, Byrne BJ, Cade WT, Pacak CA. **AAV-mediated *TAZ* gene replacement restores mitochondrial and cardioskeletal function in Barth syndrome.** Hum Gene Ther. 2018 Aug 2. doi: 10.1089/hum.2018.020. [Epub ahead of print]* *(PubMed Abstract)*

- Cole LK, Kim JH, Amoscato AA, Tyurina YY, Bayır H, Karimi B, Siddiqui TJ, Kagan VE, Hatch GM, Kauppinen TM. **Aberrant cardiolipin metabolism is associated with cognitive deficiency and hippocampal alteration in *tafazzin* knockdown mice.** Biochim Biophys Acta. 2018 Jul 25. pii: S0925-4439(18)30271-0. doi: 10.1016/j.bbadis.2018.07.022. [Epub ahead of print]* *(PubMed Abstract)*

- Johnson JM, Ferrara PJ, Verkerke ARP, Coleman CB, Wentzler EJ, Neufer PD, Kew KA, de Castro Brás LE, Funai K. **Targeted overexpression of catalase to mitochondria does not prevent cardioskeletal myopathy in Barth syndrome.** J Mol Cell Cardiol. 2018 Jul 2;121:94-102. doi: 10.1016/j.yjmcc.2018.07.001. [Epub ahead of print] *(PubMed Abstract)*

- Xu G, Liu X, Shu Y, Pillai JA, Xu Y. **A rapid and sensitive LC-MS/MS method for quantitative analysis of cardiolipin (18:2)4 in human leukocytes and mouse skeletal muscles.** J Pharm Biomed Anal. 2018 Jun 20;158:386-394. doi: 10.1016/j.jpba.2018.06.035. [Epub ahead of print] *(PubMed Abstract)*

- Schafer C, Moore V, Dasgupta N, Javadov S, James JF, Glukhov AI, Strauss AW, Khuchua Z. **The effects of PPAR stimulation on cardiac metabolic pathways in Barth syndrome mice.** Front Pharmacol. 2018 Apr 11;9:318. doi:10.3389/fphar.2018.00318. eCollection 2018. *(PubMed Abstract)**

- Kimura T, Kimura A, Ren M, Berno B, Xu Y, Schlame M, Epand RM. **Substantial decrease in plasmalogen in the heart associated with *tafazzin* deficiency.** Biochemistry. 2018 Mar 20. doi: 10.1021/acs.biochem.8b00042. [Epub ahead of print] *(PubMed Abstract)* ▼

- Chao YJ, Wu WH, Balazova M, Wu TY, Lin J, Liu YW, Hsu YH. **Chlorella diet alters mitochondrial cardiolipin contents differentially in organs of Danio rerio analyzed by a lipidomics approach.** PLoS One. 2018 Mar 1;13(3):e0193042. doi:10.1371/journal.pone.0193042. eCollection 2018. *(PubMed - Open Access)*

- Damschroder D, Reynolds C, Wessells R. **Drosophila *tafazzin* mutants have impaired exercise capacity.** Physiol Rep. 2018 Feb;6(3). doi: 10.14814/phy2.13604. *(PubMed - Open Access)*

- Sullivan EM, Fix A, Crouch MJ, Sparagna GC, Zeczycki TN, Brown DA, Shaikh SR. **Murine diet-induced obesity remodels cardiac and liver mitochondrial phospholipid acyl chains with differential effects on respiratory enzyme activity.** J Nutr Biochem. 2017 Jul;45:94-103. doi: 10.1016/j.jnutbio.2017.04.004. Epub 2017 Apr 12. *(PubMed Abstract)*

- Fajardo VA, Mikhaeil JS, Leveille CF, Saint C, LeBlanc PJ. **Cardiolipin content, linoleic acid composition, and *tafazzin* expression in response to skeletal muscle overload and unload stimuli.** Sci Rep. 2017 May 17;7(1):2060. doi:10.1038/s41598-017-02089-1. *(PubMed - Open Access)*

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access)**

- Xu Y, Phoon CK, Berno B, D'Souza K, Hoedt E, Zhang G, Neubert TA, Epand RM, Ren M, Schlame M. **Loss of protein association causes cardiolipin degradation in Barth syndrome.** Nat Chem Biol. 2016 Jun 27. doi:10.1038/nchembio.2113. [Epub ahead of print] *(PubMed Abstract)**▼

- Szczepanek K, Allegood J, Aluri H, Hu Y, Chen Q, Lesnefsky EJ. **Acquired deficiency of *tafazzin* in the adult heart: Impact on mitochondrial function and response to cardiac injury.** Biochim Biophys Acta. 2016 Apr;1861(4):294-300. doi: 10.1016/j.bbalip.2015.12.004. Epub 2015 Dec 12. *(PubMed Abstract)*

- Kokoszka JE, Waymire KG, Flierl A, Sweeney KM, Angelin A, MacGregor GR, Wallace DC. **Deficiency in the mouse mitochondrial adenine nucleotide translocator isoform 2 gene is associated with cardiac noncompaction.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Available online 24 April 2016. *(ScienceDirect Abstract)*

- Lu YW, Galbraith L, Herndon JD, Lu YL, Pras-Raves M, Vervaart M, van Kampen A, Luyf A, Koehler CM, McCaffery JM, Gottlieb E, Vaz FM, Claypool SM. **Defining functional classes of Barth syndrome mutation in humans.** Hum Mol Genet. 2016 Feb 16. pii: ddw046. [Epub ahead of print] *(PubMed Abstract)**

- Dudek J, Cheng IF, Chowdhury A, Wozny K, Balleininger M, Reinhold R, Grunau S, Callegari S, Toischer K, Wanders RJ, Hasenfuß G, Brügger B, Guan K, Rehling P. **Cardiac-specific succinate dehydrogenase deficiency in Barth syndrome.** EMBO Mol Med. 2015 Dec 23. pii: e201505644. doi: 10.15252/emmm.201505644. [Epub ahead of print] *(PubMed – Open Access)*▼

BSF does not endorse any drugs, tests, or treatments that we may report.

- Mathur A, Ma Z, Loskill P, Jeeawoody S, Healy KE. ***In vitro* cardiac tissue models: Current status and future prospects.** Adv Drug Deliv Rev. 2015 Sep 30. pii: S0169-409X(15)00214-8. doi: 10.1016/j.addr.2015.09.011. [Epub ahead of print] Review. *(PubMed Abstract)*▼

- Cadalbert LC, Ghaffar FN, Stevenson D, Bryson S, Vaz FM, Gottlieb E, Strathdee D. **Mouse *tafazzin* is required for male germ cell meiosis and spermatogenesis.** PLoS One. 2015 Jun 26;10(6):e0131066. doi:10.1371/journal.pone.0131066. eCollection 2015. (*PubMed - Open Access*)*

- Huang Y, Powers C, Madala SK, Greis KD, Haffey WD, Towbin JA, Purevjav E, Javadov S, Strauss AW, Khuchua Z. **Cardiac metabolic pathways affected in the mouse model of Barth syndrome.** PLoS One. 2015 Jun 1;10(6):e0128561. doi: 10.1371/journal.pone.0128561. eCollection 2015. *(PubMed – Open Access)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Norris FC, Wong MD, Greene ND, Scambler PJ, Weaver T, Weninger WJ, Mohun TJ, Henkelman RM, Lythgoe MF**. A coming of age: Advanced imaging technologies for characterising the developing mouse.** Trends Genet. 2013 Dec;29(12):700-11. doi: 10.1016/j.tig.2013.08.004. Epub 2013 Sep 12. *(PubMed Abstract)*

- He Q, Wang M, Harris N, Han X. ***Tafazzin* knockdown interrupts cell cycle progression in cultured neonatal ventricular fibroblasts.** Am J Physiol Heart Circ Physiol. 2013 Aug 30. [Epub ahead of print] *(PubMed – Open Access)**▼

- Strathdee D, Cadalbert L, Ghaffar FN, et al. **Generation and analysis of a mouse model for Barth syndrome.** Transgenic Research Volume: 22 Issue: 1 Pages: 237-237 Meeting Abstract: 85. *(Meeting Abstract)*

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Phoon CKL, Acehan D, Schlame M, Stokes DL, Edelman-Novemsky I, Yu D, Xu Y, Viswanathan N, Ren M. ***Tafazzin* knockdown in mice leads to a developmental cardiomyopathy with early diastolic dysfunction preceding myocardial noncompaction**. J Am Heart Assoc. 2012 Apr;1(2). doi:pii: jah3-e000455. 10.1161/JAHA.111.000455. Epub 2012 Apr 24. *(PubMed – Open Access)**

- Powers C, Huang Y, Strauss A, Khuchua Z. **Diminished exercise capacity and mitochondrial bc1 complex deficiency in *tafazzin*-knockdown mice.** Front Physiol. 2013 Apr 17;4:74. doi: 10.3389/fphys.2013.00074. Print 2013. *(PubMed – Open Access)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Kiebish MA, Yang K, Liu X, Mancuso DJ, Guan S, Zhao Z, Sims HF, Cerqua R, Cade WT, Han X, Gross RW. **Dysfunctional cardiac mitochondrial bioenergetic, lipidomic, and signaling in a murine model of Barth syndrome.** J Lipid Res. 2013 Feb 14. [Epub ahead of print] *(PubMed – Open Access)**

- Acehan D, Vaz F, Houtkooper RH, James J, Moore V, Tokunaga C, Kulik W, Wansapura J, Toth MJ, Strauss A, Khuchua Z. **Cardiac and skeletal muscle defects in a mouse model of human Barth syndrome.** J Biol Chem. 2011 Jan 14;286(2):899-908. Epub 2010 Nov 9. *(PubMed – Open Access)**

- **Mouse model of Barth syndrome.** SciBX 3(47); Dec 9 2010. *(Nature Abstract)**

- Soustek MS, Falk D, Mah C, Toth M, Schlame M, Lewin A, Byrne B. **Characterization of a transgenic shRNA induced murine model of *tafazzin* deficiency.** Hum Gene Ther. 2010 Nov 23. [Epub ahead of print] *(PubMed Abstract)*▼

- Rijken PJ. **Phosphatidylcholine-protein interactions and remodeling of cardiolipin in yeast mitochondria.** Doctoral Thesis, Scheikunde Proefschriften, 2010. *(Full Text)**

- Rea SL, Graham BH, Nakamaru-Ogiso E, Kar A, Falk MJ. **Bacteria, yeast, worms, and flies: Exploiting simple model organisms to investigate human mitochondrial diseases.** Dev Disabil Res Rev. 2010 Jun;16(2):200-18. *(PubMed Abstract)*

- Chen S, Liu D, Finley RL Jr, Greenberg ML. **Loss of mitochondrial DNA in the yeast cardiolipin synthase crd1 mutant leads to up-regulation of the protein kinase Swe1p that regulates the G2/M transition.** J Biol Chem. 2010 Apr 2;285(14):10397-407. Epub 2010 Jan 19. *(PubMed – Open Access)**

- Beranek A, Rechberger G, Knauer H, Wolinski H, Kohlwein SD, Leber R. **Identification of a cardiolipin-specific phospholipase encoded by the gene CLD1 (YGR110W) in yeast.** J Biol Chem. 2009 Apr 24;284(17):11572-8. Epub 2009 Feb 25. *(PubMed Abstract)*

- Joshi AS, Zhou J, Gohil VM, Chen S, Greenberg ML. **Cellular functions of cardiolipin in yeast.** Biochim Biophys Acta. 2009 Jan;1793(1):212-8. Epub 2008 Aug 7. *(PubMed – Open Access)**

- Chen S, He Q, Greenberg ML. **Loss of *tafazzin* in yeast leads to increased oxidative stress during respiratory growth.** Mol Microbiol. 2008 May;68(4):1061-72. *(PubMed Abstract)**

- Zhong Q, Li G, Gvozdenovic-Jeremic J, Greenberg ML. **Up-regulation of the cell integrity pathway in saccharomyces cerevisiae suppresses temperature sensitivity of the pgs1Delta mutant.** J Biol Chem. 2007 Jun 1;282(22):15946-53. Epub 2007 Apr 9. *(PubMed – Open Access)**

- Li G, Chen S, Thompson MN, Greenberg ML. **New insights into the regulation of cardiolipin biosynthesis in yeast: Implications for Barth syndrome.** Biochim Biophys Acta. 2007 Mar;1771(3):432-41. Epub 2006 Jul 8. Review. *(PubMed Abstract)**

- Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, Ren M, Schlame M. **A Drosophila model of Barth syndrome**. Proc Natl Acad Sci U S A. 2006 Aug 1;103(31):11584-8. Epub 2006 Jul 19. *(PubMed – Open Access)**

- Khuchua Z, Yue Z, Batts L, Strauss AW. **A zebrafish model of human Barth syndrome reveals the essential role of *tafazzin* in cardiac development and function.** Circ Res. 2006 Jul 21;99(2):201-8. Epub 2006 Jun 22. *(PubMed – Open Access)**

- Davey KM, Parboosingh JS, McLeod DR, Chan A, Casey R, Ferreira P, Snyder FF, Bridge PJ, Bernier FP. **Mutation of DNAJC19, a human homolog of yeast inner mitochondrial membrane co-chaperones, causes DCMA syndrome, a novel autosomal recessive Barth syndrome-like condition.** J Med Genet. 2006 May;43(5):385-93. Epub 2005 Jul 31. *(PubMed Abstract)*

- Testet E, Laroche-Traineau J, Noubhani A, Coulon D, Bunoust O, Camougrand N, Manon S, Lessire R, Bessoule JJ. **Ypr140wp, 'the yeast *tafazzin*', displays a mitochondrial lysophosphatidylcholine (lyso-PC) acyltransferase activity related to triacylglycerol and mitochondrial lipid synthesis.** Biochem J. 2005 May 1;387(Pt 3):617-26. *(PubMed Abstract)*

- Ma L, Vaz FM, Gu Z, Wanders RJ, Greenberg ML. **The human *TAZ* gene complements mitochondrial dysfunction in the yeast *taz*1delta mutant-implications for Barth syndrome**. J Biol Chem. 2004 Oct 22;279(43):44394-9. Epub 2004 Aug 10. *(PubMed – Open Access)**

- Gu Z, Valianpour F, Chen S, Vaz FM, Hakkaart GA, Wanders RJA, Greenberg ML. **Aberrant cardiolipin metabolism in the yeast *taz1* mutant: A model for Barth syndrome**. Mol Microbiol 2004 Jan; 51(1):149-158. *(PubMed Abstract)**

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NEW Van Strien J, Guerrero-Castillo S, Chatzispyrou IA, Houtkooper RH, Brandt U, Huynen MA. **COmplexome profiling aLignment (COPAL) reveals remodeling of mitochondrial protein complexes in Barth syndrome.** Bioinformatics. 2019 Jan 14. doi: 10.1093/bioinformatics/btz025. [Epub ahead of print] *(PubMed Abstract)*

NEW Gürtler S, Wolke C, Otto O, Heise N, Scholz F, Laporte A, Elsner M, Jörns A, Weinert S, Döring M, Jansing S, Gardemann A, Lendeckel U, Schild L. ***Tafazzin*-dependent cardiolipin composition in C6 glioma cells correlates with changes in mitochondrial and cellular functions, and cellular proliferation.** Biochim Biophys Acta Mol Cell Biol Lipids. 2019 Jan 9. pii: S1388-1981(19)30003-4. doi: 10.1016/j.bbalip.2019.01.006. [Epub ahead of print] *(PubMed Abstract)*

NEW Antunes D, Chowdhury A, Aich A, Saladi S, Harpaz N, Stahl M, Schuldiner M, Herrmann JM, Rehling P, Rapaport D. **Overexpression of branched-chain amino acid aminotransferases rescues the growth defects of cells lacking the Barth syndrome-related gene *TAZ*1.** J Mol Med (Berl). 2019 Jan 3. doi: 10.1007/s00109-018-1728-4. [Epub ahead of print] *(PubMed Abstract)**

NEW Stöllberger C, Finsterer J. **Understanding left ventricular hypertrabeculation/ noncompaction: pathomorphologic findings and prognostic impact of neuromuscular comorbidities.** Expert Rev Cardiovasc Ther. 2018 Dec 20. doi: 10.1080/14779072.2019.1561280. [Epub ahead of print] *(PubMed Abstract)*

NEW Steward CG, Groves SJ, Taylor CT, Maisenbacher MK, Versluys B, Newbury-Ecob RA, Ozsahin H, Damin MK, Bowen VM, McCurdy KR, Mackey MC, Bolyard AA, Dale DC. **Neutropenia in Barth syndrome: Characteristics, risks, and management.** Curr Opin Hematol. 2018 Nov 16. doi: 10.1097/MOH.0000000000000472. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Vasilescu C, Ojala TH, Brilhante V, Ojanen S, Hinterding HM, Palin E, Alastalo TP, Koskenvuo J, Hiippala A, Jokinen E, Jahnukainen T, Lohi J, Pihkala J, Tyni TA, Carroll CJ, Suomalainen A. **Genetic basis of severe childhood-onset cardiomyopathies.** J Am Coll Cardiol. 2018 Nov 6;72(19):2324-2338. doi:10.1016/j.jacc.2018.08.2171. *(PubMed Abstract)*

NEW Kim J, Lee K, Fujioka H, Tandler B, Hoppel CL. **Cardiac mitochondrial structure and function in *tafazzin*-knockdown mice.** Mitochondrion. 2018 Oct 30. pii: S1567-7249(18)30074-6. doi: 10.1016/j.mito.2018.10.005. [Epub ahead of print] *(PubMed Abstract)**▼

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NEW Szeto HH. **Stealth peptides target cellular powerhouses to fight rare and common age-related diseases.** Protein Pept Lett. 2018 Oct 31. doi: 10.2174/0929866525666181101105209. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Chowdhury A, Aich A, Jain G, Wozny K, Lüchtenborg C, Hartmann M, Bernhard O, Balleiniger M, Alfar EA, Zieseniss A, Toischer K, Guan K, Rizzoli SO, Brügger B, Fischer A, Katschinski DM, Rehling P, Dudek J. **Defective mitochondrial cardiolipin remodeling dampens HIF-1α expression in hypoxia.** Cell Rep. 2018 Oct 16;25(3):561-570.e6. doi:10.1016/j.celrep.2018.09.057. *(PubMed – Open Access)*▼

NEW Chatzispyrou IA, Guerrero-Castillo S, Held NM, Ruiter JPN, Denis SW, IJlst L, Wanders RJ, van Weeghel M, Ferdinandusse S, Vaz FM, Brandt U, Houtkooper RH. **Barth syndrome cells display widespread remodeling of mitochondrial complexes without affecting metabolic flux distribution**. Biochim Biophys Acta Mol Basis Dis. 2018 Sep 1;1864(11):3650-3658. doi: 10.1016/j.bbadis.2018.08.041. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Dudek J, Hartmann M, Rehling P. **The role of mitochondrial cardiolipin in heart function and its implication in cardiac disease.** Biochimica et Biophysica Acta (BBA) - Molecular Basis of Disease, In press, corrected proof, Available online 26 August 2018. *(ScienceDirect Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

NEW Pennington ER, Sullivan EM, Fix A, Dadoo S, Zeczycki TN, DeSantis A, Schlattner U, Coleman RA, Chicco AJ, Brown DA, Shaikh SR. **Proteolipid domains form in biomimetic and cardiac mitochondrial vesicles and are regulated by cardiolipin concentration but not monolyso-cardiolipin.** J Biol Chem. 2018 Aug 29. pii: jbc.RA118.004948. doi: 10.1074/jbc.RA118.004948. [Epub ahead of print] *(PubMed - Open Access)*

NEW Hirono K, Hata Y, Nakazawa M, Momoi N, Tsuji T, Matsuoka T, Ayusawa M, Abe Y, Hayashi T, Tsujii N, Abe T, Sakaguchi H, Wang C, Takasaki A, Takarada S, Okabe M, Miyao N, Nakaoka H, Ibuki K, Saito K, Ozawa S, Nishida N, Bowles NE, Ichida F. **Clinical and echocardiographic impact of tafazzin variants on dilated cardiomyopathy phenotype in left ventricular non-compaction patients in early infancy.** Circ J. 2018 Aug 18. doi: 10.1253/circj.CJ-18-0470. [Epub ahead of print] *(PubMed - Open Access)*

NEW Suzuki-Hatano S, Saha M, Rizzo SA, Witko RL, Gosiker BJ, Ramanathan M, Soustek MS, Jones MD, Kang PB, Byrne BJ, Cade WT, Pacak CA. **AAV-mediated *TAZ* gene replacement restores mitochondrial and cardioskeletal function in Barth syndrome.** Hum Gene Ther. 2018 Aug 2. doi: 10.1089/hum.2018.020. [Epub ahead of print]*

- Cole LK, Kim JH, Amoscato AA, Tyurina YY, Bayır H, Karimi B, Siddiqui TJ, Kagan VE, Hatch GM, Kauppinen TM. **Aberrant cardiolipin metabolism is associated with cognitive deficiency and hippocampal alteration in *tafazzin* knockdown mice.** Biochim Biophys Acta. 2018 Jul 25. pii: S0925-4439(18)30271-0. doi: 10.1016/j.bbadis.2018.07.022. [Epub ahead of print]*

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Johnson JM, Ferrara PJ, Verkerke ARP, Coleman CB, Wentzler EJ, Neufer PD, Kew KA, de Castro Brás LE, Funai K. **Targeted overexpression of catalase to mitochondria does not prevent cardioskeletal myopathy in Barth syndrome.** J Mol Cell Cardiol. 2018 Jul 2;121:94-102. doi: 10.1016/j.yjmcc.2018.07.001. [Epub ahead of print]

- Basu Ball W, Baker CD, Neff JK, Apfel GL, Lagerborg KA, Žun G, Petrovič U, Jain M, Gohil VM. **Ethanolamine ameliorates mitochondrial dysfunction in cardiolipin-deficient yeast cells.** J Biol Chem. 2018 Jun 4. pii:jbc.RA118.004014. doi:10.1074/jbc.RA118.004014. [Epub ahead of print] *(PubMed - Open Access)*

- Sudrié-Arnaud B, Marguet F, Patrier S, Martinovic J, Louillet F, Broux F, Charbonnier F, Dranguet H, Coutant S, Vezain M, Lanos R, Tebani A, Fuller M, Lamari F, Chambon P, Brehin AC, Trestard L, Tournier I, Marret S, Verspyck E, Laquerrière A, Bekri S. **Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation.** Clin Chim Acta. 2018 Jun;481:1-8. doi: 10.1016/j.cca.2018.02.023. Epub 2018 Feb 22. *(PubMed Abstract)*

- Ban T, Kohno H, Ishihara T, Ishihara N. **Relationship between OPA1 and cardiolipin in mitochondrial inner-membrane fusion.** Biochim Biophys Acta. 2018 May 28. pii: S0005-2728(18)30132-4. doi: 10.1016/j.bbabio.2018.05.016. [Epub ahead of print] *(PubMed Abstract)*

- Cade WT, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, Bashir A, Byrne BJ, Taylor CL. **Peak oxygen uptake (VO2peak) across childhood, adolescence and young adulthood in Barth syndrome: Data from cross-sectional and longitudinal studies.** PLoS One. 2018 May 24;13(5):e0197776. doi: 10.1371/journal.pone.0197776. eCollection 2018 *(PubMed - Open Access)** ▼

- Boyd KJ, Alder NN, May ER. **Molecular dynamics analysis of cardiolipin and monolysocardiolipin on bilayer properties.** Biophys J. 2018 May 8;114(9):2116-2127. doi:10.1016/j.bpj.2018.04.001. *(PubMed Abstract)*

- Lou W, Reynolds CA, Li Y, Liu J, Hüttemann M, Schlame M, Stevenson D, Strathdee D, Greenberg ML. **Loss of *tafazzin* results in decreased myoblast differentiation in C2C12 cells: A myoblast model of Barth syndrome and cardiolipin deficiency.** Biochim Biophys Acta. 2018 Apr 22. pii: S1388-1981(18)30080-5. doi:10.1016/j.bbalip.2018.04.015. [Epub ahead of print] *(PubMed Abstract)**

- Schafer C, Moore V, Dasgupta N, Javadov S, James JF, Glukhov AI, Strauss AW, Khuchua Z. **The effects of PPAR stimulation on cardiac metabolic pathways in Barth syndrome mice.** Front Pharmacol. 2018 Apr 11;9:318. doi:10.3389/fphar.2018.00318. eCollection 2018. *(PubMed Abstract)**

- Oemer G, Lackner K, Muigg K, Krumschnabel G, Watschinger K, Sailer S, Lindner H, Gnaiger E, Wortmann SB, Werner ER, Zschocke J, Keller MA. **Molecular structural diversity of mitochondrial cardiolipins.** Proc Natl Acad Sci U S A. 2018 Apr 4. pii: 201719407. doi: 10.1073/pnas.1719407115. [Epub ahead of print] *(PubMed Abstract)*

- Mejia EM, Zegallai H, Bouchard ED, Banerji V, Ravandi A, Hatch GM. **Expression of human monolysocardiolipin acyltransferase-1 improves mitochondrial function in Barth syndrome lymphoblasts.** J Biol Chem. 2018 Mar 21. pii:jbc.RA117.001024. doi:10.1074/jbc.RA117.001024. [Epub ahead of print] *(PubMed - Open Access)** ▼

- Chen WW, Chao YJ, Chang WH, Chan JF, Hsu YHH. **Phosphatidylglycerol incorporates into cardiolipin to improve mitochondrial activity and inhibits inflammation.** Sci Rep. 2018 Mar 20;8(1):4919. doi: 10.1038/s41598-018-23190-z. *(PubMed Abstract)*

- Kimura T, Kimura A, Ren M, Berno B, Xu Y, Schlame M, Epand RM. **Substantial decrease in plasmalogen in the heart associated with *tafazzin* deficiency.** Biochemistry. 2018 Mar 20. doi: 10.1021/acs.biochem.8b00042. [Epub ahead of print] *(PubMed Abstract)* ▼

- Tsujii N, Hayashi T, Hayashi T, Kimura A, Nishikubo T. **Barth syndrome associated with triple mutation.** Pediatr Int. 2018 Mar 6. doi:10.1111/ped.13517. [Epub ahead of print]. *(PubMed - No abstract available)*

- Chao YJ, Wu WH, Balazova M, Wu TY, Lin J, Liu YW, Hsu YH. **Chlorella diet alters mitochondrial cardiolipin contents differentially in organs of Danio rerio analyzed by a lipidomics approach.** PLoS One. 2018 Mar 1;13(3):e0193042. doi:10.1371/journal.pone.0193042. eCollection 2018. *(PubMed - Open Access)*

- de Taffin de Tilques M, Lasserre JP, Godard F, Sardin E, Bouhier M, Le Guedard M, Kucharczyk R, Petit PX, Testet E, di Rago JP, Tribouillard-Tanvier D. **Decreasing cytosolic translation is beneficial to yeast and human *tafazzin*-deficient cells.** Microb Cell. 2018 Feb 18;5(5):220-232. doi: 10.15698/mic2018.05.629. *(PubMed – Open Access)**

- Damschroder D, Reynolds C, Wessells R. **Drosophila *tafazzin* mutants have impaired exercise capacity.** Physiol Rep. 2018 Feb;6(3). doi: 10.14814/phy2.13604. *(PubMed - Open Access)*

- Sullivan EM, Pennington ER, Sparagna GC, Torres MJ, Neufer PD, Harris M1, Washington J, Anderson EJ, Zeczycki TN, Brown DA, Shaikh SR. **Docosahexaenoic acid lowers cardiac mitochondrial enzyme activity by replacing linoleic acid in the phospholipidome.** J Biol Chem. 2018 Jan 12;293(2):466-483. doi: 10.1074/jbc.M117.812834. Epub 2017 Nov 21. (*PubMed Abstract)*

- Ohlig T, Le DV, Gardemann A, Wolke C, Gürtler S, Peter D, Schild L, Lendeckel U. **Effects of siRNA-dependent knock-down of cardiolipin synthase and *tafazzin* on mitochondria and proliferation of glioma cells.** Biochima et Biophysica Acta. Jan 6, 2018. (*PubMed Abstract*)

- Imai-Okazaki A, Kishita Y, Kohda M, Yatsuka Y, Hirata T, Mizuno Y, Harashima H, Hirono K, Ichida F, Noguchi A, Yoshida M, Tokorodani C, Nishiuchi R, Takeda A, Nakaya A, Sakata Y, Murayama K, Ohtake A, Okazaki Y. **Barth syndrome: Different approaches to diagnosis.** J Pediatr. 2017 Dec 15. pii: S0022-3476(17)31329-X. doi: 10.1016/j.jpeds.2017.09.075. [Epub ahead of print] *(PubMed Abstract)*

- Bozelli JC Jr, Hou YH, Epand RM. **Thermodynamics of methyl-ß-cyclodextrin-induced lipid vesicle solubilization: Effect of lipid headgroup and backbone.** Langmuir. 2017 Dec 5;33(48):13882-13891. doi: 10.1021/acs.langmuir.7b03447. Epub 2017 Nov 21. (*PubMed Abstract)*

- Abe M, Sawada Y, Uno S, Chigasaki S, Oku M, Sakai Y, Miyoshi H. **Role of acyl chain composition of phosphatidylcholine in *tafazzin*-mediated remodeling of cardiolipin in liposomes.** Biochemistry. 2017 Nov 28;56(47):6268-6280. doi: 10.1021/acs.biochem.7b00941. Epub 2017 Nov 9. (*PubMed Abstract)*

- Seneviratne AK, Aristizabal Henao JJ, Xu GW, Hurren R, Kim S, MacLean N, Wang X, Xu M, Fajado V, Hao Z, Gronda M, Jeyaraju DV, Chouinard-Watkins R, Schafer C, LeBlanc P, Khuchua Z, Bazinet R, Claypool S, Stark K, Schimmer A. ***Tafazzin* (*TAZ*) regulates the differentiation of AML cells by reducing levels of the phospholipid phosphatidylethanolamine.** Blood 2017 130:788. *(Abstract)*

- Ikon N, Hsu FF, Shearer J, Forte TM, Ryan RO. **Evaluation of cardiolipin nanodisks as lipidreplacement therapy for Barth syndrome.** J Biomed Res. 2017 Nov 1. doi: 10.7555/JBR.32.20170094. [Epub ahead of print] *(PubMed - Open Access)**

- Lorca R, Rozado J, Martin M. **Miocardiopatía no compactada: Breve revisión de una miocardiopatía con controversias (Non compaction cardiomyopathy: A short review of a controversial entity.)** Medicina Clinica Nov 2017. *(ScienceDirect – Abstract)*

- Dinca AA, Chien WM, Chin MT. **Identification of novel mitochondrial localization signals in human *Tafazzin*, the cause of the inherited cardiomyopathic disorder Barth syndrome.** J Mol Cell Cardiol. 2017 Nov 9. pii: S0022-2828(17)30340-1. doi:10.1016/j.yjmcc.2017.11.005. [Epub ahead of print] *(PubMed Abstract)**

- Dibattista M, Lobasso S, Stramaglia S, Corcelli A. **Assessing olfactory functions in patients with Barth syndrome.** PLoS One. 2017 Nov 3;12(11):e0187619. doi:10.1371/journal.pone.0187619. eCollection 2017. *(PubMed - Open Access)*▼

- Giacomelli E, Mummery CL, Bellin M. **Human heart disease: Lessons from human pluripotent stem cell-derived cardiomyocytes.** Cell Mol Life Sci. 2017 Oct;74(20):3711-3739. doi:10.1007/s00018-017-2546-5. Epub 2017 Jun 1. Review. *(PubMed - Open Access)*

- Pointer CB, Klegeris A. **Cardiolipin in central nervous system physiology and pathology.** Cell Mol Neurobiol. 2017 Oct;37(7):1161-1172. doi: 10.1007/s10571-016-0458-9. Epub 2016 Dec 30. Review. *(PubMed Abstract)*

- Enns GM. **Pediatric mitochondrial diseases and the heart.** Curr Opin Pediatr. 2017 Oct;29(5):541-551. doi: 10.1097/MOP.0000000000000535. *(PubMed Abstract)*

- Dudek J. **Role of cardiolipin in mitochondrial signaling pathways.** Front Cell Dev Biol. 2017 Sep 29;5:90. doi: 10.3389/fcell.2017.00090. eCollection 2017. Review. *(PubMed - Open Access)*

- Feingold B, Mahle WT, Auerbach S, Clemens P, Domenighetti AA, Jefferies JL, Judge DP, Lal AK, Markham LW, Parks WJ, Tsuda T, Wang PJ, Yoo SJ; American Heart Association Pediatric Heart Failure Committee of the Council on Cardiovascular Disease in the Young; Council on Clinical Cardiology; Council on Cardiovascular Radiology and Intervention; Council on Functional Genomics and Translational Biology; and Stroke Council. **Management of cardiac involvement associated with neuromuscular diseases: A scientific statement from the American Heart Association.** Circulation. 2017 Sep 26;136(13):e200-e231. doi: 10.1161/CIR.0000000000000526. Epub 2017 Aug 24. *(PubMed Abstract)*

- Towbin JA, Jefferies JL. **Cardiomyopathies due to left ventricular noncompaction, mitochondrial and storage diseases, and inborn errors of metabolism.** Circ Res. 2017 Sep 15;121(7):838-854. doi: 10.1161/CIRCRESAHA.117.310987. Review. *(PubMed Abstract)*

- Reynolds S, Burgess EM, Hymowitz N, Snyder DJ, Lane SJ. **Fungiform papilla number and olfactory threshold assessment in males with and without Barth syndrome.** Chemosensory Perception, 10(3), 60-68. (Sep 2017). *(PubMed Abstract)*

- Malhotra K, Modak A, Nangia S, Daman, TH, Gunsel U, Robinson VL, Mokranjac D, May ER, Alder NN. **Cardiolipin mediates membrane and channel interactions of the mitochondrial TIM23 protein.** Sci Adv. 2017 Sep 1;3(9):e1700532. doi: 10.1126/sciadv.1700532. eCollection 2017 Sep. *(PubMed - Open Access)*

- Saneto RP. **Genetics of mitochondrial disease.** Adv Genet. 2017;98:63-116. doi:10.1016/bs.adgen.2017.06.002. Epub 2017 Sep 1. *(PubMed Abstract)*

- Wang C, Hata Y, Hirono K, Takasaki A, Ozawa SW, Nakaoka H, Saito K, Miyao N, Okabe M, Ibuki K, Nishida N, Origasa H, Yu X, Bowles NE, Ichida F; for LVNC Study Collaborators. **A wide and specific spectrum of genetic variants and genotype-phenotype correlations revealed by next-generation sequencing in patients with left ventricular noncompaction.** J Am Heart Assoc. 2017 Aug 30;6(9). pii: e006210. doi:10.1161/JAHA.117.006210. *(PubMed Abstract)*
- Zhao Y, Feng Y, Ding X, Dong S, Zhang H, Ding J, Xia X. **Identification of a novel hypertrophic cardiomyopathy-associated mutation using targeted next-generation sequencing.** Int J Mol Med. 2017 Jul;40(1):121-129. doi: 10.3892/ijmm.2017.2986. Epub 2017 May 11. *(PubMed - Open Access)*
- Sullivan EM, Fix A, Crouch MJ, Sparagna GC, Zeczycki TN, Brown DA, Shaikh SR. **Murine diet-induced obesity remodels cardiac and liver mitochondrial phospholipid acyl chains with differential effects on respiratory enzyme activity.** J Nutr Biochem. 2017 Jul;45:94-103. doi: 10.1016/j.jnutbio.2017.04.004. Epub 2017 Apr 12. *(PubMed Abstract)*
- Menshykau D. **Emerging technologies for prediction of drug candidate efficacy in the preclinical pipeline.** Drug Discov Today. 2017 May 22. pii: S1359-6446(17)30238-6. doi: 10.1016/j.drudis.2017.04.019. [Epub ahead of print] (*PubMed Abstract)*
- Sheeran FL, Pepe S. **Mitochondrial bioenergetics and dysfunction in failing heart.** Adv Exp Med Biol. 2017;982:65-80. doi: 10.1007/978-3-319-55330-6_4. (*PubMed Abstract)*
- Fajardo VA, Mikhaeil JS, Leveille CF, Saint C, LeBlanc PJ. **Cardiolipin content, linoleic acid composition, and *tafazzin* expression in response to skeletal muscle overload and unload stimuli.** Sci Rep. 2017 May 17;7(1):2060. doi:10.1038/s41598-017-02089-1. *(PubMed - Open Access)*
- de Taffin de Tilques M, Tribouillard-Tanvier D, Tétaud E1, Testet E, di Rago JP1, Lasserre JP. **Overexpression of mitochondrial oxodicarboxylate carrier (ODC1) preserves oxidative phosphorylation in a yeast model of Barth syndrome.** Dis Model Mech. 2017 Apr 1;10(4):439-450. doi: 10.1242/dmm.027540. Epub 2017 Feb 10. *(PubMed – Open Access)*
- Ikon N, Ryan RO. **Cardiolipin and mitochondrial cristae organization.** Biochim Biophys Acta. 2017 Mar 20. pii: S0005-2736(17)30095-0. doi: 10.1016/j.bbamem.2017.03.013. [Epub ahead of print] Review. (*PubMed Abstract)*
- Finsterer J, Stollberger C. **Acquired noncompaction in Barth syndrome due to the *TAZ* mutation c.481_482ins20.** J Pediatr. 2017 Mar 15. pii: S0022-3476(17)30332-3. doi: 10.1016/j.jpeds.2017.02.066. [Epub ahead of print] (*PubMed Abstract)*
- Bakšiene M, Benušiene E, Morkuniene A, Ambrozaityte L, Utkus A, Kucinskas V. **A novel intronic splice site *tafazzin* gene mutation detected prenatally in a family with Barth syndrome.** Balkan J Med Genet. 2017 Mar 8;19(2):95-100. doi: 10.1515/bjmg-2016-0043. *(PubMed - Open Access)*

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access)**

- Lloyd DF, Vara R, Mathur S. **The cardiac manifestations of inherited metabolic diseases in children.** Pediatr Int. 2017 Mar 4. doi: 10.1111/ped.13272. [Epub ahead of print] Review. (*PubMed Abstract)*

- Schlame M, Xu Y, Ren M. **The basis for acyl specificity in the *tafazzin* reaction.** J Biol Chem. 2017 Mar 31;292(13):5499-5506. doi: 10.1074/jbc.M116.769182. Epub 2017 Feb 15. (*PubMed Abstract)*

- Bashir A, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, de Las Fuentes L, Pacak CA, Byrne BJ, Cade WT**. Impaired cardiac and skeletal muscle bioenergetics in children, adolescents, and young adults with Barth syndrome.** Physiol Rep. 2017 Feb;5(3). pii: e13130. doi: 10.14814/phy2.13130. *(PubMed – Open Access)*▼

- Wang J, Guo Y, Huang M, Zhang Z, Zhu J, Liu T, Shi L, Li F, Huang H, Fu L. **Identification of *TAZ* mutations in pediatric patients with cardiomyopathy by targeted next-generation sequencing in a Chinese cohort.** Orphanet J Rare Dis. 2017 Feb 10;12(1):26. doi: 10.1186/s13023-016-0562-4. *(PubMed – Open Access)*

- Sathappa M, Mitchell W, Coscia A, Boyd K, May E, Szeto HH, Alder NN. **Investigation of the interactions of the SS-31 peptides with cardiolipin variants: A potential therapeutic for Barth syndrome.** Biophysical Journal, Volume 112, Issue 3, Supplement 1, 3 February 2017, Pages 438a. (*ScienceDirect Abstract)*

- Dudek J, Maack C. **Barth syndrome cardiomyopathy.** Cardiovasc Res. 2017 Feb 2. doi: 10.1093/cvr/cvx014. [Epub ahead of print] No abstract available. *(PubMed)*

- Kagan VE, Bayır H, Tyurina YY, Bolevich SB, Maguire JJ, Fadeel B, Balasubramanian K. **Elimination of the unnecessary: Intra- and extracellular signaling by anionic phospholipids.** Review Article. Biochemical and Biophysical Research Communications, Volume 482, Issue 3, 15 January 2017, Pages 482-490. *(PubMed Abstract)**

- Jacob ML, Johnco C, Dane BF, Collier A, Storch EA. **Psychosocial functioning in Barth syndrome: Assessment of individual and parental adjustment.** Children's Health Care, 46:1, 66-92 (2017). *(Abstract)*▼

- Borna NN, Kishita Y, Ishikawa K, Nakada K, Hayashi JI, Tokuzawa Y, Kohda M, Nyuzuki H, Yamashita-Sugahara Y, Nasu T, Takeda A, Murayama K, Ohtake A, Okazaki Y. **A novel mutation in *TAZ* causes mitochondrial respiratory chain disorder without cardiomyopathy.** J Hum Genet. 2017 Jan 26. doi: 10.1038/jhg.2016.165. [Epub ahead of print] *(PubMed Abstract)*

- Schlame M, Greenberg ML. **Biosynthesis, remodeling and turnover of mitochondrial cardiolipin.** Biochim Biophys Acta. 2017 Jan;1862(1):3-7. doi:10.1016/j.bbalip.2016.08.010. Review. *(PubMed Abstract)*

- Shen Z, Li Y, Gasparski AN, Abeliovich H, Greenberg ML. **Cardiolipin regulates mitophagy through the PKC pathway.** J Biol Chem. 2017 Jan 5. pii: jbc.M116.753574. doi:10.1074/jbc.M116.753574. [Epub ahead of print] *(PubMed - Open Access)*

- Woiewodski L, Ezon D, Cooper J, Feingold B. **Barth syndrome with late-onset cardiomyopathy: A missed opportunity for diagnosis.** The Journal of Pediatrics, In Press, Corrected Proof, Available online 18 January 2017. *(Abstract)*

- Mejia EM, Zinko JC, Hauff KD, Xu FY, Ravandi A, Hatch GM. **Glucose uptake and triacylglycerol synthesis are increased in Barth syndrome lymphoblasts.** Lipids. 2017 Jan 17. doi: 10.1007/s11745-017-4232-7. [Epub ahead of print] *(PubMed Abstract)**

- Ikon N, Ryan RO. **Barth syndrome: Connecting cardiolipin to cardiomyopathy**. Lipids. 2017 Jan 9. doi: 10.1007/s11745-016-4229-7. [Epub ahead of print] *(PubMed Abstract)*

- Avdjieva-Tzavella DM, Todorova AP, Kathom H M, Ivanova MB, Yordanova IT, Todorov TP, Litvinenko IO, Dasheva-Dimitrova AT, Tincheva RS. **Barth syndrome in male and female siblings caused by a novel mutation in the *TAZ* gene.** Genet Couns. 2016;27(4):495-501. *(PubMed Abstract)*

- Tyurina YY, Lou W, Qu F, Tyurin VA, Mohammadyani D, Liu J, Hüttemann M, Frasso MA, Wipf P, Bayir H, Greenberg ML, Kagan VE. **Lipidomics characterization of biosynthetic and remodeling pathways of cardiolipins in genetically and nutritionally manipulated yeast cells.** ACS Chem Biol. 2016 Dec 16. [Epub ahead of print] *(PubMed Abstract)**

- Dong X, Fan P, Tian T, Yang Y, Xiao Y, Yang K, Liu Y, Zhou X. **Recent advancements in the molecular genetics of left ventricular noncompaction cardiomyopathy.** Clin Chim Acta. 2016 Dec 15;465:40-44. doi:10.1016/j.cca.2016.12.013. [Epub ahead of print] Review. *(PubMed Abstract)*

- Raja V, Joshi AS, Li G, Maddipati KR, Greenberg ML. **Loss of cardiolipin leads to perturbation of acetyl-CoA synthesis.** J Biol Chem. 2016 Dec 9. pii: jbc.M116.753624. [Epub ahead of print] *(PubMed – Open Access)**

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)**▼

- Kooijman EE, Swim LA, Graber ZT, Tyurina YY, Bayir H, Kagan VE. **Magic angle spinning ^{31}P NMR spectroscopy reveals two essentially identical ionization states for the cardiolipin phosphates in phospholipid liposomes.** Biochim Biophys Acta. 2016 Oct 26. pii: S0005-2736(16)30346-7. doi:10.1016/j.bbamem.2016.10.013. [Epub ahead of print] *(PubMed Abstract)**

BSF does not endorse any drugs, tests, or treatments that we may report.

- Oláhová M, Thompson K, Hardy SA, Barbosa IA, Besse A, Anagnostou ME, White K, Davey T, Simpson MA, Champion M, Enns G, Schelley S, Lightowlers RN, Chrzanowska-Lightowlers ZM, McFarland R, Deshpande C, Bonnen PE, Taylor RW. **Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria.** J Inherit Metab Dis. 2016 Sep 30. *(PubMed Abstract)*

- Pérez-Serra A, Toro R, Sarquella-Brugada G, de Gonzalo-Calvo D, Cesar S, Carro E, Llorente-Cortes V, Iglesias A, Brugada J, Brugada R, Campuzano O. **Genetic basis of dilated cardiomyopathy.** International Journal of Cardiology, Available online 21 September 2016. *(ScienceDirect Abstract)*

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review. *(PubMed Abstract)*

- Rampelt H, Zerbes RM, van der Laan M, Pfanner N. **Role of the mitochondrial contact site and cristae organizing system in membrane architecture and dynamics.** Biochim Biophys Acta. 2016 Sep 7. pii: S0167-4889(16)30221-X. doi: 10.1016/j.bbamcr.2016.05.020. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jang S, Lewis TS, Powers C, Khuchua Z, Baines CP, Wipf P, Javadov S. **Elucidating mitochondrial ETC supercomplexes in the heart during ischemia-reperfusion.** Antioxid Redox Signal. 2016 Sep 7. [Epub ahead of print] *(PubMed Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- Schlame M, Greenberg ML. **Biosynthesis, remodeling and turnover of mitochondrial cardiolipin.** Review Article. Biochim Biophys Acta. 2016 Aug 21. pii: S1388-1981(16)30230-X. doi: 10.1016/j.bbalip.2016.08.010. [Epub ahead of print] Review. *(PubMed Abstract)*

- Maguire JJ, Tyurina YY, Mohammadyani D, Kapralov AA, Anthonymuthu TS, Qu F, Amoscato AA, Sparvero LJ, Tyurin VA, Planas-Iglesias J, He RR, Klein-Seetharaman J, Bayır H, Kagan VE. **Known unknowns of cardiolipin signaling: The best is yet to come.** Biochim Biophys Acta. 2016 Aug 4. pii: S1388-1981(16)30221-9. doi: 10.1016/j.bbalip.2016.08.001. [Epub ahead of print] *(PubMed Abstract)**

- El-Hattab AW, Scaglia F. **Mitochondrial cardiomyopathies.** Front Cardiovasc Med. 2016 Jul 25;3:25. doi: 10.3389/fcvm.2016.00025. eCollection 2016. *(PubMed - Open Access)*

- Aryal B, Rao VA. **Deficiency in cardiolipin reduces doxorubicin-induced oxidative stress and mitochondrial damage in human B-lymphocytes.** PLoS One. 2016 Jul 19;11(7):e0158376. doi: 10.1371/journal.pone.0158376. eCollection 2016. *(PubMed - Open Access)*▼

- Yoo TY, Kim MR, Son JS, Lee R, Bae SH, Chung S, Kim KS, Seong MW, Park SS. **Identification of a novel de novo mutation of the *TAZ* gene in a Korean patient with Barth syndrome.** J Cardiovasc Ultrasound. 2016 Jun;24(2):153-7. doi: 10.4250/jcu.2016.24.2.153. Epub 2016 Jun 22. *(PubMed - Open Access)*

- Xu Y, Phoon CK, Berno B, D'Souza K, Hoedt E, Zhang G, Neubert TA, Epand RM, Ren M, Schlame M. **Loss of protein association causes cardiolipin degradation in Barth syndrome.** Nat Chem Biol. 2016 Jun 27. doi:10.1038/nchembio.2113. [Epub ahead of print] *(PubMed Abstract)**▼

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)**▼

- Abe M, Hasegawa Y, Oku M, Sawada Y, Tanaka E, Sakai Y, Miyoshi H. **Mechanism for Remodeling of the acyl chain composition of cardiolipin catalyzed by saccharomyces cerevisiae *tafazzin*.** J Biol Chem June 6, 2016. doi: 10.1074/jbc.M116.718510. *(PubMed - Open Access)*

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Corcelli A, Schlame M**. Cardiolipin as key lipid of mitochondria in health and disease. 2nd Edition, Florence, Italy, September 30-October 1, 2015.** Chem Phys Lipids. 2016 Apr 27. pii: S0009-3084(16)30048-2. *(PubMed Abstract)**

- Szczepanek K, Allegood J, Aluri H, Hu Y, Chen Q, Lesnefsky EJ. **Acquired deficiency of *tafazzin* in the adult heart: Impact on mitochondrial function and response to cardiac injury.** Biochim Biophys Acta. 2016 Apr;1861(4):294-300. doi: 10.1016/j.bbalip.2015.12.004. Epub 2015 Dec 12. *(PubMed Abstract)*

- Kokoszka JE, Waymire KG, Flierl A, Sweeney KM, Angelin A, MacGregor GR, Wallace DC. **Deficiency in the mouse mitochondrial adenine nucleotide translocator isoform 2 gene is associated with cardiac noncompaction.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Available online 24 April 2016. *(ScienceDirect Abstract)*

- Bradley RM, Stark KD, Duncan RE. **Influence of tissue, diet, and enzymatic remodeling on cardiolipin fatty acyl profile.** Mol Nutr Food Res. 2016 Apr 8. doi: 10.1002/mnfr.201500966. [Epub ahead of print] *(PubMed Abstract)*

- Chanana AM, Rhee JW, Wu JC. **Human-induced pluripotent stem cell approaches to model inborn and acquired metabolic heart diseases.** Curr Opin Cardiol. 2016 Mar 26. [Epub ahead of print] *(PubMed Abstract)*

- Sandlers Y, Mercier K, Pathmasiri W, Carlson J, McRitchie S, Sumner S, Vernon HJ. **Metabolomics reveals new mechanisms for pathogenesis in Barth syndrome and introduces novel roles for cardiolipin in cellular function.** PLoS One. 2016 Mar 25;11(3):e0151802. doi: 10.1371/journal.pone.0151802. eCollection 2016. *(PubMed – Open Access)*▼

- Sathappa M, Alder NN. **The ionization properties of cardiolipin and its variants in model bilayers.** Biochim Biophys Acta. 2016 Mar 7. pii: S0005-2736(16)30084-0. doi: 10.1016/j.bbamem.2016.03.007. [Epub ahead of print] *(PubMed Abstract)**

- Dolinsky VW, Cole LK, Sparagna GC, Hatch GM. **Cardiac mitochondrial energy metabolism in heart failure: Role of cardiolipin and sirtuins.** Biochimica et Biophysica Acta (BBA) - Molecular and Cell Biology of Lipids, Available online 10 March 2016. *(ScienceDirect Abstract)*

- Joshi AS, Fei N, Greenberg ML. **Get1p and Get2p are required for maintenance of mitochondrial morphology and normal cardiolipin levels.** FEMS Yeast Res. 2016 Feb 28. pii: fow019. [Epub ahead of print] *(PubMed Abstract)**

- Dinca A, Chien WM, Chin MT. **Intracellular delivery of proteins with cell-penetrating peptides for therapeutic uses in human disease.** Int J Mol Sci. 2016 Feb 22;17(2):263. doi: 10.3390/ijms17020263. Review. *(PubMed – Open Access)**

- Lu YW, Galbraith L, Herndon JD, Lu YL, Pras-Raves M, Vervaart M, van Kampen A, Luyf A, Koehler CM, McCaffery JM, Gottlieb E, Vaz FM, Claypool SM. **Defining functional classes of Barth syndrome mutation in humans.** Hum Mol Genet. 2016 Feb 16. pii: ddw046. [Epub ahead of print] *(PubMed Abstract)**

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene** transcription. Clin Genet. 2016 Feb 8. doi:10.1111/cge.12756. [Epub ahead of print] *(PubMed Abstract)*

- Thompson WR, DeCroes B, McClellan R, Rubens J, Vaz FM, Kristaponis K, Avramopoulos D, Vernon HJ. **New targets for monitoring and therapy in Barth syndrome.** Genet Med. 2016 Feb 4. doi: 10.1038/gim.2015.204. [Epub ahead of print] *(PubMed Abstract)*▼

- Saric A, Andreau K, Armand AS, Møller IM, Petit PX. **Barth syndrome: From mitochondrial dysfunctions associated with aberrant production of reactive oxygen species to pluripotent stem cell studies.** Front Genet. 2016 Jan 20;6:359. doi: 10.3389/fgene.2015.00359. eCollection 2015. Review. *(Pub Med – Open Access)*

- Thiels C, Fleger M, Huemer M, Rodenburg RJ, Vaz FM, Houtkooper RH, Haack TB, Prokisch H, Feichtinger RG, Lücke T, Mayr JA, Wortmann SB. **Atypical clinical presentations of *TAZ* mutations: An underdiagnosed cause of growth retardation?** JIMD Reports 2016. *(PubMed Abstract)*

- Lal AK, Pruitt E, Hong BJ, Lin KY, Feingold B. **Left ventricular non-compaction cardiomyopathy in children listed for heart transplant: Analysis from the Pediatric Heart Transplant Study Group.** J Heart Lung Transplant. 2016 Apr;35(4):540-2. doi:10.1016/j.healun.2015.12.010. Epub 2016 Jan 6. *(PubMed - No abstract available)*

- Dudek J, Cheng IF, Chowdhury A, Wozny K, Balleininger M, Reinhold R, Grunau S, Callegari S, Toischer K, Wanders RJ, Hasenfuß G, Brügger B, Guan K, Rehling P. **Cardiac-specific succinate dehydrogenase deficiency in Barth syndrome.** EMBO Mol Med. 2015 Dec 23. pii: e201505644. doi: 10.15252/emmm.201505644. [Epub ahead of print] *(PubMed – Open Access)*▼

- Denning C, Borgdorff V, Crutchley J, Firth KSA, George V, Kalra S, Kondrashov A, Hoang MD, Mosqueira D, Patel A, Prodanov L, Rajamohan D, Skarnes WC, Smith JGW, Young LE. **Cardiomyocytes from human pluripotent stem cells: From laboratory curiosity to industrial biomedical platform.** Original Research Article. Biochimica et Biophysica Acta (BBA) - Molecular Cell Research, Available online 31 October 2015. *(ScienceDirect Abstract)*

- Pokorná L, Cermáková P, Horváth A, Baile MG, Claypool SM, Griac P, Malínský J, Balážová M. **Specific degradation of phosphatidylglycerol is necessary for proper mitochondrial morphology and function.** Biochim Biophys Acta. 2015 Oct 19;1857(1):34-45. doi: 10.1016/j.bbabio.2015.10.004. [Epub ahead of print]. *(PubMed Abstract)*

- Mathur A, Ma Z, Loskill P, Jeeawoody S, Healy KE. ***In vitro* cardiac tissue models: Current status and future prospects.** Adv Drug Deliv Rev. 2015 Sep 30. pii: S0169-409X(15)00214-8. doi: 10.1016/j.addr.2015.09.011. [Epub ahead of print] Review. *(PubMed Abstract)*▼

- Gaspard GJ, McMaster CR. **Cardiolipin metabolism and its causal role in the etiology of the inherited cardiomyopathy Barth syndrome.** Chem Phys Lipids. 2015 Sep 25. pii: S0009-3084(15)30054-2. doi:10.1016/j.chemphyslip.2015.09.005. [Epub ahead of print] Review. *(PubMed Abstract)**

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Shen Z, Ye C, McCain K, Greenberg ML. **The role of cardiolipin in cardiovascular health.** Biomed Res Int. 2015;2015:891707. doi: 10.1155/2015/891707. Epub 2015 Aug 2. Review. *(PubMed – Open Access)**

- Reynolds S**. Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents *TAZ* knockdown-induced apoptosis in myeloid progenitor cells.** Biochem Biophys Res Commun. 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print] *(PubMed Abstract)**

- Chin MT, Chien WM, Dinca A. **Modified *tafazzin* proteins and methods of making and using the same.** Patent Application 20150203827. July 23, 2015. *(Patent Application)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Tocchi A, Quarles EK, Basisty N, Gitari L, Rabinovitch PS. **Mitochondrial dysfunction in cardiac aging.** Biochim Biophys Acta. 2015 Jul 17. pii: S0005-2728(15)00152-8. doi: 10.1016/j.bbabio.2015.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Nagueh SF, Zoghbi WA. **Role of imaging in the evaluation of patients at risk for sudden cardiac death: Genotype–phenotype intersection.** Review Article. JACC: Cardiovascular Imaging, Volume 8, Issue 7, July 2015, Pages 828-845. *(ScienceDirect Abstract)*

- Angelini R, Lobasso S, Gorgoglione R, Bowron A, Steward CG, Corcelli A**. Cardiolipin fingerprinting of leukocytes by MALDI-TOF/MS as screening tool for Barth syndrome.** J Lipid Res. 2015 Jul 5. pii:jlr.D059824. [Epub ahead of print] *(PubMed – Open Access)**▼

- Ye Y, Carlsson G, Karlsson-Sjöberg JM, Borregaard N, Modéer TU, Andersson ML, Pütsep KL. **The antimicrobial propeptide hCAP-18 plasma levels in neutropenia of various aetiologies: A prospective study.** Sci Rep. 2015 Jun 29;5:11685. doi: 10.1038/srep11685. *(PubMed - Open Access)*

- Morita SY, Terada T. **Enzymatic measurement of phosphatidylglycerol and cardiolipin in cultured cells and mitochondria.** Sci Rep. 2015 Jun 30;5:11737. doi: 10.1038/srep11737. *(PubMed Abstract)*

- Cadalbert LC, Ghaffar FN, Stevenson D, Bryson S, Vaz FM, Gottlieb E, Strathdee D. **Mouse *tafazzin* is required for male germ cell meiosis and spermatogenesis.** PLoS One. 2015 Jun 26;10(6):e0131066. doi:10.1371/journal.pone.0131066. eCollection 2015. *(PubMed - Open Access)**

- Ryan RO. **Metabolic annotation of 2-ethylhydracrylic acid.** Clin Chim Acta. 2015 Jun 23. pii: S0009-8981(15)00295-8. doi:10.1016/j.cca.2015.06.012. [Epub ahead of print] Review. *(PubMed Abstract)**

- Huang Y, Powers C, Madala SK, Greis KD, Haffey WD, Towbin JA, Purevjav E, Javadov S, Strauss AW, Khuchua Z. **Cardiac metabolic pathways affected in the mouse model of Barth syndrome.** PLoS One. 2015 Jun 1;10(6):e0128561. doi: 10.1371/journal.pone.0128561. eCollection 2015. *(PubMed – Open Access)*

- Hijikata A, Yura K, Ohara O, Go M. **Structural and functional analyses of Barth syndrome-causing mutations and alternative splicing in the *tafazzin* acyltransferase domain.** Meta Gene. 2015 Apr 22;4:92-106. doi: 10.1016/j.mgene.2015.04.001. eCollection 2015 Jun. *(PubMed – Open Access)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Kamdar F, Klaassen Kamdar A, Koyano-Nakagawa N, Garry MG, Garry DJ. **Cardiomyopathy in a dish: Using human inducible pluripotent stem cells to model inherited cardiomyopathies.** Journal of Cardiac Failure (2015), doi: 10.1016/j.cardfail.2015.04.010. (*Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Towbin JA, Lorts A, Jefferies JL**. Left ventricular non-compaction cardiomyopathy.** Review Article. The Lancet, Available online 9 April 2015. *(ScienceDirect Abstract)*

- Lakdawala NK. **Big data for a rare disease: Examining heart transplantation for left ventricular noncompaction in the UNOS registry.** J Heart Lung Transplant, Available online 4 April 2015. *(ScienceDirect Abstract)*

- Rodier G, Kirsh O, Baraibar M, Houles T, Lacroix M, Delpech H, Hatchi E, Arnould S, Severac D, Dubois E, Caramel J, Julien E, Friguet B, Le Cam L, Sardet C. **The transcription factor E4F1 coordinates CHK1-dependent checkpoint and mitochondrial functions.** Cell Reports, Available online 2 April 2015. *(ScienceDirect Abstract)*

- Reynolds S, Kreider CM, Meeley L, Bendixen RM. **Taste perception and sensory sensitivity: Relationship to feeding problems in boys with Barth syndrome.** J Rare Disorders. March 2015.▼ *(Open Access)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Taylor D, Brady JE, Li G, Sonty N, Saroyan JM. **Characterization of pain in patients with Barth syndrome.** Children's Health Care, DOI: 10.1080/02739615.2014.996882 *(Abstract)*▼

- Wegener M, Bader A, Giri S**. How to mend a broken heart: adult and induced pluripotent stem cell therapy for heart repair and regeneration.** Review Article. Drug Discovery Today, Available online 23 February 2015. *(ScienceDirect Abstract)*

- Borkar M, Bijarnia-Mahay S, Kohli S, Juneja M,Srivastava Y, Saxena R, Verma IC. **Identification of a novel *TAZ* gene mutation in a family with X-linked dilated cardiomyopathy Barth syndrome.** Journal of Inborn Errors of Metabolism & Screening January - December 2015 3: 2326409814567131, first published on February 19, 2015 doi:10.1177/2326409814567131. *(Sage – Full Text)*

- Lu YW, Claypool SM. **Disorders of phospholipid metabolism: An emerging class of mitochondrial disease due to defects in nuclear genes.** Front Genet. 2015 Feb 3;6:3. doi:10.3389/fgene.2015.00003. eCollection 2015. Review. *(PubMed – Open Access)*

- Gaspard GJ, McMaster CR. **The mitochondrial quality control protein Yme1 is necessary to prevent defective mitophagy in a yeast model of Barth syndrome.** J Biol Chem. 2015 Feb 16. pii:jbc.M115.641878. [Epub ahead of print]* *(PubMed – Open Access)*

- Russell M, Roberts AE, Abrams DJ, Murphy AM, Towbin JA, Chung WK. **How to effectively utilize genetic testing in the care of children with cardiomyopathies.** Progress in Pediatric Cardiology, Available online 31 January 2015. *(ScienceDirect Abstract)*

- Sathappa M, Schwall CT, Greenwood MR, Baile MG, Claypool SM, Alder NA. **Investigation of the role of the phospholipid cardiolipin in activating respiratory complex activity.** Biophysical Journal, Volume 108, Issue 2, Supplement 1, 27 January 2015, Page 367a. *(ScienceDirect Abstract)*

- Desmurs M, Foti M, Raemy E, Vaz FM, Martinou JC, Bairoch A, Lane L. **C11orf83, a mitochondrial cardiolipin-binding protein involved in bc1 complex assembly and supercomplex stabilization.** Mol Cell Biol. 2015 Jan 20. pii: MCB.01047-14. [Epub ahead of print] *(PubMed Abstract)*

- Xu Y, Malhotra A, Claypool CM, Ren M, Schlame M. ***Tafazzins* from Drosophila and mammalian cells assemble in large protein complexes with a short half-life.** Mitochondrion. 2015 Jan 15. pii: S1567-7249(15)00003-3. doi: 10.1016/j.mito.2015.01.002. [Epub ahead of print] (*PubMed Abstract)*

- Saunders C, Smith L, Wibrand F, Ravn K, Bross P, Thiffault I, Christensen M, Atherton A, Farrow E, Miller N, Kingsmore, Elsebet Ostergaard. **CLPB variants associated with autosomal-recessive mitochondrial disorder with cataract, neutropenia, epilepsy, and methylglutaconic aciduria.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Wortmann SB, Zietkiewicz S, Kousi M, Szklarczyk R, Haack TB, Gersting SW, Muntau AC, Rakovic A, Renkema GH, Rodenburg RJ, Strom TM, Meitinger T, Rubio-Gozalbo ME, Chrusciel E, Distelmaier F, Golzio C, Jansen JH, van Karnebeek C, Lillquist Y, Lücke T, Õunap K, Zordania R, Yaplito-Lee J, van Bokhoven H, Spelbrink JN, Vaz FM, Pras-Raves M, Ploski R, Pronicka E, Klein C, Willemsen MAAP, de Brouwer APM, Prokisch H, Katsanis N, Wevers RA. **CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Rutter J, Hughes AL. **Power2: The power of yeast genetics applied to the powerhouse of the cell.** Trends Endocrinol Metab. 2015 Jan 12. pii: S1043-2760(14)00204-5. doi: 10.1016/j.tem.2014.12.002. [Epub ahead of print] *(PubMed Abstract)*

- Kagan VE, Tyurina YY, Tyurin V, Mohammadyani D, Angeli JP, Baranov SV, Klein-Seetharaman J, Friedlander RM, Mallampalli RK, Conrad M, Bayir H. **Cardiolipin signaling mechanisms: Collapse of asymmetry and oxidation.** Antioxid Redox Signal. 2015 Jan 8. [Epub ahead of print] *(PubMed Abstract)*

- Carrilho-Ferreira P, Almeida AG, Pinto FJ. **Non-compaction cardiomyopathy: Prevalence, prognosis, pathoetiology, genetics, and risk of cardioembolism.** Curr Heart Fail Rep. 2014 Dec;11(4):393-403. doi: 10.1007/s11897-014-0227-3. *(PubMed Abstract)*

- Frohman MA. **Role of mitochondrial lipids in guiding fission and fusion**. J Mol Med (Berl). 2014 Dec 5. [Epub ahead of print] *(PubMed Abstract)*

- Falk MJ, Shen L, Gonzalez M, Leipzig J, Lott MT, Stassen APM, Diroma MA, Navarro-Gomez D, Yeske P, Bai R, Boles RG, Brilhante B, Ralph D, Dare JT, Shelton R, Terry S, Zhang Z, Copeland WC, van Oven M, Prokisch H, Wallace DC, Attimonelli M, Krotoski D, Zuchner S, Gai X. **Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities.** Mol Genet Metab. 2014 Dec 4. pii: S1096-7192(14)00377-1. doi: 10.1016/j.ymgme.2014.11.016. [Epub ahead of print] *(PubMed Abstract)*

- Tariq M, Ware SM. **Importance of genetic evaluation and testing in pediatric cardiomyopathy.** World J Cardiol 2014 Nov 26;6(11):1156-1165. Review. *(PubMed – Open Access)*

- Ye C, Shen Z, Greenberg ML. **Cardiolipin remodeling: A regulatory hub for modulating cardiolipin metabolism and function.** J Bioenerg Biomembr. 2016 Apr;48(2):113-23. doi: 10.1007/s10863-014-9591-7. Epub 2014 Nov 29. *(PubMed – Open Access)**

- Ikeda U, Minamisawa M, Koyama J. **Isolated left ventricular non-compaction cardiomyopathy in adults.** J Cardiol. 2014 Oct 31. pii: S0914-5087(14)00297-4. doi: 10.1016/j.jjcc.2014.10.005. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jiang W, Lan F, Zhang H. **Human induced pluripotent stem cell models of inherited cardiovascular diseases.** Curr Stem Cell Res Ther. 2014 Oct 16. [Epub ahead of print] *(PubMed Abstract)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Wilson D. **Methods and compositions for the prevention or treatment of Barth syndrome.** International Publication Number W0 2014/134554 A1. 2014 Sep 4. *(Patent Application)*

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Wortmann SB, Espeel M, Almeida L, Reimer A, Bosboom D, Roels F, de Brouwer AP, Wevers RA. **Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids.** J Inherit Metab Dis. 2014 Sep 2. [Epub ahead of print] *(PubMed Abstract)*

- He Q, Harris N, Ren J, Han X. **Mitochondria-targeted antioxidant prevents cardiac dysfunction induced by *tafazzin* gene knockdown in cardiac myocytes.** Oxid Med Cell Longev. 2014;2014:654198. Epub 2014 Aug 27. *(PubMed – Open Access)**

- Donadieu J, Rigaud C, Lebre AS, Touraine R, Ottolenghi C, Chabli A, Charron P, Rio M, De Lonlay P, Bonnet D. **Syndrome de Barth : le reconnaître, le traiter. Recommandations pour la prise en charge (Barth syndrome: Guidelines for diagnosis, follow-up and medical therapy).** Revue d'Oncologie Hématologie Pédiatrique, Available online 27 August 2014. *(ScienceDirect Abstract)*

- Gilbert-Barness E. **Conduction defects/cardiomyopathies.** Adv Pediatr. 2014 Aug;61(1):127-48. doi: 10.1016/j.yapd.2014.03.001. No abstract available. *(PubMed)*

- Chi C-S. **Diagnostic approach in infants and children with mitochondrial diseases.** Pediatr Neonatol. 2014 Aug 20. pii: S1875-9572(14)00099-0. doi: 10.1016/j.pedneo.2014.03.009. [Epub ahead of print] Review. *(PubMed Abstract)*

- Debnath S, Addya S. **Mis-sense mutations in *tafazzin (TAZ)* that escort to mild clinical symptoms of Barth syndrome is owed to the minimal inhibitory effect of the mutations on the enzyme function: In-silico evidence.** Interdiscip Sci. 2014 Aug 9. [Epub ahead of print] *(PubMed Abstract)*

- Bowron A, Honeychurch J, Williams M, Tsai-Goodman B, Clayton N, Jones L, Shortland GJ, Qureshi SA, Heales SJ, Steward CG. **Barth syndrome without tetralinoleoyl cardiolipin deficiency: a possible ameliorated phenotype.** J Inherit Metab Dis. 2014 Aug 12. [Epub ahead of print] *(PubMed – Open Access)**▼

- Mayr JA. **Lipid metabolism in mitochondrial membranes.** J Inherit Metab Dis. 2014 Aug 1. [Epub ahead of print] *(PubMed Abstract)*

- Peyta L, Dumas JF, Pinault M, Guimaraes C, Jarnouen K, Lagrave J, Maillot F, Loyer P, Servais S. **Reduction of the mitochondrial content in cardiolipins decreases O2 consumption and increases ATP synthesis efficiency in human hepatocyte-like HepaRG cells.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Volume 1837, Supplement, July 2014, Pages e28–e29. *(ScienceDirect Abstract)*

- Zweigerdt R, Gruh I, Martin U. **Your heart on a chip: iPSC-based modeling of Barth-syndrome-associated cardiomyopathy.** Cell Stem Cell. 2014 Jul 3;15(1):9-11. doi: 10.1016/j.stem.2014.06.015. *(PubMed – Open Access)*

- Hwang MS, Schwall CT, Pazarentzos E, Datler C, Alder NN, Grimm S. **Mitochondrial Ca^{2+} influx targets cardiolipin to disintegrate respiratory chain complex II for cell death induction.** Cell Death Differ. 2014 Jun 20. doi: 10.1038/cdd.2014.84. [Epub ahead of print] *(PubMed Abstract)*

- Feillet-Coudray C, Fouret G, Casas F, Coudray C. **Impact of high dietary lipid intake and related metabolic disorders on the abundance and acyl composition of the unique mitochondrial phospholipid, cardiolipin.** J Bioenerg Biomembr. 2014 Jun 21. [Epub ahead of print] *(PubMed Abstract)*

- Ren M, Phoon CKL, Schlame M. **Metabolism and function of mitochondrial cardiolipin.** Prog Lipid Res. 2014 Jul;55:1-16. doi: 10.1016/j.plipres.2014.04.001. Epub 2014 Apr 24. *(PubMed Abstract)**

- Chatfield KC, Sparagna GC, Sucharov CC, Miyamoto SD, Grudis JE, Sobus RD, Hijmans J, Stauffer BL. **Dysregulation of cardiolipin biosynthesis in pediatric heart failure**. J Mol Cell Cardiol. 2014 Jun 14. pii: S0022-2828(14)00196-5. doi:10.1016/j.yjmcc.2014.06.002. [Epub ahead of print] *(PubMed Abstract)*

- Epand RM, D'Souza K, Berno B, Schlame M. **Membrane curvature modulation of protein activity determined by NMR.** Review Article. Biochimica et Biophysica Acta (BBA) - Biomembranes, Available online 13 May 2014. *(Abstract)*

- Raval KK, Kamp TJ. **Cardiomyopathy, mitochondria and Barth syndrome: iPSCs reveal a connection.** Nat Med. 2014 Jun 5;20(6):585-6. doi: 10.1038/nm.3592. *(PubMed – No abstract available).*

- Folsi V, Miglietti N, Lombardi A, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, Plebani A. **Cardiomyopathy in a male patient with neutropenia and growth delay.** Ital J Pediatr. 2014 May 12;40:45. doi:10.1186/1824-7288-40-45. *(PubMed – Open Access)*

- Richter-Dennerlein R, Korwitz A, Haag M, Tatsuta T, Dargazanli S, Baker M, Decker T, Lamkemeyer T, Rugarli EI, Langer T. **DNAJC19, a mitochondrial cochaperone associated with cardiomyopathy, forms a complex with prohibitins to regulate cardiolipin remodeling.** Cell Metab. 2014 May 21. pii: S1550-4131(14)00182-X. doi: 10.1016/j.cmet.2014.04.016. [Epub ahead of print] *(PubMed Abstract)*

- Wang G, McCain ML, Yang L, He A, Pasqualini FS, Agarwal A, Yuan H, Jiang D, Zhang D, Zangi L, Geva J, Roberts AE, Ma Q, Ding J, Chen J, Wang DZ, Li K, Wang J, Wanders RJA, Kulik W, Vaz FM, Laflamme MA, Murry CE, Chien KR, Kelley RI, Church GM, Parker K, Pu WT. **Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies.** Nat Med. 2014 Jun;20(6):616-23. doi:10.1038/nm.3545 Epub. 2014 May 11. *(PubMed – Open Access)**▼

- Mejia EM, Cole L, Hatch GM. **Cardiolipin metabolism and the role it plays in heart failure and mitochondrial supercomplex formation.** Cardiovasc Hematol Disord Drug Targets. 2014 May 5. [Epub ahead of print] *(PubMed Abstract)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Harner ME, Unger AK, Izawa T, Walther DM, Ozbalci C, Geimer S, Reggiori F, Brügger B, Mann M, Westermann B, Neupert W. **Aim24 and MICOS modulate respiratory function, *tafazzin*-related cardiolipin modification and mitochondrial architecture.** Elife. 2014 Jan 1;3:e01684. doi:10.7554/eLife.01684. *(PubMed Abstract)*

- Raja V, Greenberg ML. **The functions of cardiolipin in cellular metabolism–potential modifiers of the Barth syndrome phenotype.** Chem Phys Lipids. 2014 Jan 17. pii:S0009-3084(13)00181-3. doi: 10.1016/j.chemphyslip.2013.12.009. [Epub ahead of print] *(PubMed – Open Access)**

- Su B, Ryan RO. **Metabolic biology of 3-methylglutaconic acid-uria: A new perspective.** J Inherit Metab Dis. 2014 May;37(3):359-68. doi: 10.1007/s10545-013-9669-0. Epub 2014 Jan 10. *(PubMed Abstract)**

- Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J. **The MOGE(S) classification for a phenotype–genotype nomenclature of cardiomyopathy: Endorsed by the World Heart Federation.** Journal of the American College of Cardiology, Volume 62, Issue 22, 3 December 2013, Pgs. 2046-2072 - CORRECTION. *(ScienceDirect Abstract)*

- Digilio MC, Bernardini L, Gagliardi MG, Versacci P, Baban A, Capolino R, Dentici ML, Roberti MC, Angioni A, Novelli A, Marino b, Dallapiccola B. **Syndromic non-compaction of the left ventricle: Associated chromosomal anomalies.** Clin Genet. 2013 Oct;84(4):362-7. doi: 10.1111/cge.12069. Epub 2012 Dec 28. *(PubMed Abstract)*

- Ye C, Lou W, Li Y, Chatzispyrou IA, Huttemann M, Lee I, Houtkooper RH, Vaz FM, Chen S, Greenberg ML. **Deletion of the cardiolipin-specific phospholipase Cld1 rescues growth and lifespan defects in the *tafazzin* mutant: Implications for Barth syndrome.** J Biol Chem. 2013 Dec 8. [Epub ahead of print] *(PubMed – Open Access)**

- Baile MG, Sathappa M, Lu YW, Pryce E, Whited K, McCaffery JM, Han X, Alder NN, Claypool SM. **Unremodeled and remodeled cardiolipin are functionally indistinguishable in yeast.** J Biol Chem. 2013 Nov 27. [Epub ahead of print] *(PubMed – Open Access)*

- Kirwin SM, Manolakos A, Swain Barnett S, Gonzalez IL. ***Tafazzin* splice variants and mutations in Barth syndrome.** Mol Genet Metab. 2014 Jan;111(1):26-32. doi: 10.1016/j.ymgme.2013.11.006. Epub 2013 Nov 19. *(PubMed Abstract)**

- Mileykovskaya E, Dowhan W. **Cardiolipin-dependent formation of mitochondrial respiratory supercomplexes.** Chem Phys Lipids. 2013 Nov 9. [Epub ahead of print] *(PubMed Abstract)*

- Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J. **The MOGE(S) classification for a phenotype–genotype nomenclature of cardiomyopathy: Endorsed by the World Heart Federation.** Journal of the American College of Cardiology, Available online 18 November 2013. *(ScienceDirect Abstract)*

- He Q, Han X. **Cardiolipin remodeling in diabetic heart.** Chem Phys Lipids. 2013 Nov 1. doi:pii: S0009-3084(13)00135-7. [Epub ahead of print] *(PubMed Abstract)*

- Baile MG, Lu WY, Claypool SM. **The topology and regulation of cardiolipin biosynthesis and remodeling in yeast.** Chem Phys Lipids. 2014 Apr;179:25-31. doi: 10.1016/j.chemphyslip.2013.10.008. Epub 2013 Nov 1. *(PubMed Abstract)*

- Xu Y, Schlame M. **The turnover of glycerol and acyl moieties of cardiolipin.** Chem Phys Lipids. 2013 Nov 1. [Epub ahead of print]. *(PubMed Abstract)**

- Finsterer J, Stöllberger C. **Unclassified cardiomyopathies in neuromuscular disorders.** Wien Med Wochenschr. 2013 Oct 24. [Epub ahead of print] *(PubMed Abstract)*

- Mejia EM, Nguyen H, Hatch GM. **Mammalian cardiolipin biosynthesis.** Chem Phys Lipids. 2013 Oct 18 [Epub ahead of print] (*PubMed Abstract)**

- Mazurová S, Tesarová M, Magner M, Houštková H, Hansíková H, Augustínová J, Tomek V, Vondrácková A, Zeman J, Honzík T. **Novel mutations in the *TAZ* gene in patients with Barth syndrome.** Prague Med Rep. 2013;114(3):139-153. (*PubMed Abstract)*

- Herndon JD, Claypool SM, Koehler CM. **The *Taz*1p transacylase is imported and sorted into the outer mitochondrial membrane via a membrane anchor domain.** Eukaryot Cell. 2013 Sep 27. [Epub ahead of print] (*PubMed – Open Access)*

- Finsterer J, Stöllberger C. **Ultrastructural findings in noncompaction prevail with neuromuscular disorders.** Cardiology. 2013 Sep 21;126(4):219-223. [Epub ahead of print] (*PubMed Abstract)*

- Corcelli A, Schlame M. **Cardiolipin as key lipid of mitochondria in health and disease.** Meeting Report – September 17, 2013, Bari, Italy. Eur. J. Lipid Sci. Technol. 2013, 115, 0000–0000.*

- Agarwal A, Khandheria BK, Paterick TE, Treiber SC, Bush M, Tajik AJ. **Left ventricular noncompaction in patients with bicuspid aortic valve.** J Am Soc Echocardiogr. 2013 Sep 14. doi:pii: S0894-7317(13)00629-9. 10.1016/j.echo.2013.08.003. [Epub ahead of print] (*PubMed Abstract)*

- He Q, Wang M, Harris N, Han X. ***Tafazzin* knockdown interrupts cell cycle progression in cultured neonatal ventricular fibroblasts.** Am J Physiol Heart Circ Physiol. 2013 Aug 30. [Epub ahead of print] (*PubMed – Open Access)**▼

- Jefferies JL. **Barth syndrome.** Am J Med Genet C Semin Med Genet. 2013 Jul 10. doi: 10.1002/ajmg.c.31372. [Epub ahead of print] *(PubMed – Open Access)*▼

- Monteiro JP, Oliveira PJ, Jurado AS. **Mitochondrial membrane lipid remodeling in pathophysiology: A new target for diet and therapeutic interventions.** Prog Lipid Res. 2013 Jul 1. doi:pii: S0163-7827(13)00042-8. 10.1016/j.plipres.2013.06.002. [Epub ahead of print] (*PubMed Abstract)*

- Dudek J, Cheng IF, Balleininger M, Vaz FM, Streckfuss-Bömeke K, Hübscher D, Vukotic M, Wanders RJ, Rehling P, Guan K. **Cardiolipin deficiency affects respiratory chain function and organization in an induced pluripotent stem cell model of Barth syndrome.** Stem Cell Res. 2013 Sep;11(2):806-19. doi: 10.1016/j.scr.2013.05.005. Epub 2013 May 28. *(PubMed – Open Access)*▼

- Patil VA, Greenberg ML. **Cardiolipin-mediated cellular signaling.** Adv Exp Med Biol. 2013;991:195-213. doi: 10.1007/978-94-007-6331-9_11. (*PubMed Abstract)**

- Kim GB, Kwon BS, Bae EJ, Noh CI, Seong MW, Park SS. **A novel mutation of the *TAZ* gene in Barth syndrome: Acute exacerbation after contrast-dye injection.** J Korean Med Sci. 2013 May;28(5):784-7. doi: 10.3346/jkms.2013.28.5.784. Epub 2013 May 2. (*PubMed – Open Access)*

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Rigaud C, Lebre A, Touraine R, Beaupain B, Ottolenghi C, Chabli A, Ansquer H, Ozsahin H, Di Filippo S, De Lonlay P, Borm B, Rivier F, Vaillant M, Mathieu-Dramard M, Goldenberg A, Viot G, Charron P, Rio M, Bonnet D, Donadieu J. **Natural history of Barth syndrome: A national cohort study of 22 patients.** Orphanet J Rare Dis. 2013 May 8;8:70. doi: 10.1186/1750-1172-8-70. *(PubMed – Open Access)**▼

- Baile MG, Whited K, Claypool SM. **Deacylation on the matrix side of the mitochondrial inner membrane regulates cardiolipin remodeling.** Mol Biol Cell. 2013 May 1. [Epub ahead of print] *(PubMed – Open Access)*

- Finsterer J, Stöllberger C, Blazek G, Sehnal E. **Familal left ventricular hypertrabeculation (noncompaction) is myopathic.** Int J Cardiol. 2013 Apr 15;164(3):312-7. doi:10.1016/j.ijcard.2011.07.012. Epub 2011 Jul 23. *(PubMed Abstract)*

- Strathdee D, Cadalbert L, Ghaffar FN, et al. **Generation and analysis of a mouse model for Barth syndrome.** Transgenic Research Volume: 22 Issue: 1 Pages: 237-237 Meeting Abstract: 85. *(Meeting Abstract)*

- Powers C, Huang Y, Strauss A, Khuchua Z. **Diminished exercise capacity and mitochondrial bc1 complex deficiency in *tafazzin*-knockdown mice.** Front Physiol. 2013 Apr 17;4:74. doi: 10.3389/fphys.2013.00074. Print 2013. *(PubMed – Open Access)*

- Fan Y, Steller J, Gonzalez IL, Kulik W, Fox M, Chang R, Westerfield BA, Batra AS, Wang RY, Gallant NM, Pena LS, Wang H, Huang T, Bhuta S, Penny DJ, McCabe ER, Kimonis VE. **A novel exonic splicing mutation in the *TAZ* (G4.5) gene in a case with atypical Barth syndrome.** JIMD Rep. 2013 Apr 19. [Epub ahead of print] *(PubMed – Open Access)*

- Teekakirikul P, Kelly MA, Rehm HL, Lakdawala NK, Funke BH. **Inherited cardiomyopathies: Molecular genetics and clinical genetic testing in the postgenomic era.** J Mol Diagn. 2013 Mar;15(2):158-70. doi: 10.1016/j.jmoldx.2012.09.002. Epub 2012 Dec 27. *(PubMed Abstract)*

- Dedieu N, Giardini A, Steward CG, Fenton M, Karimova A, Hsia TY, Burch M. **Successful mechanical circulatory support for 251 days in a child with intermittent severe neutropenia due to Barth syndrome.** Pediatr Transplant. 2013 Mar;17(2):E46-9. doi: 10.1111/petr.12027. Epub 2012 Nov 28. *(PubMed Abstract)*▼

- Gonzalvez F, D'Aurelio M, Boutant M, Moustapha A, Puech JP, Landes T, Arnauré L, Vial G, Talleux N, Slomianny C, Wanders RJA, Houtkooper RH, Belenger P, Moller IM, Gottlieb E, Vaz FM, Manfredi G, Petit PX. **Barth syndrome: Cellular compensation of mitochondrial dysfunction and apoptosis inhibition due to changes in cardiolipin remodeling linked to *Tafazzin* gene mutation.** Biochim Biophys Acta. 2013 Aug;1832(8):1194-206. doi: 10.1016/j.bbadis.2013.03.005. Epub 2013 Mar 20. *(PubMed – Open Access)*

- Finsterer J, Stöllberger C, Wahbi K. **Cardiomyopathy in neurological disorders.** Cardiovascular Pathology (2013), doi.org/10.1016/j.carpath.2012.12.008 *(ScienceDirect Abstract)*

- Finsterer J, Stöllberger C, Kovacs GG, Sehnal E. **Left ventricular hypertrabeculation/ noncompaction coincidentally found in sporadic inclusion body myositis.** Int J Cardiol 2013 Sep 20;168(1):610-2. doi.org/10/1016/j.ijcard2013.01.221. Epub 2013 Feb 22. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini RR, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 14;8(1):27. doi: 10.1186/1750-1172-8-27. *(PubMed Abstract)*▼

- Kiebish MA, Yang K, Liu X, Mancuso DJ, Guan S, Zhao Z, Sims HF, Cerqua R, Cade WT, Han X, Gross RW. **Dysfunctional cardiac mitochondrial bioenergetic, lipidomic, and signaling in a murine model of Barth syndrome.** J Lipid Res. 2013 Feb 14. [Epub ahead of print] *(PubMed – Open Access)**

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Day TG, Fenton M. **Dilated cardiomyopathy in children.** Paediatrics and Child Health Volume 23, Issue 2, February 2013, Pages 59–63. *(SciVerse Abstract)*

- Stojic O, O'Leary MF, Singh K, Menzies KJ, Vainshtein A, Hood DA. **The effects of chronic muscle use and disuse on cardiolipin metabolism.** J Appl Physiol. 2013 Feb 15;114(4):444-52. doi: 10.1152/japplphysiol.01312.2012. Epub 2012 Dec 6. *(PubMed Abstract)*

- Whited K, Baile MG, Currier P, Claypool SM. **Seven functional classes of Barth syndrome mutation.** Hum Mol Genet. 2013 Feb 1;22(3):483-92. doi: 10.1093/hmg/dds447. Epub 2012 Oct 24. *(PubMed Abstract)*

- Ware SM, Towbin JA. **Nuclear genes causing mitochondrial cardiomyopathy.** Mitochondrial Disorders Caused by Nuclear Genes, 2013, pp. 319-335. *(Springer Abstract)*

- Karkucinska-Wieckowska A, Trubicka J, Werner B, Kokoszynska K, Pajdowska M, Pronicki M, Czarnowska E, Lebiedzinska M, Sykut-Cegielska J, Ziolkowska L, Jaron W, Dobrzanska A, Ciara E, Wieckowski MR, Pronicka E. **Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome.** J Inherit Metab Dis. 2013 Jan 30. [Epub ahead of print] *(PubMed Abstract)*

- Man E, Lafferty KA, Funke BH, Lun KS, Chan SY, Chau AK, Chung BH. **NGS identifies *TAZ* mutation in a family with X-linked dilated cardiomyopathy.** BMJ Case Rep. 2013 Jan 22;2013. doi:pii: bcr2012007529. 10.1136/bcr-2012-007529. *(PubMed Abstract)*

- Wortmann SB, Duran M, Anikster Y, Barth PG, Sperl W, Zschocke J, Morava E, Wevers RA. **Inborn errors of metabolism with 3-methylglutaconic aciduria as discriminative feature: Proper classification and nomenclature**. J Inherit Metab Dis. 2013 Jan 8. [Epub ahead of print] *(PubMed Abstract)*

- Lopes LR, Elliott PM. **Genetics of heart failure.** Biochim Biophys Acta. 2013 Jan 5. doi:pii: S0925-4439(13)00002-1. 10.1016/j.bbadis.2012.12.012. [Epub ahead of print] *(PubMed Abstract)*

- Finsterer JA, Frank M. **Haematological features in Barth syndrome.** Curr Opin Hematol. 2013 Jan;20(1):36-40. doi: 10.1097/MOH.0b013e32835a01d9. *(PubMed Abstract)*

- Sokolic R. **Neutropenia in primary immunodeficiency.** Curr Opin Hematol. 2013 Jan;20(1):55-65. doi:10.1097/MOH.0b013e32835aef1c. *(PubMed Abstract)*

- Patil V, Fox JL, Gohil VM, Winge DR, Greenberg ML. **Loss of cardiolipin leads to perturbation of mitochondrial and cellular iron homeostasis.** J Biol Chem. 2013 Jan 18;288(3):1696-705. doi: 10.1074/jbc.M112.428938. Epub 2012 Nov 28. *(PubMed – Open Access)**

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth syndrome in adulthood: A clinical case.** Rev Esp Cardiol (Engl Ed). 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. *(PubMed Abstact)*

- Goldstein A, Bhatia P, Vento JM. **Update on nuclear mitochondrial genes and neurologic disorders.** Semin Pediatr Neurol. 2012 Dec;19(4):181-93. doi: 10.1016/j.spen.2012.09.005. *(PubMed Abstract)*

- Boxer LA. **How to approach neutropenia.** Hematology Am Soc Hematol Educ Program. 2012;2012:174-82. doi: 10.1182/asheducation-2012.1.174. *(PubMed Abstract)*

- Kaplan JC, Hamroun D. **The 2013 version of the gene table of neuromuscular disorders (nuclear genome).** Neuromuscular Disorders, Volume 22, Issue 12, December 2012, Pages 1108-1135. *(Abstract)*

- Schlame M. **Cardiolipin remodeling and the function of *tafazzin*.** Biochim Biophys Acta. 2012 Nov 28. doi:pii: S1388-1981(12)00249-1. *(PubMed Abstract)*

- Taylor WA, Mejia EM, Mitchell RW, Choy PC, Sparagna GC, et al. **Human trifunctional protein *alpha* links cardiolipin remodeling to beta-oxidation.** PLoS. 2012;7(11):e48628. doi: 10.1371/journal.pone.0048628. Epub 2012 Nov 9. *(PubMed Abstract - Open Access)* *▼

- Roberts AE, Nixon C, Steward CG, Gauvreau K, Maisenbacher M, Fletcher M, Geva J, Byrne BJ, Spencer CT. **The Barth Syndrome Registry: Distinguishing disease characteristics and growth data from a longitudinal study.** Am J Med Genet A. 2012 Nov;158A(11):2726-32. doi: 10.1002/ajmg.a.35609. Epub 2012 Oct 8. *(PubMed – Open Access)*▼

- Bowron A, Frost R, Powers VE, Thomas PH, Heales SJ, Steward CG. **Diagnosis of Barth syndrome using a novel LC-MS/MS method for leukocyte cardiolipin analysis.** J Inherit Metab Dis. 2012 Oct 30. [Epub ahead of print] *(PubMed Abstract)*

- Gawrisch K. **Lipids: *Tafazzin* senses curvature.** Nat Chem Biol. 2012 Oct;8(10):811-2. *(PubMed – No Abstract Available)*

- Honzik T, Tesarova M, Magner M, Mayr J, Jesina P, Vesela K, Wenchich L, Szentivanyi K, Hansikova H, Sperl W, Zeman J. **Neonatal onset of mitochondrial disorders in 129 patients: clinical and laboratory characteristics and a new approach to diagnosis.** J Inherit Metab Dis. 2012 Sep;35(5):749-59. doi: 10.1007/s10545-011-9440-3. Epub 2012 Jan 10. *(PubMed Abstract)*

- Tan BK, Bogdanov M, Zhao J, Dowhan W, Raetz CR, Guan Z. **Discovery of a cardiolipin synthase utilizing phosphatidylethanolamine and phosphatidylglycerol as substrates.** Proc Natl Acad Sci U S A. 2012 Sep 17. [Epub ahead of print] *(PubMed Abstract)*

- Ronvelia D, Greenwood J, Platt J, Hakim S, Zaragoza MV. **Intrafamilial variability for novel *TAZ* gene mutation: Barth syndrome with dilated cardiomyopathy and heart failure in an infant and left ventricular noncompaction in his great-uncle.** Mol Genet Metab. 2012 Sep 18. pii: S1096-7192(12)00354-X. doi: 10.1016/j.ymgme.2012.09.013. [Epub ahead of print] *(PubMed Abstract)*

- Böttinger L, Horvath SE, Kleinschroth T, Hunte C, Daum G, Pfanner N, Becker T. **Phosphatidylethanolamine and cardiolipin differentially affect the stability of mitochondrial respiratory chain supercomplexes.** J Mol Biol. 2012 Sep 9. pii: S0022-2836(12)00720-6. doi: 10.1016/j.jmb.2012.09.001. [Epub ahead of print] *(PubMed Abstract)*

- Schlame M, Acehan D, Berno B, Xu Y, Valvo S, Ren M, Stokes DL, Epand RM. **The physical state of lipid substrates provides transacylation specificity for *tafazzin*.** Nat Chem Biol. 2012 Sep 2. doi: 10.1038/nchembio.1064. [Epub ahead of print] *(PubMed – Open Access)**

- Liu X, Ye B, Miller S, Yuan H, Zhang H, Tian L, Nie J, Imae R, Arai H, Li Y, Cheng Z, Shi Y. **Ablation of ALCAT1 mitigates hypertrophic cardiomyopathy through effects on oxidative stress and mitophagy.** Mol Cell Biol. 2012 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Lamari F, Mochel F, Sedel F, Saudubray JM. **Disorders of phospholipids, sphingolipids and fatty acids biosynthesis: Toward a new category of inherited metabolic diseases.** J Inherit Metab Dis. 2012 Jul 20. [Epub ahead of print] *(PubMed Abstract)*

- Kiebish MA, Yang K, Sims HF, Jenkins CM, Liu X, Mancuso DJ, Zhao Z, Guan S, Abendschein DR, Han X, Gross RW. **Myocardial regulation of lipidomic flux by cardiolipin synthase: Setting the beat for bioenergetic efficiency.** J Biol Chem. 2012 Jul 20;287(30):25086-97. Epub 2012 May 14. *(PubMed – Open Access)**

- Wilson LD, Al-Majid S, Rakovsky C, Schwindt CD. **Higher IL-6 and IL6:IGF ratio in patients with Barth syndrome**. J Inflamm (Lond). 2012 Jun 21;9(1):25. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S, Kreider CM, Bendixen R. **A mixed-methods investigation of sensory response patterns in Barth syndrome: A clinical phenotype?** Am J Med Genet Part A. 7 Jun 2012 Jul;158A(7):1647-53. *(PubMed Abstract)*▼

BSF does not endorse any drugs, tests, or treatments that we may report.

- Cade WT, Spencer CT, Reeds DN, Waggoner AD, O'Connor R, Maisenbacher M, Crowley JR, Byrne BJ, Peterson LR. **Substrate metabolism during basal and hyperinsulinemic conditions in adolescents and young-adults with Barth syndrome.** J Inherit Metab Dis. 2012 May 12. [Epub ahead of print] *(PubMed Abstract)*▼

- Kindel SJ, Miller EM, Gupta R, Cripe LH, Hinton RB, Spicer RL, Towbin JA, Ware SM. **Pediatric cardiomyopathy: Importance of genetic and metabolic evaluation.** J Card Fail. 2012 May;18(5):396-403. doi: 10.1016/j.cardfail.2012.01.017. Epub 2012 Mar 10. *(PubMed Abstract)*

- Raches D, Mazzocco MM. **Emergence and nature of mathematical difficulties in young children with Barth syndrome.** J Dev Behav Pediatr. 2012 May;33(4):328-35. *(PubMed Abstract)*▼

- Raja V, Greenberg ML. **The functions of cardiolipin in cellular metabolism-potential modifiers of the Barth syndrome phenotype.** Chem Phys Lipids. 2014 Apr;179:49-56. doi: 10.1016/j.chemphyslip.2013.12.009. Epub 2014 Jan 17. *(PubMed – Open Access)**

- Wan C, Yu HH, Lu MY, Lee JH, Wang LC, Lin YT, Yang YH, Chiang BL. **Clinical manifestations and outcomes of pediatric chronic neutropenia.** J Formos Med Assoc. 2012 Apr;111(4):220-7. Epub 2012 Mar 16. *(PubMed Abstract)*

- Phoon CKL, Acehan D, Schlame M, Stokes DL, Edelman-Novemsky I, Yu D, Xu Y, Viswanathan N, Ren M. ***Tafazzin* knockdown in mice leads to a developmental cardiomyopathy with early diastolic dysfunction preceding myocardial noncompaction**. J Am Heart Assoc. 2012 Apr;1(2). doi:pii: jah3-e000455. 10.1161/JAHA.111.000455. Epub 2012 Apr 24. *(PubMed – Open Access)**

- Schild L, Lendeckel U, Gardemann A, Wiswedel I, Schmidt CA, Wolke C, Walther R, Grabarczyk P, Busemann C. **Composition of molecular cardiolipin species correlates with proliferation of lymphocytes.** Exp Biol Med (Maywood). 2012 Apr;237(4):372-9. doi: 10.1258/ebm.2011.011311. Epub 2012 Apr 4. *(PubMed Abstract)*

- Hanke SP, Gardner AB, Lombardi JP, Manning PB, Nelson DP, Towbin JA, Jefferies JL, Lorts A. **Left ventricular noncompaction cardiomyopathy in Barth syndrome: An example of an undulating cardiac phenotype necessitating mechanical circulatory support as a bridge to transplantation.** Pediatr Cardiol. 2012 Mar 17. [Epub ahead of print] *(PubMed Abstract)*

- Joshi AS, Thompson MN, Fei N, Hutteman M, Greenberg ML. **Cardiolipin and mitochondrial phosphatidylethanolamine have overlapping functions in mitochondrial fusion in Saccharomyces cerevisiae.** J Biol Chem. 2012 Mar 20. [Epub ahead of print] *(PubMed – Open Access)**

- Mulligan CM, Sparagna GC, Le CH, De Mooy AB, Routh MA, Holmes MG, Hickson-Bick DL, Zarini S, Murphy RC, Xu FY, Hatch GM, McCune SA, Moore RL, Chicco AJ. **Dietary linoleate preserves cardiolipin and attenuates mitochondrial dysfunction in the failing rat heart.** Cardiovasc Res. 2012 Mar 12. [Epub ahead of print] *(PubMed Abstract)*

- Greenberg, ML. **Functions of cardiolipin as modifiers of the Barth syndrome phenotype.** ASBMB Today. March 2012. *(ASBMB Abstract)*

- Cosson L, Toutain A, Simard G, Kulik W, G Matyas, Guichet A, Blasco H, Maakaroun-Vermesse Z, Vaillant MC, Le Caignec C, Chantepie A, Labarthe F. **Barth syndrome in a female patient.** Mol Genet Metab. 2012 May;106(1):115-20. doi: 10.1016/j.ymgme.2012.01.015. Epub 2012 Jan 24. *(PubMed Abstract)*

- Schug ZT, Frezza C, Galbraith LC, Gottlieb E. **The music of lipids: How lipid composition orchestrates cellular behaviour.** Acta Oncol. 2012 Jan 30. [Epub ahead of print] *(PubMed Abstract*

- Werner B, Trubicka J, Pronicka E. **[Clinical and diagnostic aspects of Barth syndrome (X-linked cardiomyopathy)].** Kardiol Pol. 2011;69(11):1177-80. Polish. No abstract available. *(PubMed – Open Access)*

- Stanley WC, Khairallah RJ, Dabkowski ER. **Update on lipids and mitochondrial function: Impact of dietary n-3 polyunsaturated fatty acids.** Curr Opin Clin Nutr Metab Care. 2012 Jan 13. [Epub ahead of print] *(PubMed Abstract)*

- Kachewar SG, Sankaye SB, Kulkarni DS. **The role of radio-diagnosis in inborn errors of metabolism.** Journal of Clinical and Diagnostic Research [serial online] 2011 November [cited: 2012 Dec 5]; 5:1467-1472. *(Full Text)*

- Saini-Chohan HK, Mitchell RW, Vaz FM, Zelinski T, Hatch GM. **Delineating the role of alterations in lipid metabolism to the pathogenesis of inherited skeletal and cardiac muscle disorders.** J Lipid Res. 2011 Nov 7. [Epub ahead of print] *(PubMed – Open Access)**

- Makaryan V, Kulik W, Vaz FM, Allen C, Dror Y, Dale DC, Aprikyan AA**. The cellular and molecular mechanisms for neutropenia in Barth syndrome.** Eur J Haematol. 2011 Oct 24. doi: 10.1111/j.1600-0609.2011.01725.x. [Epub ahead of print] *(PubMed – Open Access)**

- Claypool SM, Koehler CM. **The complexity of cardiolipin in health and disease.** Trends Biochem Sci. 2011 Oct 17. [Epub ahead of print] *(PubMed Abstract)*

- El-Hafidi M, Meschini MC, Rizza T, Santorelli FM, Bertini E, Carrozzo R, Vázquez-Memije ME. **Cardiolipin content in mitochondria from cultured skin fibroblasts harboring mutations in the mitochondrial ATP6 gene.** J Bioenerg Biomembr. 2011 Oct 13. [Epub ahead of print] *(PubMed Abstract)*

- Momoi N, Chang B, Takeda I, Aoyagi Y, Endo K, Ichida F**. Differing clinical courses and outcomes in two siblings with Barth syndrome and left ventricular noncompaction.** Eur J Pediatr. 2011 Oct 7. [Epub ahead of print] *(PubMed Abstract)*

- Wahjudi PN, Yee J, Martinez SR, Zhang J, Teitell M, Nikolaenko L, Swerdloff R, Wang C, Lee WN. **Turn-over of non-essential fatty acid in cardiolipin in rat heart.** J Lipid Res. 2011 Sep 27. [Epub ahead of print] *(PubMed Abstract)*

- Takeda A, Sudo A, Yamada M, Yamazawa H, Izumi G, Nishino I, Ariga T**. Eponym: Barth syndrome.** Eur J Pediatr. 2011 Sep 23. [Epub ahead of print] *(PubMed Abstract)*

- Takeda A, Sudo A, Yamada M, Yamazawa H, Izumi G, Nishino I, Ariga T. **Barth syndrome diagnosed in the subclinical stage of heart failure based on the presence of lipid storage myopathy and isolated noncompaction of the ventricular myocardium.** Eur J Pediatr. 2011 Sep 20. [Epub ahead of print] *(PubMed Abstract)*

- Spencer CT, Byrne BJ, Bryant RM, Margossian R, Maisenbacher M, Breitenger P, Benni PB, Redfearn S, Marcus E, Cade WT. **Impaired cardiac reserve and severely diminished skeletal muscle oxygen utilization mediate exercise intolerance in Barth syndrome.** Am J Physiol Heart Circ Physiol. 2011 Nov;301(5):H2122-9. Epub 2011 Aug 26. *(PubMed – Open Access)**▼

- Schiff M, Ogier de Baulny H, Lombès A. **Neonatal cardiomyopathies and metabolic crises due to oxidative phosphorylation defects.** Semin Fetal Neonatal Med. 2011 Aug;16(4):216-21. doi: 10.1016/j.siny.2011.04.002. Epub 2011 May 24. *(PubMed Abstract)*

- Wortmann SB, Morava E. **3-methylglutaconic aciduria type IV: A syndrome with an evolving phenotype.** Clin Dysmorphol. 2011 Jul;20(3):168-9. No abstract available. *(PubMed Abstract)*

- Saini-Chohan HK, Dakshinamurti S, Taylor WA, Shen GX, Murphy R, Sparagna GC, Hatch GM. **Persistent pulmonary hypertension results in reduced tetralinoleoyl-cardiolipin and Complex II + III activity in neonatal pig heart.** Am J Physiol Heart Circ Physiol. 2011 Aug 12. [Epub ahead of print] *(PubMed – Open Access)**

- Jacoby D, McKenna WJ. **Genetics of inherited cardiomyopathy.** Eur Heart J. 2011 Aug 2. [Epub ahead of print] *(PubMed Abstract)*

- Swalwell H, Kirby DM, Blakely EL, Mitchell A, Salemi R, Sugiana C, Compton AG, Tucker EJ, Ke BX, Lamont PJ, Turnbull DM, McFarland R, Taylor RW, Thorburn DR. **Respiratory chain complex I deficiency caused by mitochondrial DNA mutations.** Eur J Hum Genet. 2011 Jul;19(7):769-75. doi: 10.1038/ejhg.2011.18. Epub 2011 Mar 2. *(PubMed Abstract)*

- Taylor M, Slavov D, Salcedo E, Zhu X, Ferguson D, Jirikowic J, Di Lenarda A, Sinagra G, MD, Mestroni L. ***Tafazzin* gene mutations are uncommon causes of dilated cardiomyopathy in adults.** Cardiogenetics.2011.e4 | Published: 2011-07-05. *(Abstract)**▼

- Wajner M, Goodman SI. **Disruption of mitochondrial homeostasis in organic acidurias: Insights from human and animal studies.** J Bioenerg Biomembr. 2011 Feb;43(1):31-8. *(PubMed Abstract)*

- Sandlers Y, Baumann S. **Characterization of urine metabolites in Barth syndrome patients employing a non-targeted GC/MS screening approach.** 59th ASMS Conference on Mass Spectrometry, June 5 - 9, 2011, Denver, Colorado. (*Poster*)▼

- Zhang L, Bell RJA, Kiebish MA, Seyfried TN, Han X, Gross RW, Chuang JH. **A mathematical model for the determination of steady-state cardiolipin remodeling mechanisms using lipidomic data.** PLoS One. 2011;6(6):e21170. doi: 10.1371/journal.pone.0021170. Epub 2011 Jun 10. *(PubMed – Open Access)*

- Garratt V, Riddiford D, Steward C, Tsai-Goodman B, Newbury-Ecob R. **What is Barth syndrome?** Midwives Magazine: Issue 4: 2011. *(Not peer reviewed) (Abstract)*▼

- Acehan D, Malhotra A, Xu Y, Ren M, Stokes DL, Schlame M. **Cardiolipin affects the supramolecular organization of ATP synthase in mitochondria.** Biophys J. 2011 May 4;100(9):2184-92. *(PubMed – Open Access)**

- Lisi EC, Cohn RD. **Genetic evaluation of the pediatric patient with hypotonia: Perspective from a hypotonia specialty clinic and review of the literature.** Dev Med Child Neurol. 2011 Jul;53(7):586-99. doi: 10.1111/j.1469-8749.2011.03918.x. Epub 2011 Mar 21. Review. *(PubMed Abstract)*

- Bockeria LA, Berishvili D, Baryshnikova I. **Re: Ventricular non-compaction in children: Cinical characteristics and course.** Interact Cardiovasc Thorac Surg. 2011 Mar;12(3):373. *(PubMed Abstract)*

- Rosca M, Minkler P, Hoppel CL. **Cardiac mitochondria in heart failure: Normal cardiolipin profile and increased threonine phosphorylation of complex IV**. Biochim Biophys Acta. 2011 Feb 11. [Epub ahead of print] *(PubMed Abstract)*

- Claypool SM, Whited K, Srijumnong S, Han X, Koehler CM**. Barth syndrome mutations that cause *tafazzin* complex lability.** J Cell Biol 2011 Feb 7;192(3):447-62. 192:447-462. *(PubMed Abstract)*

- Oechslin E, Jenni R. **Left ventricular non-compaction revisited: A distinct phenotype with genetic heterogeneity?** Eur Heart J. 2011 Jun;32(12):1446-56. Epub 2011 Jan 31. *(PubMed Abstract)*

- Acehan D, Vaz F, Houtkooper RH, James J, Moore V, Tokunaga C, Kulik W, Wansapura J, Toth MJ, Strauss A, Khuchua Z. **Cardiac and skeletal muscle defects in a mouse model of human Barth syndrome.** J Biol Chem. 2011 Jan 14;286(2):899-908. Epub 2010 Nov 9. *(PubMed – Open Access)**

- **Mouse model of Barth syndrome.** SciBX 3(47); Dec 9 2010. *(Nature Abstract)**

- Jalmar O, Garcia-Saez AJ, Berland L, Gonzalvez F, Petit PX. **Giant unilamellar vesicles (GUVs) as a new tool for analysis of caspase-8/Bid-FL complex binding to cardiolipin and its functional activity.** Cell Death Dis. 2010 Dec 2;1:e103. doi: 10.1038/cddis.2010.81. *(PubMed – Open Access)*▼

- Aljishi E, Ali F. **Barth syndrome: An X-linked cardiomyopathy with a novel mutation.** Indian J Pediatr. 2010 Dec;77(12):1432-3. Epub 2010 Oct 28. *(PubMed Abstract)*

- Soustek MS, Falk D, Mah C, Toth M, Schlame M, Lewin A, Byrne B. **Characterization of a transgenic shRNA induced murine model of *tafazzin* deficiency.** Hum Gene Ther. 2010 Nov 23. [Epub ahead of print] *(PubMed Abstract)*▼

- Takahashi H, Hayakawa T, Ito K, Takata M, Kobayashi T. **Small-angle and wide-angle X-ray scattering study on the bilayer structure of synthetic and bovine heart cardiolipins.** 2010 J. Phys.: Conf. Ser. 247 012021 *(Abstract)**

- Steward CG, Newbury-Ecob RA, Hastings R, Smithson SF, Tsai-Goodman B, Quarrell OW, Kulik W, Wanders R, Pennock M, Williams M, Cresswell JL, Gonzalez IL, Brennan P. **Barth syndrome: An X-linked cause of fetal cardiomyopathy and stillbirth.** Prenat Diagn. 2010 Oct;30(10):970-6. *(PubMed – Open Access)**▼

- Hauff K, Hatch GM. **Reduction in cholesterol synthesis in response to serum starvation in lymphoblasts of a patient with Barth syndrome.** Biochem Cell Biol. 2010 Aug;88(4):595-602. *(PubMed Abstract)**▼

- Rijken PJ. **Phosphatidylcholine-protein interactions and remodeling of cardiolipin in yeast mitochondria.** Doctoral Thesis, Scheikunde Proefschriften, 2010. *(Full Text)**

- Xu FY, McBride H, Acehan D, Vaz FM, Houtkooper RH, Lee RM, Mowat MA, Hatch GM. **The dynamics of cardiolipin synthesis post mitochondrial fusion.** Biochim Biophys Acta. 2010 Aug;1798(8):1577-85. Epub 2010 Apr 29. *(PubMed Abstract)**

- Kiebish MA, Bell R, Yang K, Phan T, Zhao Z, Ames W, Seyfried TN, Gross RW, Chuang JH, Han X. **Dynamic simulation of cardiolipin remodeling: Greasing the wheels for an interpretative approach to lipidomics.** J Lipid Res. 2010 Aug;51(8):2153-70. Epub 2010 Apr 21. *(PubMed Abstract)*

- He Q. ***Tafazzin* knockdown causes hypertrophy of neonatal ventricular myocytes.** Am J Physiol Heart Circ Physiol. 2010 Jul;299(1):H210-6. Epub 2010 Mar 26. *(PubMed – Open Access)**

- Rea SL, Graham BH, Nakamaru-Ogiso E, Kar A, Falk MJ. **Bacteria, yeast, worms, and flies: Exploiting simple model organisms to investigate human mitochondrial diseases.** Dev Disabil Res Rev. 2010 Jun;16(2):200-18. *(PubMed Abstract)*

- Osman C, Haag M, Wieland FT, Brügger B, Langer T. **A mitochondrial phosphatase required for cardiolipin biosynthesis: The PGP phosphatase Gep4.** EMBO J. 2010 Jun 16;29(12):1976-87. Epub 2010 May 18. *(PubMed Abstract)*

- Chang B, Momoi N, Shan L, Mitomo M, Aoyagi Y, Endo K, Takeda I, Chen R, Xing Y, Yu X, Watanabe S, Yoshida T, Kanegane H, Tsubata S, Bowles NE, Ichida F, Miyawaki T; Noncompaction study collaborators. **Gonadal mosaicism of a *TAZ* (G4.5) mutation in a Japanese family with Barth syndrome and left ventricular noncompaction.** Mol Genet Metab. 2010 Jun;100(2):198-203. Epub 2010 Mar 2. *(PubMed Abstract)*

- Zachman DK, Chicco AJ, McCune SA, Murphy RC, Moore RL, Sparagna GC. **The role of calcium-independent phospholipase A2 in cardiolipin remodeling in the spontaneously hypertensive heart failure rat heart.** J Lipid Res. 2010 Mar;51(3):525-34. Epub 2009 Sep 9. *(PubMed – Open Access)**

- Jefferies JL, Towbin JA. **Dilated cardiomyopathy.** Lancet. 2010 Feb 27;375(9716):752-62. *(PubMed Abstract)*

- Chen S, Liu D, Finley RL Jr, Greenberg ML. **Loss of mitochondrial DNA in the yeast cardiolipin synthase crd1 mutant leads to up-regulation of the protein kinase Swe1p that regulates the G2/M transition.** J Biol Chem. 2010 Apr 2;285(14):10397-407. Epub 2010 Jan 19. *(PubMed – Open Access)**

- Gebert N, Joshi AS, Kutik S, Becker T, McKenzie M, Li Guan X, Mooga VP, Stroud DA, Kulkarni G, Wenk MR, Rehling P, Meisinger C, Ryan MT, Wiedemann N, Greenberg ML, Pfanner N. **Mitochondrial cardiolipin involved in outer-membrane protein biogenesis: Implications for Barth syndrome.** Curr Biol. 2009 Dec 29;19(24):2133-9. Epub 2009 Dec 3. *(PubMed – Open Access)**

- Hauff KD, Choi SY, Frohman MA, Hatch GM. **Cardiolipin synthesis is required to support human cholesterol biosynthesis from palmitate upon serum removal in Hela cells.** Can J Physiol Pharmacol. 2009 Oct;87(10):813-20. *(PubMed – Open Access)**

- Taylor WA, Hatch GM. **Identification of the human mitochondrial linoleoyl-coenzyme a monolysocardiolipin acyltransferase (MLCL AT-1).** J Biol Chem. 2009 Oct 30;284(44):30360-71. Epub 2009 Sep 8. *(PubMed – Open Access)**

- Haines TH. **A new look at cardiolipin**. Biochim Biophys Acta. 2009 Oct;1788(10):1997-2002. *(No abstract available.)*

- Xu Y, Zhang S, Malhotra A, Edelman-Novemsky I, Ma J, Kruppa A, Cernicica C, Blais S, Neubert TA, Ren M, Schlame M. **Characterization of *tafazzin* splice variants from humans and fruit flies.** J Biol Chem. 2009 Oct 16;284(42):29230-9. Epub 2009 Aug 21. *(PubMed – Open Access)**

- Cosson L, Toutain A, Simard G, Paoli F, Kulik W, Vaz FM, Blasco H, Chantepie A, Labarthe F. **Barth syndrome in a female patient** [Abstract]. In the 11th International Congress of Inborn Errors of Metabolism Meeting, October 2009. Molecular Genetics and Metabolism 98, Issue 1 p. 89-118.

- Hastings R, Steward C, Tsai-Goodman B, Newbury-Ecob R. **Dysmorphology of Barth syndrome.** Clin Dysmorphol. 2009 Oct;18(4):185-7. *(PubMed Abstract)**▼

- Rijken PJ, Houtkooper RH, Akbari H, Brouwers JF, Koorengevel MC, de Kruijff B, Frentzen M, Vaz FM, de Kroon AI. **Cardiolipin molecular species with shorter acyl chains accumulate in S. cerevisiae mutants lacking the acyl-Coenzyme A-binding protein Acb1p. New insights into acyl chain remodeling of cardiolipin.** J Biol Chem. 2009 Oct 2;284(40):27609-19. Epub 2009 Aug 5. *(PubMed – Open Access)**

- Houtkooper RH, Turkenburg M, Poll-The BT, Karall D, Pérez-Cerdá C, Morrone A, Malvagia S, Wanders RJ, Kulik W, Vaz FM. **The enigmatic role of *tafazzin* in cardiolipin metabolism.** Biochim Biophys Acta. 2009 Oct;1788(10):2003-14. Epub 2009 Jul 18. Review. *(PubMed Abstract)**

- Koh C, Lee PW, Yung TC, Lun KS, Cheung YF. **Left ventricular noncompaction in children.** Congenit Heart Dis. 2009 Jul;4(4):288-94. *(PubMed Abstract)*

- Claypool SM. **Cardiolipin, a critical determinant of mitochondrial carrier protein assembly and function.** Biochim Biophys Acta. 2009 Oct;1788(10):2059-68. Epub 2009 May 5. Review. *(PubMed Abstract*)

- Schlattner U, Tokarska-Schlattner M, Ramirez S, Brückner A, Kay L, Polge C, Epand RF, Lee RM, Lacombe ML, Epand RM. **Mitochondrial kinases and their molecular interaction with cardiolipin.** Biochim Biophys Acta. 2009 Oct;1788(10):2032-47. Epub 2009 May 4. Review. *(PubMed – Open Access*)**

- Schlame M, Ren M. **The role of cardiolipin in the structural organization of mitochondrial membranes**. Biochim Biophys Acta. 2009 Oct;1788(10):2080-3. Epub 2009 May 4. Review. *(PubMed – Open Access)**

- Saini-Chohan HK, Holmes MG, Chicco AJ, Taylor WA, Moore RL, McCune SA, Hickson-Bick DL, Hatch GM, Sparagna GC. **Cardiolipin biosynthesis and remodeling enzymes are altered during the development of heart failure.** J Lipid Res. 2009 Aug;50(8):1600-8. Epub 2008 Nov 10. *(PubMed – Open Access)**

- Finsterer J. **Cardiogenetics, neurogenetics, and pathogenetics of left ventricular hypertrabeculation/noncompaction.** Pediatr Cardiol. 2009 Jul;30(5):659-81. Epub 2009 Jan 29. *(PubMed Abstract)*

- Zhou J, Zhong Q, Li G, Greenberg ML. **Loss of cardiolipin leads to longevity defects that are aleviated by alterations in stress response signaling.** J Biol Chem. 2009 Jul 3;284(27):18106-14. Epub 2009 Apr 28. *(PubMed – Open Access)**

- Bachou T, Giannakopoulos A, Trapali C, Vazeou A, Kattamis A. **A novel mutation in the G4.5 (*TAZ*) gene in a Greek patient with Barth syndrome.** Blood Cells Mol Dis. 2009 May-Jun;42(3):262-4. Epub 2009 Mar 3. *(PubMed Abstract)*

- Singh HR, Yang Z, Siddiqui S, Peña LS, Westerfield BH, Fan Y, Towbin JA, Vatta M. **A novel Alu-mediated Xq28 microdeletion ablates *TAZ* and partially deletes DNL1L in a patient with Barth syndrome.** Am J Med Genet A. 2009 May;149A(5):1082-5. No abstract available. *(PubMed Abstract)*

- Storch EA, Keeley M, Merlo LJ, St. Amant JB, Jacob M, Storch J, Spencer C, Byrne BJ. **Psychosocial functioning in youth with Barth syndrome.** Child Health Care. 2009 Apr;38(2):137-156. *(PubMed – Open Access)**▼

BSF does not endorse any drugs, tests, or treatments that we may report.

- Schlame M. **Formation of molecular species of mitochondrial cardiolipin. 2. A mathematical model of pattern formation by phospholipid transacylation.** Biochim Biophys Acta. 2009 Apr;1791(4):321-5. Epub 2009 Jan 31. *(PubMed Abstract)*

- Malhotra A, Xu Y, Ren M, Schlame M. **Formation of molecular species of mitochondrial cardiolipin. 1. A novel transacylation mechanism to shuttle fatty acids between sn-1 and sn-2 positions of multiple phospholipid species.** Biochim Biophys Acta. 2009 Apr;1791(4):314-20. Epub 2009 Jan 21. *(PubMed – Open Access)**

- Sparagna GC, Lesnefsky EJ. **Cardiolipin remodeling in the heart.** J Cardiovasc Pharmacol. 2009 Apr;53(4):290-301. Review. *(PubMed Abstract)**

- Beranek A, Rechberger G, Knauer H, Wolinski H, Kohlwein SD, Leber R. **Identification of a cardiolipin-specific phospholipase encoded by the gene CLD1 (YGR110W) in yeast.** J Biol Chem. 2009 Apr 24;284(17):11572-8. Epub 2009 Feb 25. *(PubMed Abstract)*

- Houtkooper RH, Rodenburg RJ, Thiels C, van Lenthe H, Stet F, Poll-The BT, Stone JE, Steward CG, Wanders RJ, Smeitink J, Kulik W, Vaz FM. **Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes and tissues using HPLC-mass spectrometry as a diagnostic test for Barth syndrome.** Anal Biochem. 2009 Apr 15;387(2):230-7. Epub 2009 Jan 31. *(PubMed Abstract)**

- Acehan D, Khuchua Z, Houtkooper RH, Malhotra A, Kaufman J, Vaz FM, Ren M, Rockman HA, Stokes DL, Schlame M. **Distinct effects of *tafazzin* deletion in differentiated and undifferentiated mitochondria.** Mitochondrion. 2009 Apr;9(2):86-95. Epub 2008 Dec 11. *(PubMed – Open Access)**

- Cortez-Dias N, Varela MG, Sargento L, Brito D, Almeida A, Cerqueira R, Lança V, Fernandes AR, Tavares P, Pereira RA, Fernandes A, Madeira H. **Left ventricular non-compaction: A new mutation predisposing to reverse remodeling?** Rev Port Cardiol. 2009 Feb;28(2):185-94. *(PubMed Abstract)*

- Malhotra A, Edelman-Novemsky I, Xu Y, Plesken H, Ma J, Schlame M, Ren M. **Role of calcium-independent phospholipase A2 in the pathogenesis of Barth syndrome.** Proc Natl Acad Sci U S A. 2009 Feb 17;106(7):2337-41. Epub 2009 Jan 21. *(PubMed – Open Access)**

- Ichida F**. Left ventricular noncompaction.** Circ J. 2009 Jan;73(1):19-26. Epub 2008 Dec 4. Review. *(PubMed Abstract*)

- van Raam BJ, Kuijpers TW. **Mitochondrial defects lie at the basis of neutropenia in Barth syndrome.** Curr Opin Hematol. 2009 Jan;16(1):14-19. *(PubMed Abstract)*

- Hauff K, Linda D, Hatch GM. **Mechanism of the elevation in cardiolipin during Hela cell entry into the S phase of the human cell cycle.** Biochem J. 2009 Jan 15;417(2):573-82. *(PubMed Abstract)*

- Joshi AS, Zhou J, Gohil VM, Chen S, Greenberg ML. **Cellular functions of cardiolipin in yeast.** Biochim Biophys Acta. 2009 Jan;1793(1):212-8. Epub 2008 Aug 7. *(PubMed – Open Access)**

- Chen S, Tarsio M, Kane PM, Greenberg ML. **Cardiolipin mediates cross-talk between mitochondria and the vacuole.** Mol Biol Cell. 2008 Dec;19(12):5047-58. Epub 2008 Sep 17. *(PubMed – Open Access)**

- Claypool SM, Boontheung P, McCaffery JM, Loo JA, Koehler CM. **The cardiolipin transacylase, *tafazzin*, associates with two distinct respiratory components providing insight into Barth syndrome.** Mol Biol Cell. 2008 Dec;19(12):5143-55. Epub 2008 Sep 17. *(PubMed Abstract)*

- Sweeney RT, Davis GJ, Noonan JA. **Cardiomyopathy of unknown etiology: Barth syndrome unrecognized.** Congenit Heart Dis. 2008 Nov;3(6):443-8. *(PubMed Abstract)*

- Scorrano L. **Caspase-8 goes cardiolipin: A new platform to provide mitochondria with microdomains of apoptotic signals?** J Cell Biol. 2008 Nov 17;183(4):579-81. Epub 2008 Nov 10. *(PubMed Abstract)*

- Gonzalvez F, Schug ZT, Houtkooper RH, Mackenzie ED, Brooks DG, Wanders RJ, Petit PX, Vaz FM, Gottlieb E. **Cardiolipin provides an essential activating platform for caspase-8 on mitochondria.** J Cell Biol. 2008 Nov 17;183(4):681-96. Epub 2008 Nov 10. *(PubMed – Open Access)**

- Huang SC, Wu ET, Chiu SN, Hwu WL, Wu MH, Wang SS. **Mitral annuloplasty in an infant with Barth syndrome and severe mitral insufficiency: First case report and determination of annular diameter.** J Thorac Cardiovasc Surg. 2008 Oct;136(4):1095-7. *(PubMed - No Abstract Available)*

- Tikhomirov E, Averyanova N, Basargina E, Degtyareva T. **Gene symbol: *TAZ*. Disease: Barth syndrome.** Hum Genet. 2008 Oct;124(3):315-6. *(PubMed - No Abstract Available)*

- McCanta AC, Chang AC, Weiner K. **Cardiomyopathy in a child with neutropenia and motor delay**. Curr Opin Pediatr. 2008 Oct;20(5):605-7. *(PubMed Abstract)*

- De Decker R, Seller N, Van der Watt G, Kulik W. **Barth syndrome: How to (easily) find a needle in a haystack.** Cardiovasc J Afr. 2008 Sep-Oct;19(5 Suppl):S9. *(PubMed - No abstract available)*

- Schlame M. **Cardiolipin synthesis for the assembly of bacterial and mitochondrial membranes**. J Lipid Res. 2008 Aug;49(8):1607-20. Epub 2007 Dec 12. *(PubMed Abstract)*

- Yen TY, Hwu WL, Chien YH, Wu MH, Lin MT, Tsao LY, Hsieh WS, Lee NC. **Acute metabolic decompensation and sudden death in Barth syndrome: Report of a family and a literature review.** Eur J Pediatr. 2008 Aug;167(8):941-4. Epub 2007 Sep 11. *(PubMed Abstract)*

- Chen S, He Q, Greenberg ML. **Loss of *tafazzin* in yeast leads to increased oxidative stress during respiratory growth.** Mol Microbiol. 2008 May;68(4):1061-72. *(PubMed Abstract)**

- Jackson SK, Abate W, Tonks AJ. **Lysophospholipid acyltransferases: Novel potential regulators of the inflammatory response and target for new drug discovery.** Pharmacol Ther. 2008 Jul;119(1):104-14. Epub 2008 Apr 23. Review. *(PubMed Abstract)*

- Boztug K, Welte K, Zeidler C, Klein C. **Congenital neutropenia syndromes.** Immunol Allergy Clin North Am 2008 May;28(2):259-75, vii-viii. Review. *(PubMed Abstract)*

- Finsterer J, Stollberger C, Blazek G. **Prevalence of Barth syndrome in adult left ventricular hypertrabeculation / noncompaction**. Scand Cardiovasc J. 2008 Apr;42(2):157-60. *(PubMed Abstract)*

- Tatu-Chitoiu A, Bradisteanu S. **A rare case of biventricular non-compaction associated with ventricular septal defect and descendent aortic stenosis in a young man.** Eur J Echocardiogr. 2008 Mar;9(2):306-8. *(PubMed Abstract)*

- Danos M, Taylor WA, Hatch GM. **Mitochondrial monolysocardiolipin acyltransferase is elevated in the surviving population of H9c2 cardiac myoblast cells exposed to 2-deoxyglucose-induced apoptosis.** Biochem Cell Biol. 2008 Feb;86(1):11-20. *(PubMed Abstract)*

- Kulik W, van Lenthe H, Stet FS, Houtkooper RH, Kemp H, Stone JE, Steward CG, Wanders RJ, Vaz FM. **Bloodspot assay using HPLC-tandem mass spectrometry for detection of Barth syndrome.** Clin Chem. 2008 Feb;54(2):371-8. Epub 2007 Dec 10. *(PubMed – Open Access)**▼

- Hullin-Matsuda F, Kawasaki K, Delton-Vandenbroucke I, Xu Y, Nishijima M, Lagarde M, Schlame M, Kobayashi T. **De novo biosynthesis of the late endosome lipid, bis(monoacylglycero)phosphate.** J Lipid Res. 2007 Sep;48(9):1997-2008. Epub 2007 Jun 8. *(PubMed – Open Access)**

- Zhong Q, Li G, Gvozdenovic-Jeremic J, Greenberg ML. **Up-regulation of the cell integrity pathway in saccharomyces cerevisiae suppresses temperature sensitivity of the pgs1Delta mutant.** J Biol Chem. 2007 Jun 1;282(22):15946-53. Epub 2007 Apr 9. *(PubMed – Open Access)**

- Mangat J, Lunnon-Wood T, Rees P, Elliott M, Burch M. **Successful cardiac transplantation in Barth syndrome - single-centre experience of four patients.** Pediatr Transplant. 2007 May;11(3):327-331. *(PubMed Abstract)*

- Marziliano N, Mannarino S, Nespoli L, Diegoli M, Pasotti M, Malattia C, Grasso M, Pilotto A, Porcu E, Raisaro A, Raineri C, Dore R, Maggio PP, Brega A, Arbustini E. **Barth syndrome associated with compound hemizygosity and heterozygosity of the *TAZ* and LDB3 genes.** Am J Med Genet A. 2007 May 1;143A(9):907-15. *(PubMed Abstract)*

- Crawford JM. **A 3D view of Barth syndrome mitochondria**. Lab Invest. (2007), 1-2. *(Abstract)*

- Kirwin SM, Vinette KM, Schwartz SB, Funanage VL, Gonzalez IL. **Multiple transmissions of Barth syndrome through an oocyte donor with a de novo *TAZ* mutation.** Fertil Steril. 2007 Apr;87(4):976.e5-7. Epub 2007 Jan 22. *(PubMed Abstract)**

- Yaplito-Lee J, Weintraub R, Jamsen K, Chow CW, Thorburn DR, Boneh A. **Cardiac manifestations in oxidative phosphorylation disorders of childhood.** J Pediatr. 2007 Apr;150(4):407-11. *(PubMed Abstract)*

- Mazzocco MM, Henry AE, Kelley RI. **Barth syndrome is associated with a cognitive phenotype.** J Dev Behav Pediatr. 2007 Feb;28(1):22-30. *(PubMed – Open Access)*▼

- Acehan D, Xu Y, Stokes DL, Schlame M. **Comparison of lymphoblast mitochondria from normal subjects and patients with Barth syndrome using electron microscopic tomography.** Lab Invest. 2007 Jan;87(1):40-8. Epub 2006 Oct 16. *(PubMed – Open Access)**▼

- Day JA, Spencer CT, Byrne B. **Grip strength in a population of boys with Barth syndrome.** Pediatric Physical Therapy Vol. 18(1)88-89 Spring 2006. *(Abstract)*▼

- Xu Y, Malhotra A, Ren M, Schlame M. **The enzymatic function of *tafazzin*.** J Biol Chem. 2006 Dec 22;281(51):39217-24. Epub 2006 Nov 2. *(PubMed Abstract)**▼

- Schlame M, Ren M. **Barth syndrome, a human disorder of cardiolipin metabolism.** FEBS Lett. 2006 Oct 9;580(23):5450-5. Epub 2006 Jul 17. *(PubMed – Open Access)**

- Donati MA, Malvagia S, Pasquini E, Morrone A, Marca GL, Garavaglia B, Toniolo D, Zammarchi E. **Barth syndrome presenting with acute metabolic decompensation in the neonatal period.** J Inherit Metab Dis. 2006 Oct;29(5):684. Epub 2006 Aug 12. *(PubMed Abstract)*

- van Werkhoven MA, Thorburn DR, Gedeon AK, Pitt JJ. **Monolysocardiolipin in cultured fibroblasts is a sensitive and specific marker for Barth syndrome.** J Lipid Res. 2006 Oct;47(10):2346-51. Epub 2006 Jul 27. *(PubMed Abstract)*

- Ances BM, Sullivan J, Weigele JB, Hwang V, Messe SR, Kasner SE, Liebeskind DS. **Stroke associated with Barth syndrome.** J Child Neurol. 2006 Sep;21(9):805-7. *(PubMed Abstract)*

- Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, Ren M, Schlame M. **A Drosophila model of Barth syndrome**. Proc Natl Acad Sci U S A. 2006 Aug 1;103(31):11584-8. Epub 2006 Jul 19. *(PubMed – Open Access)**

- Spencer CT, Bryant RM, Day J, Gonzalez IL, Colan SD, Thompson WR, Berthy J, Redfearn SP, Byrne BJ. **Cardiac and clinical phenotype in Barth syndrome.** Pediatrics. 2006 Aug;118(2):e337-46. Epub 2006 Jul 17. *(PubMed Abstract)**▼

- McKenzie M, Lazarou M, Thorburn DR, Ryan MT. **Mitochondrial respiratory chain super-complexes are destabilized in Barth syndrome patients.** J Mol Biol. 2006 Aug 18;361(3):462-9. *(PubMed Abstract)*

- Claypool SM, McCaffery JM, Koehler CM. **Mitochondrial mislocalization and altered assembly of a cluster of Barth syndrome mutant *tafazzins*.** J Cell Biol. 2006 Jul 31;174(3):379-90. *(PubMed Abstract)*

- Khuchua Z, Yue Z, Batts L, Strauss AW. **A zebrafish model of human Barth syndrome reveals the essential role of *tafazzin* in cardiac development and function.** Circ Res. 2006 Jul 21;99(2):201-8. Epub 2006 Jun 22. *(PubMed – Open Access)**

- Li G, Chen S, Thompson MN, Greenberg ML. **New insights into the regulation of cardiolipin biosynthesis in yeast: Implications for Barth syndrome.** Biochim Biophys Acta. 2007 Mar;1771(3):432-41. Epub 2006 Jul 8. Review. *(PubMed Abstract)**

- Brady AN, Shehata BM, Fernhoff PM. **X-linked fetal cardiomyopathy caused by a novel mutation in the *TAZ* gene.** Prenat Diagn. 2006 May;26(5):462-5. *(PubMed Abstract)*

- Hauff KD, Hatch, GM. **Cardiolipin metabolism and Barth syndrome.** Progress in Lipid Research 2006 Mar;45(2):91-101. Epub 2006 Jan 18. *(PubMed Abstract)**

- Schlame M, Ren M, Xu Y, Greenberg ML, Haller I. **Molecular symmetry in mitochondrial cardiolipins.** Chem Phys Lipids 2005 Dec;138(1-2):38-49. Epub 2005 Sep 7. *(PubMed Abstract)**

- Brandner K, Mick DU, Frazier AE, Taylor RD, Meisinger C, Rehling P. ***TAZ*1, an outer mitochondrial membrane protein, affects stability and assembly of inner membrane protein complexes: Implications for Barth syndrome.** Mol Biol Cell. 2005 Nov; 16(11): 5202-5214. *(PubMed Abstract)*

- Zhong Q, Greenberg ML. **Deficiency in mitochondrial anionic phospholipid synthesis impairs cell wall biogenesis.** Biochem Soc Trans. 2005 Oct;33(Pt 5):1158-61. *(PubMed Abstract)*

- DiMauro S, Gurgel-Giannetti J. **The expanding phenotype of mitochondrial myopathy.** Curr Opin Neurol. 2005 Oct;18(5):538-42. *(PubMed Abstract)*

- Spencer CT, Byrne BJ, Gewitz MH, Wechsler SB, Kao AC, Gerstenfeld EP, Merliss AD, Carboni MP, Bryant RM. **Ventricular arrhythmia in the X-linked cardiomyopathy Barth syndrome.** Pediatr Cardiol. 2005 Sep-Oct;26(5):632-7. *(PubMed Abstract)**▼

- Huhta JC, Pomerance HH, Barness EG. **Clinicopathologic conference: Barth syndrome.** Fetal Pediatr Pathol. 2005 Jul-Aug;24(4):239-54. *(PubMed Abstract)*

- Xu Y, Sutachan JJ, Plesken H, Kelley RI, Schlame M. **Characterization of lymphoblast mitochondria from patients with Barth syndrome.** Lab Invest. 2005 Jun;85(6):823-30. *(PubMed – Open Access)**

- Valianpour F, Mitsakos V, Schlemmer D, Towbin JA, Taylor JM, Ekert PG, Thorburn DR, Munnich A, Wanders RJ, Barth P, Vaz FM. **Monolysocardiolipins accumulate in Barth syndrome but do not lead to enhanced apoptosis.** J Lipid Res. 2005 Jun;46(6):1182-95. Epub 2005 Apr 1. *(PubMed – Open Access)**▼

- Bonilla FA, Bernstein IL, Khan DA, Ballas ZK, Chinen J, Frank MM, Kobrynski LJ, Levinson AI, Mazer B, Nelson RP Jr, Orange JS, Routes JM, Shearer WT, Sorensen RU; American Academy of Allergy, Asthma and Immunology; American College of Allergy, Asthma and Immunology; Joint Council of Allergy, Asthma and Immunology. **Practice parameter for the diagnosis and management of primary immunodeficiency.** Ann Allergy Asthma Immunol. 2005 May;94(5 Suppl 1):S1-63. *(PubMed Abstract)*

- Tajima T, Satoh K, Okuhara K, Tsubaki J, Fujieda K**. Hypomagnesemia in a patient with Barth syndrome.** J Pediatr Endocrinol Metab. 2005 May;18(5):523. *(PubMed Abstract)*

- Gonzalez IL. **Barth syndrome: *TAZ* gene mutations, mRNAs, and evolution.** Am J Med Genet A. 2005 May 1;134(4):409-14. *(PubMed Abstract)**

- Damin, M. **Barth Syndrome: Rare or rarely diagnosed.** Genetic Interest Group Newsletter Winter 2004/05. *(Not peer reviewed). (Full text)*

- Lu B, Kelher MR, Lee DP, Lewin TM, Coleman RA, Choy PC, Hatch GM. **Complex expression pattern of the Barth syndrome gene product *tafazzin* in human cell lines and murine tissues**. Biochem Cell Biol. 2004 Oct;82(5):569-76. *(PubMed Abstract)**

- Ma L, Vaz FM, Gu Z, Wanders RJ, Greenberg ML. **The human *TAZ* gene complements mitochondrial dysfunction in the yeast *taz*1delta mutant-implications for Barth syndrome**. J Biol Chem. 2004 Oct 22;279(43):44394-9. Epub 2004 Aug 10. *(PubMed – Open Access)**

- Valianpour F. **A mass spectrometric approach to investigate cardiolipin metabolism in Barth syndrome.** PhD Dissertation, Academic Medical Center, University of Amsterdam, September 2004. (*Not peer reviewed)*

- Soergel DG**. Highlights of the 2004 Barth Syndrome International Scientific/Medical and Family Conference.** Pediatric Cardiology Today August 2004. *(Not peer reviewed)*

- Steward CG, Martin RP, Hayes AM, Salmon AP, Williams MM, Tyfield LA, Davies SJ, Newbury-Ecob RA. **Cardiology: Barth syndrome (X-Linked cardiac and skeletal myopathy, neutropenia, and organic aciduria): Rarely recognised, frequently fatal.** Arch Dis Child 2004;89(Suppl I):A47–A48; G140. *(Abstract)*

- Hennekam RCM. Invited Comment. **The challenge in hidden treasures: A never ending story.** American Journal of Medical Genetics 2004, 126A:331-332. *(Abstract)*

- McCurdy, KR. **Responding to a genetic disorder: A case becomes a cause.** Health Advocacy Bulletin Fall 2004. *(Not peer reviewed)*

- Tusek KC. **Barth Syndrome. A review of the pathophysiology and psychosocial manifestations of this X-linked mitochondrial disorder.** Master's Thesis, Human Genetics Program, Sarah Lawrence College, May 2004. *(Not peer reviewed.)*

- Kuijpers TW, Maianski NA, Tool AT, Becker K, Plecko B, Valianpour F, Wanders RJ, Pereira R, Van Hove J, Verhoeven AJ, Roos D, Baas F, Barth PG. **Neutrophils in Barth syndrome (BTHS) avidly bind annexin-V in the absence of apoptosis.** Blood. 2004 May 15;103(10):3915-23. Epub 2004 Feb 5. *(PubMed – Open Access)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Barth PG, Valianpour F, Bowen VM, Lam J, Duran M, Vaz FM, Wanders RJ. **X-linked cardioskeletal myopathy and neutropenia (Barth syndrome): An update**. Am J Med Genet. 2004 May 1;126A(4):349-54. *(PubMed Abstract)*

- Schmidt MR, Birkebaek N, Gonzalez I, Sunde L. **Barth syndrome without 3-methylglutaconic aciduria.** Acta Paediatr. 2004 Mar;93(3):419-21. *(PubMed Abstract)*

- Maianski NA, Geissler J, Srinivasula SM, Alnemri ES, Roos D, Kuijpers TW. **Functional characterization of mitochondrial in neutrophils: A role restricted to apoptosis.** Cell Death Differ 2004 Feb; 11(2):143-53. *(PubMed Abstract)*

- Hatch GM. **Cell biology of cardiac mitochondrial phospholipids**. Biochem Cell Biol 2004 Feb;82(1):99-112. *(PubMed Abstract)*

- Gu Z, Valianpour F, Chen S, Vaz FM, Hakkaart GA, Wanders RJA, Greenberg ML. **Aberrant cardiolipin metabolism in the yeast *taz1* mutant: a model for Barth syndrome**. Mol Microbiol 2004 Jan; 51(1):149-158. *(PubMed Abstract)**

- Rugolotto S, Prioli MD, Toniolo D, Pellegrino P, Catuogno S, Burlina AB. **Long-term treatment of Barth syndrome with pantothenic acid: A retrospective study.** Mol Genet Metab 2003 Dec; 80(4):408-11. *(PubMed Abstract)*

- Schlame M, Kelley RI, Feigenbaum A, Towbin JA, Heerdt PM, Schlieble T, Wanders RJ, DiMauro S, Blanck TJ. **Phospholipid abnormalities in children with Barth syndrome**. J Am Coll Cardiol 2003 Dec 3; 42(11):1994-1999. *(PubMed Abstract)*▼

- Versluys B, Bowen V, McCurdy K, Mann S, Cantlay A, Newbury-Ecob R, Kern I, Goulden N, Steward C. **X-linked organic aciduria: An important cause of neutropenia in males**. Blood 2003 Nov 16; 102(11), #965. *(Abstract)*▼

- Bateman C. **Little known killer: Barth syndrome**. S Afr Med J 2003 Apr; 93(4):249-250. *(PubMed Abstract)*

- Khan IA, Biddle WP, Najeed SA, Abdul-Aziz S, Mehta NJ, Salaria V, Murcek AL, Harris DM. **Isolated noncompaction cardiomyopathy presenting with paroxysmal supraventricular tachycardia--case report and literature review.** Angiology. 2003 Mar-Apr;54(2):243-50. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Overmars H, Vaz FM, Barth PG, van Gennip AH. **Linoleic acid supplementation of Barth syndrome fibroblasts restores cardiolipin levels: Implications for treatment**. J Lipid Res. 2003 Mar;44(3):560-6. Epub 2002 Dec 16. *(PubMed Abstract)*▼

- Folger U, Cario H, Panis A, Keck T. **Postinflammatorial stenoses of the pharynx in a child with Barth syndrome**. Int J Pediatr Otorhinolaryngol 2003 Feb; 67(2)117-120. *(PubMed Abstract)*

- Vesel S, Stopar-Obreza M, Trebusak-Podkrajsek K, Jazbec J, Podnar T, Battelino T. **A novel mutation in the G4.5 (*TAZ*) gene in a kindred with Barth syndrome**. Eur J Hum Genet 2003 Jan; 11(1):97-101. *(PubMed Abstract)*

- Spencer CT, Colan SD, Gonzalez I, Byrne BJ. **Characterization of the cardiomyopathy associated with Barth syndrome.** January 2003.

- Chen R, Tsuji T, Ichida F, Bowles KR, Yu X, Watanabe S, Hirono K, Tsubata S, Hamamichi Y, Ohta J, Imai Y, Bowles NE, Miyawaki T, Towbin JA. Noncompaction study collaborators. **Mutation analysis of the G4.5 gene in patients with isolated left ventricular noncompaction**. Mol Genet Metab 2002 Dec; 77(4):319-325. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Overmars H, Vreken P, van Gennip AH, Baas F, Plecko B, Santer R, Becker K, Barth PG. **Cardiolipin deficiency in X-linked cardioskeletal myopathy and neutropenia (Barth syndrome, MIM 302060): A study in cultured skin fibroblasts**. J Pediatr 2002 Nov; 141(5):729-733. *(PubMed Abstract)*▼

- Chen Y. **Molecular studies on a *Dictyostelium* homolog of the *tafazzin* gene, the cause of Barth syndrome in humans**. Dr. rer. nat. dissertation; University of Kassel; November 2002. *(Not peer reviewed).*

- Kuijpers, TW. **Clinical symptoms and neutropenia: The balance of neutrophil development, functional activity, and cell death.** Eur J Pediatr 2002 Oct;161 Suppl 1:S75-82. Epub 2002 Sep 13. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Barth PG, Overmars H, van Gennip AH. **Quantitative and compositional study of cardiolipin in platelets by electrospray ionization mass spectrometry: Application for the identification of Barth syndrome patients**. Clin Chem. 2002 Sep;48(9):1390-7. *(PubMed Abstract)*

- Sakamoto O, Kitoh T, Ohura T, Ohya N, Iinuma K. **Novel missense mutation (R94S) in the *TAZ* (G4.5) gene in a Japanese patient with Barth syndrome**. J Hum Gen 2002; 47(5):229-231. *(PubMed Abstract)*

- Schlame M, Towbin JA, Heerdt PM, Jehle R, DiMauro S, Blanck TJJ. **Deficiency of tetralinoleoyl-cardiolipin in Barth syndrome**. Ann Neurol. 2002 May;51(5):634-7. *(PubMed Abstract)*▼

- Nakamura A, Ikeda S. **Barth syndrome (X-linked cardioskeletal myopathy and neutropenia)**. [Article in Japanese]. Nippon Rinsho. 2002 Apr;60 Suppl 4:389-91. *(PubMed Abstract)*

- Harbor Transcript -- A Newsletter of Cold Spring Harbor Laboratory. **Barth syndrome and "Neuwald's Hypothesis": From computer to clinic.** Winter/Spring 2002 Vol. 21 No. 1/2. *(Full text)*

- Bissler JJ, Tsoras M, Goring HHH, Hug P, Chuck G, Tombragel E, McGraw C, Schlotman J, Ralston MA, Hug G. **Infantile dilated X-linked cardiomyopathy, altered lipids, and ultrasound malformations of mitochondria in heart, liver, and skeletal muscle**. Lab Invest. 2002 Mar;82(3):335-44. *(PubMed Abstract)*

- Katsushima Y, Fujiwara I, Sakamoto O, Ohura T, Miyabayashi S, Ohnuma A, Yamaguchi S, Iinuma K. **Normal pituitary function in a Japanese patient with Barth syndrome**. Eur J Pediatr 2002 Jan; 161(1):67-68. *(PubMed Abstract)*

- Zeidler C, Barth PG, Bonilla MA, Bolyard AA, Boxer L, Cottle T, Dale DC, Donadieu J, Fier C, Freedman M, Kannourakis G, Kinsey S, Liang B, Schwinzer B, Welte K, Cham B, for the Severe Chronic Neutropenia International Registry (SCNIR). **Neutropenia in Barth syndrome: Clinical course and treatment of neutropenia**. Blood 2001; 98(11):300a.

- Sakamoto O, Ohura T, Katsushima Y, Fujiwara I, Ogawa E, Miyabayashi S, Iinuma K. **A novel intronic mutation of the *TAZ* (G4.5) gene in a patient with Barth syndrome: Creation of a 5' splice donor site with variant GC consensus and elongation of the upstream exon**. Hum Genet 2001 Nov; 109(5):559-563. *(PubMed Abstract)*

- Waggoner DJ, Brown RL, Hedrick J. **Hypoglycemia and dicarboxylic aciduria as the presenting biochemical findings in a patient with Barth syndrome.** American Journal of Human Genetics 69.4 (Oct 2001): p492. *(Abstract)*

- Cox GF. **Barth syndrome.** NORD Physician's Guide to Rare Diseases 2001.

- Mazzocco MM, Kelley RI. **Preliminary evidence for a cognitive phenotype in Barth syndrome**. Am J Med Genet 2001 Sept 1; 102(4):372-378. *(PubMed Abstract)*▼

- Makita Y. **[Barth syndrome]** [Article in Japanese]. Ryoikibetsu Shokogun Shirizu 2001; (33):254-5. *(PubMed Abstract)*

- Ronghe MD, Foot AB, Martin R, Ashworth M, Steward CG. **Non-Epstein-Barr virus-associated T-cell lymphoma following cardiac transplantation for Barth syndrome.** Acta Paediatr. 2001 May;90(5):584-6. *(PubMed Abstract)*

- Ichida F, Tsubata S, Bowles KR, Haneda N, Uses K, Miyawaki T, Dreyer WJ, Messina J, Li H, Bowles NE, Towbin JA. **Novel gene mutations in patients with left ventricular noncompaction or Barth syndrome**. Circulation. 2001 Mar 6;103(9):1256-63. *(PubMed Abstract)*

- Gonzalez IL. **Genetic disease and DNA for the families and children.** June 2002

- Rost I, Duroux A, Toniolo D, Holinski-Feder E, Kozlik-Feldman R. **Barth syndrome: X-linked dilated cardiomyopathy.** [Article in German] Monatsschrift Kinderheilkunde 2000; 148(3): 2246-250.

- Vreken P, Valianpour F, Nijtmans LG, Grivell LA, Plecko B, Wanders RJA, Barth PG. **Defective remodeling of cardiolipin and phosphatidylglycerol in Barth syndrome**. Biochem Biophys Res Commun. 2000 Dec 20;279(2):378-82. *(PubMed Abstract)*

- Cantlay AM, Shokrollahi K, Allen JT, Lunt PW, Newbury-Ecob RA, Steward CG. **Genetic analysis of the G4.5 gene in families with suspected Barth syndrome.** J Pediatr. 1999 Sep;135(3):311-5. *(PubMed Abstract)*

- Barth PG, Wanders RJ, Vreken P, Janssen EA, Lam J, Baas F. **X-linked cardioskeletal myopathy and neutropenia (Barth syndrome) (MIM 302060**). J Pediatr. 1999 Sep;135(3):273-6. *(PubMed Abstract)*

- Orstavik KH, Orstavik RE, Naumova AK, D'Adamo P, Gedeon A, Bolhuis PA, Barth PG, Toniolo D. **X chromosome inactivation in carriers of Barth syndrome**. Am J Hum Genet. 1998 Nov;63(5):1457-63. *(PubMed Abstract)*

- Neuwald, AF. **Barth syndrome may be due to an acyltransferase deficiency**. Curr Biol. 1997 Aug 1;7(8):R465-6. *(PubMed Abstract)*

- Barth PG, Van den Bogert C, Bolhuis PA, Scholte HR, van Gennip AH, Schutgens RB, Ketel AG. **X-linked cardioskeletal myopathy and neutropenia (Barth syndrome): Respiratory-chain abnormalities in cultured fibroblasts**. J Inherit Metab Dis. 1996; 19(2):157-60. *(PubMed Abstract)*

- Bione S, D'Adamo P, Maestrini E, Gedeon AK, Bolhuis PA, Toniolo D. **A novel X-linked gene, G4.5, is responsible for Barth syndrome**. Nat Genet. 1996 Apr;12(4):385-9. *(PubMed Abstract)*

- Cox GF, Pulsipher M, Rothenberg M, Korson M, Kelley RI. **Correction of neutropenia in Barth syndrome by G-CSF**. Am J Hum Genet 1995; 57:A177. *(Full Text)*

- Christodoulou J, McInnes RR, Jay V, Wilson G, Becker LE, Lehotay DC, Platt BA, Bridge PJ, Robinson BH, Clarke JT. **Barth syndrome: Clinical observations and genetic linkage studies**. Am J Med Genet. 1994 Apr 15;50(3):255-64. *(PubMed Abstract)*

- Kelly DP, Strauss AW. **Inherited cardiomyopathies.** N Engl J Med. 1994 Mar 31;330(13):913-9. No abstract available. *(PubMed)*

- Ades LC, Gedeon AK, Wilson MJ, Latham M, Partington MW, Mulley JC, Nelson J, Lui K, Sillence DO. **Barth syndrome: Clinical features and confirmation of gene localisation to distal Xq28**. Am J Med Genet. 1993 Feb 1;45(3):327-34. *(PubMed Abstract)*

- Kelley RI, Cheatham JP, Clark BJ, Nigro MA, Powell BR, Sherwood GW, Sladky JT, Swisher WP. **X-linked dilated cardiomyopathy with neutropenia, growth retardation, and 3-methylglutaconic aciduria**. J Pediatr. 1991 Nov;119(5):738-47. *(PubMed Abstract)*

- Kelley RI, Clark BJ, Morton DH, Sherwood WG. **X-linked cardiomyopathy, neutropenia, and increased urinary levels of 3-methylglutaconic and 2-ethylhydracrylic acids**. Am J Hum Genet 1989; 45(supplement):A7.

- Ino T, Sherwood WG, Cutz E, Benson LN, Rose V, Freedom RM. **Dilated cardiomyopathy with neutropenia, short stature, and abnormal carnitine metabolism**. J Pediatr. 1988 Sep;113(3):511-4. *(PubMed Abstract)*

- Barth PG, Scholte HR, Berden JA, Van der Klei-Van Moorsel JM, Luyt-Houwen IE, Van 't Veer-Korthof ET, Van der Harten JJ, Sobotka-Plojhar MA. **An X-linked mitochondrial disease affecting cardiac muscle, skeletal muscle and neutrophil leucocytes**. J Neurol Sci. 1983 Dec;62(1-3):327-55. *(PubMed Abstract)*

BARTH-LIKE DISORDERS AND ALLELES

- Neupert W. **A perspective on transport of proteins into mitochondria: A myriad of open questions**. J Mol Biol. 2015 Feb 9. pii: S0022-2836(15)00079-0. doi: 10.1016/j.jmb.2015.02.001. [Epub ahead of print] *(PubMed Abstract)*

- Zweigerdt R, Gruh I, Martin U. **Your heart on a chip: iPSC-based modeling of Barth-syndrome-associated cardiomyopathy.** Cell Stem Cell. 2014 Jul 3;15(1):9-11. doi: 10.1016/j.stem.2014.06.015. *(PubMed – Open Access)*

- Ojala T, Polinati P, Manninen T, Hiippala A, Rajantie J, Karikoski R, Suomalainen A, Tyni T. **New mutation of mitochondrial DNAJC19 causing dilated and noncompaction cardiomyopathy, anemia, ataxia, and male genital anomalies.** Pediatr Res. 2012 Oct;72(4):432-7. doi: 10.1038/pr.2012.92. Epub 2012 Jul 13. *(PubMed Abstract)*

- Pei W, Kratz LE, Bernardini I, Sood R, Yokogawa T, Dorward H, Ciccone C, Kelley RI, Anikster Y, Burgess HA, Huizing M, Feldman B. **A model of Costeff syndrome reveals metabolic and protective functions of mitochondrial OPA3.** Development 2010 Aug 1;137(15):2587-96. *(PubMed Abstract)*

- Davey KM, Parboosingh JS, McLeod DR, Chan A, Casey R, Ferreira P, Snyder FF, Bridge PJ, Bernier FP. **Mutation of DNAJC19, a human homolog of yeast inner mitochondrial membrane co-chaperones, causes DCMA syndrome, a novel autosomal recessive Barth syndrome-like condition.** J Med Genet. 2006 May;43(5):385-93. Epub 2005 Jul 31. *(PubMed Abstract)*

- De Kremer RD, Paschini-Capra A, Bacman S, Argarana C, Civallero G, Kelley RI, Guelbert N, Latini A, de Halac IN, Giner-Ayala A, Johnston J, Proujansky R, Gonzalez I, Depetris-Boldini C, Oller-Ramirez A, Angaroni C, Theaux RA, Hliba E, Juaneda E. **Barth's syndrome-like disorder: A new phenotype with a maternally inherited A3243G substitution of mitochondrial DNA (MELAS mutation)**. Am J Med Genet 2001 Mar 1; 99(2):83-93. *(PubMed Abstract)*

- Bleyl SB, Mumford BR, Thompson V, Carey JC, Pysher TJ, Chin TK, Ward K. **Neonatal, lethal noncompaction of the left ventricular myocardium is allelic with Barth syndrome**. Am J Hum Genet. 1997 Oct;61(4):868-72. *(PubMed Abstract)*

- Bleyl SB, Mumford BR, Brown-Harrison M-C, Pagotto LT, Carey JC, Pysher TJ, Ward K, Chin TK. **Xq28-linked noncompaction of the left ventricular myocardium: Prenatal diagnosis and pathologic analysis of affected individuals**. Am J Med Genet. 1997 Oct 31;72(3):257-65. *(PubMed Abstract)*

- Gedeon AK, Wilson MJ, Colley AC, Sillence DO, Mulley JC. **X-linked fatal infantile cardiomyopathy maps to Xq28 and is possibly allelic to Barth syndrome**. J Med Genet. 1995 May;32(5):383-8. *(PubMed Abstract)*

BIOCHEMISTRY

NEW Kojima R, Kakimoto Y, Furuta S, Itoh K, Sesaki H, Endo T, Tamura Y. **Maintenance of cardiolipin and crista structure requires cooperative functions of mitochondrial dynamics and phospholipid transport.** Cell Rep. 2019 Jan 15;26(3):518-528.e6. doi: 10.1016/j.celrep.2018.12.070. *(PubMed Abstract)*

NEW Van Strien J, Guerrero-Castillo S, Chatzispyrou IA, Houtkooper RH, Brandt U, Huynen MA. **COmplexome profiling aLignment (COPAL) reveals remodeling of mitochondrial protein complexes in Barth syndrome.** Bioinformatics. 2019 Jan 14. doi: 10.1093/bioinformatics/btz025. [Epub ahead of print] *(PubMed Abstract)*

NEW Gürtler S, Wolke C, Otto O, Heise N, Scholz F, Laporte A, Elsner M, Jörns A, Weinert S, Döring M, Jansing S, Gardemann A, Lendeckel U, Schild L. ***Tafazzin*-dependent cardiolipin composition in C6 glioma cells correlates with changes in mitochondrial and cellular functions, and cellular proliferation.** Biochim Biophys Acta Mol Cell Biol Lipids. 2019 Jan 9. pii: S1388-1981(19)30003-4. doi: 10.1016/j.bbalip.2019.01.006. [Epub ahead of print] *(PubMed Abstract)*

NEW Antunes D, Chowdhury A, Aich A, Saladi S, Harpaz N, Stahl M, Schuldiner M, Herrmann JM, Rehling P, Rapaport D. **Overexpression of branched-chain amino acid aminotransferases rescues the growth defects of cells lacking the Barth syndrome-related gene *TAZ*1.** J Mol Med (Berl). 2019 Jan 3. doi: 10.1007/s00109-018-1728-4. [Epub ahead of print] *(PubMed Abstract)**

NEW Kim J, Lee K, Fujioka H, Tandler B, Hoppel CL. **Cardiac mitochondrial structure and function in *tafazzin*-knockdown mice.** Mitochondrion. 2018 Oct 30. pii: S1567-7249(18)30074-6. doi: 10.1016/j.mito.2018.10.005. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Chao H, Anthonymuthu TS, Kenny EM, Amoscato AA, Cole LK, Hatch GM, Ji J, Kagan VE, Bayır H. **Disentangling oxidation/hydrolysis reactions of brain mitochondrial cardiolipins in pathogenesis of traumatic injury.** JCI Insight. 2018 Nov 2;3(21). pii: 97677. doi: 10.1172/jci.insight.97677. [Epub ahead of print] *(PubMed – Open Access)**

NEW Szeto HH. **Stealth peptides target cellular powerhouses to fight rare and common age-related diseases.** Protein Pept Lett. 2018 Oct 31. doi: 10.2174/0929866525666181101105209. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Chowdhury A, Aich A, Jain G, Wozny K, Lüchtenborg C, Hartmann M, Bernhard O, Balleiniger M, Alfar EA, Zieseniss A, Toischer K, Guan K, Rizzoli SO, Brügger B, Fischer A, Katschinski DM, Rehling P, Dudek J. **Defective mitochondrial cardiolipin remodeling dampens HIF-1α expression in hypoxia.** Cell Rep. 2018 Oct 16;25(3):561-570.e6. doi:10.1016/j.celrep.2018.09.057. *(PubMed – Open Access)*▼

NEW Li M, Hou T, Gao T, Lu X, Yang Q, Zhu Q, Li Z, Liu C, Mu G, Liu G, Bao Y, Wen H, Wang L, Wang H, Zhao Y, Gu W, Yang Y, Zhu WG. **p53 cooperates with SIRT6 to regulate cardiolipin de novo biosynthesis.** Cell Death Dis. 2018 Sep 20;9(10):941. doi: 10.1038/s41419-018-0984-0. *(PubMed - Open Access)*

NEW Pennington ER, Sullivan EM, Fix A, Dadoo S, Zeczycki TN, DeSantis A, Schlattner U, Coleman RA, Chicco AJ, Brown DA, Shaikh SR. **Proteolipid domains form in biomimetic and cardiac mitochondrial vesicles and are regulated by cardiolipin concentration but not monolyso-cardiolipin.** J Biol Chem. 2018 Aug 29. pii: jbc.RA118.004948. doi: 10.1074/jbc.RA118.004948. [Epub ahead of print] *(PubMed - Open Access)*

NEW Dudek J, Hartmann M, Rehling P. **The role of mitochondrial cardiolipin in heart function and its implication in cardiac disease.** Biochimica et Biophysica Acta (BBA) - Molecular Basis of Disease, In press, corrected proof, Available online 26 August 2018. *(ScienceDirect Abstract)*

NEW Paradies G, Paradies V, Ruggiero FM, Petrosillo G. **Mitochondrial bioenergetics and cardiolipin alterations in myocardial ischemia/reperfusion injury. Implications for pharmacological cardioprotection.** Am J Physiol Heart Circ Physiol. 2018 Aug 10. doi: 10.1152/ajpheart.00028.2018. [Epub ahead of print] *(PubMed Abstract)*

- Cole LK, Kim JH, Amoscato AA, Tyurina YY, Bayır H, Karimi B, Siddiqui TJ, Kagan VE, Hatch GM, Kauppinen TM. **Aberrant cardiolipin metabolism is associated with cognitive deficiency and hippocampal alteration in *tafazzin* knockdown mice.** Biochim Biophys Acta. 2018 Jul 25. pii: S0925-4439(18)30271-0. doi: 10.1016/j.bbadis.2018.07.022. [Epub ahead of print]*

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Xu G, Liu X, Shu Y, Pillai JA, Xu Y. **A rapid and sensitive LC-MS/MS method for quantitative analysis of cardiolipin (18:2)4 in human leukocytes and mouse skeletal muscles.** J Pharm Biomed Anal. 2018 Jun 20;158:386-394. doi: 10.1016/j.jpba.2018.06.035. [Epub ahead of print] *(PubMed Abstract)*

- Basu Ball W, Baker CD, Neff JK, Apfel GL, Lagerborg KA, Žun G, Petrovič U, Jain M, Gohil VM. **Ethanolamine ameliorates mitochondrial dysfunction in cardiolipin-deficient yeast cells.** J Biol Chem. 2018 Jun 4. pii:jbc.RA118.004014. doi:10.1074/jbc.RA118.004014. [Epub ahead of print] *(PubMed - Open Access)*

- Ban T, Kohno H, Ishihara T, Ishihara N. **Relationship between OPA1 and cardiolipin in mitochondrial inner-membrane fusion.** Biochim Biophys Acta. 2018 May 28. pii: S0005-2728(18)30132-4. doi: 10.1016/j.bbabio.2018.05.016. [Epub ahead of print] *(PubMed Abstract)*

- Boyd KJ, Alder NN, May ER. **Molecular dynamics analysis of cardiolipin and monolysocardiolipin on bilayer properties.** Biophys J. 2018 May 8;114(9):2116-2127. doi:10.1016/j.bpj.2018.04.001. *(PubMed Abstract)*

- Lou W, Reynolds CA, Li Y, Liu J, Hüttemann M, Schlame M, Stevenson D, Strathdee D, Greenberg ML. **Loss of *tafazzin* results in decreased myoblast differentiation in C2C12 cells: A myoblast model of Barth syndrome and cardiolipin deficiency.** Biochim Biophys Acta. 2018 Apr 22. pii: S1388-1981(18)30080-5. doi:10.1016/j.bbalip.2018.04.015. [Epub ahead of print] *(PubMed Abstract)**

- Oemer G, Lackner K, Muigg K, Krumschnabel G, Watschinger K, Sailer S, Lindner H, Gnaiger E, Wortmann SB, Werner ER, Zschocke J, Keller MA. **Molecular structural diversity of mitochondrial cardiolipins.** Proc Natl Acad Sci U S A. 2018 Apr 4. pii: 201719407. doi: 10.1073/pnas.1719407115. [Epub ahead of print] *(PubMed Abstract)*

- Chabi B, Fouret G, Lecomte J, Cortade F, Pessemesse L, Baati N, Coudray C, Lin L, Tong Q, Wrutniak-Cabello C, Casas F, Feillet-Coudray C. **Skeletal muscle overexpression of short isoform Sirt3 altered mitochondrial cardiolipin content and fatty acid composition.** J Bioenerg Biomembr. 2018 Mar 27. doi: 10.1007/s10863-018-9752-1. [Epub ahead of print] *(PubMed Abstract)*

- Mejia EM, Zegallai H, Bouchard ED, Banerji V, Ravandi A, Hatch GM. **Expression of human monolysocardiolipin acyltransferase-1 improves mitochondrial function in Barth syndrome lymphoblasts.** J Biol Chem. 2018 Mar 21. pii:jbc.RA117.001024. doi:10.1074/jbc.RA117.001024. [Epub ahead of print] *(PubMed - Open Access)** ▼

- Kimura T, Kimura A, Ren M, Berno B, Xu Y, Schlame M, Epand RM. **Substantial decrease in plasmalogen in the heart associated with *tafazzin* deficiency.** Biochemistry. 2018 Mar 20. doi: 10.1021/acs.biochem.8b00042. [Epub ahead of print] *(PubMed Abstract)* ▼

- Chen WW, Chao YJ, Chang WH, Chan JF, Hsu YHH. **Phosphatidylglycerol incorporates into cardiolipin to improve mitochondrial activity and inhibits inflammation.** Sci Rep. 2018 Mar 20;8(1):4919. doi: 10.1038/s41598-018-23190-z. *(PubMed Abstract)*

- Chao YJ, Wu WH, Balazova M, Wu TY, Lin J, Liu YW, Hsu YH. **Chlorella diet alters mitochondrial cardiolipin contents differentially in organs of Danio rerio analyzed by a lipidomics approach.** PLoS One. 2018 Mar 1;13(3):e0193042. doi:10.1371/journal.pone.0193042. eCollection 2018. *(PubMed - Open Access)*

- Sudrié-Arnaud B, Marguet F, Patrier S, Martinovic J, Louillet F, Broux F, Charbonnier F, Dranguet H, Coutant S, Vezain M, Lanos R, Tebani A, Fuller M, Lamari F, Chambon P, Brehin AC, Trestard L, Tournier I, Marret S, Verspyck E, Laquerrière A, Bekri S. **Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation.** Clin Chim Acta. 2018 Feb 21. pii: S0009-8981 (18)30084-6. doi: 10.1016/j.cca.2018.02.023. [Epub ahead of print] (*PubMed Abstract)*

- de Taffin de Tilques M, Lasserre JP, Godard F, Sardin E, Bouhier M, Le Guedard M, Kucharczyk R, Petit PX, Testet E, di Rago JP, Tribouillard-Tanvier D. **Decreasing cytosolic translation is beneficial to yeast and human *tafazzin*-deficient cells.** Microb Cell. 2018 Feb 18;5(5):220-232. doi: 10.15698/mic2018.05.629. *(PubMed – Open Access)**

- Gao F, McDaniel J, Chen EY, Rockwell HE, Nguyen C, Lynes MD, Tseng YH, Sarangarajan R, Narain NR, Kiebish MA. **Adapted MS/MSALL shotgun lipidomics approach for analysis of cardiolipin molecular species.** Lipids. 2018 Jan;53(1):133-142. doi: 10.1002/lipd.12004. (*PubMed Abstract)*

- Sullivan EM, Pennington ER, Sparagna GC, Torres MJ, Neufer PD, Harris M1, Washington J, Anderson EJ, Zeczycki TN, Brown DA, Shaikh SR. **Docosahexaenoic acid lowers cardiac mitochondrial enzyme activity by replacing linoleic acid in the phospholipidome.** J Biol Chem. 2018 Jan 12;293(2):466-483. doi: 10.1074/jbc.M117.812834. Epub 2017 Nov 21. (*PubMed Abstract)*

- Thong A, Tsoukanova V. **Cytochrome-c-assisted escape of cardiolipin from a model mitochondrial membrane.** Biochim Biophys Acta. 2018 Feb;1860(2):475-480. doi: 10.1016/j.bbamem.2017.10.032. Epub 2017 Nov 4. (*PubMed Abstract)*

- Serricchio M, Vissa A, Kim PK, Yip CM, McQuibban GA. **Cardiolipin synthesizing enzymes form a complex that interacts with cardiolipin-dependent membrane organizing proteins.** Biochim Biophys Acta. 2018 Jan 14. pii: S1388-1981(18)30010-6. doi:10.1016/j.bbalip.2018.01.007. [Epub ahead of print] (*PubMed Abstract)*

- Ohlig T, Le DV, Gardemann A, Wolke C, Gürtler S, Peter D, Schild L, Lendeckel U. **Effects of siRNA-dependent knock-down of cardiolipin synthase and *tafazzin* on mitochondria and proliferation of glioma cells.** Biochima et Biophysica Acta. Jan 6, 2018. (*PubMed Abstract*)

- Bozelli JC Jr, Hou YH, Epand RM. **Thermodynamics of methyl-ß-cyclodextrin-induced lipid vesicle solubilization: Effect of lipid headgroup and backbone.** Langmuir. 2017 Dec 5;33(48):13882-13891. doi: 10.1021/acs.langmuir.7b03447. Epub 2017 Nov 21. (*PubMed Abstract)*

- Abe M, Sawada Y, Uno S, Chigasaki S, Oku M, Sakai Y, Miyoshi H. **Role of acyl chain composition of phosphatidylcholine in *tafazzin*-mediated remodeling of cardiolipin in liposomes.** Biochemistry. 2017 Nov 28;56(47):6268-6280. doi: 10.1021/acs.biochem.7b00941. Epub 2017 Nov 9. (*PubMed Abstract)*

- Ikon N, Hsu FF, Shearer J, Forte TM, Ryan RO. **Evaluation of cardiolipin nanodisks as lipidreplacement therapy for Barth syndrome.** J Biomed Res. 2017 Nov 1. doi: 10.7555/JBR.32.20170094. [Epub ahead of print] *(PubMed - Open Access)**

- Seneviratne AK, Aristizabal Henao JJ, Xu GW, Hurren R, Kim S, MacLean N, Wang X, Xu M, Fajado V, Hao Z, Gronda M, Jeyaraju DV, Chouinard-Watkins R, Schafer C, LeBlanc P, Khuchua Z, Bazinet R, Claypool S, Stark K, Schimmer A. ***Tafazzin* (*TAZ*) regulates the differentiation of AML cells by reducing levels of the phospholipid phosphatidylethanolamine.** Blood 2017 130:788. *(Abstract)*

- Pointer CB, Klegeris A. **Cardiolipin in central nervous system physiology and pathology.** Cell Mol Neurobiol. 2017 Oct;37(7):1161-1172. doi: 10.1007/s10571-016-0458-9. Epub 2016 Dec 30. Review. *(PubMed Abstract)*

- Dudek J. **Role of cardiolipin in mitochondrial signaling pathways.** Front Cell Dev Biol. 2017 Sep 29;5:90. doi: 10.3389/fcell.2017.00090. eCollection 2017. Review. *(PubMed - Open Access)*

- Malhotra K, Modak A, Nangia S, Daman, TH, Gunsel U, Robinson VL, Mokranjac D, May ER, Alder NN. **Cardiolipin mediates membrane and channel interactions of the mitochondrial TIM23 protein.** Sci Adv. 2017 Sep 1;3(9):e1700532. doi: 10.1126/sciadv.1700532. eCollection 2017 Sep. *(PubMed - Open Access)*

- Chen Z, Wu Y, Ma YS, Kobayashi Y, Zhao YY, Miura Y, Chiba H, Hui SP. **Profiling of cardiolipins and their hydroperoxides in HepG2 cells by LC/MS.** Anal Bioanal Chem. 2017 Jul 31. doi: 10.1007/s00216-017-0515-3. [Epub ahead of print] *(PubMed Abstract)*

- Ramachandran R. **Mitochondrial dynamics: The dynamin superfamily and execution by collusion.** Semin Cell Dev Biol. 2017 Jul 25. pii: S1084-9521(17)30365-8. doi: 10.1016/j.semcdb.2017.07.039. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jang S, Javadov S. **Association between ROS production, swelling and the respirasome integrity in cardiac mitochondria.** Arch Biochem Biophys. 2017 Jul 20;630:1-8. doi: 10.1016/j.abb.2017.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Boyd KJ, Alder NN, May ER. **Buckling under pressure: Curvature-based lipid segregation and stability modulation in cardiolipin-containing bilayers.** Langmuir. 2017 Jul 11;33(27):6937-6946. doi: 10.1021/acs.langmuir.7b01185. Epub 2017 Jun 28. *(PubMed Abstract)*

- Fajardo VA, Mikhaeil JS, Leveille CF, Saint C, LeBlanc PJ. **Cardiolipin content, linoleic acid composition, and *tafazzin* expression in response to skeletal muscle overload and unload stimuli.** Sci Rep. 2017 May 17;7(1):2060. doi:10.1038/s41598-017-02089-1. *(PubMed - Open Access)*

- de Taffin de Tilques M, Tribouillard-Tanvier D, Tétaud E1, Testet E, di Rago JP1, Lasserre JP. **Overexpression of mitochondrial oxodicarboxylate carrier (ODC1) preserves oxidative phosphorylation in a yeast model of Barth syndrome.** Dis Model Mech. 2017 Apr 1;10(4):439-450. doi: 10.1242/dmm.027540. Epub 2017 Feb 10. *(PubMed – Open Access)*

- Sheeran FL, Pepe S. **Mitochondrial bioenergetics and dysfunction in failing heart.** Adv Exp Med Biol. 2017;982:65-80. doi: 10.1007/978-3-319-55330-6_4. (*PubMed Abstract)*

- Zhang Y, Bharathi SS, Rardin MJ, Lu J, Maringer KV, Sims-Lucas S, Prochownik EV, Gibson BW, Goetzman ES. **SIRT5 binds to cardiolipin and regulates the electron transport chain.** J Biol Chem. 2017 Apr 30. pii: jbc.M117.785022. doi: 10.1074/jbc.M117.785022. [Epub ahead of print] *(PubMed - Open Access)*

- Ikon N, Ryan RO. **Cardiolipin and mitochondrial cristae organization.** Biochim Biophys Acta. 2017 Mar 20. pii: S0005-2736(17)30095-0. doi: 10.1016/j.bbamem.2017.03.013. [Epub ahead of print] Review. (*PubMed Abstract)*

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access)**

- Schlame M, Xu Y, Ren M. **The basis for acyl specificity in the *tafazzin* reaction.** J Biol Chem. 2017 Mar 31;292(13):5499-5506. doi: 10.1074/jbc.M116.769182. Epub 2017 Feb 15. (*PubMed Abstract)*

- de Taffin de Tilques M, Tribouillard-Tanvier D, Tétaud E, Testet E, di Rago JP, Lasserre JP. **Overexpression of mitochondrial oxodicarboxylate carrier (ODC1) preserves oxidative phosphorylation in a yeast model of the Barth syndrome.** Dis Model Mech. 2017 Feb 10. pii: dmm.027540. doi: 10.1242/dmm.027540. [Epub ahead of print] *(PubMed Abstract*)

- Sathappa M, Mitchell W, Coscia A, Boyd K, May E, Szeto HH, Alder NN. **Investigation of the interactions of the SS-31 peptides with cardiolipin variants: A potential therapeutic for Barth syndrome.** Biophysical Journal, Volume 112, Issue 3, Supplement 1, 3 February 2017, Pages 438a. (*ScienceDirect Abstract)*

- Dudek J, Maack C. **Barth syndrome cardiomyopathy.** Cardiovasc Res. 2017 Feb 2. doi: 10.1093/cvr/cvx014. [Epub ahead of print] No abstract available. *(PubMed)*

- Borna NN, Kishita Y, Ishikawa K, Nakada K, Hayashi JI, Tokuzawa Y, Kohda M, Nyuzuki H, Yamashita-Sugahara Y, Nasu T, Takeda A, Murayama K, Ohtake A, Okazaki Y. **A novel mutation in *TAZ* causes mitochondrial respiratory chain disorder without cardiomyopathy.** J Hum Genet. 2017 Jan 26. doi: 10.1038/jhg.2016.165. [Epub ahead of print] *(PubMed Abstract)*

- Kagan VE, Bayır H, Tyurina YY, Bolevich SB, Maguire JJ, Fadeel B, Balasubramanian K. **Elimination of the unnecessary: Intra- and extracellular signaling by anionic phospholipids.** Review Article. Biochemical and Biophysical Research Communications, Volume 482, Issue 3, 15 January 2017, Pages 482-490. *(PubMed Abstract*)*

- Schlame M, Greenberg ML. **Biosynthesis, remodeling and turnover of mitochondrial cardiolipin.** Biochim Biophys Acta. 2017 Jan;1862(1):3-7. doi:10.1016/j.bbalip.2016.08.010. Review. *(PubMed Abstract)*

- Mejia EM, Zinko JC, Hauff KD, Xu FY, Ravandi A, Hatch GM. **Glucose uptake and triacylglycerol synthesis are increased in Barth syndrome lymphoblasts.** Lipids. 2017 Jan 17. doi: 10.1007/s11745-017-4232-7. [Epub ahead of print] *(PubMed Abstract)**

- Ikon N, Ryan RO. **Barth syndrome: Connecting cardiolipin to cardiomyopathy**. Lipids. 2017 Jan 9. doi: 10.1007/s11745-016-4229-7. [Epub ahead of print] *(PubMed Abstract)*

- Shen Z, Li Y, Gasparski AN, Abeliovich H, Greenberg ML. **Cardiolipin regulates mitophagy through the PKC pathway.** J Biol Chem. 2017 Jan 5. pii: jbc.M116.753574. doi:10.1074/jbc.M116.753574. [Epub ahead of print] *(PubMed - Open Access)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Tyurina YY, Lou W, Qu F, Tyurin VA, Mohammadyani D, Liu J, Hüttemann M, Frasso MA, Wipf P, Bayir H, Greenberg ML, Kagan VE. **Lipidomics characterization of biosynthetic and remodeling pathways of cardiolipins in genetically and nutritionally manipulated yeast cells.** ACS Chem Biol. 2016 Dec 16. [Epub ahead of print] *(PubMed Abstract)**

- Raja V, Joshi AS, Li G, Maddipati KR, Greenberg ML. **Loss of cardiolipin leads to perturbation of acetyl-CoA synthesis.** J Biol Chem. 2016 Dec 9. pii: jbc.M116.753624. [Epub ahead of print] *(PubMed – Open Access)**

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)** ▼

- Sinibaldi F, Milazzo L, Howes BD, Piro MC, Fiorucci L, Polticelli F, Ascenzi P, Coletta M, Smulevich G, Santucci R. **The key role played by charge in the interaction of cytochrome c with cardiolipin.** J Biol Inorg Chem. 2016 Nov 9. [Epub ahead of print] *(PubMed Abstract)*

- Kooijman EE, Swim LA, Graber ZT, Tyurina YY, Bayir H, Kagan VE. **Magic angle spinning ^{31}P NMR spectroscopy reveals two essentially identical ionization states for the cardiolipin phosphates in phospholipid liposomes.** Biochim Biophys Acta. 2016 Oct 26. pii: S0005-2736(16)30346-7. doi:10.1016/j.bbamem.2016.10.013. [Epub ahead of print] *(PubMed Abstract)**

- Oláhová M, Thompson K, Hardy SA, Barbosa IA, Besse A, Anagnostou ME, White K, Davey T, Simpson MA, Champion M, Enns G, Schelley S, Lightowlers RN, Chrzanowska-Lightowlers ZM, McFarland R, Deshpande C, Bonnen PE, Taylor RW. **Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria.** J Inherit Metab Dis. 2016 Sep 30. *(PubMed Abstract)*

- Pérez-Serra A, Toro R, Sarquella-Brugada G, de Gonzalo-Calvo D, Cesar S, Carro E, Llorente-Cortes V, Iglesias A, Brugada J, Brugada R, Campuzano O. **Genetic basis of dilated cardiomyopathy.** International Journal of Cardiology, Available online 21 September 2016. *(ScienceDirect Abstract)*

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review *(PubMed Abstract)*

- Rampelt H, Zerbes RM, van der Laan M, Pfanner N. **Role of the mitochondrial contact site and cristae organizing system in membrane architecture and dynamics.** Biochim Biophys Acta. 2016 Sep 7. pii: S0167-4889(16)30221-X. doi: 10.1016/j.bbamcr.2016.05.020. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jang S, Lewis TS, Powers C, Khuchua Z, Baines CP, Wipf P, Javadov S. **Elucidating mitochondrial ETC supercomplexes in the heart during ischemia-reperfusion.** Antioxid Redox Signal. 2016 Sep 7. [Epub ahead of print] *(PubMed Abstract)*

- Sparvero LJ, Amoscato AA, Fink AB, Anthonymuthu T, New LE, Kochanek PM, Watkins S, Kagan VE, Bayır H. **Imaging mass spectrometry reveals loss of polyunsaturated cardiolipins in the cortical contusion, hippocampus and thalamus after traumatic brain injury.** J Neurochem. 2016 Sep 3. doi: 10.1111/jnc.13840. [Epub ahead of print] *(PubMed Abstract)*

- Schlame M, Greenberg ML. **Biosynthesis, remodeling and turnover of mitochondrial cardiolipin.** Review Article. Biochim Biophys Acta. 2016 Aug 21. pii: S1388-1981(16)30230-X. doi: 10.1016/j.bbalip.2016.08.010. [Epub ahead of print] Review. *(PubMed Abstract)*

- Tatsuta T, Langer T. **Intramitochondrial phospholipid trafficking.** Biochim Biophys Acta. 2016 Aug 16. pii: S1388-1981(16)30226-8. doi: 10.1016/j.bbalip.2016.08.006. [Epub ahead of print] Review. *(PubMed Abstract)*

- Maguire JJ, Tyurina YY, Mohammadyani D, Kapralov AA, Anthonymuthu TS, Qu F, Amoscato AA, Sparvero LJ, Tyurin VA, Planas-Iglesias J, He RR, Klein-Seetharaman J, Bayır H, Kagan VE. **Known unknowns of cardiolipin signaling: The best is yet to come.** Biochim Biophys Acta. 2016 Aug 4. pii: S1388-1981(16)30221-9. doi: 10.1016/j.bbalip.2016.08.001. [Epub ahead of print] *(PubMed Abstract)**

- Barbot M, Meinecke M. **Reconstitutions of mitochondrial inner membrane remodeling.** J Struct Biol. 2016 Jul 25. pii: S1047-8477(16)30155-1. doi: 10.1016/j.jsb.2016.07.014. [Epub ahead of print] *(PubMed Abstract)*

- Mehdipoura AR, Hummer G. **Cardiolipin puts the seal on ATP synthase.** Proc Natl Acad Sci U S A. 2016 Aug 2;113(31):8568-70. doi: 10.1073/pnas.1609806113. Epub 2016 Jul 20. *(PubMed - No abstract available)*

- Duncan AL, Robinson AJ, Walker JE. **Cardiolipin binds selectively but transiently to conserved lysine residues in the rotor of metazoan ATP synthases.** Proc Natl Acad Sci U S A. 2016 Aug 2;113(31):8687-92. doi: 10.1073/pnas.1608396113. Epub 2016 Jul 5. *(PubMed - Open Access)*

- Aryal B, Rao VA. **Deficiency in cardiolipin reduces doxorubicin-induced oxidative stress and mitochondrial damage in human B-lymphocytes.** PLoS One. 2016 Jul 19;11(7):e0158376. doi: 10.1371/journal.pone.0158376. eCollection 2016. *(PubMed - Open Access)*▼

BSF does not endorse any drugs, tests, or treatments that we may report.

- Xu Y, Phoon CK, Berno B, D'Souza K, Hoedt E, Zhang G, Neubert TA, Epand RM, Ren M, Schlame M. **Loss of protein association causes cardiolipin degradation in Barth syndrome.** Nat Chem Biol. 2016 Jun 27. doi:10.1038/nchembio.2113. [Epub ahead of print] *(PubMed Abstract)*▼

- Abe M, Hasegawa Y, Oku M, Sawada Y, Tanaka E, Sakai Y, Miyoshi H. **Mechanism for remodeling of the acyl chain composition of cardiolipin catalyzed by saccharomyces cerevisiae *tafazzin*.** J Biol Chem June 6, 2016. doi: 10.1074/jbc.M116.718510. *(PubMed - Open Access)*

- Corcelli A, Schlame M**. Cardiolipin as key lipid of mitochondria in health and disease. 2nd Edition, Florence, Italy, September 30-October 1, 2015.** Chem Phys Lipids. 2016 Apr 27. pii: S0009-3084(16)30048-2. *(PubMed Abstract)**

- Bradley RM, Stark KD, Duncan RE. **Influence of tissue, diet, and enzymatic remodeling on cardiolipin fatty acyl profile.** Mol Nutr Food Res. 2016 Apr 8. doi: 10.1002/mnfr.201500966. [Epub ahead of print] *(PubMed Abstract)*

- Sandlers Y, Mercier K, Pathmasiri W, Carlson J, McRitchie S, Sumner S, Vernon HJ. **Metabolomics reveals new mechanisms for pathogenesis in Barth syndrome and introduces novel roles for cardiolipin in cellular function.** PLoS One. 2016 Mar 25;11(3):e0151802. doi: 10.1371/journal.pone.0151802. eCollection 2016. *(PubMed – Open Access)*▼

- Sathappa M, Alder NN. **The ionization properties of cardiolipin and its variants in model bilayers.** Biochim Biophys Acta. 2016 Mar 7. pii: S0005-2736(16)30084-0. doi: 10.1016/j.bbamem.2016.03.007. [Epub ahead of print] *(PubMed Abstract)**

- Dolinsky VW, Cole LK, Sparagna GC, Hatch GM. **Cardiac mitochondrial energy metabolism in heart failure: Role of cardiolipin and sirtuins.** Biochimica et Biophysica Acta (BBA) - Molecular and Cell Biology of Lipids, Available online 10 March 2016. *(ScienceDirect Abstract)*

- Joshi AS, Fei N, Greenberg ML. **Get1p and Get2p are required for maintenance of mitochondrial morphology and normal cardiolipin levels.** FEMS Yeast Res. 2016 Feb 28. pii: fow019. [Epub ahead of print] *(PubMed Abstract)**

- Lu YW, Galbraith L, Herndon JD, Lu YL, Pras-Raves M, Vervaart M, van Kampen A, Luyf A, Koehler CM, McCaffery JM, Gottlieb E, Vaz FM, Claypool SM. **Defining functional classes of Barth syndrome mutation in humans.** Hum Mol Genet. 2016 Feb 16. pii: ddw046. [Epub ahead of print] *(PubMed Abstract)**

- Peyta L, Jarnouen K, Pinault M, Guimaraes C, de Barros JP, Chevalier S, Dumas JF, Maillot F, Hatch GM, Loyer P, Servais S. **Reduced cardiolipin content decreases respiratory chain capacities and increases ATP synthesis yield in the human HepaRG cells.** Biochim Biophys Acta. 2016 Jan 6. pii: S0005-2728(16)00003-7. doi: 10.1016/j.bbabio.2016.01.002. [Epub ahead of print] *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Saric A, Andreau K, Armand AS, Møller IM, Petit PX. **Barth syndrome: From mitochondrial dysfunctions associated with aberrant production of reactive oxygen species to pluripotent stem cell studies.** Front Genet. 2016 Jan 20;6:359. doi: 10.3389/fgene.2015.00359. eCollection 2015. Review. *(Pub Med – Open Access)*

- Dudek J, Cheng IF, Chowdhury A, Wozny K, Balleininger M, Reinhold R, Grunau S, Callegari S, Toischer K, Wanders RJ, Hasenfuß G, Brügger B, Guan K, Rehling P. **Cardiac-specific succinate dehydrogenase deficiency in Barth syndrome.** EMBO Mol Med. 2015 Dec 23. pii: e201505644. doi: 10.15252/emmm.201505644. [Epub ahead of print] *(PubMed – Open Access)*▼

- Wang M, Wang C, Han RH, Han X. **Novel advances in shotgun lipidomics for biology and medicine.** Review Article. Prog Lipid Res. 2015 Dec 15. pii: S0163-7827(15)30011-4. doi: 10.1016/j.plipres.2015.12.002. [Epub ahead of print] *(PubMed Abstract)*

- Ruiz-Ramírez A, Barrios-Maya MA, López-Acosta O, Molina-Ortiz D, El-Hafidi M. **Cytochrome c release from rat liver mitochondria is compromised by increased saturated cardiolipin species induced by sucrose feeding.** Am J Physiol Endocrinol Metab. 2015 Nov 1;309(9):E777-86. doi: 10.1152/ajpendo.00617.2014. Epub 2015 Sep 8. *(PubMed Abstract)*

- Denning C, Borgdorff V, Crutchley J, Firth KSA, George V, Kalra S, Kondrashov A, Hoang MD, Mosqueira D, Patel A, Prodanov L, Rajamohan D, Skarnes WC, Smith JGW, Young LE. **Cardiomyocytes from human pluripotent stem cells: From laboratory curiosity to industrial biomedical platform.** Original Research Article. Biochimica et Biophysica Acta (BBA) - Molecular Cell Research, Available online 31 October 2015. *(ScienceDirect Abstract)*

- Pokorná L, Cermáková P, Horváth A, Baile MG, Claypool SM, Griac P, Malínský J, Balážová M. **Specific degradation of phosphatidylglycerol is necessary for proper mitochondrial morphology and function.** Biochim Biophys Acta. 2015 Oct 19;1857(1):34-45. doi: 10.1016/j.bbabio.2015.10.004. [Epub ahead of print]. *(PubMed Abstract)*

- Gaspard GJ, McMaster CR. **Cardiolipin metabolism and its causal role in the etiology of the inherited cardiomyopathy Barth syndrome.** Chem Phys Lipids. 2015 Sep 25. pii: S0009-3084(15)30054-2. doi:10.1016/j.chemphyslip.2015.09.005. [Epub ahead of print] Review. *(PubMed Abstract)**

- Shen Z, Ye C, McCain K, Greenberg ML. **The role of cardiolipin in cardiovascular health.** Biomed Res Int. 2015;2015:891707. doi: 10.1155/2015/891707. Epub 2015 Aug 2. Review. *(PubMed – Open Access)**

- Planas-Iglesias J, Dwarakanath H, Mohammadyani D, Yanamala N, Kagan VE, Klein-Seetharaman J. **Cardiolipin interactions with proteins**. Biophys J. 2015 Aug 20. pii: S0006-3495(15)00769-9. doi: 10.1016/j.bpj.2015.07.034. [Epub ahead of print] *(PubMed Abstract)*

- Gifford SM, Meyer P. **Enzyme function is regulated by its localization.** Comput Biol Chem. 2015 Aug 8. pii: S1476-9271(15)30112-2. doi: 10.1016/j.compbiolchem.2015.08.004. [Epub ahead of print] *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Mejia EM, Ibdah JA, Sparagna GC, Hatch GM. **Differential reduction in cardiac and liver monolysocardiolipin acyl transferase-1 and reduction in tetralinoleoyl-cardiolipin in the alpha subunit of trifunctional protein heterozygous knockout mice.** Biochem J. 2015 Aug 6. pii:BJ20150648. [Epub ahead of print] *(PubMed Abstract)*

- Tocchi A, Quarles EK, Basisty N, Gitari L, Rabinovitch PS. **Mitochondrial dysfunction in cardiac aging.** Biochim Biophys Acta. 2015 Jul 17. pii: S0005-2728(15)00152-8. doi: 10.1016/j.bbabio.2015.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Stepanyants N, Macdonald PJ, Francy CA, Mears JA, Qi X, Ramachandran R. **Cardiolipin's propensity for phase transition and its reorganization by dynamin-related protein 1 form a basis for mitochondrial membrane fission.** Mol Biol Cell. 2015 Jul 8. pii: mbc.E15-06-0330. [Epub ahead of print] *(PubMed Abstract)*

- Grevengoed TJ, Martin SA, Katunga L, Cooper DE, Anderson EJ, Murphy RC, Coleman RA. **Acyl-CoA synthetase 1 deficiency alters cardiolipin species and impairs mitochondrial function.** J Lipid Res. 2015 Aug;56(8):1572-82. doi:10.1194/jlr.M059717. Epub 2015 Jul 1. *(PubMed Abstract)*

- Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents *TAZ* knockdown-induced apoptosis in myeloid progenitor cells.** Biochem Biophys Res Commun. 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print] *(PubMed Abstract)**

- Sauerwald J, Jores T, Eisenberg-Bord M, Chuartzman SG, Schuldiner M, Rapaport D. **Genome-wide screens in yeast highlight a role for cardiolipin in biogenesis of mitochondrial outer membrane multispan proteins.** Mol Cell Biol. 2015 Jul 6. pii: MCB.00107-15. [Epub ahead of print] *(PubMed Abstract)*

- Angelini R, Lobasso S, Gorgoglione R, Bowron A, Steward CG, Corcelli A**. Cardiolipin fingerprinting of leukocytes by MALDI-TOF/MS as screening tool for Barth syndrome.** J Lipid Res. 2015 Jul 5. pii:jlr.D059824. [Epub ahead of print] *(PubMed – Open Access)**▼

- Morita SY, Terada T. **Enzymatic measurement of phosphatidylglycerol and cardiolipin in cultured cells and mitochondria.** Sci Rep. 2015 Jun 30;5:11737. doi: 10.1038/srep11737. *(PubMed Abstract)*

- Ryan RO. **Metabolic annotation of 2-ethylhydracrylic acid.** Clin Chim Acta. 2015 Jun 23. pii: S0009-8981(15)00295-8. doi:10.1016/j.cca.2015.06.012. [Epub ahead of print] Review. *(PubMed Abstract)**

- Hijikata A, Yura K, Ohara O, Go M. **Structural and functional analyses of Barth syndrome-causing mutations and alternative splicing in the *tafazzin* acyltransferase domain.** Meta Gene. 2015 Apr 22;4:92-106. doi: 10.1016/j.mgene.2015.04.001. eCollection 2015 Jun. *(PubMed – Open Access)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Casanovas A, Sprenger RR, Tarasov K, Ruckerbauer DE, Hannibal-Bach HK, Zanghellini J, Jensen ON, Ejsing CS. **Quantitative analysis of proteome and lipidome dynamics reveals functional regulation of global lipid metabolism**. Chem Biol. 2015 Mar 19;22(3):412-25. doi:10.1016/j.chembiol.2015.02.007. *(PubMed Abstract)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Borkar M, Bijarnia-Mahay S, Kohli S, Juneja M,Srivastava Y, Saxena R, Verma IC. **Identification of a novel *TAZ* gene mutation in a family with X-linked dilated cardiomyopathy Barth syndrome.** Journal of Inborn Errors of Metabolism & Screening January - December 2015 3: 2326409814567131, first published on February 19, 2015 doi:10.1177/2326409814567131. *(Sage – Full Text)*

- Schenkel LC, Bakovic M. **Formation and regulation of mitochondrial membranes.** Int J Cell Biol. 2014;2014:709828. doi: 10.1155/2014/709828. Epub 2014 Jan 22. Review. *(PubMed Abstract)*

- Lu YW, Claypool SM. **Disorders of phospholipid metabolism: An emerging class of mitochondrial disease due to defects in nuclear genes.** Front Genet. 2015 Feb 3;6:3. doi:10.3389/fgene.2015.00003. eCollection 2015. Review. *(PubMed – Open Access)*

- Gaspard GJ, McMaster CR. **The mitochondrial quality control protein Yme1 is necessary to prevent defective mitophagy in a yeast model of Barth syndrome.** J Biol Chem. 2015 Feb 16. pii:jbc.M115.641878. [Epub ahead of print]* *(PubMed – Open Access)*

- Li XX, Tsoi B, Li YF, Kurihara H, He RR. **Cardiolipin and its different properties in mitophagy and apoptosis.** J Histochem Cytochem. 2015 Feb 11. pii: 0022155415574818. [Epub ahead of print] *(PubMed - No Abstract Available)*

- Neupert W. **A perspective on transport of proteins into mitochondria: A myriad of open questions.** J Mol Biol. 2015 Feb 9. pii: S0022-2836(15)00079-0. doi: 10.1016/j.jmb.2015.02.001. [Epub ahead of print] *(PubMed Abstract)*

- Sathappa M, Schwall CT, Greenwood MR, Baile MG, Claypool SM, Alder NA. **Investigation of the role of the phospholipid cardiolipin in activating respiratory complex activity.** Biophysical Journal, Volume 108, Issue 2, Supplement 1, 27 January 2015, Page 367a. *(ScienceDirect Abstract)*

- Desmurs M, Foti M, Raemy E, Vaz FM, Martinou JC, Bairoch A, Lane L. **C11orf83, a mitochondrial cardiolipin-binding protein involved in bc1 complex assembly and supercomplex stabilization.** Mol Cell Biol. 2015 Jan 20. pii: MCB.01047-14. [Epub ahead of print] *(PubMed Abstract)*

- Xu Y, Malhotra A, Claypool CM, Ren M, Schlame M. ***Tafazzins* from Drosophila and mammalian cells assemble in large protein complexes with a short half-life.** Mitochondrion. 2015 Jan 15. pii: S1567-7249(15)00003-3. doi: 10.1016/j.mito.2015.01.002. [Epub ahead of print] (*PubMed Abstract)*

- Saunders C, Smith L, Wibrand F, Ravn K, Bross P, Thiffault I, Christensen M, Atherton A, Farrow E, Miller N, Kingsmore, Elsebet Ostergaard. **CLPB variants associated with autosomal-recessive mitochondrial disorder with cataract, neutropenia, epilepsy, and methylglutaconic aciduria.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Wortmann SB, Zietkiewicz S, Kousi M, Szklarczyk R, Haack TB, Gersting SW, Muntau AC, Rakovic A, Renkema GH, Rodenburg RJ, Strom TM, Meitinger T, Rubio-Gozalbo ME, Chrusciel E, Distelmaier F, Golzio C, Jansen JH, van Karnebeek C, Lillquist Y, Lücke T, Õunap K, Zordania R, Yaplito-Lee J, van Bokhoven H, Spelbrink JN, Vaz FM, Pras-Raves M, Ploski R, Pronicka E, Klein C, Willemsen MAAP, de Brouwer APM, Prokisch H, Katsanis N, Wevers RA. **CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Rutter J, Hughes AL**. Power2: The power of yeast genetics applied to the powerhouse of the cell.** Trends Endocrinol Metab. 2015 Jan 12. pii: S1043-2760(14)00204-5. doi: 10.1016/j.tem.2014.12.002. [Epub ahead of print] *(PubMed Abstract)*

- Kagan VE, Tyurina YY, Tyurin V, Mohammadyani D, Angeli JP, Baranov SV, Klein-Seetharaman J, Friedlander RM, Mallampalli RK, Conrad M, Bayir H. **Cardiolipin signaling mechanisms: Collapse of asymmetry and oxidation.** Antioxid Redox Signal. 2015 Jan 8. [Epub ahead of print] *(PubMed Abstract)*

- Frohman MA. **Role of mitochondrial lipids in guiding fission and fusion**. J Mol Med (Berl). 2014 Dec 5. [Epub ahead of print] *(PubMed Abstract)*

- Ye C, Shen Z, Greenberg ML. **Cardiolipin remodeling: A regulatory hub for modulating cardiolipin metabolism and function.** J Bioenerg Biomembr. 2016 Apr;48(2):113-23. doi: 10.1007/s10863-014-9591-7. Epub 2014 Nov 29. *(PubMed – Open Access)**

- Chao YJ, Chang WH, Ting HC, Chao WT, Hsu YH. **Cell cycle arrest and cell survival induce reverse trends of cardiolipin remodeling.** PLoS One. 2014 Nov 25;9(11):e113680. doi: 10.1371/journal.pone.0113680. eCollection 2014. *(PubMed Abstract)*

- Martens JC, Keilhoff G, Halangk W, Wartmann T, Gardemann A, Päge I, Schild L. **Lipidomic analysis of molecular cardiolipin species in livers exposed to ischemia/reperfusion.** Mol Cell Biochem. 2014 Nov 23. [Epub ahead of print] *(PubMed Abstract)**

- Garrett TA, Moncada RM. **The Arabidopsis thaliana lysophospholipid acyltransferase At1g78690p acylates a variety of lysophospholipids including bis(monoacylglycero)phosphate.** Biochem Biophys Res Commun. 2014 Oct 3;452(4):1022-7. doi: 10.1016/j.bbrc.2014.09.041. Epub 2014 Sep 18. *(PubMed Abstract)*

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Wortmann SB, Espeel M, Almeida L, Reimer A, Bosboom D, Roels F, de Brouwer AP, Wevers RA. **Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids.** J Inherit Metab Dis. 2014 Sep 2. [Epub ahead of print] *(PubMed Abstract)*

- Debnath S, Addya S. **Mis-sense mutations in *tafazzin (TAZ)* that escort to mild clinical symptoms of Barth syndrome is owed to the minimal inhibitory effect of the mutations on the enzyme function: In-silico evidence.** Interdiscip Sci. 2014 Aug 9. [Epub ahead of print] *(PubMed Abstract)*

- Bowron A, Honeychurch J, Williams M, Tsai-Goodman B, Clayton N, Jones L, Shortland GJ, Qureshi SA, Heales SJ, Steward CG. **Barth syndrome without tetralinoleoyl cardiolipin deficiency: A possible ameliorated phenotype.** J Inherit Metab Dis. 2014 Aug 12. [Epub ahead of print] *(PubMed – Open Access)**▼

- Mayr JA. **Lipid metabolism in mitochondrial membranes.** J Inherit Metab Dis. 2014 Aug 1. [Epub ahead of print] *(PubMed Abstract)*

- Peyta L, Dumas JF, Pinault M, Guimaraes C, Jarnouen K, Lagrave J, Maillot F, Loyer P, Servais S. **Reduction of the mitochondrial content in cardiolipins decreases O2 consumption and increases ATP synthesis efficiency in human hepatocyte-like HepaRG cells.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Volume 1837, Supplement, July 2014, Pages e28–e29. *(ScienceDirect Abstract)*

- Hwang MS, Schwall CT, Pazarentzos E, Datler C, Alder NN, Grimm S. **Mitochondrial Ca^{2+} influx targets cardiolipin to disintegrate respiratory chain complex II for cell death induction.** Cell Death Differ. 2014 Jun 20. doi: 10.1038/cdd.2014.84. [Epub ahead of print] *(PubMed Abstract)*

- Feillet-Coudray C, Fouret G, Casas F, Coudray C. **Impact of high dietary lipid intake and related metabolic disorders on the abundance and acyl composition of the unique mitochondrial phospholipid, cardiolipin.** J Bioenerg Biomembr. 2014 Jun 21. [Epub ahead of print] *(PubMed Abstract)*

- Chatfield KC, Sparagna GC, Sucharov CC, Miyamoto SD, Grudis JE, Sobus RD, Hijmans J, Stauffer BL. **Dysregulation of cardiolipin biosynthesis in pediatric heart failure**. J Mol Cell Cardiol. 2014 Jun 14. pii: S0022-2828(14)00196-5. doi:10.1016/j.yjmcc.2014.06.002. [Epub ahead of print] *(PubMed Abstract)*

- Deckers M, Balleininger M, Vukotic M, Römpler K, Bareth B, Juris L, Dudek J. **Aim24 stabilizes respiratory chain supercomplexes and is required for efficient respiration.** FEBS Lett. 2014 Aug 25;588(17):2985-92. doi: 10.1016/j.febslet.2014.06.006. Epub 2014 Jun 10. *(PubMed Abstract)*

- Zeczycki T, Whelan J, Hayden WT, Brown DA, Shaikh SR. **Increasing levels of cardiolipin differentially influence packing of phospholipids found in the mitochondrial inner membrane.** Biochem Biophys Res Commun. 2014 Jun 3. pii: S0006-291X(14)01022-5. doi:10.1016/j.bbrc.2014.05.133. [Epub ahead of print] *(PubMed Abstract)*

- Raval KK, Kamp TJ. **Cardiomyopathy, mitochondria and Barth syndrome: iPSCs reveal a connection.** Nat Med. 2014 Jun 5;20(6):585-6. doi: 10.1038/nm.3592. *(PubMed – No Abstract Available).*

- Pathak S, Meng WJ, Zhang H, Gnosa S, Nandy SK, Adell G, Holmlund B, Sun XF. ***Tafazzin* protein expression is associated with tumorigenesis and radiation response in rectal cancer: A study of Swedish clinical trial on preoperative radiotherapy.** PLoS One. 2014 May 23;9(5):e98317. doi: 10.1371/journal.pone.0098317. eCollection 2014. *(PubMed Abstract)*

- Richter-Dennerlein R, Korwitz A, Haag M, Tatsuta T, Dargazanli S, Baker M, Decker T, Lamkemeyer T, Rugarli EI, Langer T. **DNAJC19, a mitochondrial cochaperone associated with cardiomyopathy, forms a complex with prohibitins to regulate cardiolipin remodeling.** Cell Metab. 2014 May 21. pii: S1550-4131(14)00182-X. doi: 10.1016/j.cmet.2014.04.016. [Epub ahead of print] *(PubMed Abstract)*

- Epand RM, D'Souza K, Berno B, Schlame M. **Membrane curvature modulation of protein activity determined by NMR.** Biochim Biophys Acta. 2015 Jan;1848(1PB):220-228. doi: 10.1016/j.bbamem.2014.05.004. Epub 2014 May 13. Review. *(PubMed Abstract)*

- Raja V, Greenberg ML. **The functions of cardiolipin in cellular metabolism-potential modifiers of the Barth syndrome phenotype.** Chem Phys Lipids. 2014 Apr;179:49-56. doi: 10.1016/j.chemphyslip.2013.12.009. Epub 2014 Jan 17. *(PubMed – Open Access)**

- Wang G, McCain ML, Yang L, He A, Pasqualini FS, Agarwal A, Yuan H, Jiang D, Zhang D, Zangi L, Geva J, Roberts AE, Ma Q, Ding J, Chen J, Wang DZ, Li K, Wang J, Wanders RJA, Kulik W, Vaz FM, Laflamme MA, Murry CE, Chien KR, Kelley RI, Church GM, Parker K, Pu WT. **Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies.** Nat Med. 2014 Jun;20(6):616-23. doi:10.1038/nm.3545 Epub. 2014 May 11. *(PubMed – Open Access)*▼

- Mejia EM, Cole L, Hatch GM. **Cardiolipin metabolism and the role it plays in heart failure and mitochondrial supercomplex formation.** Cardiovasc Hematol Disord Drug Targets. 2014 May 5. [Epub ahead of print] *(PubMed Abstract)*

- Ren M, Phoon CKL, Schlame M. **Metabolism and function of mitochondrial cardiolipin.** Prog Lipid Res. 2014 Jul;55:1-16. doi: 10.1016/j.plipres.2014.04.001. Epub 2014 Apr 24. *(PubMed Abstract)**

- Birk AV, Chao WM, Bracken C, Warren JD, Szeto HH. **Targeting mitochondrial cardiolipin and the cytochrome c/cardiolipin complex to promote electron transport and optimize mitochondrial ATP synthesis.** Br J Pharmacol. 2014 Apr;171(8):2017-28. doi: 10.1111/bph.12468. *(PubMed Abstract)*

- Szeto HH. **First-in-class cardiolipin-protective compound as a therapeutic agent to restore mitochondrial bioenergetics.** Br J Pharmacol. 2014 Apr;171(8):2029-50. doi: 10.1111/bph.12461. Review. *(PubMed Abstract)*

- Harner ME, Unger AK, Izawa T, Walther DM, Ozbalci C, Geimer S, Reggiori F, Brügger B, Mann M, Westermann B, Neupert W. **Aim24 and MICOS modulate respiratory function, *tafazzin*-related cardiolipin modification and mitochondrial architecture.** Elife. 2014 Jan 1;3:e01684. doi:10.7554/eLife.01684. *(PubMed Abstract)*

- Chen BB, Coon TA, Glasser JR, Zou C, Ellis B, Das T, McKelvey AC, Rajbhandari S, Lear T, Kamga C, Shiva S, Li C, Pilewski JM, Callio J, Chu CT, Ray A, Ray P, Tyurina YY, Kagan VE, Mallampalli RK. **E3 Ligase subunit Fbxo15 and PINK1 kinase regulate cardiolipin synthase 1 stability and mitochondrial function in pneumonia.** Cell Rep. 2014 Apr 2. pii:S2211-1247(14)00164-8. doi: 10.1016/j.celrep.2014.02.048. [Epub ahead of print] *(PubMed Abstract)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Koob S, Reichert AS. **Novel intracellular functions of apolipoproteins: The ApoO protein family as constituents of the Mitofilin/MINOS complex determines cristae morphology in mitochondria.** Biol Chem. 2014 Mar 1;395(3):285-96. doi: 10.1515/hsz-2013-0274. *(PubMed Abstract)*

- Su B, Ryan RO. **Metabolic biology of 3-methylglutaconic acid-uria: A new perspective.** J Inherit Metab Dis. 2014 May;37(3):359-68. doi: 10.1007/s10545-013-9669-0. Epub 2014 Jan 10. *(PubMed Abstract)**

- Leung CW, Hong Y, Hanske J, Zhao E, Chen S, Pletneva EV, Tang BZ. **Superior fluorescent probe for detection of cardiolipin.** Anal Chem. 2014 Jan 21;86(2):1263-8. doi: 10.1021/ac403616c. Epub 2013 Dec 27. *(PubMed Abstract)*

- Mulligan CM, Le CH, Demooy AB, Nelson CB, Chicco AJ. **Inhibition of delta-6 desaturase reverses cardiolipin remodeling and prevents contractile dysfunction in the aged mouse heart without altering mitochondrial respiratory function.** J Gerontol A Biol Sci Med Sci. 2014 Jan 18. [Epub ahead of print] *(PubMed Abstract)*

- Raja V, Greenberg ML. **The functions of cardiolipin in cellular metabolism–potential modifiers of the Barth syndrome phenotype.** Chem Phys Lipids. 2014 Jan 17. pii:S0009-3084(13)00181-3. doi: 10.1016/j.chemphyslip.2013.12.009. [Epub ahead of print] *(PubMed – Open Access)**

- Kagan V, Epand R. **Deciphering the mysteries of cardiolipins in mitochondria.** Chem Phys Lipids. 2013 Dec 21. pii: S0009-3084(13)00169-2. [Epub ahead of print] *(PubMed – No Abstract Available)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Lai L, Wang M, Martin OJ, Leone TC, Vega RB, Han X, Kelly DP. **A role for peroxisome proliferator-activated receptor γ coactivator 1 (PGC-1) in the regulation of cardiac mitochondrial phospholipid biosynthesis.** J Biol Chem. 2014 Jan 24;289(4):2250-9. doi:10.1074/jbc.M113.523654. Epub 2013 Dec 11. *(PubMed – Open Access)*

- Schlattner U, Tokarska-Schlattner M, Rousseau D, Boissan M, Mannella C, Epand R, Lacombe ML. **Mitochondrial cardiolipin/phospholipid trafficking: The role of membrane contact site complexes and lipid transfer proteins**. Chem Phys Lipids. 2013 Dec 6. Chem Phys Lipids. 2013 Dec 6. [Epub ahead of print] *(PubMed Abstract)*

- Kagan VE, Chu CT, Tyurina YY, Cheikhi A, Bayir H. **Cardiolipin asymmetry, oxidation and signaling.** Chem Phys Lipids. 2013 Dec 1.[Epub ahead of print] *(PubMed – Open Access)*

- Ye C, Lou W, Li Y, Chatzispyrou IA, Huttemann M, Lee I, Houtkooper RH, Vaz FM, Chen S, Greenberg ML. **Deletion of the cardiolipin-specific phospholipase Cld1 rescues growth and lifespan defects in the *tafazzin* mutant: Implications for Barth syndrome.** J Biol Chem. 2013 Dec 8. [Epub ahead of print] *(PubMed – Open Access)**

- Baile MG, Sathappa M, Lu YW, Pryce E, Whited K, McCaffery JM, Han X, Alder NN, Claypool SM. **Unremodeled and remodeled cardiolipin are functionally indistinguishable in yeast.** J Biol Chem. 2013 Nov 27. [Epub ahead of print] *(PubMed – Open Access)*

- Mileykovskaya E, Dowhan W. **Cardiolipin-dependent formation of mitochondrial respiratory supercomplexes.** Chem Phys Lipids. 2013 Nov 9. [Epub ahead of print] *(PubMed Abstract)*

- Thomas E, Roman E, Claypool S, Manzoor N, Pla J, Panwar SL. **Mitochondria influence CDR1 efflux pump activity, Hog1-mediated oxidative stress pathway, iron homeostasis, and ergosterol levels in candida albicans.** Antimicrob. Agents Chemother. 2013, 57(11):5580. *(PubMed Abstract)*

- He Q, Han X. **Cardiolipin remodeling in diabetic heart.** Chem Phys Lipids. 2013 Nov 1. doi:pii: S0009-3084(13)00135-7. [Epub ahead of print] *(PubMed Abstract)*

- Baile MG, Lu WY, Claypool SM. **The topology and regulation of cardiolipin biosynthesis and remodeling in yeast.** Chem Phys Lipids. 2014 Apr;179:25-31. doi: 10.1016/j.chemphyslip.2013.10.008. Epub 2013 Nov 1. *(PubMed Abstract)*

- Xu Y, Schlame M. **The turnover of glycerol and acyl moieties of cardiolipin.** Chem Phys Lipids. 2013 Nov 1. [Epub ahead of print]. *(PubMed Abstract)**

- Finsterer J, Stöllberger C. **Unclassified cardiomyopathies in neuromuscular disorders.** Wien Med Wochenschr. 2013 Oct 24. [Epub ahead of print] *(PubMed Abstract)*

- Mejia EM, Nguyen H, Hatch GM. **Mammalian cardiolipin biosynthesis.** Chem Phys Lipids. 2013 Oct 18 [Epub ahead of print] *(PubMed Abstract)**

- D'Souza K, Epand RM. **Enrichment of phosphatidylinositols with specific acyl chains.** Biochim Biophys Acta. 2013 Oct 10. [Epub ahead of print] *(PubMed Abstract)*

- Herndon JD, Claypool SM, Koehler CM. **The *Taz*1p transacylase is imported and sorted into the outer mitochondrial membrane via a membrane anchor domain.** Eukaryot Cell. 2013 Sep 27. [Epub ahead of print] (*PubMed – Open Access)*

- Corcelli A, Schlame M. **Cardiolipin as key lipid of mitochondria in health and disease.** Meeting Report – September 17, 2013, Bari, Italy. Eur. J. Lipid Sci. Technol. 2013, 115, 0000–0000.*

- Horvath SE, Daum G. **Lipids of mitochondria.** Progress in Lipid Research, Available online 2 September 2013. *(Abstract)*

- He Q, Wang M, Harris N, Han X. ***Tafazzin* knockdown interrupts cell cycle progression in cultured neonatal ventricular fibroblasts.** Am J Physiol Heart Circ Physiol. 2013 Aug 30. [Epub ahead of print] *(PubMed – Open Access)**▼

- Unsay J, Cosentino K, Subburaj Y, Garcia-Saez A. **Cardiolipin effects on membrane structure and dynamics.** Langmuir. 2013 Aug 20. [Epub ahead of print] (*PubMed Abstract)*

- Birk AV, Liu S, Soong Y, Mills W, Singh P, Warren JD, Seshan SV, Pardee JD, Szeto HH. **The mitochondrial-targeted compound SS-31 re-energizes ischemic mitochondria by interacting with cardiolipin.** J Am Soc Nephrol. 2013 Jun 27. [Epub ahead of print] *(PubMed Abstract)*

- Brown DA, Sabbah HN, Shaikh SR. **Mitochondrial inner membrane lipids and proteins as targets for decreasing cardiac ischemia/reperfusion injury.** Pharmacol Ther. 2013 Dec;140(3):258-66. doi: 10.1016/j.pharmthera.2013.07.005. Epub 2013 Jul 15. *(PubMed Abstract)*

- Jefferies JL. **Barth syndrome.** Am J Med Genet C Semin Med Genet. 2013 Jul 10. doi: 10.1002/ajmg.c.31372. [Epub ahead of print] *(PubMed – Open Access)*▼

- Monteiro JP, Oliveira PJ, Jurado AS. **Mitochondrial membrane lipid remodeling in pathophysiology: A new target for diet and therapeutic interventions.** Prog Lipid Res. 2013 Jul 1. doi:pii: S0163-7827(13)00042-8. 10.1016/j.plipres.2013.06.002. [Epub ahead of print] (*PubMed Abstract)*

- Dudek J, Cheng IF, Balleininger M, Vaz FM, Streckfuss-Bömeke K, Hübscher D, Vukotic M, Wanders RJ, Rehling P, Guan K. **Cardiolipin deficiency affects respiratory chain function and organization in an induced pluripotent stem cell model of Barth syndrome.** Stem Cell Res. 2013 Sep;11(2):806-19. doi: 10.1016/j.scr.2013.05.005. Epub 2013 May 28. *(PubMed – Open Access)*▼

- Patil VA, Greenberg ML. **Cardiolipin-mediated cellular signaling.** Adv Exp Med Biol. 2013;991:195-213. doi: 10.1007/978-94-007-6331-9_11. (*PubMed Abstract)**

- Kim GB, Kwon BS, Bae EJ, Noh CI, Seong MW, Park SS. **A novel mutation of the *TAZ* gene in Barth syndrome: Acute exacerbation after contrast-dye injection.** J Korean Med Sci. 2013 May;28(5):784-7. doi: 10.3346/jkms.2013.28.5.784. Epub 2013 May 2. (*PubMed – Open Access)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Baile MG, Whited K, Claypool SM. **Deacylation on the matrix side of the mitochondrial inner membrane regulates cardiolipin remodeling.** Mol Biol Cell. 2013 May 1. [Epub ahead of print] *(PubMed – Open Access)*

- Grudis JE, Chatfield KC, Sparagna GC, Hijmans J, Sobus RD, Miyamoto SD, Stauffer BL. **Expression of cardiolipin biosynthesis and remodeling enzymes in adult heart failure.** J Heart and Lung Transplantation, April 2013, Vol. 32, Iss 4, Pgs. S97-S98. *(ScienceDirect Abstract)*

- Becker T, Horvath SE, Bottinger L, Gebert N, Daum G, Pfanner N. **Role of phosphatidylethanolamine in the biogenesis of mitochondrial outer membrane proteins.** J Biol Chem. 2013 Apr 26. [Epub ahead of print] *(PubMed – Open-Access)*

- Powers C, Huang Y, Strauss A, Khuchua Z. **Diminished exercise capacity and mitochondrial bc1 complex deficiency in *tafazzin*-knockdown mice.** Front Physiol. 2013 Apr 17;4:74. doi: 10.3389/fphys.2013.00074. Print 2013. *(PubMed – Open Access)*

- Luévano-Martínez LA, Appolinario P, Miyamoto S, Uribe-Carvajal S, Kowaltowski AJ. **Deletion of the transcriptional regulator Opi1p decreases cardiolipin content and disrupts mitochondrial metabolism in saccharomyces cerevisiae.** Fungal Genetics and Biology, Available online 8 April 2013. *(PubMed Abstract)*

- Hsu YH, Dumlao DS, Cao J, Dennis EA. **Assessing phospholipase A2 Activity toward cardiolipin by mass spectrometry.** PLoS One. 2013;8(3):e59267. doi: 10.1371/journal.pone.0059267. Epub 2013 Mar 22. *(PubMed – Open Access)*

- Gonzalvez F, D'Aurelio M, Boutant M, Moustapha A, Puech JP, Landes T, Arnauré L, Vial G, Talleux N, Slomianny C, Wanders RJA, Houtkooper RH, Belenger P, Moller IM, Gottlieb E, Vaz FM, Manfredi G, Petit PX. **Barth syndrome: Cellular compensation of mitochondrial dysfunction and apoptosis inhibition due to changes in cardiolipin remodeling linked to *Tafazzin* gene mutation.** Biochim Biophys Acta. 2013 Aug;1832(8):1194-206. doi: 10.1016/j.bbadis.2013.03.005. Epub 2013 Mar 20. *(PubMed – Open Access)*

- Perhirin A, Kraffe E, Marty Y, Quentel F, Elies P, Gloaguen F. **Electrochemistry of cytochrome c immobilized on cardiolipin-modified electrodes: A probe for protein - lipid interactions.** Biochim Biophys Acta. 2013 Mar;1830(3):2798-803. *(PubMed Abstract)*

- Arnarez C, Marrink SJ, Periole X. **ERRATUM: Identification of cardiolipin binding sites on cytochrome c oxidase at the entrance of proton channels.** Sci Rep. 2013 Feb 22;3:1343. doi: 10.1038/srep01343. *(PubMed - No abstract available)*

- Jalmar O, François-Moutal L, García-Sáez AJ, Perry M, Granjon T, Gonzalvez F, Gottlieb E, Ayala-Sanmartin J, Klösgen B, Schwille P, Petit PX. **Caspase-8 binding to cardiolipin in giant unilamellar vesicles provides a functional docking platform for Bid.** PLoS One. 2013;8(2):e55250. Epub 2013 Feb 13. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini RR, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 14;8(1):27. doi: 10.1186/1750-1172-8-27. *(PubMed Abstract)*▼

- Ostojic O, O'Leary MF, Singh K, Menzies KJ, Vainshtein A, Hood DA. **The effects of chronic muscle use and disuse on cardiolipin metabolism.** J Appl Physiol. 2013 Feb 15;114(4):444-52. doi: 10.1152/japplphysiol.01312.2012. Epub 2012 Dec 6. *(PubMed Abstract)*

- Arnarez C, Marrink SJ, Periole X. **Identification of cardiolipin binding sites on cytochrome c oxidase at the entrance of proton channels.** Sci Rep. 2013;3:1263. doi: 10.1038/srep01263. Epub 2013 Feb 12. *(PubMed Abstract)*

- Arnarez C, Mazat JP, Elezgaray J, Marrink SJ, Periole X. **Evidence for cardiolipin binding sites on the membrane-exposed surface of the cytochrome bc1.** J Am Chem Soc. 2013 Jan 30. [Epub ahead of print] *(PubMed Abstract)*

- Agassandian M, Mallampalli RK. **Surfactant phospholipid metabolism.** Biochim Biophys Acta. 2013 Mar;1831(3):612-25. doi: 10.1016/j.bbalip.2012.09.010. Epub 2012 Sep 29. Review. *(PubMed – Open Access)*

- Wortmann SB, Duran M, Anikster Y, Barth PG, Sperl W, Zschocke J, Morava E, Wevers RA. **Inborn errors of metabolism with 3-methylglutaconic aciduria as discriminative feature: Proper classification and nomenclature**. J Inherit Metab Dis. 2013 Jan 8. [Epub ahead of print] *(PubMed Abstract)*

- Patil V, Fox JL, Gohil VM, Winge DR, Greenberg ML. **Loss of cardiolipin leads to perturbation of mitochondrial and cellular iron homeostasis.** J Biol Chem. 2013 Jan 18;288(3):1696-705. doi: 10.1074/jbc.M112.428938. Epub 2012 Nov 28. *(PubMed – Open Access)**

- Kim J, Hoppel CL. **Comprehensive approach to the quantitative analysis of mitochondrial phospholipids by HPLC-MS.** J Chromatogr B Analyt Technol Biomed Life Sci. 2012 Nov 5;912C:105-114. doi: 10.1016/j.jchromb.2012.10.036. [Epub ahead of print] *(PubMed Abstract)*

- Boxer LA. **How to approach neutropenia.** Hematology Am Soc Hematol Educ Program. 2012;2012:174-82. doi: 10.1182/asheducation-2012.1.174. *(PubMed Abstract)*

- Cortie CH, Else PL. **Dietary docosahexaenoic Acid (22:6) incorporates into cardiolipin at the expense of linoleic Acid (18:2): Analysis and potential implications.** Int J Mol Sci. 2012 Nov 21;13(11):15447-63. doi: 10.3390/ijms131115447. *(PubMed Abstract)*

- Schlame M. **Cardiolipin remodeling and the function of *tafazzin*.** Biochim Biophys Acta. 2012 Nov 28. doi:pii: S1388-1981(12)00249-1. *(PubMed Abstract)*

- Taylor WA, Mejia EM, Mitchell RW, Choy PC, Sparagna GC, et al. **Human trifunctional protein *alpha* links cardiolipin remodeling to beta-oxidation.** PLoS. 2012;7(11):e48628. doi: 10.1371/journal.pone.0048628. Epub 2012 Nov 9. *(PubMed – Open Access)* *▼

- Gawrisch K. **Lipids: *Tafazzin* senses curvature.** Nat Chem Biol. 2012 Oct;8(10):811-2. *(PubMed Abstract – No Abstract Available.)*

- Bowron A, Frost R, Powers VE, Thomas PH, Heales SJ, Steward CG. **Diagnosis of Barth syndrome using a novel LC-MS/MS method for leukocyte cardiolipin analysis.** J Inherit Metab Dis. 2012 Oct 30. [Epub ahead of print] *(PubMed Abstract)*

- Carman GM. **An unusual phosphatidylethanolamine-utilizing cardiolipin synthase is discovered in bacteria.** Proc Natl Acad Sci U S A. 2012 Sep 25. [Epub ahead of print] No abstract available. *(PubMed Abstract)*

- Tan BK, Bogdanov M, Zhao J, Dowhan W, Raetz CR, Guan Z. **Discovery of a cardiolipin synthase utilizing phosphatidylethanolamine and phosphatidylglycerol as substrates.** Proc Natl Acad Sci U S A. 2012 Sep 17. [Epub ahead of print] *(PubMed Abstract)*

- Ronvelia D, Greenwood J, Platt J, Hakim S, Zaragoza MV. **Intrafamilial variability for novel *TAZ* gene mutation: Barth syndrome with dilated cardiomyopathy and heart failure in an infant and left ventricular noncompaction in his great-uncle.** Mol Genet Metab. 2012 Sep 18. pii: S1096-7192(12)00354-X. doi: 10.1016/j.ymgme.2012.09.013. [Epub ahead of print] *(PubMed Abstract)*

- Böttinger L, Horvath SE, Kleinschroth T, Hunte C, Daum G, Pfanner N, Becker T. **Phosphatidylethanolamine and cardiolipin differentially affect the stability of mitochondrial respiratory chain supercomplexes.** J Mol Biol. 2012 Sep 9. pii: S0022-2836(12)00720-6. doi: 10.1016/j.jmb.2012.09.001. [Epub ahead of print] *(PubMed Abstract)*

- Schlame M, Acehan D, Berno B, Xu Y, Valvo S, Ren M, Stokes DL, Epand RM. **The physical state of lipid substrates provides transacylation specificity for *tafazzin*.** Nat Chem Biol. 2012 Sep 2. doi: 10.1038/nchembio.1064. [Epub ahead of print] *(PubMed – Open Access)**

- Liu X, Ye B, Miller S, Yuan H, Zhang H, Tian L, Nie J, Imae R, Arai H, Li Y, Cheng Z, Shi Y. **Ablation of ALCAT1 mitigates hypertrophic cardiomyopathy through effects on oxidative stress and mitophagy.** Mol Cell Biol. 2012 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Lamari F, Mochel F, Sedel F, Saudubray JM. **Disorders of phospholipids, sphingolipids and fatty acids biosynthesis: Toward a new category of inherited metabolic diseases.** J Inherit Metab Dis. 2012 Jul 20. [Epub ahead of print] *(PubMed Abstract)*

- Zhuravleva E, Gut H, Hynx D, Marcellin D, Bleck CK, Genoud C, Cron P, Keusch JJ, Dummler B, Esposti MD, Hemmings BA. **Acyl coenzyme A thioesterase Them5/Acot15 is involved in cardiolipin remodeling and fatty liver development.** Mol Cell Biol. 2012 Jul;32(14):2685-97. Epub 2012 May 14. *(PubMed Abstract)*

- Kiebish MA, Yang K, Sims HF, Jenkins CM, Liu X, Mancuso DJ, Zhao Z, Guan S, Abendschein DR, Han X, Gross RW. **Myocardial regulation of lipidomic flux by cardiolipin synthase: Setting the beat for bioenergetic efficiency.** J Biol Chem. 2012 Jul 20;287(30):25086-97. Epub 2012 May 14. *(PubMed – Open Access)**

- Mulligan CM, Sparagna GC, Le CH, De Mooy AB, Routh MA, Holmes MG, Hickson-Bick DL, Zarini S, Murphy RC, Xu FY, Hatch GM, McCune SA, Moore RL, Chicco AJ. **Dietary linoleate preserves cardiolipin and attenuates mitochondrial dysfunction in the failing rat heart.** Cardiovasc Res. 2012 Jun 1;94(3):460-8. Epub 2012 Mar 12. *(PubMed Abstract)*

- Wilson LD, Al-Majid S, Rakovsky C, Schwindt CD. **Higher IL-6 and IL6:IGF ratio in patients with Barth syndrome**. J Inflamm (Lond). 2012 Jun 21;9(1):25. [Epub ahead of print] *(PubMed Abstract)*▼

- Khairallah RJ, Kim J, O'Shea KM, O'Connell KA, Brown BH, Galvao T, Daneault C, Des Rosiers C, Polster BM, Hoppel CL, Stanley WC. **Improved mitochondrial function with diet-induced increase in either docosahexaenoic acid or arachidonic acid in membrane phospholipids.** PLoS One. 2012;7(3):e34402. Epub 2012 Mar 30. *(PubMed Abstract)*

- Samhan-Arias AK, Ji J, Demidova OM, Sparvero LJ, Feng W, Tyurin V, Tyurina YY, Epperly MW, Shvedova AA, Greenberger JS, Bayır H, Kagan VE, Amoscato AA. **Oxidized phospholipids as biomarkers of tissue and cell damage with a focus on cardiolipin.** Biochim Biophys Acta. 2012 Mar 23. [Epub ahead of print] *(PubMed Abstract)*

- Schild L, Lendeckel U, Gardemann A, Wiswedel I, Schmidt CA, Wolke C, Walther R, Grabarczyk P, Busemann C. **Composition of molecular cardiolipin species correlates with proliferation of lymphocytes.** Exp Biol Med (Maywood). 2012 Apr;237(4):372-9. doi: 10.1258/ebm.2011.011311. Epub 2012 Apr 4. *(PubMed Abstract)*

- Schlame M, Blais S, Edelman-Novemsky I, Xu Y, Montecillo F, Phoon CK, Ren M, Neubert TA. **Comparison of cardiolipins from Drosophila strains with mutations in putative remodeling enzymes.** Chem Phys Lipids. 2012 Mar 23. [Epub ahead of print] *(PubMed Abstract)*

- Joshi AS, Thompson MN, Fei N, Hutteman M, Greenberg ML. **Cardiolipin and mitochondrial phosphatidylethanolamine have overlapping functions in mitochondrial fusion in Saccharomyces cerevisiae.** J Biol Chem. 2012 Mar 20. [Epub ahead of print] *(PubMed – Open Access)**

- Tian HF, Feng JM, Wen JF. **The evolution of cardiolipin biosynthesis and maturation pathways and its implications for the evolution of eukaryotes.** BMC Evol Biol. 2012 Mar 13;12(1):32. [Epub ahead of print] *(Pubmed – Open Access)*

- Mulligan CM, Sparagna GC, Le CH, De Mooy AB, Routh MA, Holmes MG, Hickson-Bick DL, Zarini S, Murphy RC, Xu FY, Hatch GM, McCune SA, Moore RL, Chicco AJ. **Dietary linoleate preserves cardiolipin and attenuates mitochondrial dysfunction in the failing rat heart.** Cardiovasc Res. 2012 Mar 12. [Epub ahead of print] *(PubMed Abstract)*

- Greenberg, ML. **Functions of cardiolipin as modifiers of the Barth syndrome phenotype.** ASBMB Today. March 2012. *(ASBMB Abstract)*

- Schug ZT, Frezza C, Galbraith LC, Gottlieb E. **The music of lipids: How lipid composition orchestrates cellular behaviour.** Acta Oncol. 2012 Jan 30. [Epub ahead of print] *(PubMed Abstract)*

- Karo J, Peterson P, Vendelin M. **Molecular dynamics simulations of creatine kinase and adenine nucleotide translocase in mitochondrial membrane patch.** J Biol Chem. 2012 Jan 12. [Epub ahead of print] *(PubMed Abstract)*

- Saini-Chohan HK, Mitchell RW, Vaz FM, Zelinski T, Hatch GM. **Delineating the role of alterations in lipid metabolism to the pathogenesis of inherited skeletal and cardiac muscle disorders.** J Lipid Res. 2011 Nov 7. [Epub ahead of print] *(PubMed – Open Access)**

- Makaryan V, Kulik W, Vaz FM, Allen C, Dror Y, Dale DC, Aprikyan AA**. The cellular and molecular mechanisms for neutropenia in Barth syndrome.** Eur J Haematol. 2011 Oct 24. doi: 10.1111/j.1600-0609.2011.01725.x. [Epub ahead of print] *(PubMed – Open Access)**

- Claypool SM, Koehler CM. **The complexity of cardiolipin in health and disease.** Trends Biochem Sci. 2011 Oct 17. [Epub ahead of print] *(PubMed Abstract)*

- Dashti M, Kulik W, Hoek F, Veerman EC, Peppelenbosch MP, Rezaee F. **A phospholipidomic analysis of all defined human plasma lipoproteins.** Sci. Rep. 1, 139; DOI:10.1038/srep00139 (2011). *(PubMed – Open Access)*

- El-Hafidi M, Meschini MC, Rizza T, Santorelli FM, Bertini E, Carrozzo R, Vázquez-Memije ME. **Cardiolipin content in mitochondria from cultured skin fibroblasts harboring mutations in the mitochondrial ATP6 gene.** J Bioenerg Biomembr. 2011 Oct 13. [Epub ahead of print] *(PubMed Abstract)*

- Wahjudi PN, Yee J, Martinez SR, Zhang J, Teitell M, Nikolaenko L, Swerdloff R, Wang C, Lee WN. **Turn-over of non-essential fatty acid in cardiolipin in rat heart.** J Lipid Res. 2011 Sep 27. [Epub ahead of print] *(PubMed Abstract)*

- Saini-Chohan HK, Dakshinamurti S, Taylor WA, Shen GX, Murphy R, Sparagna GC, Hatch GM. **Persistent pulmonary hypertension results in reduced tetralinoleoyl-cardiolipin and Complex II + III activity in neonatal pig heart.** Am J Physiol Heart Circ Physiol. 2011 Aug 12. [Epub ahead of print] *(PubMed – Open Access)**

- Cavallo A, Gnoni A, Conte E, Siculella L, Zanotti F, Papa S, Gnoni GV**. 3,5-Diiodo-L-Thyronine increases F(o)F (1)-ATP synthase activity and cardiolipin level in liver mitochondria of hypothyroid rats.** J Bioenerg Biomembr. 2011 Jul 8. [Epub ahead of print] *(PubMed Abstract)*

- Khalifat N, Fournier JB, Angelova MI, Puff N. **Lipid packing variations induced by pH in cardiolipin-containing bilayers: The driving force for the cristae-like shape instability.** Biochim Biophys Acta. 2011 Jul 22. [Epub ahead of print] *(PubMed Abstract)*

- Xiao J, Engel JL, Zhang J, Chen MJ, Manning G, Dixon JE**. Structural and functional analysis of PTPMT1, a phosphatase required for cardiolipin synthesis.** Proc Natl Acad Sci U S A. 2011 Jul 19;108(29):11860-5. Epub 2011 Jul 5. *(PubMed Abstract)*

- Christie DA, Lemke CD, Elias IM, Chau LA, Kirchhof MG, Li B, Ball EH, Dunn SD, Hatch GM, Madrenas J. **Stomatin-like protein 2 binds cardiolipin and regulates mitochondrial biogenesis and function.** Mol Cell Biol. 2011 Jul 11. [Epub ahead of print] *(PubMed Abstract)*

- Xiao J, Engel JL, Zhang J, Chen MJ, Manning G, Dixon JE. **Structural and functional analysis of PTPMT1, a phosphatase required for cardiolipin synthesis.** Proc Natl Acad Sci U S A. 2011 Jul 5. [Epub ahead of print] *(PubMed Abstract)*

- Sandlers Y, Baumann S. **Characterization of urine metabolites in Barth syndrome patients employing a non-targeted GC/MS screening approach.** 59th ASMS Conference on Mass Spectrometry, June 5 - 9, 2011, Denver, Colorado. (*Poster*)▼

- Zhang L, Bell RJA, Kiebish MA, Seyfried TN, Han X, Gross RW, Chuang JH. **A mathematical model for the determination of steady-state cardiolipin remodeling mechanisms using lipidomic data.** PLoS One. 2011;6(6):e21170. doi: 10.1371/journal.pone.0021170. Epub 2011 Jun 10. *(PubMed – Open Access)*

- Acehan D, Malhotra A, Xu Y, Ren M, Stokes DL, Schlame M. **Cardiolipin affects the supramolecular organization of ATP synthase in mitochondria.** Biophys J. 2011 May 4;100(9):2184-92. *(PubMed – Open Access)**

- Kuroda T, Tani M, Moriguchi A, Tokunaga S, Higuchi T, Kitada S, Kuge O. **FMP30 is required for the maintenance of a normal cardiolipin level and mitochondrial morphology in the absence of mitochondrial phosphatidylethanolamine synthesis.** Mol Microbiol. 2011 Apr;80(1):248-65. Epub 2011 Feb 24. *(PubMed Abstract)*

- Wajner M, Goodman SI. **Disruption of mitochondrial homeostasis in organic acidurias: Insights from human and animal studies.** J Bioenerg Biomembr. 2011 Feb;43(1):31-8. *(PubMed Abstract)*

- Rosca M, Minkler P, Hoppel CL. **Cardiac mitochondria in heart failure: Normal cardiolipin profile and increased threonine phosphorylation of complex IV**. Biochim Biophys Acta. 2011 Feb 11. [Epub ahead of print] *(PubMed Abstract)*

- Claypool SM, Whited K, Srijumnong S, Han X, Koehler CM**. Barth syndrome mutations that cause *tafazzin* complex lability.** J Cell Biol 2011 Feb 7;192(3):447-62. 192:447-462. *(PubMed Abstract)*

- Maniti O, Cheniour M, Lecompte MF, Marcillat O, Buchet R, Vial C, Granjon T. **Acyl chain composition determines cardiolipin clustering induced by mitochondrial creatine kinase binding to monolayers.** Biochim Biophys Acta. 2011 Jan 19. [Epub ahead of print] *(PubMed Abstract)*

- Osman C, Voelker DR, Langer T. **Making heads or tails of phospholipids in mitochondria.** J Cell Biol. 2011 Jan 10;192(1):7-16. *(PubMed Abstract)*

- Paradies G, Petrosillo G, Paradies V, Reiter RJ, Ruggiero FM. **Melatonin, cardiolipin and mitochondrial bioenergetics in health and disease.** J Pineal Res. 2010 May;48(4):297-310. Review. *(PubMed Abstract)*

- Bird SS, Marur VR, Sniatynski MJ, Greenberg HK, Kristal BSF. **Lipodomics profiling by high-resolution LC-MS and high-energy collisional dissociation fragmentation: Focus on characterization of mitochondrial cardiolipins and monolysocardiolipins.** Anal Chem. 2010 Dec 30. [Epub ahead of print] *(PubMed Abstract)*

- Jalmar O, Garcia-Saez AJ, Berland L, Gonzalvez F, Petit PX. **Giant unilamellar vesicles (GUVs) as a new tool for analysis of caspase-8/Bid-FL complex binding to cardiolipin and its functional activity.** Cell Death Dis. 2010 Dec 2;1:e103. doi: 10.1038/cddis.2010.81. *(PubMed – Open Access)*▼

- Soustek MS, Falk D, Mah C, Toth M, Schlame M, Lewin A, Byrne B. **Characterization of a transgenic shRNA induced murine model of *tafazzin* deficiency.** Hum Gene Ther. 2010 Nov 23. [Epub ahead of print] *(PubMed Abstract)*▼

- Takahashi H, Hayakawa T, Ito K, Takata M, Kobayashi T. **Small-angle and wide-angle X-ray scattering study on the bilayer structure of synthetic and bovine heart cardiolipins.** 2010 J. Phys.: Conf. Ser. 247 012021 *(Abstract)**

- Kim J, Hoppel CL. **Monolysocardiolipin: Improved preparation with high yield.** J Lipid Res. 2010 Oct 19. [Epub ahead of print] *(PubMed Abstract)*

- Scherer M, Schmitz G, Liebisch G. **Simultaneous quantification of cardiolipin, Bis(monoacylglycero)phosphate and their precursors by hydrophilic interaction LC-MS/MS including correction of isotopic overlap.** Anal Chem. 2010 Oct 14. [Epub ahead of print]. *(PubMed Abstract)*

- Yoda E, Hachisu K, Taketomi Y, Yoshida K, Nakamura M, Ikeda K, Taguchi R, Nakatani Y, Kuwata H, Murakami M, Kudo I, Hara S. **Mitochondrial dysfunction and reduced prostaglandin synthesis in skeletal muscle of Group VIB Ca2+-independent phospholipase A2gamma-deficient mice.** J Lipid Res. 2010 Oct;51(10):3003-15. Epub 2010 Jul 12. *(PubMed Abstract)*

- Zhao W, Waisum O, Fung Y, Cheung MP. **Analysis of mitochondria by capillary electrophoresis: Cardiolipin levels decrease in response to carbonyl cyanide 4-(trifluoromethoxy) phenylhydrazone.** European Journal of Lipid Science and Technology Special Issue: Focus Issue Euro Fed Lipid Highlights Volume 112, Issue 9, pages 1058–1066, September 2010. *(Wiley Abstract)*

- Crimi M, Esposti MD. **Apoptosis-induced changes in mitochondrial lipids.** Biochim Biophys Acta. 2010 Sep 29. [Epub ahead of print] *(PubMed Abstract)*

- Trusova VM, Gorbenko GP, Molotkovsky JG, Kinnunen PK. **Cytochrome c-lipid interactions: New insights from resonance energy transfer.** Biophys J. 2010 Sep 22;99(6):1754-63. *(PubMed Abstract)*

- Kim J, Minkler PE, Salomon RG, Anderson VE, Hoppel CL. **Cardiolipin: Characterization of distinct oxidized molecular species.** J Lipid Res. 2010 Sep 20. [Epub ahead of print] *(PubMed Abstract)*

- Gebert N, Ryan MT, Pfanner N, Wiedemann N, Stojanovski D. **Mitochondrial protein import machineries and lipids: A functional connection.** Biochim Biophys Acta. 2010 Aug 7. [Epub ahead of print] *(PubMed Abstract)*

- Hauff K, Hatch GM. **Reduction in cholesterol synthesis in response to serum starvation in lymphoblasts of a patient with Barth syndrome.** Biochem Cell Biol. 2010 Aug;88(4):595-602. *(PubMed Abstract)** ▼

- Xu FY, McBride H, Acehan D, Vaz FM, Houtkooper RH, Lee RM, Mowat MA, Hatch GM. **The dynamics of cardiolipin synthesis post mitochondrial fusion.** Biochim Biophys Acta. 2010 Aug;1798(8):1577-85. Epub 2010 Apr 29. *(PubMed Abstract)**

- Kiebish MA, Bell R, Yang K, Phan T, Zhao Z, Ames W, Seyfried TN, Gross RW, Chuang JH, Han X. **Dynamic simulation of cardiolipin remodeling: Greasing the wheels for an interpretative approach to lipidomics.** J Lipid Res. 2010 Aug;51(8):2153-70. Epub 2010 Apr 21. *(PubMed Abstract)*

- Macchioni L, Corazzi T, Davidescu M, Francescangeli E, Roberti R, Corazzi L. **Cytochrome c redox state influences the binding and release of cytochrome c in model membranes and in brain mitochondria.** Mol Cell Biochem. 2010 Aug;341(1-2):149-57. Epub 2010 Mar 30. *(PubMed Abstract)*

- Pei W, Kratz LE, Bernardini I, Sood R, Yokogawa T, Dorward H, Ciccone C, Kelley RI, Anikster Y, Burgess HA, Huizing M, Feldman B. **A model of Costeff syndrome reveals metabolic and protective functions of mitochondrial OPA3.** Development 2010 Aug 1;137(15):2587-96. *(PubMed Abstract)*

- Lu B, Xu FY, Taylor WA, Feingold KR, Hatch GM. **Cardiolipin synthase-1 mRNA expression does not correlate with endogenous cardiolipin synthase enzyme activity in vitro and in vivo in mammalian lipopolysaccharide models of inflammation.** Inflammation. 2010 Jul 23. [Epub ahead of print] *(PubMed Abstract)*

- Osman C, Haag M, Wieland FT, Brügger B, Langer T. **A mitochondrial phosphatase required for cardiolipin biosynthesis: The PGP phosphatase Gep4.** EMBO J. 2010 Jun 16;29(12):1976-87. Epub 2010 May 18. *(PubMed Abstract)*

- Rijken PJ. **Phosphatidylcholine-protein interactions and remodeling of cardiolipin in yeast mitochondria.** Doctoral Thesis, Scheikunde Proefschriften, 2010. *(Full Text)**

- Hasson SA, Damoiseaux R, Glavin JD, Dabir DV, Walker SS, Koehler CM. **Substrate specificity of the TIM22 mitochondrial import pathway revealed with small molecule inhibitor of protein translocation.** Proc Natl Acad Sci U S A. 2010 May 25;107(21):9578-83. Epub 2010 May 10. *(PubMed Abstract)*

- Raja M. **The Role of phosphatidic acid and cardiolipin in stability of the tetrameric assembly of potassium channel KcsA.** J Membr Biol. 2010 Apr;234(3):235-40. Epub 2010 Mar 30. *(PubMed Abstract)*

- Lenaz G, Genova ML. **Structure and organization of mitochondrial respiratory complexes: A new understanding of an old subject.** Antioxid Redox Signal. 2010 Apr 15;12(8):961-1008. Review. *(PubMed Abstract)*

- Chen S, Liu D, Finley RL Jr, Greenberg ML. **Loss of mitochondrial DNA in the yeast cardiolipin synthase crd1 mutant leads to up-regulation of the protein kinase Swe1p that regulates the G2/M transition.** J Biol Chem. 2010 Apr 2;285(14):10397-407. Epub 2010 Jan 19. *(PubMed – Open Access)**

- Nie J, Hao X, Chen D, Han X, Chang Z, Shi Y. **A novel function of the human CLS1 in phosphatidylglycerol synthesis and remodeling.** Biochim Biophys Acta. 2010 Apr;1801(4):438-45. Epub 2009 Dec 16. *(PubMed Abstract)*

- Zachman DK, Chicco AJ, McCune SA, Murphy RC, Moore RL, Sparagna GC. **The role of calcium-independent phospholipase A2 in cardiolipin remodeling in the spontaneously hypertensive heart failure rat heart.** J Lipid Res. 2010 Mar;51(3):525-34. Epub 2009 Sep 9. *(PubMed – Open Access)**

- Corcelli A, Saponetti MS, Zaccagnino P, Lopalco P, Mastrodonato MG, Liquori GE, Lorusso M. **Mitochondria isolated in nearly isotonic KCl buffer: Focus on cardiolipin and organelle morphology.** Biochim Biophys Acta. 2010 Mar;1798(3):681-7. Epub 2010 Jan 20. *(PubMed Abstract)*

- Gonzalvez F, Pariselli F, Jalmar O, Dupaigne P, Sureau F, Dellinger M, Hendrickson EA, Bernard S, Petit PX. **Mechanistic issues of the interaction of the hairpin-forming domain of tBid with mitochondrial cardiolipin.** PLoS One. 2010 Feb 22;5(2):e9342. *(PubMed Abstract)*

- Wiswedel I, Gardemann A, Storch A, Peter D, Schild L. **Degradation of phospholipids by oxidative stress-exceptional significance of cardiolipin**. Free Radic Res. 2010 Feb;44(2):135-45. *(PubMed Abstract)*

- van Gestel RA, Rijken PJ, Surinova S, O'Flaherty M, Heck AJ, Killian JA, de Kroon AI, Slijper M. **The influence of the acyl chain composition of cardiolipin on the stability of mitochondrial complexes: An unexpected effect of cardiolipin in alpha-ketoglutarate dehydrogenase and prohibitin complexes.** J Proteomics. 2010 Feb 10;73(4):806-14. Epub 2009 Nov 24. *(PubMed Abstract)*

- Gebert N, Joshi AS, Kutik S, Becker T, McKenzie M, Li Guan X, Mooga VP, Stroud DA, Kulkarni G, Wenk MR, Rehling P, Meisinger C, Ryan MT, Wiedemann N, Greenberg ML, Pfanner N. **Mitochondrial cardiolipin involved in outer-membrane protein biogenesis: Implications for Barth syndrome.** Curr Biol. 2009 Dec 29;19(24):2133-9. Epub 2009 Dec 3. *(PubMed – Open Access)**

- Mancuso DJ, Kotzbauer PT, Wozniak DF, Sims HF, Jenkins CM, Guan S, Han X, Yang K, Sun G, Malik I, Conyers S, Green KG, Schmidt RE, Gross RW. **Genetic ablation of calcium-independent phospholipase A2{gamma} leads to alterations in hippocampal cardiolipin content and molecular species distribution, mitochondrial degeneration, autophagy and cognitive dysfunction.** J Biol Chem. 2009 Dec 18;284(51):35632-44. Epub. *(PubMed Abstract)*

- Hauff KD, Choi SY, Frohman MA, Hatch GM. **Cardiolipin synthesis is required to support human cholesterol biosynthesis from palmitate upon serum removal in Hela cells.** Can J Physiol Pharmacol. 2009 Oct;87(10):813-20. *(PubMed – Open Access)**

- Haines TH. **A new look at cardiolipin**. Biochim Biophys Acta. 2009 Oct;1788(10):1997-2002. *(PubMed - No abstract available)*

- Taylor WA, Hatch GM. **Identification of the human mitochondrial linoleoyl-coenzyme a monolysocardiolipin acyltransferase (MLCL AT-1).** J Biol Chem. 2009 Oct 30;284(44):30360-71. Epub 2009 Sep 8. *(PubMed – Open Access)**

- Xu Y, Zhang S, Malhotra A, Edelman-Novemsky I, Ma J, Kruppa A, Cernicica C, Blais S, Neubert TA, Ren M, Schlame M. **Characterization of *tafazzin* splice variants from humans and fruit flies.** J Biol Chem. 2009 Oct 16;284(42):29230-9. Epub 2009 Aug 21. *(PubMed – Open Access)**

- Rijken PJ, Houtkooper RH, Akbari H, Brouwers JF, Koorengevel MC, de Kruijff B, Frentzen M, Vaz FM, de Kroon AI. **Cardiolipin molecular species with shorter acyl chains accumulate in S. cerevisiae mutants lacking the acyl-Coenzyme A-binding protein Acb1p. New insights into acyl chain remodeling of cardiolipin.** J Biol Chem. 2009 Oct 2;284(40):27609-19. Epub 2009 Aug 5. *(PubMed – Open Access)**

- Houtkooper RH, Turkenburg M, Poll-The BT, Karall D, Pérez-Cerdá C, Morrone A, Malvagia S, Wanders RJ, Kulik W, Vaz FM. **The enigmatic role of *tafazzin* in cardiolipin metabolism.** Biochim Biophys Acta. 2009 Oct;1788(10):2003-14. Epub 2009 Jul 18. Review. *(PubMed Abstract)**

- Mitchell RW, Hatch GM. **Regulation of cardiolipin biosynthesis by fatty acid transport Protein-1 in HEK 293 cells.** Biochim Biophys Acta. 2009 Oct;1788(10):2015-21. Epub 2009 Jun 11. *(PubMed Abstract)*

- Schug ZY, Gottlieb E. **Cardiolipin acts as a mitochondrial signalling platform to launch apoptosis.** Biochim Biophys Acta. 2009 Oct;1788(10):2022-31. Epub 2009 May 18. Review. *(PubMed Abstract)*

- Claypool SM. **Cardiolipin, a critical determinant of mitochondrial carrier protein assembly and function.** Biochim Biophys Acta. 2009 Oct;1788(10):2059-68. Epub 2009 May 5. Review. *(PubMed Abstract*)

- Schlattner U, Tokarska-Schlattner M, Ramirez S, Brückner A, Kay L, Polge C, Epand RF, Lee RM, Lacombe ML, Epand RM. **Mitochondrial kinases and their molecular interaction with cardiolipin.** Biochim Biophys Acta. 2009 Oct;1788(10):2032-47. Epub 2009 May 4. Review. *(PubMed – Open Access*)*

- Schlame M, Ren M. **The role of cardiolipin in the structural organization of mitochondrial membranes**. Biochim Biophys Acta. 2009 Oct;1788(10):2080-3. Epub 2009 May 4. Review. *(PubMed – Open Access)**

- Mileykovskaya E, Dowhan W. **Cardiolipin membrane domains in prokaryotes and eukaryotes.** Biochim Biophys Acta. 2009 Oct;1788(10):2084-91. Epub 2009 Apr 14. Review. *(PubMed Abstract)*

- Johns MK, Yin MX, Conway SJ, Robinson DEJE, Wong LSM, Bamert R, Wettenhall REH, Holmes AB. **Synthesis and biological evaluation of a novel cardiolipin affinity matrix.** Org Biomol Chem. 2009 Sep 21;7(18):3691-7. Epub 2009 Jul 14. *(PubMed Abstract)*

- Saini-Chohan HK, Holmes MG, Chicco AJ, Taylor WA, Moore RL, McCune SA, Hickson-Bick DL, Hatch GM, Sparagna GC. **Cardiolipin biosynthesis and remodeling enzymes are altered during the development of heart failure.** J Lipid Res. 2009 Aug;50(8):1600-8. Epub 2008 Nov 10. *(PubMed – Open Access)**

- Sorice M, Manganelli V, Matarrese P, Tinari A, Misasi R, Malorni W, Garofalo T. **Cardiolipin-enriched raft-like microdomains are essential activating platforms for apoptotic signals on mitochondria.** FEBS Lett. 2009 Aug 6;583(15):2447-50. Epub 2009. *(PubMed Abstract)*

- Finsterer J. **Cardiogenetics, neurogenetics, and pathogenetics of left ventricular hypertrabeculation/noncompaction.** Pediatr Cardiol. 2009 Jul;30(5):659-81. Epub 2009 Jan 29. *(PubMed Abstract)*

- Zhou J, Zhong Q, Li G, Greenberg ML. **Loss of cardiolipin leads to longevity defects that are alleviated by alterations in stress response signaling.** J Biol Chem. 2009 Jul 3;284(27):18106-14. Epub 2009 Apr 28. *(PubMed – Open Access)**

- Paradies G, Petrosillo G, Paradies V, Ruggiero FM. **Role of cardiolipin peroxidation and Ca(2+) in mitochondrial dysfunction and disease.** Cell Calcium. 2009 Jun;45(6):643-50. Epub 2009 Apr 15. Review. *(PubMed Abstract)*

- Wittig I, Schägger H. **Supramolecular organization of ATP synthase and respiratory chain in mitochondrial membranes.** Biochim Biophys Acta. 2009 Jun;1787(6):672-80. Epub 2009 Jan 8. Review. *(PubMed Abstract)*

- Wendel AA, Lewin TM, Coleman RA. **Glycerol-3-phosphate acyltransferases: Rate limiting enzymes of triacylglycerol biosynthesis.** Biochim Biophys Acta. 2009 Jun;1791(6):501-6. Epub 2008 Nov 7. Review. *(PubMed Abstract)*

- Schlame M. **Formation of molecular species of mitochondrial cardiolipin. 2. A mathematical model of pattern formation by phospholipid transacylation.** Biochim Biophys Acta. 2009 Apr;1791(4):321-5. Epub 2009 Jan 31. *(PubMed Abstract)*

- Rosca MG, Hoppel CL. **New aspects of impaired mitochondrial function in heart failure.** J Bioenerg Biomembr. 2009 Apr;41(2):107-12. Review. *(PubMed Abstract)*

- Malhotra A, Xu Y, Ren M, Schlame M. **Formation of molecular species of mitochondrial cardiolipin. 1. A novel transacylation mechanism to shuttle fatty acids between sn-1 and sn-2 positions of multiple phospholipid species.** Biochim Biophys Acta. 2009 Apr;1791(4):314-20. Epub 2009 Jan 21. *(PubMed – Open Access)**

- Sparagna GC, Lesnefsky EJ. **Cardiolipin remodeling in the heart.** J Cardiovasc Pharmacol. 2009 Apr;53(4):290-301. Review. *(PubMed Abstract)**

- Houtkooper RH, Rodenburg RJ, Thiels C, van Lenthe H, Stet F, Poll-The BT, Stone JE, Steward CG, Wanders RJ, Smeitink J, Kulik W, Vaz FM. **Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes and tissues using HPLC-mass spectrometry as a diagnostic test for Barth syndrome.** Anal Biochem. 2009 Apr 15;387(2):230-7. Epub 2009 Jan 31. *(PubMed Abstract)**

- Rog T, Martinez-Seara H, Munck N, Karttunen M, Vattulainen I. **Role of cardiolipins in the inner mitochondrial membrane: Insight gained through atom-scale simulations.** J Phys Chem B. 2009 Mar 19;113(11):3413-22. *(PubMed Abstract)*

- Beranek A, Rechberger G, Knauer H, Wolinski H, Kohlwein SD, Leber R. **Identification of a cardiolipin-specific phospholipase encoded by the gene CLD1 (YGR110W) in yeast.** J Biol Chem. 2009 Apr 24;284(17):11572-8. Epub 2009 Feb 25. *(PubMed Abstract)*

- Acehan D, Khuchua Z, Houtkooper RH, Malhotra A, Kaufman J, Vaz FM, Ren M, Rockman HA, Stokes DL, Schlame M. **Distinct effects of *tafazzin* deletion in differentiated and undifferentiated mitochondria.** Mitochondrion. 2009 Apr;9(2):86-95. Epub 2008 Dec 11. *(PubMed – Open Access)**

- Gohil VM, Greenberg ML. **Mitochondrial membrane biogenesis: Phospholipids and proteins go hand in hand.** J Cell Biol. 2009 Feb 23;184(4):469-72. *(PubMed Abstract)*

- Osman C, Haag M, Potting C, Rodenfels J, Dip PV, Wieland FT, Brügger B, Westermann B, Langer T. **The genetic interactome of prohibitins: Coordinated control of cardiolipin and phosphatidylethanolamine by conserved regulators in mitochondria.** J Cell Biol. 2009 Feb 23;184(4):583-96. Epub 2009 Feb 16. *(PubMed Abstract)*

- Malhotra A, Edelman-Novemsky I, Xu Y, Plesken H, Ma J, Schlame M, Ren M. **Role of calcium-independent phospholipase A2 in the pathogenesis of Barth syndrome.** Proc Natl Acad Sci U S A. 2009 Feb 17;106(7):2337-41. Epub 2009 Jan 21. *(PubMed – Open Access)**

- Joshi AS, Zhou J, Gohil VM, Chen S, Greenberg ML. **Cellular functions of cardiolipin in yeast.** Biochim Biophys Acta. 2009 Jan;1793(1):212-8. Epub 2008 Aug 7. *(PubMed – Open Access)**

- Hauff K, Linda D, Hatch GM. **Mechanism of the elevation in cardiolipin during Hela cell entry into the S phase of the human cell cycle.** Biochem J. 2009 Jan 15;417(2):573-82. *(PubMed Abstract)*

- Kutik S, Rissler M, Guan XL, Guiard B, Shui G, Gebert N, Heacock PN, Rehling P, Dowhan W, Wenk MR, Pfanner N, Wiedemann N. **The translocator maintenance protein Tam41 is required for mitochondrial cardiolipin biosynthesis.** J Cell Biol. 2008 Dec 29;183(7):1213-21. *(PubMed Abstract)*

- Chen S, Tarsio M, Kane PM, Greenberg ML. **Cardiolipin mediates cross-talk between mitochondria and the vacuole.** Mol Biol Cell. 2008 Dec;19(12):5047-58. Epub 2008 Sep 17. *(PubMed – Open Access)**

- Claypool SM, Boontheung P, McCaffery JM, Loo JA, Koehler CM. **The cardiolipin transacylase, *tafazzin*, associates with two distinct respiratory components providing insight into Barth syndrome.** Mol Biol Cell. 2008 Dec;19(12):5143-55. Epub 2008 Sep 17. *(PubMed Abstract)*

- Scorrano L. **Caspase-8 goes cardiolipin: A new platform to provide mitochondria with microdomains of apoptotic signals?** J Cell Biol. 2008 Nov 17;183(4):579-81. Epub 2008 Nov 10. *(PubMed Abstract)*

- Gonzalvez F, Schug ZT, Houtkooper RH, Mackenzie ED, Brooks DG, Wanders RJ, Petit PX, Vaz FM, Gottlieb E. **Cardiolipin provides an essential activating platform for caspase-8 on mitochondria.** J Cell Biol. 2008 Nov 17;183(4):681-96. Epub 2008 Nov 10. *(PubMed – Open Access)**

- Kaewsuya P, Miller JD, Danielson ND, Sanjeevi J, James PF. **Comparison of N-alkyl acridine orange dyes as fluorescence probes for the determination of cardiolipin.** Anal Chim Acta. 2008 Sep 26;626(2):111-8. Epub 2008 Aug 15. *(PubMed Abstract)*

- Claypool SM, Oktay Y, Boontheung P, Loo JA, Koehler CM. **Cardiolipin defines the interactome of the major ADP/ATP carrier protein of the mitochondrial inner membrane.** J Cell Biol. 2008 Sep 8;182(5):937-50. *(PubMed Abstract)*

- Houtkooper RH, Vaz FM. **Cardiolipin, the heart of mitochondrial metabolism.** Cell Mol Life Sci. 2008 Aug;65(16):2493-506. *(PubMed Abstract)*

- Schlame M. **Cardiolipin synthesis for the assembly of bacterial and mitochondrial membranes**. J Lipid Res. 2008 Aug;49(8):1607-20. Epub 2007 Dec 12. *(PubMed Abstract)*

- Cheng H, Mancuso DJ, Jiang X, Guan S, Yang J, Yang K, Sun G, Gross RW, Han X. **Shotgun lipidomics reveals the temporally dependent, highly diversified cardiolipin profile in the mammalian brain: Temporally coordinated postnatal diversification of cardiolipin molecular species with neuronal remodeling.** Biochemistry. 2008 May 27;47(21):5869-80. Epub 2008 May 3. *(PubMed Abstract)*

- Chen S, He Q, Greenberg ML. **Loss of *tafazzin* in yeast leads to increased oxidative stress during respiratory growth.** Mol Microbiol. 2008 May;68(4):1061-72. *(PubMed Abstract)**

- Danos M, Taylor WA, Hatch GM. **Mitochondrial monolysocardiolipin acyltransferase is elevated in the surviving population of H9c2 cardiac myoblast cells exposed to 2-deoxyglucose-induced apoptosis.** Biochem Cell Biol. 2008 Feb;86(1):11-20. *(PubMed Abstract)*

- Garrett TA, Kordestani R, Raetz CR. **Quantification of cardiolipin by liquid chromatography-electrospray ionization mass spectrometry.** Methods Enzymol. 2007;433:213-30. *(PubMed Abstract)*

- Mancuso DJ, Sims HF, Han X, Jenkins CM, Guan SP, Yang K, Moon SH, Pietka T, Abumrad NA, Schlesinger PH, Gross RW. **Genetic ablation of calcium-independent phospholipase A2gamma leads to alterations in mitochondrial lipid metabolism and function resulting in a deficient mitochondrial bioenergetic phenotype.** J Biol Chem. 2007 Nov 30;282(48):34611-22. Epub 2007 Oct 8. *(PubMed Abstract)*

- Hullin-Matsuda F, Kawasaki K, Delton-Vandenbroucke I, Xu Y, Nishijima M, Lagarde M, Schlame M, Kobayashi T. **De novo biosynthesis of the late endosome lipid, bis(monoacylglycero)phosphate.** J Lipid Res. 2007 Sep;48(9):1997-2008. Epub 2007 Jun 8. *(PubMed – Open Access)**

- Philipp U, Broschk C, Vollmar A, Distl O. **Evaluation of *tafazzin* as candidate for dilated cardiomyopathy in Irish wolfhounds.** J Hered. 2007 Sep-Oct;98(5):506-9. Epub 2007 Jul 9. *(PubMed Abstract)*

- Sparagna GC, Chicco AJ, Murphy RC, Bristow MR, Johnson CA, Rees ML, Maxey ML, McCune SA, Moore RL. **Loss of cardiac tetralinoleoyl cardiolipin in human and experimental heart failure.** J Lipid Res. 2007 Jul;48(7):1559-70. Epub 2007 Apr 10. *(PubMed Abstract)*

- Zhong Q, Li G, Gvozdenovic-Jeremic J, Greenberg ML. **Up-regulation of the cell integrity pathway in saccharomyces cerevisiae suppresses temperature sensitivity of the pgs1Delta mutant.** J Biol Chem. 2007 Jun 1;282(22):15946-53. Epub 2007 Apr 9. *(PubMed – Open Access)**

- Han X, Yang J, Yang K, Zhao Z, Abendschein DR, Gross RW. **Alterations in myocardial cardiolipin content and composition occur at the very earliest stages of diabetes: A shotgun lipidomics study.** Biochemistry. 2007 May 29;46(21):6417-28. Epub 2007 May 8. *(PubMed Abstract)*

- Postle AD, Wilton DC, Hunt AN, Attard GS. **Probing phospholipid dynamics by electrospray ionisation mass spectrometry.** Prog Lipid Res. 2007 May-Jul;46(3-4):200-24. Epub 2007 Apr 24. *(PubMed Abstract)*

- Marziliano N, Mannarino S, Nespoli L, Diegoli M, Pasotti M, Malattia C, Grasso M, Pilotto A, Porcu E, Raisaro A, Raineri C, Dore R, Maggio PP, Brega A, Arbustini E. **Barth syndrome associated with compound hemizygosity and heterozygosity of the *TAZ* and LDB3 genes.** Am J Med Genet A. 2007 May 1;143A(9):907-15. *(PubMed Abstract)*

- Chicco AJ, Sparagna GC. **Role of cardiolipin alterations in mitochondrial dysfunction and disease.** Am J Physiol Cell Physiol. 2007 Jan;292(1):C33-44. Epub 2006 Aug 9. *(PubMed Abstract)*

- Xu Y, Malhotra A, Ren M, Schlame M. **The enzymatic function of *tafazzin*.** J Biol Chem. 2006 Dec 22;281(51):39217-24. Epub 2006 Nov 2. *(PubMed Abstract)**▼

BSF does not endorse any drugs, tests, or treatments that we may report.

- Alessandri C, Sorice M, Bombardieri M, Conigliaro P, Longo A, Garofalo T, Manganelli V, Conti F, Degli Esposti M, Valesini G. **Anti-phospholipid reactivity against cardiolipin metabolites occurring during endothelial cell apoptosis.** Arthritis Res Ther. 2006 Dec 6;8(6):R180. *(PubMed Abstract)**

- Tuma RS. **Sitting in—not through—a membrane.** J. Cell Biol., Jul 2006; 174: 318. *(Full text)*

- Schlame M, Ren M. **Barth syndrome, a human disorder of cardiolipin metabolism.** FEBS Lett. 2006 Oct 9;580(23):5450-5. Epub 2006 Jul 17. *(PubMed – Open Access)**

- Chen D, Zhang XY, Shi Y. **Identification and functional characterization of hCLS1, a human cardiolipin synthase localized in mitochondria.** Biochem J. 2006 Sep 1;398(2):169-76. *(PubMed Abstract)*

- Lu B, Xu FY, Jiang YJ, Choy PC, Hatch GM, Grunfeld C, Feingold KR. **Cloning and characterization of a cDNA encoding human cardiolipin synthase (hCLS1).** J Lipid Res. 2006 Jun;47(6):1140-5. Epub 2006 Mar 18. *(PubMed Abstract)*

- Houtkooper RH, Akbari H, van Lenthe H, Kulik W, Wanders RJ, Frentzen M, Vaz FM. **Identification and characterization of human cardiolipin synthase.** FEBS Lett. 2006 May 29;580(13):3059-64. Epub 2006 Apr 27. *(PubMed Abstract)*

- Li G, Chen S, Thompson MN, Greenberg ML. **New insights into the regulation of cardiolipin biosynthesis in yeast: Implications for Barth syndrome.** Biochim Biophys Acta. 2007 Mar;1771(3):432-41. Epub 2006 Jul 8. Review. *(PubMed Abstract)**

- van Werkhoven MA, Thorburn DR, Gedeon AK, Pitt JJ. **Monolysocardiolipin in cultured fibroblasts is a sensitive and specific marker for Barth syndrome.** J Lipid Res. 2006 Oct;47(10):2346-51. Epub 2006 Jul 27. *(PubMed Abstract)*

- Claypool SM, McCaffery JM, Koehler CM. **Mitochondrial mislocalization and altered assembly of a cluster of Barth syndrome mutant *tafazzins*.** J Cell Biol. 2006 Jul 31;174(3):379-90. *(PubMed Abstract)*

- Han X, Yang K, Yang J, Cheng H, Gross RW. **Shotgun lipidomics of cardiolipin molecular species in lipid extracts of biological samples**. J Lipid Res. 2006 Apr;47(4):864-79. Epub 2006 Jan 31. *(PubMed Abstract)*

- Hauff KD, Hatch, GM. **Cardiolipin metabolism and Barth syndrome**. Prog Lipid Res. 2006 Mar;45(2):91-101. Epub 2006 Jan 18. *(PubMed Abstract)**

- Brandner K, Mick DU, Frazier AE, Taylor RD, Meisinger C, Rehling P. ***TAZ*1, an outer mitochondrial membrane protein, affects stability and assembly of inner membrane protein complexes: Implications for Barth syndrome.** Mol Biol Cell. 2005 Nov;16(11):5202-14. Epub 2005 Aug 31. *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Gonzalvez F, Pariselli F, Dupaigne P, Budihardjo I, Lutter M, Antonsson B, Diolez P, Manon S, Martinou J-C, Goubern M, Wang X, Bernard S, Petit PX. **tBid interaction with cardiolipin primarily orchestrates mitochondrial dysfunctions and subsequently activates Bax and Bak**. Cell Death Differ. 2005 Jun;12(6):614-26. *(PubMed Abstract)*

- Gonzalvez F, Bessoule J-J, Rocchiccioli F, Manon S, Petit PX. **Role of cardiolipin on tBid and tBid/Bax synergistic effects on yeast mitochondria.** Cell Death Differ 2005 Jun;12(6):659-67. *(PubMed Abstract)*

- Xu Y, Sutachan JJ, Plesken H, Kelley RI, Schlame M. **Characterization of lymphoblast mitochondria from patients with Barth syndrome.** Lab Invest. 2005 Jun;85(6):823-30. *(PubMed – Open Access)**

- Valianpour F, Mitsakos V, Schlemmer D, Towbin JA, Taylor JM, Ekert PG, Thorburn DR, Munnich A, Wanders RJ, Barth P, Vaz FM. **Monolysocardiolipins accumulate in Barth syndrome but do not lead to enhanced apoptosis.** J Lipid Res. 2005 Jun;46(6):1182-95. Epub 2005 Apr 1. *(PubMed – Open Access)*▼

- Testet E, Laroche-Traineau J, Noubhani A, Coulon D, Bunoust O, Camougrand N, Manon S, Lessire R, Bessoule JJ. **Ypr140wp, 'the yeast *tafazzin*', displays a mitochondrial lysophosphatidylcholine (lyso-PC) acyltransferase activity related to triacylglycerol and mitochondrial lipid synthesis.** Biochem J. 2005 May 1;387(Pt 3):617-26. *(PubMed Abstract)*

- Appelt U, Sheriff A, Gaipl US, Kalden JR, Voll RE, Herrmann M. **Viable, apoptotic and necrotic monocytes expose phosphatidylserine: Cooperative binding of the ligand Annexin V to dying but not viable cells and implications for PS-dependent clearance.** Cell Death Differ. 2005 Feb;12(2):194-6. *(PubMed Abstract)*

- Valianpour F. **A mass spectrometric approach to investigate cardiolipin metabolism in Barth syndrome.** PhD Dissertation, Academic Medical Center, University of Amsterdam, September 2004. *(Not peer reviewed).*

- Fadeel B, Karpova MB, Enoksson M, Orrenius S, Kuijpers TW**. Phosphatidylserine externalization in cardiolipin-deficient cells.** Blood. 2004 Sep 1;104(5):1582-3; author reply 1583-4. *(PubMed Abstract)*

- Fadeel B, Karpova MB, Enoksson M, Orrenius S. Correspondence: **To the editor: Phosphatidylserine externalization in cardiolipin-deficient cells.** Blood. 2004 May 15;103(10):3915-23. *(Full text)*

- Lu B, Kelher MR, Lee DP, Lewin TM, Coleman RA, Choy PC, Hatch GM. **Complex expression pattern of the Barth syndrome gene product *tafazzin* in human cell lines and murine tissues.** Biochem Cell Biol. 2004 Oct;82(5):569-76. *(PubMed Abstract)**

- Schaefer U, Hurlstone A, Degli Esposti M. **The mitochondria-dependent death of neurosensory cells *in vivo*** [abstract]. Presented at Fall 2004 Conference in Glasgow, UK.

- Schaefer U, Goonesinghe A, Hurlstone A, Degli Esposti M. **Swimming to death: A sniff into developmental apoptosis** [abstract]. In European Cell Death Organization (ECDO) 12th Euroconference on Apoptosis; 2004 Sept. 17-20; Chania, Crete, Greece.

- Ma L, Vaz FM, Gu Z, Wanders RJ, Greenberg ML. **The human *TAZ* gene complements mitochondrial dysfunction in the yeast *taz*1delta mutant-implications for Barth syndrome**. J Biol Chem. 2004 Oct 22;279(43):44394-9. Epub 2004 Aug 10. *(PubMed – Open Access)**

- Sorice M, Circella A, Cristea IM, Garfalo T, Di Renzo L, Alessandri C, Valesini G, Degli Esposti M. **Cardiolipin and its metabolites move from mitochondria to other cellular membranes during death receptor-medicated apoptosis**. Cell Death Differ. 2004 Oct;11(10):1133-45. *(PubMed – Open Access)**

- Cao J, Liu Y, Lockwood J, Burn P, Shi Y. **A novel cardiolipin-remodeling pathway revealed by a gene encoding an endoplasmic reticulum-associated acyl-CoA:lysocardiolipin acyltransferase (ALCAT1) in mouse.** J Biol Chem. 2004 Jul 23;279(30):31727-34. Epub 2004 May 19. *(PubMed Abstract)*

- Degli Esposti M. **Mitochondria in apoptosis: Past, present and future.** Biochem Soc Trans. 2004 Jun;32(Pt3):493-5. *(PubMed Abstract)*

- Cristea IM, Degli Esposti M. **Membrane lipids and cell death: An overview**. Chem Phys Lipids. 2004 May;129(2):133-60. *(PubMed Abstract)*▼

- Schmidt MR, Birkebaek N, Gonzalez I, Sunde L. **Barth syndrome without 3-methylglutaconic aciduria**. Acta Paediatr 2004 Mar; 93(3):419-421. *(PubMed Abstract)*

- Hatch GM. **Cell biology of cardiac mitochondrial phospholipids**. Biochem Cell Biol 2004 Feb; 82(1):99-112. *(PubMed Abstract)*

- Maianski NA, Maianski AN, Kuijpers TW, Roos D. **Apoptosis of neutrophils.** Acta Haematol 2004; 111(1-2):56-66. *(PubMed Abstract)*

- Gu Z, Valianpour F, Chen S, Vaz FM, Hakkaart GA, Wanders RJA, Greenberg ML. **Aberrant cardiolipin metabolism in the yeast *taz1* mutant: A model for Barth syndrome**. Mol Microbiol. 2004 Jan; 51(1):149-58. *(PubMed Abstract)**

- Xu Y, Kelley RI, Blanck TJ, Schlame M. **Remodeling of cardiolipin by phospholipid transacylation.** J Biol Chem. 2003 Dec 19;278(51):51380-5. Epub 2003 Oct 9. *(PubMed Abstract)*

- Schlame M, Kelley RI, Feigenbaum A, Towbin JA, Heerdt PM, Schlieble T, Wanders RJ, DiMauro S, Blanck TJ**. Phospholipid abnormalities in children with Barth syndrome**. J Am Coll Cardiol 2003 Dec 3; 42(11):1994-1999. *(PubMed Abstract)*▼

- Degli Esposti M, Cristea IM, Gaskell SJ, Nakao Y, Dive C. **Proapoptotic Bid binds to monolysocardiolipin, a new molecular connection between mitochondrial membranes and cell death**. Cell Death Differ. 2003 Dec;10(12):1300-9. *(PubMed Abstract)*

- Vaz FM, Houtkooper RH, Valianpour F, Barth PG, Wanders RJ. **Only one splice variant of the human *TAZ* gene encodes a functional protein with a role in cardiolipin metabolism.** J Biol Chem. 2003 Oct 31;278(44):43089-94. Epub 2003 Aug 20. *(PubMed – Open Access)**

- Degli Esposti M. **The mitochondrial battlefield and membrane lipids during cell death signalling.** Ital J Biochem. 2003 Mar;52(1):43-50. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Overmars H, Vaz FM, Barth PG, van Gennip AH. **Linoleic acid supplementation of Barth syndrome fibroblasts restores cardiolipin levels: Implications for treatment**. J Lipid Res. 2003 Mar;44(3):560-6. Epub 2002 Dec 16. *(PubMed Abstract)*▼

- McMillin JB, Dowhan W. **Cardiolipin and apoptosis**. Biochem Biophys Acta 2002 Dec; 1585(2-3);97-107. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Overmars H, Vreken P, van Gennip AH, Baas F, Plecko B, Santer R, Becker K, Barth PG. **Cardiolipin deficiency in X-linked cardioskeletal myopathy and neutropenia (Barth syndrome, MIM 302060): A study in cultured skin fibroblasts**. J Pediatr. 2002 Nov; 141(5):729-33. *(PubMed Abstract)*▼

- Chen Y. Molecular studies on a *Dictyostelium* homolog of the *taffazin* gene, the cause of Barth syndrome in humans. Dr. rer. nat. dissertation; University of Kassel; November 2002. *(Not peer reviewed).*

- Gu Z, Gohil V, Zhong Q, Schlame M, Greenberg M. **The biosynthesis and remodeling of cardiolipin**. Research Signpost 37/661(2):67-84.

- Valianpour F, Wanders RJA, Barth PG, Overmars H, van Gennip AH. **Quantitative and compositional study of cardiolipin in platelets by electrospray ionization mass spectrometry: Application for the identification of Barth syndrome patients**. Clin Chem. 2002 Sep;48(9):1390-7. *(PubMed Abstract)*

- Kato A, Kawamata N, Tamayose K, Egashira M, Miura R, Fujimura T, Murayama K, Oshimi K. **Ancient ubiquitous protein 1 binds to the conserved membraneproximal sequence of the cytoplasmic tail of the integrin α subunits that plays a crucial role in the inside-out signaling of $\alpha_{IIb}\beta_3$.** J Biol Chem. 2002 Aug 9;277(32):28934-41. Epub 2002 May 31. *(PubMed Abstract)*

- Schlame M, Towbin JA, Heerdt PM, Jehle R, DiMauro S, Blanck TJJ. **Deficiency of tetralinoleoyl-cardiolipin in Barth syndrome**. Ann Neur 2002 May; 51(5):634-637. *(PubMed Abstract)*▼

- Koshkin V, Greenberg ML. **Cardiolipin prevents rate-dependent uncoupling and provides osmotic stabilitiy in yeast mitochondria.** Biochem J 2002 May 15;364(Pt 1):317-22. *(PubMed Abstract)*

- Waggoner DJ, Brown RL, Hedrick J. **Hypoglycemia and dicarboxylic aciduria as the presenting biochemical findings in a patient with Barth syndrome.** American Journal of Human Genetics 69.4 (Oct 2001): p492. *(Abstract)*

- Kelher MR, Coleman RA. ***Tafazzin*: Biochemical function and relationship to mitochondrial energy metabolism [**Symposium abstract #101]. In 11th Annual Institute of Nutrition Research Symposium; UNC-CH; 2000 Oct 13.

- Vreken P, Valianpour F, Nijtmans LG, Grivell LA, Plecko B, Wanders RJA, Barth PG. **Defective remodeling of cardiolipin and phosphatidylglycerol in Barth syndrome**. Biochem Biophys Res Commun. 2000 Dec 20;279(2):378-82. *(PubMed Abstract)*

- Schlame M, Rua D, Greenberg ML. **The biosynthesis and functional role of cardiolipin**. Prog Lipid Res. 2000 May;39(3):257-88. *(PubMed Abstract)*

- Mutter T, Dolinsky VW, Ma BJ, Taylor WA, Hatch GM. **Thyroxine regulation of monolysocardiolipin acyltransferase activity in rat heart**. Biochem J. 2000 Mar 1;346 Pt 2:403-6. *(PubMed Abstract)*

- Schlame M, Shanske S, Doty S, Konig T, Sculco T, DiMauro S, Blanck TJ. **Microanalysis of cardiolipin in small biopsies including skeletal muscle from patients with mitochondrial disease**. J Lipid Res. 1999 Sep;40(9):1585-92. *(PubMed Abstract)*

- Ma, BJ, Taylor WA, Dolinsky VW, Hatch GM. **Acylation of monolysocardiolipin in rat heart**. J Lipid Res. 1999 Oct;40(10):1837-45. *(PubMed Abstract)*

- Walsh R, Conway H, Roche G, Mayne PD. **What is the origin of 3-methylglutaconic acid?** J Inherit Metab Dis. 1999 May;22(3):251-5. *(PubMed Abstract)*

- Nikolaeva EA, Leontieva IV, Semenov V, Chentsova T, Sebeleva IA, Kazantseva LZ. **A case of Barth syndrome with massive 3-methylglutaconic and 3-methylglutaric aciduria**.

- Hatch GM. **Cardiolipin: Biosynthesis, remodeling and trafficking in the heart and mammalian cells** (Review). Int J Mol Med 1998 Jan; 1(1):33-41. *(PubMed Abstract)*

- Neuwald, AF. **Barth syndrome may be due to an acyltransferase deficiency**. Curr Biol. 1997 Aug 1;7(8):R465-6. *(PubMed Abstract)*

- Hatch GM, McClarty G. **Regulation of cardiolipin biosynthesis in H9c2 cardiac myoblasts by cytidine 5-triphosphate**. J Biol Chem. 1996 Oct 18;271(42):25810-6. *(PubMed Abstract)*

- Hatch GM. **Regulation of cardiolipin biosynthesis in the heart.** Mol Cell Biochem. 1996 Jun 21;159(2):139-48. *(PubMed Abstract)*

- Barth PG, Van den Bogert C, Bolhuis PA, Scholte HR, van Gennip AH, Schutgens RB, Ketel AG. **X-linked cardioskeletal myopathy and neutropenia (Barth syndrome): Respiratory-chain abnormalities in cultured fibroblasts**. Inherit Metab Dis. 1996;19(2):157-60. *(PubMed Abstract)*

- Gibson KM, Sherwood WG, Hoffmann GF, Stumpf DA, Dianzani I, Schutgens RBH, Barth PG, Weisman U, Bachmann C, Schrynemackers-Pitance P, Verloes A, Narisawa K, Mino M, Ohya N, Kelley RI. **Phenotypic heterogeneity in the syndromes of 3-methylglutaconic aciduria**. J Pediatr 1991 June; 118(6):885-890. *(PubMed Abstract)*

CARDIOMYOPATHY, HEART TRANSPLANTATION, AND ARRHYTHMIAS

NEW Stöllberger C, Finsterer J. **Understanding left ventricular hypertrabeculation/ noncompaction: pathomorphologic findings and prognostic impact of neuromuscular comorbidities.** Expert Rev Cardiovasc Ther. 2018 Dec 20. doi: 10.1080/14779072.2019.1561280. [Epub ahead of print] *(PubMed Abstract)*

NEW Vasilescu C, Ojala TH, Brilhante V, Ojanen S, Hinterding HM, Palin E, Alastalo TP, Koskenvuo J, Hiippala A, Jokinen E, Jahnukainen T, Lohi J, Pihkala J, Tyni TA, Carroll CJ, Suomalainen A. **Genetic basis of severe childhood-onset cardiomyopathies.** J Am Coll Cardiol. 2018 Nov 6;72(19):2324-2338. doi:10.1016/j.jacc.2018.08.2171. *(PubMed Abstract)*

NEW Kim J, Lee K, Fujioka H, Tandler B, Hoppel CL. **Cardiac mitochondrial structure and function in *tafazzin*-knockdown mice.** Mitochondrion. 2018 Oct 30. pii: S1567-7249(18)30074-6. doi: 10.1016/j.mito.2018.10.005. [Epub ahead of print] *(PubMed Abstract)**▼

NEW Chowdhury A, Aich A, Jain G, Wozny K, Lüchtenborg C, Hartmann M, Bernhard O, Balleiniger M, Alfar EA, Zieseniss A, Toischer K, Guan K, Rizzoli SO, Brügger B, Fischer A, Katschinski DM, Rehling P, Dudek J. **Defective mitochondrial cardiolipin remodeling dampens HIF-1α expression in hypoxia.** Cell Rep. 2018 Oct 16;25(3):561-570.e6. doi:10.1016/j.celrep.2018.09.057. *(PubMed – Open Access)*▼

NEW Hirono K, Hata Y, Nakazawa M, Momoi N, Tsuji T, Matsuoka T, Ayusawa M, Abe Y, Hayashi T, Tsujii N, Abe T, Sakaguchi H, Wang C, Takasaki A, Takarada S, Okabe M, Miyao N, Nakaoka H, Ibuki K, Saito K, Ozawa S, Nishida N, Bowles NE, Ichida F. **Clinical and echocardiographic impact of tafazzin variants on dilated cardiomyopathy phenotype in left ventricular non-compaction patients in early infancy.** Circ J. 2018 Aug 18. doi: 10.1253/circj.CJ-18-0470. [Epub ahead of print] *(PubMed Abstract - Open Access)*

NEW Paradies G, Paradies V, Ruggiero FM, Petrosillo G. **Mitochondrial bioenergetics and cardiolipin alterations in myocardial ischemia/reperfusion injury. Implications for pharmacological cardioprotection.** Am J Physiol Heart Circ Physiol. 2018 Aug 10. doi: 10.1152/ajpheart.00028.2018. [Epub ahead of print] *(PubMed Abstract)*

NEW Suzuki-Hatano S, Saha M, Rizzo SA, Witko RL, Gosiker BJ, Ramanathan M, Soustek MS, Jones MD, Kang PB, Byrne BJ, Cade WT, Pacak CA. **AAV-mediated *TAZ* gene replacement restores mitochondrial and cardioskeletal function in Barth syndrome.** Hum Gene Ther. 2018 Aug 2. doi: 10.1089/hum.2018.020. [Epub ahead of print]*

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Johnson JM, Ferrara PJ, Verkerke ARP, Coleman CB, Wentzler EJ, Neufer PD, Kew KA, de Castro Brás LE, Funai K. **Targeted overexpression of catalase to mitochondria does not prevent cardioskeletal myopathy in Barth syndrome.** J Mol Cell Cardiol. 2018 Jul 2;121:94-102. doi: 10.1016/j.yjmcc.2018.07.001. [Epub ahead of print] *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Cade WT, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, Bashir A, Byrne BJ, Taylor CL. **Peak oxygen uptake (VO2peak) across childhood, adolescence and young adulthood in Barth syndrome: Data from cross-sectional and longitudinal studies.** PLoS One. 2018 May 24;13(5):e0197776. doi: 10.1371/journal.pone.0197776. eCollection 2018 *(PubMed - Open Access)** ▼

- Schafer C, Moore V, Dasgupta N, Javadov S, James JF, Glukhov AI, Strauss AW, Khuchua Z. **The effects of PPAR stimulation on cardiac metabolic pathways in Barth syndrome mice.** Front Pharmacol. 2018 Apr 11;9:318. doi:10.3389/fphar.2018.00318. eCollection 2018. *(PubMed Abstract)**

- Kimura T, Kimura A, Ren M, Berno B, Xu Y, Schlame M, Epand RM. **Substantial decrease in plasmalogen in the heart associated with *tafazzin* deficiency.** Biochemistry. 2018 Mar 20. doi: 10.1021/acs.biochem.8b00042. [Epub ahead of print] *(PubMed Abstract)* ▼

- Tsujii N, Hayashi T, Hayashi T, Kimura A, Nishikubo T. **Barth syndrome associated with triple mutation.** Pediatr Int. 2018 Mar 6. doi:10.1111/ped.13517. [Epub ahead of print]. *(PubMed - No abstract available)*

- Seo SH, Kim SY, Cho SI, Park H, Lee S, Choi JM, Kim MJ, Lee JS, Ahn KJ, Song MK, Bae EJ, Park SS, Seong MW. **Application of multigene panel sequencing in patients with prolonged rate-corrected QT interval and no pathogenic variants detected in KCNQ1, KCNH2, and SCN5A.** Ann Lab Med. 2018 Jan;38(1):54-58. doi: 10.3343/alm.2018.38.1.54. *(PubMed - Open Access)*

- Imai-Okazaki A, Kishita Y, Kohda M, Yatsuka Y, Hirata T, Mizuno Y, Harashima H, Hirono K, Ichida F, Noguchi A, Yoshida M, Tokorodani C, Nishiuchi R, Takeda A, Nakaya A, Sakata Y, Murayama K, Ohtake A, Okazaki Y. **Barth syndrome: Different approaches to diagnosis.** J Pediatr. 2017 Dec 15. pii: S0022-3476(17)31329-X. doi: 10.1016/j.jpeds.2017.09.075. [Epub ahead of print] *(PubMed Abstract)*

- Lorca R, Rozado J, Martin M. **Miocardiopatía no compactada: Breve revisión de una miocardiopatía con controversias (Non compaction cardiomyopathy: A short review of a controversial entity.)** Medicina Clinica Nov 2017. *(ScienceDirect Abstract)*

- Dinca AA, Chien WM, Chin MT. **Identification of novel mitochondrial localization signals in human *Tafazzin*, the cause of the inherited cardiomyopathic disorder Barth syndrome.** J Mol Cell Cardiol. 2017 Nov 9. pii: S0022-2828(17)30340-1. doi:10.1016/j.yjmcc.2017.11.005. [Epub ahead of print] *(PubMed Abstract)**

- Giacomelli E, Mummery CL, Bellin M. **Human heart disease: Lessons from human pluripotent stem cell-derived cardiomyocytes.** Cell Mol Life Sci. 2017 Oct;74(20):3711-3739. doi:10.1007/s00018-017-2546-5. Epub 2017 Jun 1. Review. *(PubMed - Open Access)*

- Enns GM. **Pediatric mitochondrial diseases and the heart.** Curr Opin Pediatr. 2017 Oct;29(5):541-551. doi: 10.1097/MOP.0000000000000535. *(PubMed Abstract)*

- Feingold B, Mahle WT, Auerbach S, Clemens P, Domenighetti AA, Jefferies JL, Judge DP, Lal AK, Markham LW, Parks WJ, Tsuda T, Wang PJ, Yoo SJ; American Heart Association Pediatric Heart Failure Committee of the Council on Cardiovascular Disease in the Young; Council on Clinical Cardiology; Council on Cardiovascular Radiology and Intervention; Council on Functional Genomics and Translational Biology; and Stroke Council. **Management of cardiac involvement associated with neuromuscular diseases: A scientific statement from the American Heart Association.** Circulation. 2017 Sep 26;136(13):e200-e231. doi: 10.1161/CIR.0000000000000526. Epub 2017 Aug 24. *(PubMed Abstract)*

- Towbin JA, Jefferies JL. **Cardiomyopathies due to left ventricular noncompaction, mitochondrial and storage diseases, and inborn errors of metabolism.** Circ Res. 2017 Sep 15;121(7):838-854. doi: 10.1161/CIRCRESAHA.117.310987. Review. *(PubMed Abstract)*

- Wang C, Hata Y, Hirono K, Takasaki A, Ozawa SW, Nakaoka H, Saito K, Miyao N, Okabe M, Ibuki K, Nishida N, Origasa H, Yu X, Bowles NE, Ichida F; for LVNC Study Collaborators. **A wide and specific spectrum of genetic variants and genotype-phenotype correlations revealed by next-generation sequencing in patients with left ventricular noncompaction.** J Am Heart Assoc. 2017 Aug 30;6(9). pii: e006210. doi:10.1161/JAHA.117.006210. *(PubMed Abstract)*

- Zhao Y, Feng Y, Ding X, Dong S, Zhang H, Ding J, Xia X. **Identification of a novel hypertrophic cardiomyopathy-associated mutation using targeted next-generation sequencing.** Int J Mol Med. 2017 Jul;40(1):121-129. doi: 10.3892/ijmm.2017.2986. Epub 2017 May 11. *(PubMed - Open Access)*

- Sullivan EM, Fix A, Crouch MJ, Sparagna GC, Zeczycki TN, Brown DA, Shaikh SR. **Murine diet-induced obesity remodels cardiac and liver mitochondrial phospholipid acyl chains with differential effects on respiratory enzyme activity.** J Nutr Biochem. 2017 Jul;45:94-103. doi: 10.1016/j.jnutbio.2017.04.004. Epub 2017 Apr 12. *(PubMed Abstract)*

- Sheeran FL, Pepe S. **Mitochondrial bioenergetics and dysfunction in failing heart.** Adv Exp Med Biol. 2017;982:65-80. doi: 10.1007/978-3-319-55330-6_4. (*PubMed Abstract)*

- Finsterer J, Stollberger C. **Acquired noncompaction in Barth syndrome due to the *TAZ* mutation c.481_482ins20.** J Pediatr. 2017 Mar 15. pii: S0022-3476(17)30332-3. doi: 10.1016/j.jpeds.2017.02.066. [Epub ahead of print] (*PubMed Abstract)*

- Bakšiene M, Benušiene E, Morkuniene A, Ambrozaityte L, Utkus A, Kucinskas V. **A novel intronic splice site *tafazzin* gene mutation detected prenatally in a family with Barth syndrome.** Balkan J Med Genet. 2017 Mar 8;19(2):95-100. doi: 10.1515/bjmg-2016-0043. *(PubMed - Open Access)*

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access)**

- Lloyd DF, Vara R, Mathur S. **The cardiac manifestations of inherited metabolic diseases in children.** Pediatr Int. 2017 Mar 4. doi: 10.1111/ped.13272. [Epub ahead of print] Review. (*PubMed Abstract)*

- Bashir A, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, de Las Fuentes L, Pacak CA, Byrne BJ, Cade WT**. Impaired cardiac and skeletal muscle bioenergetics in children, adolescents, and young adults with Barth syndrome.** Physiol Rep. 2017 Feb;5(3). pii: e13130. doi: 10.14814/phy2.13130. *(PubMed – Open Access)*▼

- Wang J, Guo Y, Huang M, Zhang Z, Zhu J, Liu T, Shi L, Li F, Huang H, Fu L. **Identification of *TAZ* mutations in pediatric patients with cardiomyopathy by targeted next-generation sequencing in a Chinese cohort.** Orphanet J Rare Dis. 2017 Feb 10;12(1):26. doi: 10.1186/s13023-016-0562-4. *(PubMed – Open Access)*

- Dudek J, Maack C. **Barth syndrome cardiomyopathy.** Cardiovasc Res. 2017 Feb 2. doi: 10.1093/cvr/cvx014. [Epub ahead of print] No abstract available. *(PubMed)*

- Borna NN, Kishita Y, Ishikawa K, Nakada K, Hayashi JI, Tokuzawa Y, Kohda M, Nyuzuki H, Yamashita-Sugahara Y, Nasu T, Takeda A, Murayama K, Ohtake A, Okazaki Y. **A novel mutation in *TAZ* causes mitochondrial respiratory chain disorder without cardiomyopathy.** J Hum Genet. 2017 Jan 26. doi: 10.1038/jhg.2016.165. [Epub ahead of print] *(PubMed Abstract)*

- Woiewodski L, Ezon D, Cooper J, Feingold B. **Barth syndrome with late-onset cardiomyopathy: A missed opportunity for diagnosis.** The Journal of Pediatrics, In Press, Corrected Proof, Available online 18 January 2017. *(Abstract)*

- Ikon N, Ryan RO. **Barth syndrome: Connecting cardiolipin to cardiomyopathy**. Lipids. 2017 Jan 9. doi: 10.1007/s11745-016-4229-7. [Epub ahead of print] *(PubMed Abstract)*

- Avdjieva-Tzavella DM, Todorova AP, Kathom H M, Ivanova MB, Yordanova IT, Todorov TP, Litvinenko IO, Dasheva-Dimitrova AT, Tincheva RS. **Barth syndrome in male and female siblings caused by a novel mutation in the *TAZ* gene.** Genet Couns. 2016;27(4):495-501. *(PubMed Abstract)*

- Dong X, Fan P, Tian T, Yang Y, Xiao Y, Yang K, Liu Y, Zhou X. **Recent advancements in the molecular genetics of left ventricular noncompaction cardiomyopathy.** Clin Chim Acta. 2016 Dec 15;465:40-44. doi:10.1016/j.cca.2016.12.013. [Epub ahead of print] Review. *(PubMed Abstract)*

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)**▼

- Oláhová M, Thompson K, Hardy SA, Barbosa IA, Besse A, Anagnostou ME, White K, Davey T, Simpson MA, Champion M, Enns G, Schelley S, Lightowlers RN, Chrzanowska-Lightowlers ZM, McFarland R, Deshpande C, Bonnen PE, Taylor RW. **Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria.** J Inherit Metab Dis. 2016 Sep 30. *(PubMed Abstract)*

- Pérez-Serra A, Toro R, Sarquella-Brugada G, de Gonzalo-Calvo D, Cesar S, Carro E, Llorente-Cortes V, Iglesias A, Brugada J, Brugada R, Campuzano O. **Genetic basis of dilated cardiomyopathy.** International Journal of Cardiology, Available online 21 September 2016. *(ScienceDirect Abstract)*

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review. *(PubMed Abstract)*

- Jang S, Lewis TS, Powers C, Khuchua Z, Baines CP, Wipf P, Javadov S. **Elucidating mitochondrial ETC supercomplexes in the heart during ischemia-reperfusion.** Antioxid Redox Signal. 2016 Sep 7. [Epub ahead of print] *(PubMed Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- El-Hattab AW, Scaglia F. **Mitochondrial cardiomyopathies.** Front Cardiovasc Med. 2016 Jul 25;3:25. doi: 10.3389/fcvm.2016.00025. eCollection 2016. *(PubMed - Open Access)*

- Yoo TY, Kim MR, Son JS, Lee R, Bae SH, Chung S, Kim KS, Seong MW, Park SS. **Identification of a novel de novo mutation of the *TAZ* gene in a Korean patient with Barth syndrome.** J Cardiovasc Ultrasound. 2016 Jun;24(2):153-7. doi: 10.4250/jcu.2016.24.2.153. Epub 2016 Jun 22. *(PubMed - Open Access)*

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)**▼

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Szczepanek K, Allegood J, Aluri H, Hu Y, Chen Q, Lesnefsky EJ. **Acquired deficiency of *tafazzin* in the adult heart: Impact on mitochondrial function and response to cardiac injury.** Biochim Biophys Acta. 2016 Apr;1861(4):294-300. doi: 10.1016/j.bbalip.2015.12.004. Epub 2015 Dec 12. *(PubMed Abstract)*

- Kokoszka JE, Waymire KG, Flierl A, Sweeney KM, Angelin A, MacGregor GR, Wallace DC. **Deficiency in the mouse mitochondrial adenine nucleotide translocator isoform 2 gene is associated with cardiac noncompaction.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Available online 24 April 2016. *(ScienceDirect Abstract)*

- Chanana AM, Rhee JW, Wu JC. **Human-induced pluripotent stem cell approaches to model inborn and acquired metabolic heart diseases.** Curr Opin Cardiol. 2016 Mar 26. [Epub ahead of print] *(PubMed Abstract)*

- Finsterer J, Stöllberger C. **Heart disease in disorders of muscle, neuromuscular transmission, and the nerves.** Korean Circ J. 2016 Mar;46(2):117-34. doi: 10.4070/kcj.2016.46.2.117. Epub 2016 Mar 21. Review. *(PubMed Abstract)*

- Dolinsky VW, Cole LK, Sparagna GC, Hatch GM. **Cardiac mitochondrial energy metabolism in heart failure: Role of cardiolipin and sirtuins.** Biochimica et Biophysica Acta (BBA) - Molecular and Cell Biology of Lipids, Available online 10 March 2016. *(ScienceDirect Abstract)*

- Thompson WR, DeCroes B, McClellan R, Rubens J, Vaz FM, Kristaponis K, Avramopoulos D, Vernon HJ. **New targets for monitoring and therapy in Barth syndrome.** Genet Med. 2016 Feb 4. doi: 10.1038/gim.2015.204. [Epub ahead of print] *(PubMed Abstract)*▼

- Lal AK, Pruitt E, Hong BJ, Lin KY, Feingold B. **Left ventricular non-compaction cardiomyopathy in children listed for heart transplant: Analysis from the Pediatric Heart Transplant Study Group.** J Heart Lung Transplant. 2016 Apr;35(4):540-2. doi:10.1016/j.healun.2015.12.010. Epub 2016 Jan 6. *(PubMed - No abstract available)*

- Dudek J, Cheng IF, Chowdhury A, Wozny K, Balleininger M, Reinhold R, Grunau S, Callegari S, Toischer K, Wanders RJ, Hasenfuß G, Brügger B, Guan K, Rehling P. **Cardiac-specific succinate dehydrogenase deficiency in Barth syndrome.** EMBO Mol Med. 2015 Dec 23. pii: e201505644. doi: 10.15252/emmm.201505644. [Epub ahead of print] *(PubMed – Open Access)*▼

- Denning C, Borgdorff V, Crutchley J, Firth KSA, George V, Kalra S, Kondrashov A, Hoang MD, Mosqueira D, Patel A, Prodanov L, Rajamohan D, Skarnes WC, Smith JGW, Young LE. **Cardiomyocytes from human pluripotent stem cells: From laboratory curiosity to industrial biomedical platform.** Original Research Article. Biochimica et Biophysica Acta (BBA) - Molecular Cell Research, Available online 31 October 2015. *(ScienceDirect Abstract)*

- Mathur A, Ma Z, Loskill P, Jeeawoody S, Healy KE. ***In vitro* cardiac tissue models: Current status and future prospects.** Adv Drug Deliv Rev. 2015 Sep 30. pii: S0169-409X(15)00214-8. doi: 10.1016/j.addr.2015.09.011. [Epub ahead of print] Review. *(PubMed Abstract)*▼

- Gaspard GJ, McMaster CR. **Cardiolipin metabolism and its causal role in the etiology of the inherited cardiomyopathy Barth syndrome.** Chem Phys Lipids. 2015 Sep 25. pii: S0009-3084(15)30054-2. doi:10.1016/j.chemphyslip.2015.09.005. [Epub ahead of print] Review. *(PubMed Abstract)**

BSF does not endorse any drugs, tests, or treatments that we may report.

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents *TAZ* knockdown-induced apoptosis in myeloid progenitor cells.** Biochem Biophys Res Commun. 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print] *(PubMed Abstract)**

- Nagueh SF, Zoghbi WA. **Role of imaging in the evaluation of patients at risk for sudden cardiac death: Genotype–phenotype intersection.** Review Article. JACC: Cardiovascular Imaging, Volume 8, Issue 7, July 2015, Pages 828-845. *(ScienceDirect Abstract)*

- Tocchi A, Quarles EK, Basisty N, Gitari L, Rabinovitch PS. **Mitochondrial dysfunction in cardiac aging.** Biochim Biophys Acta. 2015 Jul 17. pii: S0005-2728(15)00152-8. doi: 10.1016/j.bbabio.2015.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Angelini R, Lobasso S, Gorgoglione R, Bowron A, Steward CG, Corcelli A. **Cardiolipin fingerprinting of leukocytes by MALDI-TOF/MS as screening tool for Barth syndrome.** J Lipid Res. 2015 Jul 5. pii:jlr.D059824. [Epub ahead of print] *(PubMed – Open Access)**▼

- Ryan RO. **Metabolic annotation of 2-ethylhydracrylic acid.** Clin Chim Acta. 2015 Jun 23. pii: S0009-8981(15)00295-8. doi:10.1016/j.cca.2015.06.012. [Epub ahead of print] Review. *(PubMed Abstract)**

- Huang Y, Powers C, Madala SK, Greis KD, Haffey WD, Towbin JA, Purevjav E, Javadov S, Strauss AW, Khuchua Z. **Cardiac metabolic pathways affected in the mouse model of Barth syndrome.** PLoS One. 2015 Jun 1;10(6):e0128561. doi: 10.1371/journal.pone.0128561. eCollection 2015. *(PubMed – Open Access)*

- Kamdar F, Klaassen Kamdar A, Koyano-Nakagawa N, Garry MG, Garry DJ. **Cardiomyopathy in a dish: Using human inducible pluripotent stem cells to model inherited cardiomyopathies.** Journal of Cardiac Failure (2015), doi: 10.1016/j.cardfail.2015.04.010. (*Abstract)*

- Miyake CY, Kim JJ. **Arrhythmias in left ventricular noncompaction.** Review Article. Cardiac Electrophysiology Clinics, Available online 10 April 2015. *(ScienceDirect Abstract)*

- Towbin JA, Lorts A, Jefferies JL. **Left ventricular non-compaction cardiomyopathy.** Review Article. The Lancet, Available online 9 April 2015. *(ScienceDirect Abstract)*

- Lakdawala NK. **Big data for a rare disease: Examining heart transplantation for left ventricular noncompaction in the UNOS registry.** J Heart Lung Transplant, Available online 4 April 2015. *(ScienceDirect Abstract)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Wegener M, Bader A, Giri S**. How to mend a broken heart: Adult and induced pluripotent stem cell therapy for heart repair and regeneration.** Review Article. Drug Discovery Today, Available online 23 February 2015. *(ScienceDirect Abstract)*

- Russell M, Roberts AE, Abrams DJ, Murphy AM, Towbin JA, Chung WK. **How to effectively utilize genetic testing in the care of children with cardiomyopathies.** Progress in Pediatric Cardiology, Available online 31 January 2015. *(ScienceDirect Abstract)*

- Carrilho-Ferreira P, Almeida AG, Pinto FJ. **Non-compaction cardiomyopathy: Prevalence, prognosis, pathoetiology, genetics, and risk of cardioembolism.** Curr Heart Fail Rep. 2014 Dec;11(4):393-403. doi: 10.1007/s11897-014-0227-3. *(PubMed Abstract)*

- Tariq M, Ware SM. **Importance of genetic evaluation and testing in pediatric cardiomyopathy.** World J Cardiol 2014 Nov 26;6(11):1156-1165. Review. *(PubMed – Open Access)*

- Ikeda U, Minamisawa M, Koyama J. **Isolated left ventricular non-compaction cardiomyopathy in adults.** J Cardiol. 2014 Oct 31. pii: S0914-5087(14)00297-4. doi: 10.1016/j.jjcc.2014.10.005. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jiang W, Lan F, Zhang H. **Human induced pluripotent stem cell models of inherited cardiovascular diseases.** Curr Stem Cell Res Ther. 2014 Oct 16. [Epub ahead of print] *(PubMed Abstract)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)* ▼

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Zhao Y, Feric NT, Thavandiran N, Nunes SS, Radisic M. **The role of tissue engineering and biomaterials in cardiac regenerative medicine.** Can J Cardiol. 2014 Nov;30(11):1307-1322. doi: 10.1016/j.cjca.2014.08.027. Epub 2014 Sep 4. Review. *(PubMed Abstract)*

- Zhao Q, Cai H, Zhan Y, Li B, Sun N. **Applications of human induced pluripotent stem cells in the investigation of inherited cardiomyopathy.** Int J Cardiol. 2014 Dec 15;177(2):604-6. doi: 10.1016/j.ijcard.2014.08.135. Epub 2014 Aug 30. *(PubMed – No Abstract Available)*

- He Q, Harris N, Ren J, Han X. **Mitochondria-targeted antioxidant prevents cardiac dysfunction induced by *tafazzin* gene knockdown in cardiac myocytes.** Oxid Med Cell Longev. 2014;2014:654198. Epub 2014 Aug 27. *(PubMed – Open Access)**

- Gilbert-Barness E. **Conduction defects/cardiomyopathies.** Adv Pediatr. 2014 Aug;61(1):127-48. doi: 10.1016/j.yapd.2014.03.001. No abstract available. *(PubMed – No Abstract Available)*

- Zweigerdt R, Gruh I, Martin U. **Your heart on a chip: iPSC-based modeling of Barth-syndrome-associated cardiomyopathy.** Cell Stem Cell. 2014 Jul 3;15(1):9-11. doi: 10.1016/j.stem.2014.06.015. *(PubMed – Open Access)*

- Chatfield KC, Sparagna GC, Sucharov CC, Miyamoto SD, Grudis JE, Sobus RD, Hijmans J, Stauffer BL. **Dysregulation of cardiolipin biosynthesis in pediatric heart failure**. J Mol Cell Cardiol. 2014 Jun 14. pii: S0022-2828(14)00196-5. doi:10.1016/j.yjmcc.2014.06.002. [Epub ahead of print] *(PubMed Abstract)*

- Raval KK, Kamp TJ. **Cardiomyopathy, mitochondria and Barth syndrome: iPSCs reveal a connection.** Nat Med. 2014 Jun 5;20(6):585-6. doi: 10.1038/nm.3592. *(PubMed – No Abstract Available).*

- Folsi V, Miglietti N, Lombardi A, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, Plebani A. **Cardiomyopathy in a male patient with neutropenia and growth delay.** Ital J Pediatr. 2014 May 12;40:45. doi:10.1186/1824-7288-40-45. *(PubMed – Open Access)*

- Richter-Dennerlein R, Korwitz A, Haag M, Tatsuta T, Dargazanli S, Baker M, Decker T, Lamkemeyer T, Rugarli EI, Langer T. **DNAJC19, a mitochondrial cochaperone associated with cardiomyopathy, forms a complex with prohibitins to regulate cardiolipin remodeling.** Cell Metab. 2014 May 21. pii: S1550-4131(14)00182-X. doi: 10.1016/j.cmet.2014.04.016. [Epub ahead of print] *(PubMed Abstract)*

- Wang G, McCain ML, Yang L, He A, Pasqualini FS, Agarwal A, Yuan H, Jiang D, Zhang D, Zangi L, Geva J, Roberts AE, Ma Q, Ding J, Chen J, Wang DZ, Li K, Wang J, Wanders RJA, Kulik W, Vaz FM, Laflamme MA, Murry CE, Chien KR, Kelley RI, Church GM, Parker K, Pu WT. **Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies.** Nat Med. 2014 Jun;20(6):616-23. doi:10.1038/nm.3545 Epub. 2014 May 11. *(PubMed – Open Access)*▼

- Mulligan CM, Le CH, Demooy AB, Nelson CB, Chicco AJ. **Inhibition of delta-6 desaturase reverses cardiolipin remodeling and prevents contractile dysfunction in the aged mouse heart without altering mitochondrial respiratory function.** J Gerontol A Biol Sci Med Sci. 2014 Jan 18. [Epub ahead of print] *(PubMed Abstract)*

- Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J. **The MOGE(S) classification for a phenotype–genotype nomenclature of cardiomyopathy: Endorsed by the World Heart Federation.** Journal of the American College of Cardiology, Volume 62, Issue 22, 3 December 2013, Pgs. 2046-2072 - CORRECTION. *(ScienceDirect Abstract)*

- Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J. **The MOGE(S) classification for a phenotype–genotype nomenclature of cardiomyopathy: Endorsed by the World Heart Federation.** Journal of the American College of Cardiology, Available online 18 November 2013. *(ScienceDirect Abstract)*

- Digilio MC, Bernardini L, Gagliardi MG, Versacci P, Baban A, Capolino R, Dentici ML, Roberti MC, Angioni A, Novelli A, Marino b, Dallapiccola B. **Syndromic non-compaction of the left ventricle: Associated chromosomal anomalies.** Clinical Genet 2013:84:362-367. *(PubMed Abstract)*

- Finsterer J, Stöllberger C. **Unclassified cardiomyopathies in neuromuscular disorders.** Wien Med Wochenschr. 2013 Oct 24. [Epub ahead of print] *(PubMed Abstract)*

- Mazurová S, Tesarová M, Magner M, Houštková H, Hansíková H, Augustínová J, Tomek V, Vondrácková A, Zeman J, Honzík T. **Novel mutations in the *TAZ* gene in patients with Barth syndrome.** Prague Med Rep. 2013;114(3):139-153. (*PubMed Abstract)*

- Finsterer J, Stöllberger C. **Ultrastructural findings in noncompaction prevail with neuromuscular disorders.** Cardiology. 2013 Sep 21;126(4):219-223. [Epub ahead of print] (*PubMed Abstract)*

- Agarwal A, Khandheria BK, Paterick TE, Treiber SC, Bush M, Tajik AJ. **Left ventricular noncompaction in patients with bicuspid aortic valve.** J Am Soc Echocardiogr. 2013 Sep 14. doi:pii: S0894-7317(13)00629-9. 10.1016/j.echo.2013.08.003. [Epub ahead of print] (*PubMed Abstract)*

- Jefferies JL. **Barth syndrome.** Am J Med Genet C Semin Med Genet. 2013 Jul 10. doi: 10.1002/ajmg.c.31372. [Epub ahead of print] *(PubMed – Open Access)*

- Monteiro JP, Oliveira PJ, Jurado AS. **Mitochondrial membrane lipid remodeling in pathophysiology: A new target for diet and therapeutic interventions.** Prog Lipid Res. 2013 Jul 1. doi:pii: S0163-7827(13)00042-8. 10.1016/j.plipres.2013.06.002. [Epub ahead of print] (*PubMed Abstract)*

- Dudek J, Cheng IF, Balleininger M, Vaz FM, Streckfuss-Bömeke K, Hübscher D, Vukotic M, Wanders RJ, Rehling P, Guan K. **Cardiolipin deficiency affects respiratory chain function and organization in an induced pluripotent stem cell model of Barth syndrome.** Stem Cell Res. 2013 Sep;11(2):806-19. doi: 10.1016/j.scr.2013.05.005. Epub 2013 May 28. *(PubMed – Open Access)*▼

- Kim GB, Kwon BS, Bae EJ, Noh CI, Seong MW, Park SS. **A novel mutation of the *TAZ* gene in Barth syndrome: Acute exacerbation after contrast-dye injection.** J Korean Med Sci. 2013 May;28(5):784-7. doi: 10.3346/jkms.2013.28.5.784. Epub 2013 May 2. (*PubMed – Open Access)*

- Rigaud C, Lebre A, Touraine R, Beaupain B, Ottolenghi C, Chabli A, Ansquer H, Ozsahin H, Di Filippo S, De Lonlay P, Borm B, Rivier F, Vaillant M, Mathieu-Dramard M, Goldenberg A, Viot G, Charron P, Rio M, Bonnet D, Donadieu J. **Natural history of Barth syndrome: A national cohort study of 22 patients.** Orphanet J Rare Dis. 2013 May 8;8:70. doi: 10.1186/1750-1172-8-70. *(PubMed – Open Access)**▼

- Grudis JE, Chatfield KC, Sparagna GC, Hijmans J, Sobus RD, Miyamoto SD, Stauffer BL. **Expression of cardiolipin biosynthesis and remodeling enzymes in adult heart failure.** J Heart and Lung Transplantation, April 2013, Vol. 32, Iss 4, Pgs. S97-S98. *(ScienceDirect Abstract)*

- Finsterer J, Stöllberger C, Blazek G, Sehnal E. **Familal left ventricular hypertrabeculation (noncompaction) is myopathic.** Int J Cardiol. 2013 Apr 15;164(3):312-7. doi:10.1016/j.ijcard.2011.07.012. Epub 2011 Jul 23. *(PubMed Abstract)*

- Fan Y, Steller J, Gonzalez IL, Kulik W, Fox M, Chang R, Westerfield BA, Batra AS, Wang RY, Gallant NM, Pena LS, Wang H, Huang T, Bhuta S, Penny DJ, McCabe ER, Kimonis VE. **A novel exonic splicing mutation in the *TAZ* (G4.5) gene in a case with atypical Barth syndrome.** JIMD Rep. 2013 Apr 19. [Epub ahead of print] *(PubMed – Open Access)*

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth syndrome in adulthood: A clinical case.** [Article in English, Spanish] Rev Esp Cardiol. 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. *(PubMed Abstract)*

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Teekakirikul P, Kelly MA, Rehm HL, Lakdawala NK, Funke BH. **Inherited cardiomyopathies: Molecular genetics and clinical genetic testing in the postgenomic era.** J Mol Diagn. 2013 Mar;15(2):158-70. doi: 10.1016/j.jmoldx.2012.09.002. Epub 2012 Dec 27. *(PubMed Abstract)*

- Finsterer J, Stöllberger C, Wahbi K. **Cardiomyopathy in neurological disorders.** Cardiovascular Pathology (2013), doi.org/10.1016/j.carpath.2012.12.008 *(ScienceDirect Abstract)*

- Finsterer J, Stöllberger C, Kovacs GG, Sehnal E. **Left ventricular hypertrabeculation/noncompaction coincidentally found in sporadic inclusion body myositis.** Int J Cardiol 2013 Sep 20;168(1):610-2. doi.org/10/1016/j.ijcard2013.01.221. Epub 2013 Feb 22. *(PubMed Abstract)*

- Liu S, Bai Y, Huang J, Zhao H, Zhang X, Hu S, Wei Y. **Do mitochondria contribute to left ventricular non-compaction cardiomyopathy? New findings from myocardium of patients with left ventricular non-compaction cardiomyopathy.** Mol Genet Metab. 2013 May;109(1):100-6. doi: 10.1016/j.ymgme.2013.02.004. Epub 2013 Feb 13. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini RR, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 14;8(1):27. doi: 10.1186/1750-1172-8-27. *(PubMed Abstract)*▼

- Kiebish MA, Yang K, Liu X, Mancuso DJ, Guan S, Zhao Z, Sims HF, Cerqua R, Cade WT, Han X, Gross RW. **Dysfunctional cardiac mitochondrial bioenergetic, lipidomic, and signaling in a murine model of Barth syndrome.** J Lipid Res. 2013 Feb 14. [Epub ahead of print] *(PubMed – Open Access)**

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Day TG, Fenton M. **Dilated cardiomyopathy in children.** Paediatrics and Child Health Volume 23, Issue 2, February 2013, Pages 59–63. *(SciVerse Abstract)*

- Pulgaron ER, Wile D, Schneider K, Young ML, Delamater AM. **Quality of life and psychosocial functioning of children with cardiac arrhythmias.** Cardiol Young. 2013 Feb;23(1):82-8. doi: 10.1017/S1047951112000388. Epub 2012 Apr 18. *(PubMed Abstract)*

- Kohlmann S, Kilbert MS, Ziegler K, Schulz KH. **Supportive care needs in patients with cardiovascular disorders.** Patient Education and Counseling xxx (2013) xxx-xxx. *(Abstract)*

- Mestroni L, Taylor MRG. **Genetics and genetic testing of dilated cardiomyopathy: A new perspective.** Discov Med 2013 Jan;15(80):43-49. *(PubMed – Open Access)*

- Ware SM, Towbin JA. **Nuclear genes causing mitochondrial cardiomyopathy.** Mitochondrial Disorders Caused by Nuclear Genes, 2013, pp. 319-335. *(Springer Abstract)*

- Karkucinska-Wieckowska A, Trubicka J, Werner B, Kokoszynska K, Pajdowska M, Pronicki M, Czarnowska E, Lebiedzinska M, Sykut-Cegielska J, Ziolkowska L, Jaron W, Dobrzanska A, Ciara E, Wieckowski MR, Pronicka E. **Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome.** J Inherit Metab Dis. 2013 Jan 30. [Epub ahead of print] *(PubMed Abstract)*

- Man E, Lafferty KA, Funke BH, Lun KS, Chan SY, Chau AK, Chung BH. **NGS identifies *TAZ* mutation in a family with X-linked dilated cardiomyopathy.** BMJ Case Rep. 2013 Jan 22;2013. doi:pii: bcr2012007529. 10.1136/bcr-2012-007529. *(PubMed Abstract)*

- Lopes LR, Elliott PM. **Genetics of heart failure.** Biochim Biophys Acta. 2013 Jan 5. doi:pii: S0925-4439(13)00002-1. 10.1016/j.bbadis.2012.12.012. [Epub ahead of print] *(PubMed Abstract)*

- Piga A, Longo F, Musallam KM, Veltri A, Ferroni F, Chiribiri A, Bonamini R. **Left ventricular noncompaction in patients with β-thalassemia: Uncovering a previously unrecognized abnormality.** Am J Hematol. 2012 Dec;87(12):1079-83. doi: 10.1002/ajh.23323. Epub 2012 Sep 11. *(PubMed Abstract)*

- Whited K, Baile MG, Currier P, Claypool SM. **Seven functional classes of Barth syndrome mutation.** Hum Mol Genet. 2013 Feb 1;22(3):483-92. doi: 10.1093/hmg/dds447. Epub 2012 Oct 24. *(PubMed Abstract)*

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth Syndrome in adulthood: A clinical case.** Rev Esp Cardiol (Engl Ed). 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. (*PubMed Abstract*)

- Roberts AE, Nixon C, Steward CG, Gauvreau K, Maisenbacher M, Fletcher M, Geva J, Byrne BJ, Spencer CT. **The Barth Syndrome Registry: Distinguishing disease characteristics and growth data from a longitudinal study.** Am J Med Genet A. 2012 Nov;158A(11):2726-32. doi: 10.1002/ajmg.a.35609. Epub 2012 Oct 8. *(PubMed – Open Access)**▼

- Ojala T, Polinati P, Manninen T, Hiippala A, Rajantie J, Karikoski R, Suomalainen A, Tyni T. **New mutation of mitochondrial DNAJC19 causing dilated and noncompaction cardiomyopathy, anemia, ataxia, and male genital anomalies.** Pediatr Res. 2012 Oct;72(4):432-7. doi: 10.1038/pr.2012.92. Epub 2012 Jul 13. *(PubMed Abstract)*

- Ronvelia D, Greenwood J, Platt J, Hakim S, Zaragoza MV. **Intrafamilial variability for novel *TAZ* gene mutation: Barth syndrome with dilated cardiomyopathy and heart failure in an infant and left ventricular noncompaction in his great-uncle.** Mol Genet Metab. 2012 Sep 18. pii: S1096-7192(12)00354-X. doi: 10.1016/j.ymgme.2012.09.013. [Epub ahead of print] *(PubMed Abstract)*

- Liu X, Ye B, Miller S, Yuan H, Zhang H, Tian L, Nie J, Imae R, Arai H, Li Y, Cheng Z, Shi Y. **Ablation of ALCAT1 mitigates hypertrophic cardiomyopathy through effects on oxidative stress and mitophagy.** Mol Cell Biol. 2012 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Cade WT, Spencer CT, Reeds DN, Waggoner AD, O'Connor R, Maisenbacher M, Crowley JR, Byrne BJ, Peterson LR. **Substrate metabolism during basal and hyperinsulinemic conditions in adolescents and young-adults with Barth syndrome.** J Inherit Metab Dis. 2012 May 12. [Epub ahead of print] *(PubMed Abstract)**▼

- Kindel SJ, Miller EM, Gupta R, Cripe LH, Hinton RB, Spicer RL, Towbin JA, Ware SM. **Pediatric cardiomyopathy: Importance of genetic and metabolic evaluation.** J Card Fail. 2012 May;18(5):396-403. doi: 10.1016/j.cardfail.2012.01.017. Epub 2012 Mar 10. *(PubMed Abstract)*

- Phoon CKL, Acehan D, Schlame M, Stokes DL, Edelman-Novemsky I, Yu D, Xu Y, Viswanathan N, Ren M. ***Tafazzin* knockdown in mice leads to a developmental cardiomyopathy with early diastolic dysfunction preceding myocardial noncompaction**. J Am Heart Assoc. 2012 Apr;1(2). doi:pii: jah3-e000455. 10.1161/JAHA.111.000455. Epub 2012 Apr 24. *(PubMed – Open Access)* *

- Hanke SP, Gardner AB, Lombardi JP, Manning PB, Nelson DP, Towbin JA, Jefferies JL, Lorts A. **Left ventricular noncompaction cardiomyopathy in Barth syndrome: An example of an undulating cardiac phenotype necessitating mechanical circulatory support as a bridge to transplantation.** Pediatr Cardiol. 2012 Mar 17. [Epub ahead of print] *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Cosson L, Toutain A, Simard G, Kulik W, G Matyas, Guichet A, Blasco H, Maakaroun-Vermesse Z, Vaillant MC, Le Caignec C, Chantepie A, Labarthe F. **Barth syndrome in a female patient.** Mol Genet Metab. 2012 May;106(1):115-20. doi: 10.1016/j.ymgme.2012.01.015. Epub 2012 Jan 24. *(PubMed Abstract)*

- Werner B, Trubicka J, Pronicka E. **[Clinical and diagnostic aspects of Barth syndrome (X-linked cardiomyopathy)].** Kardiol Pol. 2011;69(11):1177-80. Polish.. *(PubMed – Open Access)*

- Momoi N, Chang B, Takeda I, Aoyagi Y, Endo K, Ichida F**. Differing clinical courses and outcomes in two siblings with Barth syndrome and left ventricular noncompaction.** Eur J Pediatr. 2011 Oct 7. [Epub ahead of print] *(PubMed Abstract)*

- Takeda A, Sudo A, Yamada M, Yamazawa H, Izumi G, Nishino I, Ariga T. **Barth syndrome diagnosed in the subclinical stage of heart failure based on the presence of lipid storage myopathy and isolated noncompaction of the ventricular myocardium.** Eur J Pediatr. 2011 Sep 20. [Epub ahead of print] *(PubMed Abstract)*

- Spencer CT, Byrne BJ, Bryant RM, Margossian R, Maisenbacher M, Breitenger P, Benni PB, Redfearn S, Marcus E, Cade WT. **Impaired cardiac reserve and severely diminished skeletal muscle oxygen utilization mediate exercise intolerance in Barth syndrome.** Am J Physiol Heart Circ Physiol. 2011 Nov;301(5):H2122-9. Epub 2011 Aug 26. *(PubMed – Open Access)**▼

- Jacoby D, McKenna WJ. **Genetics of inherited cardiomyopathy.** Eur Heart J. 2011 Aug 2. [Epub ahead of print] *(PubMed Abstract)*

- Taylor M, Slavov D, Salcedo E, Zhu X, Ferguson D, Jirikowic J, Di Lenarda A, Sinagra G, MD, Mestroni L. ***Tafazzin* gene mutations are uncommon causes of dilated cardiomyopathy in adults.** Cardiogenetics.2011.e4 | Published: 2011-07-05. *(Abstract)**▼

- Alvarez JA, Orav EJ, Wilkinson JD, Fleming LE, Lee DJ, Sleeper LA, Rusconi PG, Colan SD, Hsu DT, Canter CE, Webber SA, Cox GF, Jefferies JL, Towbin JA, Lipshultz SE; for the Pediatric Cardiomyopathy Registry Investigators. **Competing risks for death and cardiac transplantation in children with dilated cardiomyopathy: Results from the Pediatric Cardiomyopathy Registry.** Circulation. 2011 Jul 25. [Epub ahead of print] *(PubMed Abstract)*

- Bockeria LA, Berishvili D, Baryshnikova I. **Re: Ventricular non-compaction in children: Clinical characteristics and course.** Interact Cardiovasc Thorac Surg. 2011 Mar;12(3):373. *(PubMed Abstract)*

- Acehan D, Vaz F, Houtkooper RH, James J, Moore V, Tokunaga C, Kulik W, Wansapura J, Toth MJ, Strauss A, Khuchua Z. **Cardiac and skeletal muscle defects in a mouse model of human Barth syndrome.** J Biol Chem. 2011 Jan 14;286(2):899-908. Epub 2010 Nov 9. *(PubMed – Open Access)**

- Oechslin E, Jenni R. **Left ventricular non-compaction revisited: A distinct phenotype with genetic heterogeneity?** Eur Heart J. 2011 Jun;32(12):1446-56. Epub 2011 Jan 31. *(PubMed Abstract)*

- Schiff M, Ogier de Baulny H, Lombès A. **Neonatal cardiomyopathies and metabolic crises due to oxidative phosphorylation defects.** Semin Fetal Neonatal Med. 2011 Aug;16(4):216-21. doi: 10.1016/j.siny.2011.04.002. Epub 2011 May 24. *(PubMed Abstract)*

- Lisi EC, Cohn RD. **Genetic evaluation of the pediatric patient with hypotonia: Perspective from a hypotonia specialty clinic and review of the literature.** Dev Med Child Neurol. 2011 Jul;53(7):586-99. doi: 10.1111/j.1469-8749.2011.03918.x. Epub 2011 Mar 21. Review. *(PubMed Abstract)*

- Aljishi E, Ali F. **Barth syndrome: An X-linked cardiomyopathy with a novel mutation.** Indian J Pediatr. 2010 Dec;77(12):1432-3. Epub 2010 Oct 28. *(PubMed Abstract)*

- Hershberger RE, Morales A, Siegfried JD. **Clinical and genetic issues in dilated cardiomyopathy: A review for genetics professionals**. Genet Med. 2010 Nov;12(11):655-67. *(PubMed Abstract)*

- Wilkinson JD, Landy DC, Colan SD, Towbin JA, Sleeper LA, Oray EJ, Cox GF, Canter CE, Hsu DT, Webber SA, Lipshultz SE. **The Pediatric Cardiomyopathy Registry and heart failure: Key results from the first 15 years.** Heart Fail Clin. 2010 Oct;6(4):401-13, vii. *(PubMed Abstract)*

- Steward CG, Newbury-Ecob RA, Hastings R, Smithson SF, Tsai-Goodman B, Quarrell OW, Kulik W, Wanders R, Pennock M, Williams M, Cresswell JL, Gonzalez IL, Brennan P. **Barth syndrome: An X-linked cause of fetal cardiomyopathy and stillbirth.** Prenat Diagn. 2010 Oct;30(10):970-6. *(PubMed – Open Access)** ▼

- He Q. ***Tafazzin* knockdown causes hypertrophy of neonatal ventricular myocytes.** Am J Physiol Heart Circ Physiol. 2010 Jul;299(1):H210-6. Epub 2010 Mar 26. *(PubMed – Open Access)**

- Koh C, Lee PW, Yung TC, Lun KS, Cheung YF. **Left ventricular noncompaction in children.** Congenit Heart Dis. 2009 Jul;4(4):288-94. *(PubMed Abstract)*

- Towbin JA, Sleeper L, Jefferies JL, Colan S, Webber SA, Canter CE, Hsu DT, Ware SM, Wilkinson JD, Orav EJ, Lipshultz SE. **Genetic and viral genome analysis of childhood cardiomyopathy: The PCMR/PCSR experience** [abstract]. In American College of Cardiology 59th Annual Scientific Session; 2010 Mar 14-16; Altanta, GA: J. Am. Coll. Cardiol. 2010;55;A43.E409. *(Abstract)*

- Chang B, Momoi N, Shan L, Mitomo M, Aoyagi Y, Endo K, Takeda I, Chen R, Xing Y, Yu X, Watanabe S, Yoshida T, Kanegane H, Tsubata S, Bowles NE, Ichida F, Miyawaki T; Noncompaction study collaborators. **Gonadal mosaicism of a *TAZ* (G4.5) mutation in a Japanese family with Barth syndrome and left ventricular noncompaction.** Mol Genet Metab. 2010 Jun;100(2):198-203. Epub 2010 Mar 2. *(PubMed Abstract)*

- Zimmerman RS, Cox S, Lakdawala NK, Cirino A, Mancini-DiNardo D, Clark E, Leon A, Duffy E, White E, Baxter S, Alaamery M, Farwell L, Weiss S, Seidman CE, Seidman JG, Ho CY, Rehm HL, Funke BH. **A novel custom resequencing array for dilated cardiomyopathy.** Genet Med. 2010 May;12(5):268-78. *(PubMed Abstract)*

- Zachman DK, Chicco AJ, McCune SA, Murphy RC, Moore RL, Sparagna GC. **The role of calcium-independent phospholipase A2 in cardiolipin remodeling in the spontaneously hypertensive heart failure rat heart.** J Lipid Res. 2010 Mar;51(3):525-34. Epub 2009 Sep 9. *(PubMed – Open Access)**

- Jefferies JL, Towbin JA. **Dilated cardiomyopathy.** Lancet. 2010 Feb 27;375(9716):752-62. *(PubMed Abstract)*

- Ashworth M. **Cardiomyopathy in childhood: Histopathological and genetic features.** Pathology Journal, 2010, 4, 80-93. *(Open Access/Bentham)*

- Shah KB, Duda MK, O'Shea KM, Sparagna GC, Chess DJ, Khairallah RJ, Robillard-Frayne I, Xu W, Murphy RC, Des Rosiers C, Stanley WC. **The cardioprotective effects of fish oil during pressure overload are blocked by high fat intake. Role of cardiac phospholipid remodeling.** Hypertension. 2009 Sep;54(3):605-11. Epub 2009 Jul 13. *(PubMed Abstract)*

- Saini-Chohan HK, Holmes MG, Chicco AJ, Taylor WA, Moore RL, McCune SA, Hickson-Bick DL, Hatch GM, Sparagna GC. **Cardiolipin biosynthesis and remodeling enzymes are altered during the development of heart failure.** J Lipid Res. 2009 Aug;50(8):1600-8. Epub 2008 Nov 10. *(PubMed – Open Access)**

- Sparagna GC, Lesnefsky EJ. **Cardiolipin remodeling in the heart.** J Cardiovasc Pharmacol. 2009 Apr;53(4):290-301. Review. *(PubMed Abstract)**

- Cortez-Dias N, Varela MG, Sargento L, Brito D, Almeida A, Cerqueira R, Lança V, Fernandes AR, Tavares P, Pereira RA, Fernandes A, Madeira H. **Left ventricular non-compaction: A new mutation predisposing to reverse remodeling?** Rev Port Cardiol. 2009 Feb;28(2):185-94. *(PubMed Abstract)*

- Ichida F**. Left ventricular noncompaction.** Circ J. 2009 Jan;73(1):19-26. Epub 2008 Dec 4. Review. *(PubMed Abstract*)

- Sweeney RT, Davis GJ, Noonan JA. **Cardiomyopathy of unknown etiology: Barth syndrome unrecognized.** Congenit Heart Dis. 2008 Nov;3(6):443-8. *(PubMed Abstract)*

- McCanta AC, Chang AC, Weiner K. **Cardiomyopathy in a child with neutropenia and motor delay**. Curr Opin Pediatr. 2008 Oct;20(5):605-7. *(PubMed Abstract)*

- Finsterer J, Stöllberger C. **Atrial fibrillation/flutter in myopathies.** Int J Cardiol. 2008 Aug 29;128(3):304-10. Epub 2008 Mar 17. *(PubMed Abstract)*

- Moric-Janiszewska E, Markiewicz-Łoskot G. **Genetic heterogeneity of left-ventricular noncompaction cardiomyopathy.** Clin Cardiol. 2008 May;31(5):201-4. *(PubMed Abstract)*

- Towbin JA. **Molecular mechanisms of pediatric cardiomyopathies and new targeted therapies.** Progress in Pediatric Cardiology, Volume 25, Issue 1, April 2008, Pages 3-21. *(Abstract)*

- Menon SC, Olson TM, Michels VV. **Genetics of familial dilated cardiomyopathy.** Progress in Pediatric Cardiology Volume 25, Issue 1, April 2008, Pages 57-67. *(Abstract)*

- Wilkinson JD, Sleeper LA, Alvarez JA, Bublik N, Lipshultz SE and for the Pediatric Cardiomyopathy Study Group. **The Pediatric Cardiomyopathy Registry: 1995-2007.** Progress in Pediatric Cardiology Volume 25, Issue 1, April 2008, Pages 31-36. *(Abstract)*

- Bublik N, Alvarez JA, Lipshultz SE. **Pediatric cardiomyopathy as a chronic disease: A perspective on comprehensive care programs.** Progress in Pediatric Cardiology, Volume 25, Issue 1, April 2008, Pages 103-111. *(Abstract)*

- Finsterer J, Stollberger C, Blazek G. **Prevalence of Barth syndrome in adult left ventricular hypertrabeculation/noncompaction**. Scand Cardiovasc J. 2008 Apr;42(2):157-60. *(PubMed Abstract)*

- Tatu-Chitoiu A, Bradisteanu S. **A rare case of biventricular non-compaction associated with ventricular septal defect and descendent aortic stenosis in a young man.** Eur J Echocardiogr. 2008 Mar;9(2):306-8. *(PubMed Abstract)*

- Finsterer J, Stöllberger C. **Primary myopathies and the heart.** Scand Cardiovasc J. 2008 Feb;42(1):9-24. *(PubMed Abstract)*

- Andrews RE, Fenton MJ, Ridout DA, Burch M; British Congenital Cardiac Association. **New-onset heart failure due to heart muscle disease in childhood: A prospective study in the United Kingdom and Ireland.** Circulation. 2008 Jan 1;117(1):79-84. Epub 2007 Dec 17. Comment in: Circulation. 2008 Jan 1;117(1):11-2. *(PubMed Abstract)*

- Cox GF. **Diagnostic approaches to pediatric cardiomyopathy of metabolic genetic etiologies and their relation to therapy.** Prog Pediatr Cardiol. 2007;24(1):15-25. Prog Pediatr Cardiol. 2007;24(1):15-25. *(PubMed Abstract)*

- Markiewicz-Loskot G, Moric-Janiszewska E, Loskot M, Szydlowski L, Weglarz L, Hollek A. **The letter of Finsterer and Stollberger was shown to the authors who replied.** Europace. 2007 Apr;9(4):256-7. Epub 2007 Mar 8. *(PubMed - No Abstract Available)*

- Sparagna GC, Chicco AJ, Murphy RC, Bristow MR, Johnson CA, Rees ML, Maxey ML, McCune SA, Moore RL. **Loss of cardiac tetralinoleoyl cardiolipin in human and experimental heart failure.** J Lipid Res. 2007 Jul;48(7):1559-70. Epub 2007 Apr 10. *(PubMed Abstract)*

- Mangat J, Lunnon-Wood T, Rees P, Elliott M, Burch M. **Successful cardiac transplantation in Barth syndrome - single-centre experience of four patients.** Pediatr Transplant. 2007 May;11(3):327-331. *(PubMed Abstract)*

- Yaplito-Lee J, Weintraub R, Jamsen K, Chow CW, Thorburn DR, Boneh A. **Cardiac manifestations in oxidative phosphorylation disorders of childhood.** J Pediatr. 2007 Apr;150(4):407-11. *(PubMed Abstract)*

- Szulik M, Lenarczyk A, Rycaj J, Bialkowski J, Dziubek B, Kalarus Z, Kukulski T. **Isolated non-compaction of the left ventricular myocardium in a neonate - a case report.].** Kardiol Pol. 2006 Dec;64(12):1422-5. *(PubMed Abstract)*

- Cox GF, Sleeper LA, Lowe AM, Towbin JA, Colan SD, Orav EJ, Lurie PR, Messere JE, Wilkinson JD, Lipshultz SE. **Factors associated with establishing a causal diagnosis for children with cardiomyopathy.** Pediatrics. 2006 Oct;118(4):1519-31. *(PubMed Abstract)*

- Towbin JA, Lowe AM, Colan SD, Sleeper LA, Orav EJ, Clunie S, Messere J, Cox GF, Lurie PR, Hsu D, Canter C, Wilkinson JD, Lipshultz SE. **Incidence, causes, and outcomes of dilated cardiomyopathy in children.** JAMA. 2006 Oct 18;296(15):1867-76. *(PubMed Abstract)*

- Gilbert-Barness E, Barness LA. 2006. Festschrift for Dr. John M. Opitz. **Pathogenesis of cardiac conduction disorders in children genetic and histopathologic aspects.** Am J Med Genet A. 2006 Oct 1;140(19):1993-2006. *(PubMed Abstract)*

- Spencer CT, Bryant RM, Day J, Gonzalez IL, Colan SD, Thompson WR, Berthy J, Redfearn SP, Byrne BJ. **Cardiac and clinical phenotype in Barth syndrome.** Pediatrics 2006 Aug;118(2):e337-46. Epub 2006 Jul 17. *(PubMed Abstract)**▼

- Johnson MT, Zhang S, Gilkeson R, Ameduri R, Siwik E, Patel CR, Chebotarev O, Kenton AB, Bowles KR, Towbin JA, Robin NH, Brozovich F, Hoit BD. **Intrafamilial variability of noncompaction of the ventricular myocardium.** Am Heart J. 2006 May;151(5):1012.e7-14. *(PubMed Abstract)*

- Xing Y, Ichida F, Matsuoka T, Isobe T, Ikemoto Y, Higaki T, Tsuji T, Haneda N, Kuwabara A, Chen R, Futatani T, Tsubata S, Watanabe S, Watanabe K, Hirono K, Uese K, Miyawaki T, Bowles KR, Bowles NE, Towbin JA. **Genetic analysis in patients with left ventricular noncompaction and evidence for genetic heterogeneity.** Mol Genet Metab. 2006 May;88(1):71-7. Epub 2006 Jan 19. *(PubMed Abstract)*

- Ellinor PT, Milan DJ, MacRae CA. **Metabolic gene defects and risk of arrhythmia.** Heart Metab. 2006;33:9-12. *(Abstract)*

- Brady AN, Shehata BM, Fernhoff PM. **X-linked fetal cardiomyopathy caused by a novel mutation in the *TAZ* gene.** Prenat Diagn. 2006 May;26(5):462-5. *(PubMed Abstract)*

- Spencer CT, Byrne BJ, Gewitz MH, Wechsler SB, Kao AC, Gerstenfeld EP, Merliss AD, Carboni MP, Bryant RM. **Ventricular arrhythmia in the X-linked cardiomyopathy Barth syndrome.** Pediatr Cardiol. 2005 Sep-Oct;26(5):632-7. *(PubMed Abstract)**▼

- Stollberger C, Finsterer J. **Cardiologic and neurologic findings in left ventricular hypertrabeculation/non-compaction related to wall thickness, size and systolic function.** Eur J Heart Fail. 2005. Jan;7(1):95-7. *(PubMed Abstract)*

- Murphy RT, Thaman R, Blanes JG, Ward D, Sevdalis E, Papra E, Kiotsekolglou A, Tome MT, Pellerin D, McKenna WJ, Elliott PM. **Natural history and familial characteristics of isolated left ventricular non-compaction.** Eur Heart J. 2005 Jan;26(2):187-92. Epub 2004 Nov 30. *(PubMed Abstract)*

- Steward CG, Martin RP, Hayes AM, Salmon AP, Williams MM, Tyfield LA, Davies SJ, Newbury-Ecob RA. **Cardiology: Barth syndrome (X-Linked cardiac and skeletal myopathy, neutropenia, and organic aciduria): Rarely recognised, frequently fatal.** Arch Dis Child 2004;89(Suppl I):A47–A48; G140. (*Abstract)*

- Khositseth, A, Michels VV. **Familial dilated cardiomyopathy.** Cardiovasc Rev Rep 25(5):200-202, 2004. *(Abstract)*

- Gilbert-Barness E. **Review: Metabolic cardiomyopathy and conduction system defects in children.** Ann Clin Lab Sci. 2004 Winter;34(1):15-34. *(PubMed Abstract)*

- Cohen N, Muntoni F. **Multiple pathogenetic mechanisms in X-linked dilated cardiomyopathy.** Heart. 2004 Aug;90(8):835-41. *(PubMed Abstract)*

- Kenton AB, Sanchez X, Coveler KJ, Makar KA, Jimenez S, Ichida F, Murphy RT, Elliott PM, McKenna W, Bowles NE, Towbin JA, Bowles KR. **Isolated left ventricular noncompaction is rarely caused by mutations in G4.5, alpha-dystrobrevin and FK Binding Protein-12.** Mol Genet Metab 2004 Jun; 82: 162-6. *(PubMed Abstract)*

- Lee DS, Mamdani MM, Austin PC, Gong Y, Liu PP, Rouleau JL, Tu JV. **Trends in heart failure outcomes and pharmacotherapy: 1992 to 2000.** Am J Med 2004 May 1;116(9):581-589. *(PubMed Abstract)*

- DiLenarda A, Pinamonti B, Mestroni L, Salvi A, Sabbadini G, Gregori D, Perkan A, Zecchin M, Carniel E, Bussani R, Silvestri F, Morgera T, Camerini F, Sinagra G; Gruppo di Studio sulle Malattie del Miocardio. **The natural history of dilated cardiomyopathy: A review of the Heart Muscle Disease Registry of Trieste.** Ital Heart J Suppl 2004 Apr;5(4):253-266. *(PubMed Abstract)*

- Doolan A, Langlois N, Semsarian C. **Causes of sudden cardiac death in young Australians**. Med J Aust. 2004 Feb 2;180(3):110-2. *(PubMed Abstract)*

- Pignatelli RH, McMahon CJ, Dreyer WJ, Denfield SW, Price J, Belmont JW, Craigen WJ, Wu J, El Said H, Bezold LI, Clunie S, Fernbach S, Bowles NE, Towbin JA. **Clinical characterization of left ventricular noncompaction in children: A relatively common form of cardiomyopathy.** Circulation 2003 Nov 25;108(21):2672-2678. *(PubMed Abstract)*

- Borges AC, Kivelitz D, Baumann G. **Isolated left ventricular non-compaction: cardiomyopathy with homogeneous transmural and heterogeneous segmental perfusion.** Heart. 2003 Aug;89(8):e21. *(PubMed Abstract)*

- Khan IA, Biddle WP, Najeed SA, Abdul-Aziz S, Mehta NJ, Salaria V, Murcek AL, Harris DM. **Isolated noncompaction cardiomyopathy presenting with paroxysmal supraventricular tachycardia-case report and literature review.** Angiology. 2003 Mar-Apr;54(2):243-50. *(PubMed Abstract)*

- Spencer CT, Colan SD, Gonzalez I, Byrne BJ. **Characterization of the cardiomyopathy associated with Barth syndrome.** January 2003.

- DiMarco JP. **Implantable cardioverter-defibrillators**. (Review Article) N Engl J Med 2003 Nov 6; 349(19):1836-1847. *(PubMed Abstract)*

- Hohnloser SH, Klingenheben T, Bloomfield D, Dabbous O, Cohen RJ. **Usefulness of microvolt t-wave alternans for prediction of ventricular tachyarrhythmic events in patients with dilated cardiomyopathy: Results from a prospective observational study.** J Am Coll Cardiol 2003 June 18; 41(12):2220-2224. *(PubMed Abstract)*

- Sasse-Klaassen S, Gerull B, Oechslin E, Jenni R, Thierfelder L. **Isolated noncompaction of the left ventricular myocardium in the adult is an autosomal dominant disorder in the majority of patients**. Am J Med Genet 2003 June 1; 119A(2):162-167. *(PubMed Abstract)*

- Strauss A, Lock JE. **Pediatric cardiomyopathy – a long way to go**. Editorial. N Engl J Med 2003 April 24; 348(17):1703-1705. *(PubMed Abstract)*

- Lipshultz SE, Sleeper LA, Towbin JA, Lowe AM, Orav EJ, Cox GF, Lurie PR, McCoy K, Messere J, Orfaly RA, Colan SD. **The incidence of pediatric cardiomyopathy: The prospective Pediatric Cardiomyopathy Registry in two regions of the United States**. N Engl J Med 2003 April 24; 348(17):1647-1655. *(PubMed Abstract)*

- Nugent AW, Daubeney PEF, Chondros P, Carlin JB, Cheung M, Wilkinson LC, Davis AM, Kahler SG, Chow CW, Wilkinson JL, Weintraub RG. **The epidemiology of childhood cardiomyopathy in Australia**. N Engl J Med 2003 April 24; 348(17):1639-1646. *(PubMed Abstract)*

- Chen R, Tsuji T, Ichida F, Bowles KR, Yu X, Watanabe S, Hirono K, Tsubata S, Hamamichi Y, Ohta J, Imai Y, Bowles NE, Miyawaki T, Towbin JA. **Mutation analysis of the G4.5 gene in patients with isolated left ventricular noncompaction**. Mol Genet Metab 2002 Dec; 77(4):319-325. *(PubMed Abstract)*

- Osterziel KJ, Perrot A. **Molecular basis of heart failure and dilated cardiomyopathy**. Herz 2002 Dec; 27(8):816-818. *(PubMed Abstract)*

- Burch M. **Heart failure in the young.** Heart. 2002 Aug;88(2):198-202. *(PubMed Abstract)*

- Pasotti M, Repetto A, Pisani A, Arbustini E. **Diagnosi genetica di cardiomiopatia dilatavia familiare**. Ital Heart J Suppl. 2002 Apr;3(4):386-93. *(PubMed Abstract)*

- Miura K, Nakagawa H, Morikawa Y, Sasayama S, Matsumori A, Hasegawa K, Ohno Y, Tamakoshi A, Kawamura T, Inaba Y. **Epidemiology of idiopathic cardiomyopathy in Japan: Results from a nationwide survey.** Heart. 2002 Feb;87(2):126-30. *(PubMed Abstract)*

- Towbin JA, Bowles NE. **The failing heart**. Nature. 2002 Jan 10;415(6868):227-33. *(PubMed Abstract)*

- Klingenheben T, Gronefeld G, Li YG, Hohnloser SH. **Effect of metoprolol and *d,l*-sotalol on microvolt-level t-wave alternans. Results of a prospective, double-blind, randomized study.** J Am Coll Cardiol 2001 Dec; 38(7):2013-2019. *(PubMed Abstract)*

- Schonberger J, Seidman CE. **Many roads lead to a broken heart: The genetics of dilated cardiomyopathy**. Am J Hum Genet. 2001 Aug;69(2):249-60. Epub 2001 Jul 6. *(PubMed Abstract)*

- Osterziel KJ, Scheffold T, Perrot A, Dietz R; Netzwerk Myokardiale Erkrankungen. **The genetics of dilated cardiomyopathy.** [Article in German] Z Kardiol. 2001 Jul; 90(7):461-9. *(PubMed Abstract)*

- Lipshultz SE, Sleeper LA, Towbin JA, Lowe AM, Orav EJ, Cox GF, Lurie PR, McCoy KL, McDonald MA, Messere JE, Colan SD. **The incidence of pediatric cardiomyopathy in two regions of the United States**. N Engl J Med. 2003 Apr 24;348(17):1647-55. *(PubMed Abstract)*

- Cox GF, Sleeper LA, Lowe AM, Orav EJ, Lurie PR, McCoy KL, Messere JE, Orfaly RA, Towbin JA, Colan SD, Lipshultz SE. **Variables associated with a known etiology of cardiomyopathy in children: the Pediatric Cardiomyopathy Registry (PCMR) from 1990-1995**. Circ Supp 2001; 84:II-588. *(Abstract)*

- Ronghe MD, Foot AB, Martin R, Ashworth M, Steward CG. **Non-Epstein-Barr virus-associated T-cell lymphoma following cardiac transplantation for Barth syndrome**. Acta Paediatr 2001 May; 90(5):584-586. *(PubMed Abstract)*

- Bruns LA, Chrisant MK, Lamour JM, Shaddy RE, Pahl E, Blume ED, Hallowell Ss, Addonizio LJ, Canter CE. **Carvedilol as therapy in pediatric heart failure: An initial multicenter experience.** J Pediatr 2001 Apr; 138(4):505-511. *(PubMed Abstract)*

- Towbin JA, Bowles NE. **Molecular genetics of left ventricular dysfunction**. Curr Mol Med. 2001 Mar;1(1):81-90. *(PubMed Abstract)*

- Ichida F, Tsubata S, Bowles KR, Haneda N, Uses K, Miyawaki T, Dreyer WJ, Messina J, Li H, Bowles NE, Towbin JA. **Novel gene mutations in patients with left ventricular noncompaction or Barth syndrome**. Circulation. 2001 Mar 6;103(9):1256-63. *(PubMed Abstract)*

- Seidman JG, Seidman C. **The genetic basis for cardiomyopathy: From mutation identification to mechanistic paradigms**. Cell. 2001 Feb 23; 104(4):557-67. *(PubMed Abstract)*

- Shaddy RE. **Cardiomyopathies in adolescents: Dilated, hypertrophic, and restrictive**. Adolesc Med 2001 Feb; 12(1):35-45. *(PubMed Abstract)*

- Lubiszewska B, Hoffman P, Ruzyllo W. **Isolated noncompaction of the ventricular myocardium. Case report and review of literature.** Kardiol. Pol. 2001, 55, 443.

- Daubeney P, Nugent A, Chondros P, Wilkinson L, Davis AM, Kleinert S, Chow CW, Wilkinson JL, Weintraub R. **Incidence and natural history of left ventricular noncompaction presenting during childhood** [abstract]. In 2000 Annual Meeting of Cardiac Society of Australia and New Zealand – 48th ASM abstracts. *(Abstract)*

- Seliem MA, Mansara KB, Palileo M, Ye X, Zhang Z, Benson DW. **Evidence of autosomal recessive inheritance of infantile dilated cardiomyopathy: Studies from the Eastern Province of Saudi Arabia.** Pediatr Res. 2000 Dec;48(6):770-5. *(PubMed Abstract)*

- Komajda, M. **Genetics of dilated cardiomyopathy: A molecular maze?** Heart. 2000 Nov;84(5):463-4. *(PubMed Abstract)*

- Tsubata S, Bowles KR, Vatta M, Zintz C, Titus J, Muhonen L, Bowles NE, Towbin JA. **Mutations in the human δ-*sarcoglycan* gene in familial and sporadic dilated cardiomyopathy.** J Clin Invest. 2000 Sep;106(5):655-62. *(PubMed Abstract)*

- Finsterer J, Stollberger C. **Cardiac involvement in primary myopathies**. Cardiology 2000; 94(1):1-11. *(PubMed Abstract)*

- Arbustini E, Morbini P, Pilotto A, Gavazzi A, Tavazzi L. **Genetics of idiopathic dilated cardiomyopathy**. Herz. 2000 May; 25(3):156-60. *(PubMed Abstract)*

- Felker GM, Thompson RE, Hare JM, Hruban RH, Clemetson DE, Howard DL, Baughman KL, Kasper EK. **Underlying causes and long-term survival in patients with initially unexplained cardiomyopathy**. N Engl J Med. 2000 Apr 13;342(15):1077-84. *(PubMed Abstract)*

- Buonanno C, Variola A, Dander B, Gabaldo S, Marafioti V. **Isolated noncompaction of the myocardium: An exceedingly rare cardiomyopathy. A case report.** Ital Heart J. 2000 Apr;1(4):301-5. *(PubMed Abstract)*

- Grenier MA, Osganian SK, Cox GF, Towbin JA, Colan SD, Lurie PR, Sleeper LA, Orav EJ, Lipshultz SE. **Design and implementation of the North American Pediatric Cardiomyopathy Registry**. Am Heart J. 2000 Feb;139(2 Pt 3):S86-95. *(PubMed Abstract)*

- Priori SG, Barhanin J, Hauer RNW, Haverkamp W, Jongsma HJ, Kleber AG, McKenna WJ, Roden DM, Rudy Y, Schwartz K, Schwartz PJ, Towbin JA, Wilde AM. **Genetic and molecular basis of cardiac arrhythmias: Impact on clinical management Parts I and II.** Circulation. 1999 Feb 2;99(4):518-28. *(PubMed Abstract)*

- D'Adamo P, Fassone L, Gedeon A, Janssen EA, Bione S, Bolhuis PA, Barth PG, Wilson M, Haan E, Orstavik KH, Patton MA, Green AJ, Zammarchi E, Donati MA, Toniolo D. **The X-linked gene G4.5 is responsible for different infantile dilated cardiomyopathies**. Am J Hum Genet. 1997 Oct;61(4):862-7. *(PubMed Abstract)*

- Adwani SS, Whitehead BF, Rees PG, Morris A, Turnball DM, Elliott MJ, de Leval MR. **Heart transplantation for Barth syndrome**. Pediatr Cardiol. 1997 Mar-Apr;18(2):143-5. *(PubMed Abstract)*

- Schwartz ML, Cox GF, Lin AE, Korson MS, Perez-Atayde A, Lacro RV, Lipshultz SE. **Clinical approach to genetic cardiomyopathy in children**. Circulation. 1996 Oct 15;94(8):2021-38. *(PubMed Abstract)*

- Kelly DP, Strauss AW. **Inherited cardiomyopathies.** N Engl J Med. 1994 Mar 31;330(13):913-9. *(PubMed – No Abstract Available.)*

- Hodgson S, Child A, Dyson M. **Endocardial fibroelastosis: Possible X linked inheritance.** J Med Genet. 1987 Apr;24(4):210-4. *(PubMed Abstract)*

- Neustein HB, Lurie PR, Dahms B, Takahashi M. **An X-linked recessive cardiomyopathy with abnormal mitochondria.** Pediatrics. 1979 Jul;64(1):24-9. *(PubMed Abstract)*

CARRIERS

- Finsterer J, Stollberger C. **Acquired noncompaction in Barth syndrome due to the *TAZ* mutation c.481_482ins20.** J Pediatr. 2017 Mar 15. pii: S0022-3476(17)30332-3. doi: 10.1016/j.jpeds.2017.02.066. [Epub ahead of print] (*PubMed Abstract)*

- Bakšiene M, Benušiene E, Morkuniene A, Ambrozaityte L, Utkus A, Kucinskas V. **A novel intronic splice site *tafazzin* gene mutation detected prenatally in a family with Barth syndrome.** Balkan J Med Genet. 2017 Mar 8;19(2):95-100. doi: 10.1515/bjmg-2016-0043. *(PubMed - Open Access)*

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)**▼

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth syndrome in adulthood: A clinical case.** [Article in English, Spanish] Rev Esp Cardiol. 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. *(PubMed Abstract)*

- Garratt V, Riddiford D, Steward C, Tsai-Goodman B, Newbury-Ecob R. **What is Barth syndrome?** Midwives Magazine: Issue 4: 2011. *(Not peer reviewed) (Abstract)*▼

- Aljishi E, Ali F. **Barth syndrome: An X-linked cardiomyopathy with a novel mutation.** Indian J Pediatr. 2010 Dec;77(12):1432-3. Epub 2010 Oct 28. *(PubMed Abstract)*

- Chang B, Momoi N, Shan L, Mitomo M, Aoyagi Y, Endo K, Takeda I, Chen R, Xing Y, Yu X, Watanabe S, Yoshida T, Kanegane H, Tsubata S, Bowles NE, Ichida F, Miyawaki T; Noncompaction study collaborators. **Gonadal mosaicism of a *TAZ* (G4.5) mutation in a Japanese family with Barth syndrome and left ventricular noncompaction.** Mol Genet Metab. 2010 Jun;100(2):198-203. Epub 2010 Mar 2. *(PubMed Abstract)*

- Bachou T, Giannakopoulos A, Trapali C, Vazeou A, Kattamis A. **A novel mutation in the G4.5 (*TAZ*) gene in a Greek patient with Barth syndrome.** Blood Cells Mol Dis. 2009 May-Jun;42(3):262-4. Epub 2009 Mar 3. *(PubMed Abstract)*

- Brady AN, Shehata BM, Fernhoff PM. **X-linked fetal cardiomyopathy caused by a novel mutation in the *TAZ* gene.** Prenat Diagn. 2006 May;26(5):462-5. *(PubMed Abstract)*

- Orstavik KH, Orstavik RE, Naumova AK, D'Adamo P, Gedeon A, Bolhuis PA, Barth PG, Toniolo D. **X chromosome inactivation in carriers of Barth syndrome**. Am J Hum Genet. 1998 Nov;63(5):1457-63. *(PubMed Abstract)*

- Cardonick EH, Kuhlman K, Ganz E, Pagotto LT. **Prenatal clinical expression of 3-methylglutaconic aciduria: Barth syndrome**. Prenat Diagn. 1997 Oct;17(10):983-8. *(PubMed Abstract)*

- Ruitenbeck W, Wendel U, Hamel BC, Trijbels JM. **Genetic counselling and prenatal diagnosis in disorders of the mitochondrial energy metabolism**. J Inherit Metab Dis 1996; 19(4):581-587. *(PubMed Abstract)*

- Grifo JA, Tang YX, Munne S, Alikani M, Cohen J, Rosenwaks Z. **Healthy deliveries from biopsied human embryos**. Hum Reprod 1994 May; 9(5):912-916. *(PubMed Abstract)*

CHRONIC ILLNESS

- Jacob ML, Johnco C, Dane BF, Collier A, Storch EA. **Psychosocial functioning in Barth syndrome: Assessment of individual and parental adjustment.** Children's Health Care, 46:1, 66-92 (2015). *(Abstract)*▼

- Folsi V, Miglietti N, Lombardi A, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, Plebani A. **Cardiomyopathy in a male patient with neutropenia and growth delay.** Ital J Pediatr. 2014 May 12;40:45. doi:10.1186/1824-7288-40-45. *(PubMed – Open Access)*

- Rigaud C, Lebre A, Touraine R, Beaupain B, Ottolenghi C, Chabli A, Ansquer H, Ozsahin H, Di Filippo S, De Lonlay P, Borm B, Rivier F, Vaillant M, Mathieu-Dramard M, Goldenberg A, Viot G, Charron P, Rio M, Bonnet D, Donadieu J. **Natural history of Barth syndrome: A national cohort study of 22 patients.** Orphanet J Rare Dis. 2013 May 8;8:70. doi: 10.1186/1750-1172-8-70. *(PubMed – Open Access)**▼

- Kachewar SG, Sankaye SB, Kulkarni DS. **The role of radio-diagnosis in inborn errors of metabolism.** Journal of Clinical and Diagnostic Research [serial online] 2011 November [cited: 2012 Dec 5]; 5:1467-1472. *(Full Text)*

- Wan C, Yu HH, Lu MY, Lee JH, Wang LC, Lin YT, Yang YH, Chiang BL. **Clinical manifestations and outcomes of pediatric chronic neutropenia.** J Formos Med Assoc. 2012 Apr;111(4):220-7. Epub 2012 Mar 16. *(PubMed Abstract)*

- UK Department of Health. **The expert patient: A new approach to chronic disease management for the 21st century**. Aug 2001.

- National Association of School Psychologists. **Mini-series: Promoting school success in children with chronic medical conditions**. School Psychology Review 1999; Vol.28, No.2. *(Abstract)*

EDUCATION

- Cole LK, Kim JH, Amoscato AA, Tyurina YY, Bayır H, Karimi B, Siddiqui TJ, Kagan VE, Hatch GM, Kauppinen TM. **Aberrant cardiolipin metabolism is associated with cognitive deficiency and hippocampal alteration in *tafazzin* knockdown mice.** Biochim Biophys Acta. 2018 Jul 25. pii: S0925-4439(18)30271-0. doi: 10.1016/j.bbadis.2018.07.022. [Epub ahead of print]*

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Raches D, Mazzocco MM. **Emergence and nature of mathematical difficulties in young children with Barth syndrome.** J Dev Behav Pediatr. 2012 May;33(4):328-35. *(PubMed Abstract)**▼

- Mancuso DJ, Kotzbauer PT, Wozniak DF, Sims HF, Jenkins CM, Guan S, Han X, Yang K, Sun G, Malik I, Conyers S, Green KG, Schmidt RE, Gross RW. **Genetic ablation of calcium-independent phospholipase A2{gamma} leads to alterations in hippocampal cardiolipin content and molecular species distribution, mitochondrial degeneration, autophagy and cognitive dysfunction.** J Biol Chem. 2009 Dec 18;284(51):35632-44. Epub. *(PubMed Abstract)*

- Storch EA, Keeley M, Merlo LJ, St. Amant JB, Jacob M, Storch J, Spencer C, Byrne BJ. **Psychosocial functioning in youth with Barth syndrome.** Child Health Care. 2009 Apr;38(2):137-156. *(PubMed – Open Access)**▼

- Mazzocco MM, Henry AE, Kelly RI. **Barth syndrome is associated with a cognitive phenotype.** J Dev Behav Pediatr. 2007 Feb;28(1):22-30. *(PubMed – Open Access)**▼

- Mazzocco MM, Kelley RI. **Preliminary evidence for a cognitive phenotype in Barth syndrome.** Am J Med Genet. 2001 Sep 1;102(4):372-8. *(PubMed Abstract)*▼

- National Association of School Psychologists. **Mini-series: Promoting school success in children with chronic medical conditions**. School Psychology Review 1999; Vol.28, No.2.

ENDOCRINOLOGY

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Katsushima Y, Fujiwara I, Sakamoto O, Ohura T, Miyabayashi S, Ohnuma A, Yamaguchi S, Iinuma K. **Normal pituitary function in a Japanese patient with Barth syndrome**. Eur J Pediatr. 2002 Jan;161(1):67-8. *(PubMed Abstract)*

FREQUENCY STATISTICS

- Garratt V, Riddiford D, Steward C, Tsai-Goodman B, Newbury-Ecob R. **What is Barth syndrome?** Midwives Magazine: Issue 4: 2011. *(Not peer reviewed) (Abstract)*▼

- Strauss A, Lock JE. **Pediatric cardiomyopathy – a long way to go**. N Engl J Med 2003 April 24; 348(17):1703-1705. *(PubMed Abstract)*

- Lipshultz SE, Sleeper LA, Towbin JA. Lowe AM, Gray EJ, Cox GF, Lurie PR, McCoy KL, McDonald MA, Messere JE, Colan SD. **The incidence of pediatric cardiomyopathy in two regions of the United States**. N Engl J Med 2003 April 24; 348(17):1647-1655. *(PubMed Abstract)*

- Nugent AW, Daubeney PEF, Chondros P, Carlin JB, Cheung M, Wilkinson LC, Davis AM, Kahler SG, Chow CW, Wilkinson JL, Weintraub RG. **The epidemiology of childhood cardiomyopathy in Australia**. N Engl J Med 2003 April 24; 348(17):1639-1646 . *(PubMed Abstract)*

- Miura K, Nakagawa H, Morikawa Y, Sasayama S, Matsumori A, Hasegawa K, Ohno Y, Tamakoshi A, Kawamura T, Inaba Y. **Epidemiology of idiopathic cardiomyopathy in Japan: Results from a nationwide survey.** Heart. 2002 Feb;87(2):126-30. *(PubMed Abstract)*

- Cantlay AM, Shokrollahi K, Allen JT, Lunt PW, Newbury-Ecob RA, Steward CG. **Genetic analysis of the G4.5 gene in families with suspected Barth syndrome**. J Pediatr. 1999 Sep;135(3):311-5. *(PubMed Abstract)*

GENETICS

NEW Vasilescu C, Ojala TH, Brilhante V, Ojanen S, Hinterding HM, Palin E, Alastalo TP, Koskenvuo J, Hiippala A, Jokinen E, Jahnukainen T, Lohi J, Pihkala J, Tyni TA, Carroll CJ, Suomalainen A. **Genetic basis of severe childhood-onset cardiomyopathies.** J Am Coll Cardiol. 2018 Nov 6;72(19):2324-2338. doi:10.1016/j.jacc.2018.08.2171. *(PubMed Abstract)*

NEW Chatzispyrou IA, Guerrero-Castillo S, Held NM, Ruiter JPN, Denis SW, IJlst L, Wanders RJ, van Weeghel M, Ferdinandusse S, Vaz FM, Brandt U, Houtkooper RH. **Barth syndrome cells display widespread remodeling of mitochondrial complexes without affecting metabolic flux distribution**. Biochim Biophys Acta Mol Basis Dis. 2018 Sep 1;1864(11):3650-3658. doi: 10.1016/j.bbadis.2018.08.041. [Epub ahead of print] *(PubMed Abstract)*▼

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. *(PubMed - Open Access)**▼

- Sudrié-Arnaud B, Marguet F, Patrier S, Martinovic J, Louillet F, Broux F, Charbonnier F, Dranguet H, Coutant S, Vezain M, Lanos R, Tebani A, Fuller M, Lamari F, Chambon P, Brehin AC, Trestard L, Tournier I, Marret S, Verspyck E, Laquerrière A, Bekri S. **Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation.** Clin Chim Acta. 2018 Jun;481:1-8. doi: 10.1016/j.cca.2018.02.023. Epub 2018 Feb 22. *(PubMed Abstract)*

- Mejia EM, Zegallai H, Bouchard ED, Banerji V, Ravandi A, Hatch GM. **Expression of human monolysocardiolipin acyltransferase-1 improves mitochondrial function in Barth syndrome lymphoblasts.** J Biol Chem. 2018 Mar 21. pii:jbc.RA117.001024. doi:10.1074/jbc.RA117.001024. [Epub ahead of print] *(PubMed - Open Access)**▼

- Tsujii N, Hayashi T, Hayashi T, Kimura A, Nishikubo T. **Barth syndrome associated with triple mutation.** Pediatr Int. 2018 Mar 6. doi:10.1111/ped.13517. [Epub ahead of print]. *(PubMed - mo abstract available)*

- Damschroder D, Reynolds C, Wessells R. **Drosophila *tafazzin* mutants have impaired exercise capacity.** Physiol Rep. 2018 Feb;6(3). doi: 10.14814/phy2.13604. *(PubMed - Open Access)*

- Seo SH, Kim SY, Cho SI, Park H, Lee S, Choi JM, Kim MJ, Lee JS, Ahn KJ, Song MK, Bae EJ, Park SS, Seong MW. **Application of multigene panel sequencing in patients with prolonged rate-corrected QT interval and no pathogenic variants detected in KCNQ1, KCNH2, and SCN5A.** Ann Lab Med. 2018 Jan;38(1):54-58. doi: 10.3343/alm.2018.38.1.54. *(PubMed - Open Access)*

- Imai-Okazaki A, Kishita Y, Kohda M, Yatsuka Y, Hirata T, Mizuno Y, Harashima H, Hirono K, Ichida F, Noguchi A, Yoshida M, Tokorodani C, Nishiuchi R, Takeda A, Nakaya A, Sakata Y, Murayama K, Ohtake A, Okazaki Y. **Barth syndrome: Different approaches to diagnosis.** J Pediatr. 2017 Dec 15. pii: S0022-3476(17)31329-X. doi: 10.1016/j.jpeds.2017.09.075. [Epub ahead of print] *(PubMed Abstract)*

- Abe M, Sawada Y, Uno S, Chigasaki S, Oku M, Sakai Y, Miyoshi H. **Role of acyl chain composition of phosphatidylcholine in *tafazzin*-mediated remodeling of cardiolipin in liposomes.** Biochemistry. 2017 Nov 28;56(47):6268-6280. doi: 10.1021/acs.biochem.7b00941. Epub 2017 Nov 9. (*PubMed Abstract)*

- Seneviratne AK, Aristizabal Henao JJ, Xu GW, Hurren R, Kim S, MacLean N, Wang X, Xu M, Fajado V, Hao Z, Gronda M, Jeyaraju DV, Chouinard-Watkins R, Schafer C, LeBlanc P, Khuchua Z, Bazinet R, Claypool S, Stark K, Schimmer A. ***Tafazzin* (*TAZ*) regulates the differentiation of AML cells by reducing levels of the phospholipid phosphatidylethanolamine.** Blood 2017 130:788. *(Abstract)*

- Giacomelli E, Mummery CL, Bellin M. **Human heart disease: Lessons from human pluripotent stem cell-derived cardiomyocytes.** Cell Mol Life Sci. 2017 Oct;74(20):3711-3739. doi:10.1007/s00018-017-2546-5. Epub 2017 Jun 1. Review. *(PubMed - Open Access)*

- Saneto RP. **Genetics of mitochondrial disease.** Adv Genet. 2017;98:63-116. doi:10.1016/bs.adgen.2017.06.002. Epub 2017 Sep 1. *(PubMed Abstract)*

- Wang C, Hata Y, Hirono K, Takasaki A, Ozawa SW, Nakaoka H, Saito K, Miyao N, Okabe M, Ibuki K, Nishida N, Origasa H, Yu X, Bowles NE, Ichida F; for LVNC Study Collaborators. **A wide and specific spectrum of genetic variants and genotype-phenotype correlations revealed by next-generation sequencing in patients with left ventricular noncompaction.** J Am Heart Assoc. 2017 Aug 30;6(9). pii: e006210. doi:10.1161/JAHA.117.006210. *(PubMed Abstract)*

- Zhao Y, Feng Y, Ding X, Dong S, Zhang H, Ding J, Xia X. **Identification of a novel hypertrophic cardiomyopathy-associated mutation using targeted next-generation sequencing.** Int J Mol Med. 2017 Jul;40(1):121-129. doi: 10.3892/ijmm.2017.2986. Epub 2017 May 11. *(PubMed - Open Access)*

- Finsterer J, Stollberger C. **Acquired noncompaction in Barth syndrome due to the *TAZ* mutation c.481_482ins20.** J Pediatr. 2017 Mar 15. pii: S0022-3476(17)30332-3. doi: 10.1016/j.jpeds.2017.02.066. [Epub ahead of print] (*PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Bakšiene M, Benušiene E, Morkuniene A, Ambrozaityte L, Utkus A, Kucinskas V. **A novel intronic splice site *tafazzin* gene mutation detected prenatally in a family with Barth syndrome.** Balkan J Med Genet. 2017 Mar 8;19(2):95-100. doi: 10.1515/bjmg-2016-0043. *(PubMed - Open Access)*

- Lloyd DF, Vara R, Mathur S. **The cardiac manifestations of inherited metabolic diseases in children.** Pediatr Int. 2017 Mar 4. doi: 10.1111/ped.13272. [Epub ahead of print] Review. (*PubMed Abstract)*

- Schlame M, Xu Y, Ren M. **The basis for acyl specificity in the *tafazzin* reaction.** J Biol Chem. 2017 Mar 31;292(13):5499-5506. doi: 10.1074/jbc.M116.769182. Epub 2017 Feb 15. (*PubMed Abstract)*

- Borna NN, Kishita Y, Ishikawa K, Nakada K, Hayashi JI, Tokuzawa Y, Kohda M, Nyuzuki H, Yamashita-Sugahara Y, Nasu T, Takeda A, Murayama K, Ohtake A, Okazaki Y. **A novel mutation in *TAZ* causes mitochondrial respiratory chain disorder without cardiomyopathy.** J Hum Genet. 2017 Jan 26. doi: 10.1038/jhg.2016.165. [Epub ahead of print] *(PubMed Abstract)*

- Wang J, Guo Y, Huang M, Zhang Z, Zhu J, Liu T, Shi L, Li F, Huang H, Fu L. **Identification of *TAZ* mutations in pediatric patients with cardiomyopathy by targeted next-generation sequencing in a Chinese cohort.** Orphanet J Rare Dis. 2017 Feb 10;12(1):26. doi: 10.1186/s13023-016-0562-4. *(PubMed Abstract*)

- Woiewodski L, Ezon D, Cooper J, Feingold B. **Barth syndrome with late-onset cardiomyopathy: A missed opportunity for diagnosis.** The Journal of Pediatrics, In Press, Corrected Proof, Available online 18 January 2017. *(Abstract)*

- Dong X, Fan P, Tian T, Yang Y, Xiao Y, Yang K, Liu Y, Zhou X. **Recent advancements in the molecular genetics of left ventricular noncompaction cardiomyopathy.** Clin Chim Acta. 2016 Dec 15;465:40-44. doi:10.1016/j.cca.2016.12.013. [Epub ahead of print] Review. *(PubMed Abstract)*

- Avdjieva-Tzavella DM, Todorova AP, Kathom H M, Ivanova MB, Yordanova IT, Todorov TP, Litvinenko IO, Dasheva-Dimitrova AT, Tincheva RS. **Barth syndrome in male and female siblings caused by a novel mutation in the *TAZ* gene.** Genet Couns. 2016;27(4):495-501. *(PubMed Abstract)*

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)**▼

- Pérez-Serra A, Toro R, Sarquella-Brugada G, de Gonzalo-Calvo D, Cesar S, Carro E, Llorente-Cortes V, Iglesias A, Brugada J, Brugada R, Campuzano O. **Genetic basis of dilated cardiomyopathy.** International Journal of Cardiology, Available online 21 September 2016. *(ScienceDirect Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- Yoo TY, Kim MR, Son JS, Lee R, Bae SH, Chung S, Kim KS, Seong MW, Park SS. **Identification of a novel de novo mutation of the *TAZ* gene in a Korean patient with Barth syndrome.** J Cardiovasc Ultrasound. 2016 Jun;24(2):153-7. doi: 10.4250/jcu.2016.24.2.153. Epub 2016 Jun 22. *(PubMed - Open Access)*

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Szczepanek K, Allegood J, Aluri H, Hu Y, Chen Q, Lesnefsky EJ. **Acquired deficiency of *tafazzin* in the adult heart: Impact on mitochondrial function and response to cardiac injury.** Biochim Biophys Acta. 2016 Apr;1861(4):294-300. doi: 10.1016/j.bbalip.2015.12.004. Epub 2015 Dec 12. *(PubMed Abstract)*

- Lu YW, Galbraith L, Herndon JD, Lu YL, Pras-Raves M, Vervaart M, van Kampen A, Luyf A, Koehler CM, McCaffery JM, Gottlieb E, Vaz FM, Claypool SM. **Defining functional classes of Barth syndrome mutation in humans.** Hum Mol Genet. 2016 Feb 16. pii: ddw046. [Epub ahead of print] *(PubMed Abstract)**

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene** transcription. Clin Genet. 2016 Feb 8. doi:10.1111/cge.12756. [Epub ahead of print] *(PubMed Abstract)*

- Thompson WR, DeCroes B, McClellan R, Rubens J, Vaz FM, Kristaponis K, Avramopoulos D, Vernon HJ. **New targets for monitoring and therapy in Barth syndrome.** Genet Med. 2016 Feb 4. doi: 10.1038/gim.2015.204. [Epub ahead of print] *(PubMed Abstract)*▼

- Thiels C, Fleger M, Huemer M, Rodenburg RJ, Vaz FM, Houtkooper RH, Haack TB, Prokisch H, Feichtinger RG, Lücke T, Mayr JA, Wortmann SB. **Atypical clinical presentations of *TAZ* mutations: An underdiagnosed cause of growth retardation?** JIMD Reports 2016. *(PubMed Abstract)*

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Nagueh SF, Zoghbi WA. **Role of imaging in the evaluation of patients at risk for sudden cardiac death: Genotype–phenotype intersection.** Review Article. JACC: Cardiovascular Imaging, Volume 8, Issue 7, July 2015, Pages 828-845. *(ScienceDirect Abstract)*

- Cadalbert LC, Ghaffar FN, Stevenson D, Bryson S, Vaz FM, Gottlieb E, Strathdee D. **Mouse *tafazzin* is required for male germ cell meiosis and spermatogenesis.** PLoS One. 2015 Jun 26;10(6):e0131066. doi:10.1371/journal.pone.0131066. eCollection 2015. *(PubMed - Open Access)**

- Rodier G, Kirsh O, Baraibar M, Houles T, Lacroix M, Delpech H, Hatchi E, Arnould S, Severac D, Dubois E, Caramel J, Julien E, Friguet B, Le Cam L, Sardet C. **The transcription factor E4F1 coordinates CHK1-dependent checkpoint and mitochondrial functions.** Cell Reports, Available online 2 April 2015. *(ScienceDirect Abstract)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Borkar M, Bijarnia-Mahay S, Kohli S, Juneja M,Srivastava Y, Saxena R, Verma IC. **Identification of a novel *TAZ* gene mutation in a family with X-linked dilated cardiomyopathy Barth syndrome.** Journal of Inborn Errors of Metabolism & Screening January - December 2015 3: 2326409814567131, first published on February 19, 2015 doi:10.1177/2326409814567131. *(Sage – Full Text)*

- Lu YW, Claypool SM. **Disorders of phospholipid metabolism: An emerging class of mitochondrial disease due to defects in nuclear genes.** Front Genet. 2015 Feb 3;6:3. doi:10.3389/fgene.2015.00003. eCollection 2015. Review. *(PubMed – Open Access)*

- Russell M, Roberts AE, Abrams DJ, Murphy AM, Towbin JA, Chung WK. **How to effectively utilize genetic testing in the care of children with cardiomyopathies.** Progress in Pediatric Cardiology, Available online 31 January 2015. *(ScienceDirect Abstract)*

- Rutter J, Hughes AL**. Power2: The power of yeast genetics applied to the powerhouse of the cell.** Trends Endocrinol Metab. 2015 Jan 12. pii: S1043-2760(14)00204-5. doi: 10.1016/j.tem.2014.12.002. [Epub ahead of print] *(PubMed Abstract)*

- Carrilho-Ferreira P, Almeida AG, Pinto FJ. **Non-compaction cardiomyopathy: Prevalence, prognosis, pathoetiology, genetics, and risk of cardioembolism.** Curr Heart Fail Rep. 2014 Dec;11(4):393-403. doi: 10.1007/s11897-014-0227-3. *(PubMed Abstract)*

- Falk MJ, Shen L, Gonzalez M, Leipzig J, Lott MT, Stassen APM, Diroma MA, Navarro-Gomez D, Yeske P, Bai R, Boles RG, Brilhante B, Ralph D, Dare JT, Shelton R, Terry S, Zhang Z, Copeland WC, van Oven M, Prokisch H, Wallace DC, Attimonelli M, Krotoski D, Zuchner S, Gai X. **Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities.** Mol Genet Metab. 2014 Dec 4. pii: S1096-7192(14)00377-1. doi: 10.1016/j.ymgme.2014.11.016. [Epub ahead of print] *(PubMed Abstract)*

- Tariq M, Ware SM. **Importance of genetic evaluation and testing in pediatric cardiomyopathy.** World J Cardiol 2014 Nov 26;6(11):1156-1165. Review. *(PubMed – Open Access)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Bowron A, Honeychurch J, Williams M, Tsai-Goodman B, Clayton N, Jones L, Shortland GJ, Qureshi SA, Heales SJ, Steward CG. **Barth syndrome without tetralinoleoyl cardiolipin deficiency: A possible ameliorated phenotype.** J Inherit Metab Dis. 2014 Aug 12. [Epub ahead of print] *(PubMed – Open Access)**▼

- Debnath S, Addya S. **Mis-sense mutations in *tafazzin (TAZ)* that escort to mild clinical symptoms of Barth syndrome is owed to the minimal inhibitory effect of the mutations on the enzyme function: In-silico evidence.** Interdiscip Sci. 2014 Aug 9. [Epub ahead of print] *(PubMed Abstract)*

- Folsi V, Miglietti N, Lombardi AM, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, Plebani A. **Cardiomyopathy in a male patient with neutropenia and growth delay.** Italian Journal of Pediatrics 2014, 40:45 doi:10.1186/1824-7288-40-45. *(PubMed – Open Access)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Kirwin SM, Manolakos A, Swain Barnett S, Gonzalez IL. ***Tafazzin* splice variants and mutations in Barth syndrome.** Mol Genet Metab. 2014 Jan;111(1):26-32. doi: 10.1016/j.ymgme.2013.11.006. Epub 2013 Nov 19. *(PubMed Abstract)**

- Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J. **The MOGE(S) classification for a phenotype–genotype nomenclature of cardiomyopathy: Endorsed by the World Heart Federation.** Journal of the American College of Cardiology, Volume 62, Issue 22, 3 December 2013, Pgs. 2046-2072 - CORRECTION. *(ScienceDirect Abstract)*

- Arbustini E, Narula N, Dec GW, Reddy KS, Greenberg B, Kushwaha S, Marwick T, Pinney S, Bellazzi R, Favalli V, Kramer C, Roberts R, Zoghbi WA, Bonow R, Tavazzi L, Fuster V, Narula J. **The MOGE(S) classification for a phenotype–genotype nomenclature of cardiomyopathy: Endorsed by the World Heart Federation.** Journal of the American College of Cardiology, Available online 18 November 2013. *(ScienceDirect Abstract)*

- Digilio MC, Bernardini L, Gagliardi MG, Versacci P, Baban A, Capolino R, Dentici ML, Roberti MC, Angioni A, Novelli A, Marino b, Dallapiccola B. **Syndromic non-compaction of the left ventricle: Associated chromosomal anomalies.** Clin Genet. 2013 Oct;84(4):362-7. doi: 10.1111/cge.12069. Epub 2012 Dec 28. *(PubMed Abstract)*

- Finsterer J, Stöllberger C. **Unclassified cardiomyopathies in neuromuscular disorders.** Wien Med Wochenschr. 2013 Oct 24. [Epub ahead of print] *(PubMed Abstract)*

- Mazurová S, Tesarová M, Magner M, Houštková H, Hansíková H, Augustínová J, Tomek V, Vondrácková A, Zeman J, Honzík T. **Novel mutations in the *TAZ* gene in patients with Barth syndrome.** Prague Med Rep. 2013;114(3):139-153. (*PubMed Abstract)*

- Kim GB, Kwon BS, Bae EJ, Noh CI, Seong MW, Park SS. **A novel mutation of the *TAZ* gene in Barth syndrome: Acute exacerbation after contrast-dye injection.** J Korean Med Sci. 2013 May;28(5):784-7. doi: 10.3346/jkms.2013.28.5.784. Epub 2013 May 2. (*PubMed – Open Access)*

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Rigaud C, Lebre A, Touraine R, Beaupain B, Ottolenghi C, Chabli A, Ansquer H, Ozsahin H, Di Filippo S, De Lonlay P, Borm B, Rivier F, Vaillant M, Mathieu-Dramard M, Goldenberg A, Viot G, Charron P, Rio M, Bonnet D, Donadieu J. **Natural history of Barth syndrome: A national cohort study of 22 patients.** Orphanet J Rare Dis. 2013 May 8;8:70. doi: 10.1186/1750-1172-8-70. *(PubMed – Open Access)**▼

- Finsterer J, Stöllberger C, Blazek G, Sehnal E. **Familal left ventricular hypertrabeculation (noncompaction) is myopathic.** Int J Cardiol. 2013 Apr 15;164(3):312-7. doi:10.1016/j.ijcard.2011.07.012. Epub 2011 Jul 23. *(PubMed Abstract)*

- Fan Y, Steller J, Gonzalez IL, Kulik W, Fox M, Chang R, Westerfield BA, Batra AS, Wang RY, Gallant NM, Pena LS, Wang H, Huang T, Bhuta S, Penny DJ, McCabe ER, Kimonis VE. **A novel exonic splicing mutation in the *TAZ* (G4.5) gene in a case with atypical Barth syndrome.** JIMD Rep. 2013 Apr 19. [Epub ahead of print] *(PubMed – Open Access)*

- Teekakirikul P, Kelly MA, Rehm HL, Lakdawala NK, Funke BH. **Inherited cardiomyopathies: Molecular genetics and clinical genetic testing in the postgenomic era.** J Mol Diagn. 2013 Mar;15(2):158-70. doi: 10.1016/j.jmoldx.2012.09.002. Epub 2012 Dec 27. *(PubMed Abstract)*

- Gonzalvez F, D'Aurelio M, Boutant M, Moustapha A, Puech JP, Landes T, Arnauré L, Vial G, Talleux N, Slomianny C, Wanders RJA, Houtkooper RH, Belenger P, Moller IM, Gottlieb E, Vaz FM, Manfredi G, Petit PX. **Barth syndrome: Cellular compensation of mitochondrial dysfunction and apoptosis inhibition due to changes in cardiolipin remodeling linked to *Tafazzin* gene mutation.** Biochim Biophys Acta. 2013 Aug;1832(8):1194-206. doi: 10.1016/j.bbadis.2013.03.005. Epub 2013 Mar 20. *(PubMed – Open Access)*

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth syndrome in adulthood: A clinical case.** [Article in English, Spanish] Rev Esp Cardiol. 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini RR, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 14;8(1):27. doi: 10.1186/1750-1172-8-27. *(PubMed Abstract)*▼

- Whited K, Baile MG, Currier P, Claypool SM. **Seven functional classes of Barth syndrome mutation.** Hum Mol Genet. 2013 Feb 1;22(3):483-92. doi: 10.1093/hmg/dds447. Epub 2012 Oct 24. *(PubMed Abstract)*

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Ware SM, Towbin JA. **Nuclear genes causing mitochondrial cardiomyopathy.** Mitochondrial Disorders Caused by Nuclear Genes, 2013, pp. 319-335. *(Springer Abstract)*

- Man E, Lafferty KA, Funke BH, Lun KS, Chan SY, Chau AK, Chung BH. **NGS identifies *TAZ* mutation in a family with X-linked dilated cardiomyopathy.** BMJ Case Rep. 2013 Jan 22;2013. doi:pii: bcr2012007529. 10.1136/bcr-2012-007529. *(PubMed Abstract)*

- Mestroni L, Taylor MRG. **Genetics and genetic testing of dilated cardiomyopathy: A new perspective.** Discov Med 2013 Jan;15(80):43-49. *(PubMed – Open Access)*

- Wortmann SB, Duran M, Anikster Y, Barth PG, Sperl W, Zschocke J, Morava E, Wevers RA. **Inborn errors of metabolism with 3-methylglutaconic aciduria as discriminative feature: Proper classification and nomenclature**. J Inherit Metab Dis. 2013 Jan 8. [Epub ahead of print] *(PubMed Abstract)*

- Lopes LR, Elliott PM. **Genetics of heart failure.** Biochim Biophys Acta. 2013 Jan 5. doi:pii: S0925-4439(13)00002-1. 10.1016/j.bbadis.2012.12.012. [Epub ahead of print] *(PubMed Abstract)*

- Falk MJ, Pierce EA, Consugar M, Xie MH, Guadalupe M, Hardy O, Rappaport EF, Wallace DC, LeProust E, Gai X. **Mitochondrial disease genetic diagnostics: Optimized whole-exome analysis for all mitocarta muclear genes and the mitochondrial genome.** Discov Med 2012 Dec 14(79):389-399. *(PubMed – Open Access)*

- Goldstein A, Bhatia P, Vento JM. **Update on nuclear mitochondrial genes and neurologic disorders.** Semin Pediatr Neurol. 2012 Dec;19(4):181-93. doi: 10.1016/j.spen.2012.09.005. *(PubMed Abstract)*

- Ojala T, Polinati P, Manninen T, Hiippala A, Rajantie J, Karikoski R, Suomalainen A, Tyni T. **New mutation of mitochondrial DNAJC19 causing dilated and noncompaction cardiomyopathy, anemia, ataxia, and male genital anomalies.** Pediatr Res. 2012 Oct;72(4):432-7. doi: 10.1038/pr.2012.92. Epub 2012 Jul 13. *(PubMed Abstract)*

- Ronvelia D, Greenwood J, Platt J, Hakim S, Zaragoza MV. **Intrafamilial variability for novel *TAZ* gene mutation: Barth syndrome with dilated cardiomyopathy and heart failure in an infant and left ventricular noncompaction in his great-uncle.** Mol Genet Metab. 2012 Sep 18. pii: S1096-7192(12)00354-X. doi: 10.1016/j.ymgme.2012.09.013. [Epub ahead of print] *(PubMed Abstract)*

- Kindel SJ, Miller EM, Gupta R, Cripe LH, Hinton RB, Spicer RL, Towbin JA, Ware SM. **Pediatric cardiomyopathy: Importance of genetic and metabolic evaluation.** J Card Fail. 2012 May;18(5):396-403. doi: 10.1016/j.cardfail.2012.01.017. Epub 2012 Mar 10. *(PubMed Abstract)*

- Cosson L, Toutain A, Simard G, Kulik W, G Matyas, Guichet A, Blasco H, Maakaroun-Vermesse Z, Vaillant MC, Le Caignec C, Chantepie A, Labarthe F. **Barth syndrome in a female patient.** Mol Genet Metab. 2012 May;106(1):115-20. doi: 10.1016/j.ymgme.2012.01.015. Epub 2012 Jan 24. *(PubMed Abstract)*

- El-Hafidi M, Meschini MC, Rizza T, Santorelli FM, Bertini E, Carrozzo R, Vázquez-Memije ME. **Cardiolipin content in mitochondria from cultured skin fibroblasts harboring mutations in the mitochondrial ATP6 gene.** J Bioenerg Biomembr. 2011 Oct 13. [Epub ahead of print] *(PubMed Abstract)*

- Jacoby D, McKenna WJ. **Genetics of inherited cardiomyopathy.** Eur Heart J. 2011 Aug 2. [Epub ahead of print] *(PubMed Abstract)*

- Swalwell H, Kirby DM, Blakely EL, Mitchell A, Salemi R, Sugiana C, Compton AG, Tucker EJ, Ke BX, Lamont PJ, Turnbull DM, McFarland R, Taylor RW, Thorburn DR. **Respiratory chain complex I deficiency caused by mitochondrial DNA mutations.** Eur J Hum Genet. 2011 Jul;19(7):769-75. doi: 10.1038/ejhg.2011.18. Epub 2011 Mar 2. *(PubMed Abstract)*

- Schiff M, Ogier de Baulny H, Lombès A. **Neonatal cardiomyopathies and metabolic crises due to oxidative phosphorylation defects.** Semin Fetal Neonatal Med. 2011 Aug;16(4):216-21. doi: 10.1016/j.siny.2011.04.002. Epub 2011 May 24. *(PubMed Abstract)*

- Taylor M, Slavov D, Salcedo E, Zhu X, Ferguson D, Jirikowic J, Di Lenarda A, Sinagra G, MD, Mestroni L. ***Tafazzin* gene mutations are uncommon causes of dilated cardiomyopathy in adults.** Cardiogenetics.2011.e4 | Published: 2011-07-05. *(Abstract)**▼

- Garratt V, Riddiford D, Steward C, Tsai-Goodman B, Newbury-Ecob R. **What is Barth syndrome?** Midwives Magazine: Issue 4: 2011. *(Not peer reviewed) (Abstract)*▼

- Oechslin E, Jenni R. **Left ventricular non-compaction revisited: A distinct phenotype with genetic heterogeneity?** Eur Heart J. 2011 Jun;32(12):1446-56. Epub 2011 Jan 31. *(PubMed Abstract)*

- Lisi EC, Cohn RD. **Genetic evaluation of the pediatric patient with hypotonia: Perspective from a hypotonia specialty clinic and review of the literature.** Dev Med Child Neurol. 2011 Jul;53(7):586-99. doi: 10.1111/j.1469-8749.2011.03918.x. Epub 2011 Mar 21. Review. *(PubMed Abstract)*

- Aljishi E, Ali F. **Barth syndrome: An X-linked cardiomyopathy with a novel mutation.** Indian J Pediatr. 2010 Dec;77(12):1432-3. Epub 2010 Oct 28. *(PubMed Abstract)*

- Hersberger RE, Morales A, Siegfried JD. **Clinical and genetic issues in dilated cardiomyopathy: A review for genetics professionals**. Genet Med. 2010 Nov;12(11):655-67. *(PubMed Abstract)*

- Koh C, Lee PW, Yung TC, Lun KS, Cheung YF. **Left ventricular noncompaction in children.** Congenit Heart Dis. 2009 Jul;4(4):288-94. *(PubMed Abstract)*

- Chang B, Momoi N, Shan L, Mitomo M, Aoyagi Y, Endo K, Takeda I, Chen R, Xing Y, Yu X, Watanabe S, Yoshida T, Kanegane H, Tsubata S, Bowles NE, Ichida F, Miyawaki T; Noncompaction study collaborators. **Gonadal mosaicism of a *TAZ* (G4.5) mutation in a Japanese family with Barth syndrome and left ventricular noncompaction.** Mol Genet Metab. 2010 Jun;100(2):198-203. Epub 2010 Mar 2. *(PubMed Abstract)*

- Zimmerman RS, Cox S, Lakdawala NK, Cirino A, Mancini-DiNardo D, Clark E, Leon A, Duffy E, White E, Baxter S, Alaamery M, Farwell L, Weiss S, Seidman CE, Seidman JG, Ho CY, Rehm HL, Funke BH. **A novel custom resequencing array for dilated cardiomyopathy.** Genet Med. 2010 May;12(5):268-78. *(PubMed Abstract)*

- Towbin JA, Sleeper L, Jefferies JL, Colan S, Webber SA, Canter CE, Hsu DT, Ware SM, Wilkinson JD, Orav EJ, Lipshultz SE. **Genetic and viral genome analysis of childhood cardiomyopathy: The PCMR/PCSR experience** [abstract]. In American College of Cardiology 59th Annual Scientific Session; 2010 Mar 14-16; Altanta, GA: J. Am. Coll. Cardiol. 2010;55;A43.E409. *(Abstract)*

- Ashworth M. **Cardiomyopathy in childhood: Histopathological and genetic features.** Pathology Journal, 2010, 4, 80-93. *(Open Access/Bentham)*

- Orstavik KH. **X-chromosome inactivation in clinical practice.** Hum Genet. 2009 Sep;126(3):363-73. Epub 2009 Apr 25. Review. *(PubMed Abstract)*

- Cosson L, Toutain A, Simard G, Paoli F, Kulik W, Vaz FM, Blasco H, Chantepie A, Labarthe F. **Barth syndrome in a female patient** [Abstract]. In the 11th International Congress of Inborn Errors of Metabolism Meeting, October 2009. Molecular Genetics and Metabolism 98, Issue 1 p. 89-118.

- Singh HR, Yang Z, Siddiqui S, Peña LS, Westerfield BH, Fan Y, Towbin JA, Vatta M. **A novel Alu-mediated Xq28 microdeletion ablates *TAZ* and partially deletes DNL1L in a patient with Barth syndrome.** Am J Med Genet A. 2009 May;149A(5):1082-5. *(PubMed – No Abstract Available)*

- Bachou T, Giannakopoulos A, Trapali C, Vazeou A, Kattamis A. **A novel mutation in the G4.5 (*TAZ*) gene in a Greek patient with Barth syndrome.** Blood Cells Mol Dis. 2009 May-Jun;42(3):262-4. Epub 2009 Mar 3. *(PubMed Abstract)*

- Osman C, Haag M, Potting C, Rodenfels J, Dip PV, Wieland FT, Brügger B, Westermann B, Langer T. **The genetic interactome of prohibitins: Coordinated control of cardiolipin and phosphatidylethanolamine by conserved regulators in mitochondria.** J Cell Biol. 2009 Feb 23;184(4):583-96. Epub 2009 Feb 16. *(PubMed Abstract)*

- Cortez-Dias N, Varela MG, Sargento L, Brito D, Almeida A, Cerqueira R, Lança V, Fernandes AR, Tavares P, Pereira RA, Fernandes A, Madeira H. **Left ventricular non-compaction: A new mutation predisposing to reverse remodeling?** Rev Port Cardiol. 2009 Feb;28(2):185-94. *(PubMed Abstract)*

- Tikhomirov E, Averyanova N, Basargina E, Degtyareva T. **Gene symbol: *TAZ*. Disease: Barth syndrome.** Hum Genet. 2008 Oct;124(3):315-6. *(PubMed - No Abstract Available)*

- Menon SC, Olson TM, Michels VV. **Genetics of familial dilated cardiomyopathy.** Progress in Pediatric Cardiology Volume 25, Issue 1, April 2008, Pages 57-67. *(Abstract)*

- Cox GF. **Diagnostic approaches to pediatric cardiomyopathy of metabolic genetic etiologies and their relation to therapy.** Prog Pediatr Cardiol. 2007;24(1):15-25. Prog Pediatr Cardiol. 2007;24(1):15-25. *(PubMed Abstract)*

- Kirwin SM, Vinette KM, Schwartz SB, Funanage VL, Gonzalez IL. **Multiple transmissions of Barth syndrome through an oocyte donor with a de novo *TAZ* mutation.** Fertil Steril. 2007 Apr;87(4):976.e5-7. Epub 2007 Jan 22. *(PubMed Abstract)**

- van Werkhoven MA, Thorburn DR, Gedeon AK, Pitt JJ. **Monolysocardiolipin in cultured fibroblasts is a sensitive and specific marker for Barth syndrome.** J Lipid Res. 2006 Oct;47(10):2346-51. Epub 2006 Jul 27. *(PubMed Abstract)*

- Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, Ren M, Schlame M. **A Drosophila model of Barth syndrome**. Proc Natl Acad Sci U S A. 2006 Aug 1;103(31):11584-8. Epub 2006 Jul 19. *(PubMed – Open Access)**

- Khuchua Z, Yue Z, Batts L, Strauss AW. **A zebrafish model of human Barth syndrome reveals the essential role of *tafazzin* in cardiac development and function.** Circ Res. 2006 Jul 21;99(2):201-8. Epub 2006 Jun 22. *(PubMed – Open Access)**

- Xing Y, Ichida F, Matsuoka T, Isobe T, Ikemoto Y, Higaki T, Tsuji T, Haneda N, Kuwabara A, Chen R, Futatani T, Tsubata S, Watanabe S, Watanabe K, Hirono K, Uese K, Miyawaki T, Bowles KR, Bowles NE, Towbin JA. **Genetic analysis in patients with left ventricular noncompaction and evidence for genetic heterogeneity.** Mol Genet Metab. 2006 May;88(1):71-7. Epub 2006 Jan 19. *(PubMed Abstract)*

- Ellinor PT, Milan DJ, MacRae CA. **Metabolic gene defects and risk of arrhythmia.** Heart Metab. 2006;33:9-12. *(Abstract)*

- Ahmad, F, Seidman, JG, Seidman, C. **The genetic basis for cardiac remodeling.** Annu. Rev. Genomics Hum. Genet. 2005.6:185-216. *(PubMed Abstract)*

- Gonzalez IL. **Barth syndrome: *TAZ* gene mutations, mRNAs, and evolution**. Am J Med Genet A. 2005 May 1;134(4):409-14. *(PubMed Abstract)**

- Gonzalez IL. ***TAZ* mRNAs in Barth syndrome subjects, alternative splicing and exon evolution** [abstract]. In American Society of Human Genetics Annual Meeting, October 26-30, 2004, Toronto, Canada. *(Abstract)*

- Vaz FM, Houtkooper RH, Valianpour F, Barth PG, Wanders RJ. **Only one splice variant of the human *TAZ* gene encodes a functional protein with a role in cardiolipin metabolism.** J Biol Chem. 2004 Jun 18;279(25):26802. *(Additions and Corrections) (PubMed Abstract)**

- Kenton AB, Sanchez X, Coveler KJ, Makar KA, Jimenez S, Ichida F, Murphy RT, Elliott PM, McKenna W, Bowles NE, Towbin JA, Bowles KR. **Isolated left ventricular noncompaction is rarely caused by mutations in G4.5, alpha-dystrobrevin and FK binding protein-12.** Mol Genet Metab. 2004 Jun; 82(2):162-6. *(PubMed Abstract)*

- Vaz FM, Houtkooper RH, Valianpour F, Barth PG, Wanders RJ. **Only one splice variant of the human *TAZ* gene encodes a functional protein with a role in cardiolipin metabolism.** J Biol Chem 2003 Oct 31; 278(44):43089-94. Epub 2003 Aug 20. *(PubMed – Open Access)**

- Vesel S, Stopar-Obreza M, Trebusak-Podkrajsek K, Jazbec J, Podnar T, Battelino T. **A novel mutation in the G4.5 (*TAZ*) gene in a kindred with Barth syndrome**. Eur J Hum Genet 2003 Jan; 11(1):97-101. *(PubMed Abstract)*

- Chen R, Tsuji T, Ichida F, Bowles KR, Yu X, Watanabe S, Hirono K, Tsubata S, Hamamichi Y, Ohta J, Imai Y, Bowles NE, Miyawaki T, Towbin JA. **Mutation analysis of the G4.5 gene in patients with isolated left ventricular noncompaction**. Mol Genet Metab 2002 Dec; 77(4):319-325. *(PubMed Abstract)*

- Chen Y. Molecular studies on a *Dictyostelium* homolog of the *taffazin* gene, the cause of Barth syndrome in humans. Dr. rer. nat. dissertation; University of Kassel; November 2002. *(Not peer reviewed).*

- Gonzalez IL. **Genetic disease and DNA for the families and children.** June 2002.

- Sakamoto O, Kitoh T, Ohura T, Ohya N, Iinuma K. **Novel missense mutation (R94S) in the *TAZ* (G4.5) gene in a Japanese patient with Barth syndrome**. J Hum Genet. 2002;47(5):229-31. *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Sakamoto O, Ohura T, Katsushima Y, Fujiwara I, Ogawa E, Miyabayashi S, Iinuma K. **A novel intronic mutation of the *TAZ* (G4.5) gene in a patient with Barth syndrome: Creation of a 5' splice donor site with variant GC consensus and elongation of the upstream exon**. Hum Genet 2001 Nov; 109(5):559-563. *(PubMed Abstract)*

- DiMauro S, Schon EA. **Mitochondrial DNA mutations in human disease**. Am J Med Genet. 2001 Spring; 106(1):18-26. *(PubMed Abstract)*

- DeKremer RD, Paschini-Capra A, Bacman S, Argarana C, Civallero G, Kelley RI, Guelbert N, Latini A, deHalac IN, Giner-Ayala A, Johnston J, Proujansky R, Gonzalez I, Depetris-Boldini C, Oller-Ramirez A, Angaroni C, Theaux RA, Hliba E, Juaneda E. **Barth's syndrome-like disorder: A new phenotype with a maternally inherited A3243G substitution of mitochondrial DNA (MELAS mutation)**. Am J Med Genet. 2001 Mar 1;99(2):83-93. *(PubMed Abstract)*

- Ichida F, Tsubata S, Bowles KR, Haneda N, Uses K, Miyawaki T, Dreyer WJ, Messina J, Li H, Bowles NE, Towbin JA. **Novel gene mutations in patients with left ventricular noncompaction or Barth syndrome**. Circulation. 2001 Mar 6;103(9):1256-63. *(PubMed Abstract)*

- Towbin JA, Bowles NE. **Molecular genetics of left ventricular dysfunction.** Curr Mol Med. 2001 Mar;1(1):81-90. *(PubMed Abstract)*

- Frolov MV, Benevolenskaya EV, Birchler JA. **The oxen gene of Drosophila encodes a homolog of subunit 9 of yeast ubiquinol-cytochrome c oxidoreductase complex: Evidence for modulation of gene expression in response to mitochondrial activity.** Genetics. 2000 Dec;156(4):1727-36. *(PubMed Abstract)*

- Arbustini E, Morbini P, Pilotto A, Gavazzi A, Tavazzi L. **Genetics of idiopathic dilated cardiomyopathy.** Herz. 2000 May;25(3):156-60. *(PubMed Abstract)*

- Gonzalez IL, Johnston J, Funanage V, Proujansky R. **Frequency of new mutations and allele drop-out in Barth syndrome.** Am J Hum Genet 2000; 67(supplement):173. *(Abstract)*

- Seliem MA, Mansara KB, Palileo M, Ye X, Zhang Z, Benson WD. **Evidence of autosomal recessive inheritance of infantile dilated cardiomyopathy: Studies from the Eastern Province of Saudi Arabia.** Pediatr Res. 2000 Dec;48(6):770-5. *(PubMed Abstract)*

- Komajda, M (2000). **Genetics of dilated cardiomyopathy: A molecular maze?** Heart. 2000 Nov;84(5):463-4. *(PubMed Abstract)*

- Cantlay AM, Shokrollahi K, Allen JT, Lunt PW, Newbury-Ecob RA, Steward CG. **Genetic analysis of the G4.5 gene in families with suspected Barth syndrome**. J Pediatr. 1999 Sep;135(3):311-5. *(PubMed Abstract)*

- Orstavik KH, Orstavik RE, Naumova AK, D'Adamo P, Gedeon A, Bolhuis PA, Barth PG, Toniolo D. **X chromosome inactivation in carriers of Barth syndrome**. Am J Hum Genet. 1998 Nov;63(5):1457-63. *(PubMed Abstract)*

- Johnston J, Kelley RI, Feigenbaum A, Cox GF, Iyer GS, Funanage VL, Proujansky R. **Mutation characterization and genotype-phenotype correlation in Barth syndrome**. Am J Hum Genet. 1997 Nov;61(5):1053-8. *(PubMed Abstract)*

- D'Adamo P, Fassone L, Gedeon A, Janssen EA, Bione S, Bolhuis PA, Barth PG, Wilson M, Haan E, Orstavik KH, Patton MA, Green AJ, Zammarchi E, Donati MA, Toniolo D. **The X-linked gene G4.5 is responsible for different infantile dilated cardiomyopathies**. Am J Hum Genet. 1997 Oct;61(4):862-7. *(PubMed Abstract)*

- Bleyl SB, Mumford BR, Thompson V, Carey JC, Pysher TJ, Chin TK, Ward K. **Neonatal, lethal noncompaction of the left ventricular myocardium is allelic with Barth syndrome**. Am J Hum Genet. 1997 Oct;61(4):868-72. *(PubMed Abstract)*

- Sandoval N, Bauer D, Brenner V, Coy JF, Drescher B, Kioschis P, Korn B, Nyakatura G, Poustka A, Reichwald K, Rosenthal A, Platzer M. **The genomic organization of a human creatine transporter (CRTR) gene located in Xq28**. Genomics. 1996 Jul 15;35(2):383-5. *(PubMed Abstract)*

- Heiss NS, Rogner UC, Kioschis P, Korn B, Poustka A. **Transcription mapping in a 700-kb region around the DXS52 locus in Xq28: Isolation of six novel transcripts and a novel ATPase isoform (hPMCA5)**. Genome Res. 1996 Jun;6(6):478-91. *(PubMed Abstract)*

- Bione S, D'Adamo P, Maestrini E, Gedeon AK, Bolhuis PA, Toniolo D. **A novel X-linked gene, G4.5, is responsible for Barth syndrome**. Nat Genet 1996 Apr; 12(4):385-389. *(PubMed Abstract)*

- The American Society of Human Genetics Board of Directors and The American College of Medical Genetics Board of Directors**. ASHG/ACMG Report – Points to consider: Ethical, legal, and psychosocial implications of genetic testing in children and adolescents**. Am J Hum Genet. 1995 Nov;57(5):1233-41. *(PubMed Abstract)*

- Gedeon AK, Wilson MJ, Colley AC, Sillence DO, Mulley JC. **X linked fatal infantile cardiomyopathy maps to Xq28 and is possibly allelic to Barth syndrome**. J Med Genet. 1995 May;32(5):383-8. *(PubMed Abstract)*

- Kelly DP, Strauss AW. **Inherited cardiomyopathies.** N Engl J Med. 1994 Mar 31;330(13):913-9. No abstract available. *(PubMed Abstract)*

- Christodoulou J, McInnes RR, Jay V, Wilson G, Becker LE, Lehotay DC, Platt BA, Bridge PJ, Robinson BH, Clarke JT. **Barth syndrome: Clinical observations and genetic linkage studies**. Am J Med Genet. 1994 Apr 15;50(3):255-64. *(PubMed Abstract)*

- Orstavik KH, Skjorten F, Hellenbostad M, Haga P, Langslet A. **Possible X linked congenital mitochondrial cardiomyopathy in three families**. J Med Genet. 1993 Apr;30(4):269-72. *(PubMed Abstract)*

- Ades LC, Gedeon AK, Wilson MJ, Latham M, Partington MW, Mulley JC, Nelson J, Lui K, Sillence DO. **Barth syndrome: Clinical features and confirmation of gene localisation to distal Xq28**. Am J Med Genet. 1993 Feb 1;45(3):327-34. *(PubMed Abstract)*

- Bolhuis PA, Hensels GW, Hulsebos TJ, Baas F, Barth PG. **Mapping of the locus for X-linked cardioskeletal myopathy with neutropenia and abnormal mitochondria (Barth syndrome) to Xq28**. Am J Hum Genet. 1991 Mar;48(3):481-5. *(PubMed Abstract)*

- Hodgson S, Child A, Dyson M. **Endocardial fibroelastosis: Possible X linked inheritance.** J Med Genet. 1987 Apr;24(4):210-4. *(PubMed Abstract)*

MITOCHONDRIAL DISORDERS

NEW Kojima R, Kakimoto Y, Furuta S, Itoh K, Sesaki H, Endo T, Tamura Y. **Maintenance of cardiolipin and crista structure requires cooperative functions of mitochondrial dynamics and phospholipid transport.** Cell Rep. 2019 Jan 15;26(3):518-528.e6. doi: 10.1016/j.celrep.2018.12.070. *(PubMed Abstract)*

NEW Van Strien J, Guerrero-Castillo S, Chatzispyrou IA, Houtkooper RH, Brandt U, Huynen MA. **COmplexome profiling aLignment (COPAL) reveals remodeling of mitochondrial protein complexes in Barth syndrome.** Bioinformatics. 2019 Jan 14. doi: 10.1093/bioinformatics/btz025. [Epub ahead of print] *(PubMed Abstract)*

NEW Gürtler S, Wolke C, Otto O, Heise N, Scholz F, Laporte A, Elsner M, Jörns A, Weinert S, Döring M, Jansing S, Gardemann A, Lendeckel U, Schild L. ***Tafazzin*-dependent cardiolipin composition in C6 glioma cells correlates with changes in mitochondrial and cellular functions, and cellular proliferation.** Biochim Biophys Acta Mol Cell Biol Lipids. 2019 Jan 9. pii: S1388-1981(19)30003-4. doi: 10.1016/j.bbalip.2019.01.006. [Epub ahead of print] *(PubMed Abstract)*

NEW Antunes D, Chowdhury A, Aich A, Saladi S, Harpaz N, Stahl M, Schuldiner M, Herrmann JM, Rehling P, Rapaport D. **Overexpression of branched-chain amino acid aminotransferases rescues the growth defects of cells lacking the Barth syndrome-related gene *TAZ*1.** J Mol Med (Berl). 2019 Jan 3. doi: 10.1007/s00109-018-1728-4. [Epub ahead of print] *(PubMed Abstract)**

NEW Kim J, Lee K, Fujioka H, Tandler B, Hoppel CL. **Cardiac mitochondrial structure and function in *tafazzin*-knockdown mice.** Mitochondrion. 2018 Oct 30. pii: S1567-7249(18)30074-6. doi: 10.1016/j.mito.2018.10.005. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Chao H, Anthonymuthu TS, Kenny EM, Amoscato AA, Cole LK, Hatch GM, Ji J, Kagan VE, Bayır H. **Disentangling oxidation/hydrolysis reactions of brain mitochondrial cardiolipins in pathogenesis of traumatic injury.** JCI Insight. 2018 Nov 2;3(21). pii: 97677. doi: 10.1172/jci.insight.97677. [Epub ahead of print] *(PubMed – Open Access)**

NEW Szeto HH. **Stealth peptides target cellular powerhouses to fight rare and common age-related diseases.** Protein Pept Lett. 2018 Oct 31. doi: 10.2174/0929866525666181101105209. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Chowdhury A, Aich A, Jain G, Wozny K, Lüchtenborg C, Hartmann M, Bernhard O, Balleiniger M, Alfar EA, Zieseniss A, Toischer K, Guan K, Rizzoli SO, Brügger B, Fischer A, Katschinski DM, Rehling P, Dudek J. **Defective mitochondrial cardiolipin remodeling dampens HIF-1α expression in hypoxia.** Cell Rep. 2018 Oct 16;25(3):561-570.e6. doi:10.1016/j.celrep.2018.09.057. *(PubMed – Open Access)*▼

NEW Li M, Hou T, Gao T, Lu X, Yang Q, Zhu Q, Li Z, Liu C, Mu G, Liu G, Bao Y, Wen H, Wang L, Wang H, Zhao Y, Gu W, Yang Y, Zhu WG. **p53 cooperates with SIRT6 to regulate cardiolipin de novo biosynthesis.** Cell Death Dis. 2018 Sep 20;9(10):941. doi: 10.1038/s41419-018-0984-0. *(PubMed - Open Access)*

NEW Chatzispyrou IA, Guerrero-Castillo S, Held NM, Ruiter JPN, Denis SW, IJlst L, Wanders RJ, van Weeghel M, Ferdinandusse S, Vaz FM, Brandt U, Houtkooper RH. **Barth syndrome cells display widespread remodeling of mitochondrial complexes without affecting metabolic flux distribution**. Biochim Biophys Acta Mol Basis Dis. 2018 Sep 1;1864(11):3650-3658. doi: 10.1016/j.bbadis.2018.08.041. [Epub ahead of print] *(PubMed Abstract)*▼

NEW Pennington ER, Sullivan EM, Fix A, Dadoo S, Zeczycki TN, DeSantis A, Schlattner U, Coleman RA, Chicco AJ, Brown DA, Shaikh SR. **Proteolipid domains form in biomimetic and cardiac mitochondrial vesicles and are regulated by cardiolipin concentration but not monolyso-cardiolipin.** J Biol Chem. 2018 Aug 29. pii: jbc.RA118.004948. doi: 10.1074/jbc.RA118.004948. [Epub ahead of print] *(PubMed - Open Access)*

NEW Dudek J, Hartmann M, Rehling P. **The role of mitochondrial cardiolipin in heart function and its implication in cardiac disease.** Biochimica et Biophysica Acta (BBA) - Molecular Basis of Disease, In press, corrected proof, Available online 26 August 2018. *(ScienceDirect Abstract)*

NEW Paradies G, Paradies V, Ruggiero FM, Petrosillo G. **Mitochondrial bioenergetics and cardiolipin alterations in myocardial ischemia/reperfusion injury. Implications for pharmacological cardioprotection.** Am J Physiol Heart Circ Physiol. 2018 Aug 10. doi: 10.1152/ajpheart.00028.2018. [Epub ahead of print] *(PubMed Abstract)*

NEW Suzuki-Hatano S, Saha M, Rizzo SA, Witko RL, Gosiker BJ, Ramanathan M, Soustek MS, Jones MD, Kang PB, Byrne BJ, Cade WT, Pacak CA. **AAV-mediated *TAZ* gene replacement restores mitochondrial and cardioskeletal function in Barth syndrome.** Hum Gene Ther. 2018 Aug 2. doi: 10.1089/hum.2018.020. [Epub ahead of print]*

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Johnson JM, Ferrara PJ, Verkerke ARP, Coleman CB, Wentzler EJ, Neufer PD, Kew KA, de Castro Brás LE, Funai K. **Targeted overexpression of catalase to mitochondria does not prevent cardioskeletal myopathy in Barth syndrome.** J Mol Cell Cardiol. 2018 Jul 2;121:94-102. doi: 10.1016/j.yjmcc.2018.07.001. [Epub ahead of print]

- Xu G, Liu X, Shu Y, Pillai JA, Xu Y. **A rapid and sensitive LC-MS/MS method for quantitative analysis of cardiolipin (18:2)4 in human leukocytes and mouse skeletal muscles.** J Pharm Biomed Anal. 2018 Jun 20;158:386-394. doi: 10.1016/j.jpba.2018.06.035. [Epub ahead of print] *(PubMed Abstract)*

- Basu Ball W, Baker CD, Neff JK, Apfel GL, Lagerborg KA, Žun G, Petrovič U, Jain M, Gohil VM. **Ethanolamine ameliorates mitochondrial dysfunction in cardiolipin-deficient yeast cells.** J Biol Chem. 2018 Jun 4. pii:jbc.RA118.004014. doi:10.1074/jbc.RA118.004014. [Epub ahead of print] *(PubMed - Open Access)*

- Ban T, Kohno H, Ishihara T, Ishihara N. **Relationship between OPA1 and cardiolipin in mitochondrial inner-membrane fusion.** Biochim Biophys Acta. 2018 May 28. pii: S0005-2728(18)30132-4. doi: 10.1016/j.bbabio.2018.05.016. [Epub ahead of print] *(PubMed Abstract)*

- Cade WT, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, Bashir A, Byrne BJ, Taylor CL. **Peak oxygen uptake (VO2peak) across childhood, adolescence and young adulthood in Barth syndrome: Data from cross-sectional and longitudinal studies.** PLoS One. 2018 May 24;13(5):e0197776. doi: 10.1371/journal.pone.0197776. eCollection 2018 *(PubMed - Open Access)**▼

- Oemer G, Lackner K, Muigg K, Krumschnabel G, Watschinger K, Sailer S, Lindner H, Gnaiger E, Wortmann SB, Werner ER, Zschocke J, Keller MA. **Molecular structural diversity of mitochondrial cardiolipins.** Proc Natl Acad Sci U S A. 2018 Apr 4. pii: 201719407. doi: 10.1073/pnas.1719407115. [Epub ahead of print] *(PubMed Abstract)*

- Chabi B, Fouret G, Lecomte J, Cortade F, Pessemesse L, Baati N, Coudray C, Lin L, Tong Q, Wrutniak-Cabello C, Casas F, Feillet-Coudray C. **Skeletal muscle overexpression of short isoform Sirt3 altered mitochondrial cardiolipin content and fatty acid composition.** J Bioenerg Biomembr. 2018 Mar 27. doi: 10.1007/s10863-018-9752-1. [Epub ahead of print] *(PubMed Abstract)*

- Mejia EM, Zegallai H, Bouchard ED, Banerji V, Ravandi A, Hatch GM. **Expression of human monolysocardiolipin acyltransferase-1 improves mitochondrial function in Barth syndrome lymphoblasts.** J Biol Chem. 2018 Mar 21. pii:jbc.RA117.001024. doi:10.1074/jbc.RA117.001024. [Epub ahead of print] *(PubMed - Open Access)**▼

- Chen WW, Chao YJ, Chang WH, Chan JF, Hsu YHH. **Phosphatidylglycerol incorporates into cardiolipin to improve mitochondrial activity and inhibits inflammation.** Sci Rep. 2018 Mar 20;8(1):4919. doi: 10.1038/s41598-018-23190-z. *(PubMed – Abstract)*

- Sudrié-Arnaud B, Marguet F, Patrier S, Martinovic J, Louillet F, Broux F, Charbonnier F, Dranguet H, Coutant S, Vezain M, Lanos R, Tebani A, Fuller M, Lamari F, Chambon P, Brehin AC, Trestard L, Tournier I, Marret S, Verspyck E, Laquerrière A, Bekri S. **Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation.** Clin Chim Acta. 2018 Feb 21. pii: S0009-8981 (18)30084-6. doi: 10.1016/j.cca.2018.02.023. [Epub ahead of print] (*PubMed Abstract)*

- Thong A, Tsoukanova V. **Cytochrome-c-assisted escape of cardiolipin from a model mitochondrial membrane.** Biochim Biophys Acta. 2018 Feb;1860(2):475-480. doi: 10.1016/j.bbamem.2017.10.032. Epub 2017 Nov 4. (*PubMed Abstract)*

- Sullivan EM, Pennington ER, Sparagna GC, Torres MJ, Neufer PD, Harris M1, Washington J, Anderson EJ, Zeczycki TN, Brown DA, Shaikh SR. **Docosahexaenoic acid lowers cardiac mitochondrial enzyme activity by replacing linoleic acid in the phospholipidome.** J Biol Chem. 2018 Jan 12;293(2):466-483. doi: 10.1074/jbc.M117.812834. Epub 2017 Nov 21. (*PubMed Abstract)*

- Serricchio M, Vissa A, Kim PK, Yip CM, McQuibban GA. **Cardiolipin synthesizing enzymes form a complex that interacts with cardiolipin-dependent membrane organizing proteins.** Biochim Biophys Acta. 2018 Jan 14. pii: S1388-1981(18)30010-6. doi:10.1016/j.bbalip.2018.01.007. [Epub ahead of print] (*PubMed Abstract)*

- Ohlig T, Le DV, Gardemann A, Wolke C, Gürtler S, Peter D, Schild L, Lendeckel U. **Effects of siRNA-dependent knock-down of cardiolipin synthase and *tafazzin* on mitochondria and proliferation of glioma cells.** Biochima et Biophysica Acta. Jan 6, 2018. (*PubMed Abstract*)

- Imai-Okazaki A, Kishita Y, Kohda M, Yatsuka Y, Hirata T, Mizuno Y, Harashima H, Hirono K, Ichida F, Noguchi A, Yoshida M, Tokorodani C, Nishiuchi R, Takeda A, Nakaya A, Sakata Y, Murayama K, Ohtake A, Okazaki Y. **Barth syndrome: Different approaches to diagnosis.** J Pediatr. 2017 Dec 15. pii: S0022-3476(17)31329-X. doi: 10.1016/j.jpeds.2017.09.075. [Epub ahead of print] *(PubMed Abstract)*

- Dinca AA, Chien WM, Chin MT. **Identification of novel mitochondrial localization signals in human *tafazzin*, the cause of the inherited cardiomyopathic disorder Barth syndrome.** J Mol Cell Cardiol. 2017 Nov 9. pii: S0022-2828(17)30340-1. doi:10.1016/j.yjmcc.2017.11.005. [Epub ahead of print] *(PubMed Abstract)**

- Pointer CB, Klegeris A. **Cardiolipin in central nervous system physiology and pathology.** Cell Mol Neurobiol. 2017 Oct;37(7):1161-1172. doi: 10.1007/s10571-016-0458-9. Epub 2016 Dec 30. Review. *(PubMed Abstract)*

- Enns GM. **Pediatric mitochondrial diseases and the heart.** Curr Opin Pediatr. 2017 Oct;29(5):541-551. doi: 10.1097/MOP.0000000000000535. *(PubMed Abstract)*

- Dudek J. **Role of cardiolipin in mitochondrial signaling pathways.** Front Cell Dev Biol. 2017 Sep 29;5:90. doi: 10.3389/fcell.2017.00090. eCollection 2017. Review. *(PubMed - Open Access)*

- Towbin JA, Jefferies JL. **Cardiomyopathies due to left ventricular noncompaction, mitochondrial and storage diseases, and inborn errors of metabolism.** Circ Res. 2017 Sep 15;121(7):838-854. doi: 10.1161/CIRCRESAHA.117.310987. Review. *(PubMed Abstract)*

- Malhotra K, Modak A, Nangia S, Daman, TH, Gunsel U, Robinson VL, Mokranjac D, May ER, Alder NN. **Cardiolipin mediates membrane and channel interactions of the mitochondrial TIM23 protein.** Sci Adv. 2017 Sep 1;3(9):e1700532. doi: 10.1126/sciadv.1700532. eCollection 2017 Sep. *(PubMed - Open Access)*

- Saneto RP. **Genetics of mitochondrial disease.** Adv Genet. 2017;98:63-116. doi:10.1016/bs.adgen.2017.06.002. Epub 2017 Sep 1. *(PubMed Abstract)*

- Chen Z, Wu Y, Ma YS, Kobayashi Y, Zhao YY, Miura Y, Chiba H, Hui SP. **Profiling of cardiolipins and their hydroperoxides in HepG2 cells by LC/MS.** Anal Bioanal Chem. 2017 Jul 31. doi: 10.1007/s00216-017-0515-3. [Epub ahead of print] *(PubMed Abstract)*

- Ramachandran R. **Mitochondrial dynamics: The dynamin superfamily and execution by collusion.** Semin Cell Dev Biol. 2017 Jul 25. pii: S1084-9521(17)30365-8. doi: 10.1016/j.semcdb.2017.07.039. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jang S, Javadov S. **Association between ROS production, swelling and the respirasome integrity in cardiac mitochondria.** Arch Biochem Biophys. 2017 Jul 20;630:1-8. doi: 10.1016/j.abb.2017.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Sullivan EM, Fix A, Crouch MJ, Sparagna GC, Zeczycki TN, Brown DA, Shaikh SR. **Murine diet-induced obesity remodels cardiac and liver mitochondrial phospholipid acyl chains with differential effects on respiratory enzyme activity.** J Nutr Biochem. 2017 Jul;45:94-103. doi: 10.1016/j.jnutbio.2017.04.004. Epub 2017 Apr 12. *(PubMed Abstract)*

- Fajardo VA, Mikhaeil JS, Leveille CF, Saint C, LeBlanc PJ. **Cardiolipin content, linoleic acid composition, and *tafazzin* expression in response to skeletal muscle overload and unload stimuli.** Sci Rep. 2017 May 17;7(1):2060. doi:10.1038/s41598-017-02089-1. *(PubMed - Open Access)*

- de Taffin de Tilques M, Tribouillard-Tanvier D, Tétaud E1, Testet E, di Rago JP1, Lasserre JP. **Overexpression of mitochondrial oxodicarboxylate carrier (ODC1) preserves oxidative phosphorylation in a yeast model of Barth syndrome.** Dis Model Mech. 2017 Apr 1;10(4):439-450. doi: 10.1242/dmm.027540. Epub 2017 Feb 10. *(PubMed – Open Access)*

- Sheeran FL, Pepe S. **Mitochondrial bioenergetics and dysfunction in failing heart.** Adv Exp Med Biol. 2017;982:65-80. doi: 10.1007/978-3-319-55330-6_4. (*PubMed Abstract)*

- Zhang Y, Bharathi SS, Rardin MJ, Lu J, Maringer KV, Sims-Lucas S, Prochownik EV, Gibson BW, Goetzman ES. **SIRT5 binds to cardiolipin and regulates the electron transport chain.** J Biol Chem. 2017 Apr 30. pii: jbc.M117.785022. doi: 10.1074/jbc.M117.785022. [Epub ahead of print] *(PubMed - Open Access)*

- Ikon N, Ryan RO. **Cardiolipin and mitochondrial cristae organization.** Biochim Biophys Acta. 2017 Mar 20. pii: S0005-2736(17)30095-0. doi: 10.1016/j.bbamem.2017.03.013. [Epub ahead of print] Review. (*PubMed Abstract)*

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access*)*

BSF does not endorse any drugs, tests, or treatments that we may report.

- Lloyd DF, Vara R, Mathur S. **The cardiac manifestations of inherited metabolic diseases in children.** Pediatr Int. 2017 Mar 4. doi: 10.1111/ped.13272. [Epub ahead of print] Review. (*PubMed Abstract)*

- Schlame M, Xu Y, Ren M. **The basis for acyl specificity in the *tafazzin* reaction.** J Biol Chem. 2017 Mar 31;292(13):5499-5506. doi: 10.1074/jbc.M116.769182. Epub 2017 Feb 15. (*PubMed Abstract)*

- Bashir A, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, de Las Fuentes L, Pacak CA, Byrne BJ, Cade WT**. Impaired cardiac and skeletal muscle bioenergetics in children, adolescents, and young adults with Barth syndrome.** Physiol Rep. 2017 Feb;5(3). pii: e13130. doi: 10.14814/phy2.13130. *(PubMed – Open Access)*▼

- de Taffin de Tilques M, Tribouillard-Tanvier D, Tétaud E, Testet E, di Rago JP, Lasserre JP. **Overexpression of mitochondrial oxodicarboxylate carrier (ODC1) preserves oxidative phosphorylation in a yeast model of the Barth syndrome.** Dis Model Mech. 2017 Feb 10. pii: dmm.027540. doi: 10.1242/dmm.027540. [Epub ahead of print] *(PubMed – Open Access*)

- Sathappa M, Mitchell W, Coscia A, Boyd K, May E, Szeto HH, Alder NN. **Investigation of the interactions of the SS-31 peptides with cardiolipin variants: A potential therapeutic for Barth syndrome.** Biophysical Journal, Volume 112, Issue 3, Supplement 1, 3 February 2017, Pages 438a. (*ScienceDirect Abstract)*

- Dudek J, Maack C. **Barth syndrome cardiomyopathy.** Cardiovasc Res. 2017 Feb 2. di: 10.1093/cvr/cvx014. [Epub ahead of print] No abstract available. *(PubMed)*

- Borna NN, Kishita Y, Ishikawa K, Nakada K, Hayashi JI, Tokuzawa Y, Kohda M, Nyuzuki H, Yamashita-Sugahara Y, Nasu T, Takeda A, Murayama K, Ohtake A, Okazaki Y. **A novel mutation in *TAZ* causes mitochondrial respiratory chain disorder without cardiomyopathy.** J Hum Genet. 2017 Jan 26. doi: 10.1038/jhg.2016.165. [Epub ahead of print] *(PubMed Abstract)*

- Kagan VE, Bayır H, Tyurina YY, Bolevich SB, Maguire JJ, Fadeel B, Balasubramanian K. **Elimination of the unnecessary: Intra- and extracellular signaling by anionic phospholipids.** Review Article. Biochemical and Biophysical Research Communications, Volume 482, Issue 3, 15 January 2017, Pages 482-490. *(PubMed Abstract*)*

- Schlame M, Greenberg ML. **Biosynthesis, remodeling and turnover of mitochondrial cardiolipin.** Biochim Biophys Acta. 2017 Jan;1862(1):3-7. doi:10.1016/j.bbalip.2016.08.010. Review. *(PubMed Abstract)*

- Shen Z, Li Y, Gasparski AN, Abeliovich H, Greenberg ML. **Cardiolipin regulates mitophagy through the PKC pathway.** J Biol Chem. 2017 Jan 5. pii: jbc.M116.753574. doi:10.1074/jbc.M116.753574. [Epub ahead of print] *(PubMed - Open Access)*

- Mejia EM, Zinko JC, Hauff KD, Xu FY, Ravandi A, Hatch GM. **Glucose uptake and triacylglycerol synthesis are increased in Barth syndrome lymphoblasts.** Lipids. 2017 Jan 17. doi: 10.1007/s11745-017-4232-7. [Epub ahead of print] *(PubMed Abstract)**

- Woiewodski L, Ezon D, Cooper J, Feingold B. **Barth syndrome with late-onset cardiomyopathy: A missed opportunity for diagnosis.** The Journal of Pediatrics, In Press, Corrected Proof, Available online 18 January 2017. *(Abstract)*

- Ikon N, Ryan RO. **Barth syndrome: Connecting cardiolipin to cardiomyopathy**. Lipids. 2017 Jan 9. doi: 10.1007/s11745-016-4229-7. [Epub ahead of print] *(PubMed Abstract)*

- Dong X, Fan P, Tian T, Yang Y, Xiao Y, Yang K, Liu Y, Zhou X. **Recent advancements in the molecular genetics of left ventricular noncompaction cardiomyopathy.** Clin Chim Acta. 2016 Dec 15;465:40-44. doi:10.1016/j.cca.2016.12.013. [Epub ahead of print] Review. *(PubMed Abstract)*

- Raja V, Joshi AS, Li G, Maddipati KR, Greenberg ML. **Loss of cardiolipin leads to perturbation of acetyl-CoA synthesis.** J Biol Chem. 2016 Dec 9. pii: jbc.M116.753624. [Epub ahead of print] *(PubMed - Open Access)**

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)*▼

- Kooijman EE, Swim LA, Graber ZT, Tyurina YY, Bayir H, Kagan VE. **Magic angle spinning ^{31}P NMR spectroscopy reveals two essentially identical ionization states for the cardiolipin phosphates in phospholipid liposomes.** Biochim Biophys Acta. 2016 Oct 26. pii: S0005-2736(16)30346-7. doi:10.1016/j.bbamem.2016.10.013. [Epub ahead of print] *(PubMed Abstract)**

- Oláhová M, Thompson K, Hardy SA, Barbosa IA, Besse A, Anagnostou ME, White K, Davey T, Simpson MA, Champion M, Enns G, Schelley S, Lightowlers RN, Chrzanowska-Lightowlers ZM, McFarland R, Deshpande C, Bonnen PE, Taylor RW. **Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria.** J Inherit Metab Dis. 2016 Sep 30. *(PubMed Abstract)*

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review. *(PubMed Abstract)*

- Rampelt H, Zerbes RM, van der Laan M, Pfanner N. **Role of the mitochondrial contact site and cristae organizing system in membrane architecture and dynamics.** Biochim Biophys Acta. 2016 Sep 7. pii: S0167-4889(16)30221-X. doi: 10.1016/j.bbamcr.2016.05.020. [Epub ahead of print] Review. *(PubMed Abstract)*

- Jang S, Lewis TS, Powers C, Khuchua Z, Baines CP, Wipf P, Javadov S. **Elucidating mitochondrial ETC supercomplexes in the heart during ischemia-reperfusion.** Antioxid Redox Signal. 2016 Sep 7. [Epub ahead of print] *(PubMed Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- Tatsuta T, Langer T. **Intramitochondrial phospholipid trafficking.** Biochim Biophys Acta. 2016 Aug 16. pii: S1388-1981(16)30226-8. doi: 10.1016/j.bbalip.2016.08.006. [Epub ahead of print] Review. *(PubMed Abstract)*

- Schlame M, Greenberg ML. **Biosynthesis, remodeling and turnover of mitochondrial cardiolipin.** Review Article. Biochim Biophys Acta. 2016 Aug 21. pii: S1388-1981(16)30230-X. doi: 10.1016/j.bbalip.2016.08.010. [Epub ahead of print] Review. *(PubMed Abstract)*

- El-Hattab AW, Scaglia F. **Mitochondrial cardiomyopathies.** Front Cardiovasc Med. 2016 Jul 25;3:25. doi: 10.3389/fcvm.2016.00025. eCollection 2016. *(PubMed - Open Access)*

- Barbot M, Meinecke M. **Reconstitutions of mitochondrial inner membrane remodeling.** J Struct Biol. 2016 Jul 25. pii: S1047-8477(16)30155-1. doi: 10.1016/j.jsb.2016.07.014. [Epub ahead of print] *(PubMed Abstract)*

- Aryal B, Rao VA. **Deficiency in cardiolipin reduces doxorubicin-induced oxidative stress and mitochondrial damage in human B-lymphocytes.** PLoS One. 2016 Jul 19;11(7):e0158376. doi: 10.1371/journal.pone.0158376. eCollection 2016. *(PubMed - Open Access)*▼

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)**▼

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Corcelli A, Schlame M**. Cardiolipin as key lipid of mitochondria in health and disease. 2nd Edition, Florence, Italy, September 30-October 1, 2015.** Chem Phys Lipids. 2016 Apr 27. pii: S0009-3084(16)30048-2. *(PubMed Abstract)**

- Szczepanek K, Allegood J, Aluri H, Hu Y, Chen Q, Lesnefsky EJ. **Acquired deficiency of *tafazzin* in the adult heart: Impact on mitochondrial function and response to cardiac injury.** Biochim Biophys Acta. 2016 Apr;1861(4):294-300. doi: 10.1016/j.bbalip.2015.12.004. Epub 2015 Dec 12. *(PubMed Abstract)*

- Kokoszka JE, Waymire KG, Flierl A, Sweeney KM, Angelin A, MacGregor GR, Wallace DC. **Deficiency in the mouse mitochondrial adenine nucleotide translocator isoform 2 gene is associated with cardiac noncompaction.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Available online 24 April 2016. *(ScienceDirect Abstract)*

- Bradley RM, Stark KD, Duncan RE. **Influence of tissue, diet, and enzymatic remodeling on cardiolipin fatty acyl profile.** Mol Nutr Food Res. 2016 Apr 8. doi: 10.1002/mnfr.201500966. [Epub ahead of print] *(PubMed Abstract)*

- Sandlers Y, Mercier K, Pathmasiri W, Carlson J, McRitchie S, Sumner S, Vernon HJ. **Metabolomics reveals new mechanisms for pathogenesis in Barth syndrome and introduces novel roles for cardiolipin in cellular function.** PLoS One. 2016 Mar 25;11(3):e0151802. doi: 10.1371/journal.pone.0151802. eCollection 2016. *(PubMed – Open Access)*▼

- Sathappa M, Alder NN. **The ionization properties of cardiolipin and its variants in model bilayers.** Biochim Biophys Acta. 2016 Mar 7. pii: S0005-2736(16)30084-0. doi: 10.1016/j.bbamem.2016.03.007. [Epub ahead of print] *(PubMed Abstract)**

- Dolinsky VW, Cole LK, Sparagna GC, Hatch GM. **Cardiac mitochondrial energy metabolism in heart failure: Role of cardiolipin and sirtuins.** Biochimica et Biophysica Acta (BBA) - Molecular and Cell Biology of Lipids, Available online 10 March 2016. *(ScienceDirect Abstract)*

- Joshi AS, Fei N, Greenberg ML. **Get1p and Get2p are required for maintenance of mitochondrial morphology and normal cardiolipin levels.** FEMS Yeast Res. 2016 Feb 28. pii: fow019. [Epub ahead of print] *(PubMed Abstract)**

- Dinca A, Chien WM, Chin MT. **Intracellular delivery of proteins with cell-penetrating peptides for therapeutic uses in human disease.** Int J Mol Sci. 2016 Feb 22;17(2):263. doi: 10.3390/ijms17020263. Review. *(PubMed – Open Access)**

- Lu YW, Galbraith L, Herndon JD, Lu YL, Pras-Raves M, Vervaart M, van Kampen A, Luyf A, Koehler CM, McCaffery JM, Gottlieb E, Vaz FM, Claypool SM. **Defining functional classes of Barth syndrome mutation in humans.** Hum Mol Genet. 2016 Feb 16. pii: ddw046. [Epub ahead of print] *(PubMed Abstract)**

- Saric A, Andreau K, Armand AS, Møller IM, Petit PX. **Barth syndrome: From mitochondrial dysfunctions associated with aberrant production of reactive oxygen species to pluripotent stem cell studies.** Front Genet. 2016 Jan 20;6:359. doi: 10.3389/fgene.2015.00359. eCollection 2015. Review. *(Pub Med – Open Access)*

- Dudek J, Cheng IF, Chowdhury A, Wozny K, Balleininger M, Reinhold R, Grunau S, Callegari S, Toischer K, Wanders RJ, Hasenfuß G, Brügger B, Guan K, Rehling P. **Cardiac-specific succinate dehydrogenase deficiency in Barth syndrome.** EMBO Mol Med. 2015 Dec 23. pii: e201505644. doi: 10.15252/emmm.201505644. [Epub ahead of print] *(PubMed – Open Access)*▼

- Ruiz-Ramírez A, Barrios-Maya MA, López-Acosta O, Molina-Ortiz D, El-Hafidi M. **Cytochrome c release from rat liver mitochondria is compromised by increased saturated cardiolipin species induced by sucrose feeding.** Am J Physiol Endocrinol Metab. 2015 Nov 1;309(9):E777-86. doi: 10.1152/ajpendo.00617.2014. Epub 2015 Sep 8. *(PubMed Abstract)*

- Denning C, Borgdorff V, Crutchley J, Firth KSA, George V, Kalra S, Kondrashov A, Hoang MD, Mosqueira D, Patel A, Prodanov L, Rajamohan D, Skarnes WC, Smith JGW, Young LE. **Cardiomyocytes from human pluripotent stem cells: From laboratory curiosity to industrial biomedical platform.** Original Research Article. Biochimica et Biophysica Acta (BBA) - Molecular Cell Research, Available online 31 October 2015. *(ScienceDirect Abstract)*

- Pokorná L, Cermáková P, Horváth A, Baile MG, Claypool SM, Griac P, Malínský J, Balážová M. **Specific degradation of phosphatidylglycerol is necessary for proper mitochondrial morphology and function.** Biochim Biophys Acta. 2015 Oct 19;1857(1):34-45. doi: 10.1016/j.bbabio.2015.10.004. [Epub ahead of print]. *(PubMed Abstract)*

- Gaspard GJ, McMaster CR. **Cardiolipin metabolism and its causal role in the etiology of the inherited cardiomyopathy Barth syndrome.** Chem Phys Lipids. 2015 Sep 25. pii: S0009-3084(15)30054-2. doi:10.1016/j.chemphyslip.2015.09.005. [Epub ahead of print] Review. *(PubMed Abstract)**

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S**. Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Tocchi A, Quarles EK, Basisty N, Gitari L, Rabinovitch PS. **Mitochondrial dysfunction in cardiac aging.** Biochim Biophys Acta. 2015 Jul 17. pii: S0005-2728(15)00152-8. doi: 10.1016/j.bbabio.2015.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Stepanyants N, Macdonald PJ, Francy CA, Mears JA, Qi X, Ramachandran R. **Cardiolipin's propensity for phase transition and its reorganization by dynamin-related protein 1 form a basis for mitochondrial membrane fission.** Mol Biol Cell. 2015 Jul 8. pii: mbc.E15-06-0330. [Epub ahead of print] *(PubMed Abstract)*

- Grevengoed TJ, Martin SA, Katunga L, Cooper DE, Anderson EJ, Murphy RC, Coleman RA. **Acyl-CoA synthetase 1 deficiency alters cardiolipin species and impairs mitochondrial function.** J Lipid Res. 2015 Aug;56(8):1572-82. doi:10.1194/jlr.M059717. Epub 2015 Jul 1. *(PubMed Abstract)*

- Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents *TAZ* knockdown-induced apoptosis in myeloid progenitor cells.** Biochem Biophys Res Commun. 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print] *(PubMed Abstract)**

- Sauerwald J, Jores T, Eisenberg-Bord M, Chuartzman SG, Schuldiner M, Rapaport D. **Genome-wide screens in yeast highlight a role for cardiolipin in biogenesis of mitochondrial outer membrane multispan proteins.** Mol Cell Biol. 2015 Jul 6. pii: MCB.00107-15. [Epub ahead of print] *(PubMed Abstract)*

- Angelini R, Lobasso S, Gorgoglione R, Bowron A, Steward CG, Corcelli A**. Cardiolipin fingerprinting of leukocytes by MALDI-TOF/MS as screening tool for Barth syndrome.** J Lipid Res. 2015 Jul 5. pii:jlr.D059824. [Epub ahead of print] *(PubMed – Open Access)**▼

- Morita SY, Terada T. **Enzymatic measurement of phosphatidylglycerol and cardiolipin in cultured cells and mitochondria.** Sci Rep. 2015 Jun 30;5:11737. doi: 10.1038/srep11737. *(PubMed Abstract)*

- Ryan RO. **Metabolic annotation of 2-ethylhydracrylic acid.** Clin Chim Acta. 2015 Jun 23. pii: S0009-8981(15)00295-8. doi:10.1016/j.cca.2015.06.012. [Epub ahead of print] Review. *(PubMed Abstract)**

- Melser S, Lavie J, Bénard G. **Mitochondrial degradation and energy metabolism.** Biochim Biophys Acta. 2015 May 12. pii: S0167-4889(15)00156-1. doi: 10.1016/j.bbamcr.2015.05.010. [Epub ahead of print] Review. *(PubMed Abstract)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Rodier G, Kirsh O, Baraibar M, Houles T, Lacroix M, Delpech H, Hatchi E, Arnould S, Severac D, Dubois E, Caramel J, Julien E, Friguet B, Le Cam L, Sardet C. **The transcription factor E4F1 coordinates CHK1-dependent checkpoint and mitochondrial functions.** Cell Reports, Available online 2 April 2015. *(ScienceDirect Abstract)*

- Casanovas A, Sprenger RR, Tarasov K, Ruckerbauer DE, Hannibal-Bach HK, Zanghellini J, Jensen ON, Ejsing CS. **Quantitative analysis of proteome and lipidome dynamics reveals functional regulation of global lipid metabolism**. Chem Biol. 2015 Mar 19;22(3):412-25. doi:10.1016/j.chembiol.2015.02.007. *(PubMed Abstract)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Wegener M, Bader A, Giri S. **How to mend a broken heart: Adult and induced pluripotent stem cell therapy for heart repair and regeneration.** Review Article. Drug Discovery Today, Available online 23 February 2015. *(ScienceDirect Abstract)*

- Borkar M, Bijarnia-Mahay S, Kohli S, Juneja M, Srivastava Y, Saxena R, Verma IC. **Identification of a novel *TAZ* gene mutation in a family with X-linked dilated cardiomyopathy Barth syndrome.** Journal of Inborn Errors of Metabolism & Screening January - December 2015 3: 2326409814567131, first published on February 19, 2015 doi:10.1177/2326409814567131. *(Sage – Full Text)*

- Lu YW, Claypool SM. **Disorders of phospholipid metabolism: An emerging class of mitochondrial disease due to defects in nuclear genes.** Front Genet. 2015 Feb 3;6:3. doi:10.3389/fgene.2015.00003. eCollection 2015. Review. *(PubMed – Open Access)*

- Gaspard GJ, McMaster CR. **The mitochondrial quality control protein Yme1 is necessary to prevent defective mitophagy in a yeast model of Barth syndrome.** J Biol Chem. 2015 Feb 16. pii:jbc.M115.641878. [Epub ahead of print]* *(PubMed – Open Access)*

- Li XX, Tsoi B, Li YF, Kurihara H, He RR. **Cardiolipin and its different properties in mitophagy and apoptosis.** J Histochem Cytochem. 2015 Feb 11. pii: 0022155415574818. [Epub ahead of print] *(PubMed - No Abstract Available)*

- Neupert W. **A perspective on transport of proteins into mitochondria: A myriad of open question**s. J Mol Biol. 2015 Feb 9. pii: S0022-2836(15)00079-0. doi: 10.1016/j.jmb.2015.02.001. [Epub ahead of print] *(PubMed Abstract)*

- Xu Y, Malhotra A, Claypool CM, Ren M, Schlame M. ***Tafazzins* from Drosophila and mammalian cells assemble in large protein complexes with a short half-life.** Mitochondrion. 2015 Jan 15. pii: S1567-7249(15)00003-3. doi: 10.1016/j.mito.2015.01.002. [Epub ahead of print] (*PubMed Abstract)*

- Saunders C, Smith L, Wibrand F, Ravn K, Bross P, Thiffault I, Christensen M, Atherton A, Farrow E, Miller N, Kingsmore, Elsebet Ostergaard. **CLPB variants associated with autosomal-recessive mitochondrial disorder with cataract, neutropenia, epilepsy, and methylglutaconic aciduria.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Wortmann SB, Zietkiewicz S, Kousi M, Szklarczyk R, Haack TB, Gersting SW, Muntau AC, Rakovic A, Renkema GH, Rodenburg RJ, Strom TM, Meitinger T, Rubio-Gozalbo ME, Chrusciel E, Distelmaier F, Golzio C, Jansen JH, van Karnebeek C, Lillquist Y, Lücke T, Õunap K, Zordania R, Yaplito-Lee J, van Bokhoven H, Spelbrink JN, Vaz FM, Pras-Raves M, Ploski R, Pronicka E, Klein C, Willemsen MAAP, de Brouwer APM, Prokisch H, Katsanis N, Wevers RA. **CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Frohman MA. **Role of mitochondrial lipids in guiding fission and fusion**. J Mol Med (Berl). 2014 Dec 5. [Epub ahead of print] *(PubMed Abstract)*

- Falk MJ, Shen L, Gonzalez M, Leipzig J, Lott MT, Stassen APM, Diroma MA, Navarro-Gomez D, Yeske P, Bai R, Boles RG, Brilhante B, Ralph D, Dare JT, Shelton R, Terry S, Zhang Z, Copeland WC, van Oven M, Prokisch H, Wallace DC, Attimonelli M, Krotoski D, Zuchner S, Gai X. **Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities.** Mol Genet Metab. 2014 Dec 4. pii: S1096-7192(14)00377-1. doi: 10.1016/j.ymgme.2014.11.016. [Epub ahead of print] *(PubMed Abstract)*

- Ye C, Shen Z, Greenberg ML. **Cardiolipin remodeling: A regulatory hub for modulating cardiolipin metabolism and function.** J Bioenerg Biomembr. 2016 Apr;48(2):113-23. doi: 10.1007/s10863-014-9591-7. Epub 2014 Nov 29. *(PubMed – Open Access)**

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Wortmann SB, Espeel M, Almeida L, Reimer A, Bosboom D, Roels F, de Brouwer AP, Wevers RA. **Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids.** J Inherit Metab Dis. 2014 Sep 2. [Epub ahead of print] *(PubMed Abstract)*

- He Q, Harris N, Ren J, Han X. **Mitochondria-targeted antioxidant prevents cardiac dysfunction induced by *tafazzin* gene knockdown in cardiac myocytes.** Oxid Med Cell Longev. 2014;2014:654198. Epub 2014 Aug 27. *(PubMed – Open Access)**

- Chi C-S. **Diagnostic approach in infants and children with mitochondrial diseases.** Pediatr Neonatol. 2014 Aug 20. pii: S1875-9572(14)00099-0. doi: 10.1016/j.pedneo.2014.03.009. [Epub ahead of print] Review. *(PubMed Abstract)*

- Mayr JA. **Lipid metabolism in mitochondrial membranes.** J Inherit Metab Dis. 2014 Aug 1. [Epub ahead of print] *(PubMed Abstract)*

- Peyta L, Dumas JF, Pinault M, Guimaraes C, Jarnouen K, Lagrave J, Maillot F, Loyer P, Servais S. **Reduction of the mitochondrial content in cardiolipins decreases O2 consumption and increases ATP synthesis efficiency in human hepatocyte-like HepaRG cells.** Biochimica et Biophysica Acta (BBA) - Bioenergetics, Volume 1837, Supplement, July 2014, Pages e28–e29. *(ScienceDirect Abstract)*

- Deckers M, Balleininger M, Vukotic M, Römpler K, Bareth B, Juris L, Dudek J. **Aim24 stabilizes respiratory chain supercomplexes and is required for efficient respiration.** FEBS Lett. 2014 Aug 25;588(17):2985-92. doi: 10.1016/j.febslet.2014.06.006. Epub 2014 Jun 10. *(PubMed Abstract)*

- Ren M, Phoon CKL, Schlame M. **Metabolism and function of mitochondrial cardiolipin.** Prog Lipid Res. 2014 Jul;55:1-16. doi: 10.1016/j.plipres.2014.04.001. Epub 2014 Apr 24. *(PubMed Abstract)**

- Birk AV, Chao WM, Bracken C, Warren JD, Szeto HH. **Targeting mitochondrial cardiolipin and the cytochrome c/cardiolipin complex to promote electron transport and optimize mitochondrial ATP synthesis.** Br J Pharmacol. 2014 Apr;171(8):2017-28. doi: 10.1111/bph.12468. *(PubMed Abstract)*

- Szeto HH. **First-in-class cardiolipin-protective compound as a therapeutic agent to restore mitochondrial bioenergetics.** Br J Pharmacol. 2014 Apr;171(8):2029-50. doi: 10.1111/bph.12461. Review. *(PubMed Abstract)*

- Aires V, Delmas D, Le Bachelier C, Latruffe N, Schlemmer D, Benoist JF, Djouadi F, Bastin J. **Stilbenes and resveratrol metabolites improve mitochondrial fatty acid oxidation defects in human fibroblasts.** Orphanet J Rare Dis. 2014 Jun 5;9:79. doi: 10.1186/1750-1172-9-79. *(PubMed Abstract)*

- Hwang MS, Schwall CT, Pazarentzos E, Datler C, Alder NN, Grimm S. **Mitochondrial Ca^{2+} influx targets cardiolipin to disintegrate respiratory chain complex II for cell death induction.** Cell Death Differ. 2014 Jun 20. doi: 10.1038/cdd.2014.84. [Epub ahead of print] *(PubMed Abstract)*

- Feillet-Coudray C, Fouret G, Casas F, Coudray C. **Impact of high dietary lipid intake and related metabolic disorders on the abundance and acyl composition of the unique mitochondrial phospholipid, cardiolipin.** J Bioenerg Biomembr. 2014 Jun 21. [Epub ahead of print] *(PubMed Abstract)*

- Raval KK, Kamp TJ. **Cardiomyopathy, mitochondria and Barth syndrome: iPSCs reveal a connection.** Nat Med. 2014 Jun 5;20(6):585-6. doi: 10.1038/nm.3592. *(PubMed – No Abstract Available).*

- Zeczycki T, Whelan J, Hayden WT, Brown DA, Shaikh SR. **Increasing levels of cardiolipin differentially influence packing of phospholipids found in the mitochondrial inner membrane.** Biochem Biophys Res Commun. 2014 Jun 3. pii: S0006-291X(14)01022-5. doi:10.1016/j.bbrc.2014.05.133. [Epub ahead of print] *(PubMed Abstract)*

- Richter-Dennerlein R, Korwitz A, Haag M, Tatsuta T, Dargazanli S, Baker M, Decker T, Lamkemeyer T, Rugarli EI, Langer T. **DNAJC19, a mitochondrial cochaperone associated with cardiomyopathy, forms a complex with prohibitins to regulate cardiolipin remodeling.** Cell Metab. 2014 May 21. pii: S1550-4131(14)00182-X. doi: 10.1016/j.cmet.2014.04.016. [Epub ahead of print] *(PubMed Abstract)*

- Wang G, McCain ML, Yang L, He A, Pasqualini FS, Agarwal A, Yuan H, Jiang D, Zhang D, Zangi L, Geva J, Roberts AE, Ma Q, Ding J, Chen J, Wang DZ, Li K, Wang J, Wanders RJA, Kulik W, Vaz FM, Laflamme MA, Murry CE, Chien KR, Kelley RI, Church GM, Parker K, Pu WT. **Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies.** Nat Med. 2014 Jun;20(6):616-23. doi:10.1038/nm.3545 Epub. 2014 May 11. *(PubMed – Open Access)**▼

- Mejia EM, Cole L, Hatch GM. **Cardiolipin metabolism and the role it plays in heart failure and mitochondrial supercomplex formation.** Cardiovasc Hematol Disord Drug Targets. 2014 May 5. [Epub ahead of print] *(PubMed Abstract)*

- Chen BB, Coon TA, Glasser JR, Zou C, Ellis B, Das T, McKelvey AC, Rajbhandari S, Lear T, Kamga C, Shiva S, Li C, Pilewski JM, Callio J, Chu CT, Ray A, Ray P, Tyurina YY, Kagan VE, Mallampalli RK. **E3 Ligase subunit Fbxo15 and PINK1 kinase regulate cardiolipin synthase 1 stability and mitochondrial function in pneumonia.** Cell Rep. 2014 Apr 2. pii:S2211-1247(14)00164-8. doi: 10.1016/j.celrep.2014.02.048. [Epub ahead of print] *(PubMed Abstract)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Koob S, Reichert AS. **Novel intracellular functions of apolipoproteins: The ApoO protein family as constituents of the Mitofilin/MINOS complex determines cristae morphology in mitochondria.** Biol Chem. 2014 Mar 1;395(3):285-96. doi: 10.1515/hsz-2013-0274. *(PubMed Abstract)*

- Su B, Ryan RO. **Metabolic biology of 3-methylglutaconic acid-uria: A new perspective.** J Inherit Metab Dis. 2014 May;37(3):359-68. doi: 10.1007/s10545-013-9669-0. Epub 2014 Jan 10. *(PubMed Abstract)**

- Parikh S, Goldstein A, Koenig MK, Scaglia F, Enns GM, Saneto R; Mitochondrial Medicine Society Clinical Directors Working Group; MMS Clinical Director's Work Group, Anselm I, Collins A, Cohen BH, DeBrosse SD, Dimmock D, Falk MJ, Ganesh J, Greene C, Gropman AL, Haas R, Kahler SG, Kamholz J, Kendall F, Korson MS, Mattman A, Milone M, Niyazov D, Pearl PL, Reimschisel T, Salvarinova-Zivkovic R, Sims K, Tarnopolsky M, Tsao CY, van Hove J, Walsh L, Wolfe LA. **Practice patterns of mitochondrial disease physicians in North America. Part 1: Diagnostic and clinical challenges.** Mitochondrion. 2014 Jan;14(1):26-33. doi:10.1016/j.mito.2013.07.116. Epub 2013 Jul 26. *(PubMed Abstract)*

- Mulligan CM, Le CH, Demooy AB, Nelson CB, Chicco AJ. **Inhibition of delta-6 desaturase reverses cardiolipin remodeling and prevents contractile dysfunction in the aged mouse heart without altering mitochondrial respiratory function.** J Gerontol A Biol Sci Med Sci. 2014 Jan 18. [Epub ahead of print] *(PubMed Abstract)*

- Schenkel LC, Bakovic M. **Formation and regulation of mitochondrial membranes.** Int J Cell Biol. 2014;2014:709828. doi: 10.1155/2014/709828. Epub 2014 Jan 22. Review. *(PubMed Abstract)*

- Harner ME, Unger AK, Izawa T, Walther DM, Ozbalci C, Geimer S, Reggiori F, Brügger B, Mann M, Westermann B, Neupert W. **Aim24 and MICOS modulate respiratory function, *tafazzin*-related cardiolipin modification and mitochondrial architecture.** Elife. 2014 Jan 1;3:e01684. doi:10.7554/eLife.01684. *(PubMed Abstract)*

- Kagan V, Epand R. **Deciphering the mysteries of cardiolipins in mitochondria.** Chem Phys Lipids. 2013 Dec 21. pii: S0009-3084(13)00169-2. [Epub ahead of print] *(PubMed – No Abstract Available)*

- Lai L, Wang M, Martin OJ, Leone TC, Vega RB, Han X, Kelly DP. **A role for peroxisome proliferator-activated receptor γ coactivator 1 (PGC-1) in the regulation of cardiac mitochondrial phospholipid biosynthesis.** J Biol Chem. 2014 Jan 24;289(4):2250-9. doi:10.1074/jbc.M113.523654. Epub 2013 Dec 11. *(PubMed – Open Access)*

- Schlattner U, Tokarska-Schlattner M, Rousseau D, Boissan M, Mannella C, Epand R, Lacombe ML. **Mitochondrial cardiolipin/phospholipid trafficking: The role of membrane contact site complexes and lipid transfer proteins**. Chem Phys Lipids. 2013 Dec 6. Chem Phys Lipids. 2013 Dec 6. [Epub ahead of print] *(PubMed Abstract)*

- Thomas E, Roman E, Claypool S, Manzoor N, Pla J, Panwar SL. **Mitochondria influence CDR1 efflux pump activity, Hog1-mediated oxidative stress pathway, iron homeostasis, and ergosterol levels in candida albicans.** Antimicrob. Agents Chemother. 2013, 57(11):5580. (*PubMed Abstract)*

- Mileykovskaya E, Dowhan W. **Cardiolipin-dependent formation of mitochondrial respiratory supercomplexes.** Chem Phys Lipids. 2013 Nov 9. [Epub ahead of print] *(PubMed Abstract)*

- Mazurová S, Tesarová M, Magner M, Houštková H, Hansíková H, Augustínová J, Tomek V, Vondrácková A, Zeman J, Honzík T. **Novel mutations in the *TAZ* gene in patients with Barth syndrome.** Prague Med Rep. 2013;114(3):139-153. (*PubMed Abstract)*

- Herndon JD, Claypool SM, Koehler CM. **The *Taz*1p transacylase is imported and sorted into the outer mitochondrial membrane via a membrane anchor domain.** Eukaryot Cell. 2013 Sep 27. [Epub ahead of print] (*PubMed – Open Access)*

- Brown DA, Sabbah HN, Shaikh SR. **Mitochondrial inner membrane lipids and proteins as targets for decreasing cardiac ischemia/reperfusion injury.** Pharmacol Ther. 2013 Dec;140(3):258-66. doi: 10.1016/j.pharmthera.2013.07.005. Epub 2013 Jul 15. *(PubMed Abstract)*

- Jefferies JL. **Barth syndrome.** Am J Med Genet C Semin Med Genet. 2013 Jul 10. doi: 10.1002/ajmg.c.31372. [Epub ahead of print] *(PubMed – Open Access)*▼

- Monteiro JP, Oliveira PJ, Jurado AS. **Mitochondrial membrane lipid remodeling in pathophysiology: A new target for diet and therapeutic interventions.** Prog Lipid Res. 2013 Jul 1. doi:pii: S0163-7827(13)00042-8. 10.1016/j.plipres.2013.06.002. [Epub ahead of print] (*PubMed Abstract)*

- Birk AV, Liu S, Soong Y, Mills W, Singh P, Warren JD, Seshan SV, Pardee JD, Szeto HH. **The mitochondrial-targeted compound SS-31 re-energizes ischemic mitochondria by interacting with cardiolipin.** J Am Soc Nephrol. 2013 Jun 27. [Epub ahead of print] *(PubMed Abstract)*

- Dudek J, Cheng IF, Balleininger M, Vaz FM, Streckfuss-Bömeke K, Hübscher D, Vukotic M, Wanders RJ, Rehling P, Guan K. **Cardiolipin deficiency affects respiratory chain function and organization in an induced pluripotent stem cell model of Barth syndrome.** Stem Cell Res. 2013 Sep;11(2):806-19. doi: 10.1016/j.scr.2013.05.005. Epub 2013 May 28. *(PubMed – Open Access)*▼

- Rigaud C, Lebre A, Touraine R, Beaupain B, Ottolenghi C, Chabli A, Ansquer H, Ozsahin H, Di Filippo S, De Lonlay P, Borm B, Rivier F, Vaillant M, Mathieu-Dramard M, Goldenberg A, Viot G, Charron P, Rio M, Bonnet D, Donadieu J. **Natural history of Barth syndrome: A national cohort study of 22 patients.** Orphanet J Rare Dis. 2013 May 8;8:70. doi: 10.1186/1750-1172-8-70. *(PubMed – Open Access)**▼

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Baile MG, Whited K, Claypool SM. **Deacylation on the matrix side of the mitochondrial inner membrane regulates cardiolipin remodeling.** Mol Biol Cell. 2013 May 1. [Epub ahead of print] *(PubMed – Open Access)*

- Becker T, Horvath SE, Bottinger L, Gebert N, Daum G, Pfanner N. **Role of phosphatidylethanolamine in the biogenesis of mitochondrial outer membrane proteins.** J Biol Chem. 2013 Apr 26. [Epub ahead of print] *(PubMed – Open-Access)*

- Powers C, Huang Y, Strauss A, Khuchua Z. **Diminished exercise capacity and mitochondrial bc1 complex deficiency in *tafazzin*-knockdown mice.** Front Physiol. 2013 Apr 17;4:74. doi: 10.3389/fphys.2013.00074. Print 2013. *(PubMed – Open Access)*

- Luévano-Martínez LA, Appolinario P, Miyamoto S, Uribe-Carvajal S, Kowaltowski AJ. **Deletion of the transcriptional regulator Opi1p decreases cardiolipin content and disrupts mitochondrial metabolism in saccharomyces cerevisiae.** Fungal Genetics and Biology, Available online 8 April 2013. *(PubMed Abstract)*

- Fan Y, Steller J, Gonzalez IL, Kulik W, Fox M, Chang R, Westerfield BA, Batra AS, Wang RY, Gallant NM, Pena LS, Wang H, Huang T, Bhuta S, Penny DJ, McCabe ER, Kimonis VE. **A novel exonic splicing mutation in the *TAZ* (G4.5) gene in a case with atypical Barth syndrome.** JIMD Rep. 2013 Apr 19. [Epub ahead of print] *(PubMed – Open Access)*

- Gonzalvez F, D'Aurelio M, Boutant M, Moustapha A, Puech JP, Landes T, Arnauré L, Vial G, Talleux N, Slomianny C, Wanders RJA, Houtkooper RH, Belenger P, Moller IM, Gottlieb E, Vaz FM, Manfredi G, Petit PX. **Barth syndrome: Cellular compensation of mitochondrial dysfunction and apoptosis inhibition due to changes in cardiolipin remodeling linked to *Tafazzin* gene mutation.** Biochim Biophys Acta. 2013 Aug;1832(8):1194-206. doi: 10.1016/j.bbadis.2013.03.005. Epub 2013 Mar 20. *(PubMed – Open Access)*

- Liu S, Bai Y, Huang J, Zhao H, Zhang X, Hu S, Wei Y. **Do mitochondria contribute to left ventricular non-compaction cardiomyopathy? New findings from myocardium of patients with left ventricular non-compaction cardiomyopathy.** Mol Genet Metab. 2013 May;109(1):100-6. doi: 10.1016/j.ymgme.2013.02.004. Epub 2013 Feb 13. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini RR, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 14;8(1):27. doi: 10.1186/1750-1172-8-27. *(PubMed Abstract)*▼

- Kiebish MA, Yang K, Liu X, Mancuso DJ, Guan S, Zhao Z, Sims HF, Cerqua R, Cade WT, Han X, Gross RW. **Dysfunctional cardiac mitochondrial bioenergetic, lipidomic, and signaling in a murine model of Barth syndrome.** J Lipid Res. 2013 Feb 14. [Epub ahead of print] *(PubMed – Open Access)**

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)*▼

- Ware SM, Towbin JA. **Nuclear genes causing mitochondrial cardiomyopathy.** Mitochondrial Disorders Caused by Nuclear Genes, 2013, pp. 319-335. *(Springer Abstract)*

- Karkucinska-Wieckowska A, Trubicka J, Werner B, Kokoszynska K, Pajdowska M, Pronicki M, Czarnowska E, Lebiedzinska M, Sykut-Cegielska J, Ziolkowska L, Jaron W, Dobrzanska A, Ciara E, Wieckowski MR, Pronicka E. **Left ventricular noncompaction (LVNC) and low mitochondrial membrane potential are specific for Barth syndrome.** J Inherit Metab Dis. 2013 Jan 30. [Epub ahead of print] *(PubMed Abstract)*

- Wortmann SB, Duran M, Anikster Y, Barth PG, Sperl W, Zschocke J, Morava E, Wevers RA. **Inborn errors of metabolism with 3-methylglutaconic aciduria as discriminative feature: Proper classification and nomenclature**. J Inherit Metab Dis. 2013 Jan 8. [Epub ahead of print] *(PubMed Abstract)*

- Ostojic O, O'Leary MF, Singh K, Menzies KJ, Vainshtein A, Hood DA. **The effects of chronic muscle use and disuse on cardiolipin metabolism.** J Appl Physiol. 2013 Feb 15;114(4):444-52. doi: 10.1152/japplphysiol.01312.2012. Epub 2012 Dec 6. *(PubMed Abstract)*

- Boxer LA. **How to approach neutropenia.** Hematology Am Soc Hematol Educ Program. 2012;2012:174-82. doi: 10.1182/asheducation-2012.1.174. *(PubMed Abstract)*

- Falk MJ, Pierce EA, Consugar M, Xie MH, Guadalupe M, Hardy O, Rappaport EF, Wallace DC, LeProust E, Gai X. **Mitochondrial disease genetic diagnostics: Optimized whole-exome analysis for all mitocarta nuclear genes and the mitochondrial genome.** Discov Med 2012 Dec 14(79):389-399. *(PubMed – Open Access)*

- Goldstein A, Bhatia P, Vento JM. **Update on nuclear mitochondrial genes and neurologic disorders.** Semin Pediatr Neurol. 2012 Dec;19(4):181-93. doi: 10.1016/j.spen.2012.09.005. *(PubMed Abstract)*

- Cortie CH, Else PL. **Dietary docosahexaenoic Acid (22:6) incorporates into cardiolipin at the expense of linoleic Acid (18:2): Analysis and potential implications.** Int J Mol Sci. 2012 Nov 21;13(11):15447-63. doi: 10.3390/ijms131115447. *(PubMed Abstract)*

- Ojala T, Polinati P, Manninen T, Hiippala A, Rajantie J, Karikoski R, Suomalainen A, Tyni T. **New mutation of mitochondrial DNAJC19 causing dilated and noncompaction cardiomyopathy, anemia, ataxia, and male genital anomalies.** Pediatr Res. 2012 Oct;72(4):432-7. doi: 10.1038/pr.2012.92. Epub 2012 Jul 13. *(PubMed Abstract)*

- Honzik T, Tesarova M, Magner M, Mayr J, Jesina P, Vesela K, Wenchich L, Szentivanyi K, Hansikova H, Sperl W, Zeman J. **Neonatal onset of mitochondrial disorders in 129 patients: Clinical and laboratory characteristics and a new approach to diagnosis.** J Inherit Metab Dis. 2012 Sep;35(5):749-59. doi: 10.1007/s10545-011-9440-3. Epub 2012 Jan 10. *(PubMed Abstract)*

- Böttinger L, Horvath SE, Kleinschroth T, Hunte C, Daum G, Pfanner N, Becker T. **Phosphatidylethanolamine and cardiolipin differentially affect the stability of mitochondrial respiratory chain supercomplexes.** J Mol Biol. 2012 Sep 9. pii: S0022-2836(12)00720-6. doi: 10.1016/j.jmb.2012.09.001. [Epub ahead of print] *(PubMed Abstract)*

- Schlame M, Acehan D, Berno B, Xu Y, Valvo S, Ren M, Stokes DL, Epand RM. **The physical state of lipid substrates provides transacylation specificity for *tafazzin*.** Nat Chem Biol. 2012 Sep 2. doi: 10.1038/nchembio.1064. [Epub ahead of print] *(PubMed – Open Access)**

- Lamari F, Mochel F, Sedel F, Saudubray JM. **Disorders of phospholipids, sphingolipids and fatty acids biosynthesis: Toward a new category of inherited metabolic diseases.** J Inherit Metab Dis. 2012 Jul 20. [Epub ahead of print] *(PubMed Abstract)*

- Kiebish MA, Yang K, Sims HF, Jenkins CM, Liu X, Mancuso DJ, Zhao Z, Guan S, Abendschein DR, Han X, Gross RW. **Myocardial regulation of lipidomic flux by cardiolipin synthase: Setting the beat for bioenergetic efficiency.** J Biol Chem. 2012 Jul 20;287(30):25086-97. Epub 2012 May 14. *(PubMed – Open Access)**

- Mulligan CM, Sparagna GC, Le CH, De Mooy AB, Routh MA, Holmes MG, Hickson-Bick DL, Zarini S, Murphy RC, Xu FY, Hatch GM, McCune SA, Moore RL, Chicco AJ. **Dietary linoleate preserves cardiolipin and attenuates mitochondrial dysfunction in the failing rat heart.** Cardiovasc Res. 2012 Jun 1;94(3):460-8. Epub 2012 Mar 12. *(PubMed Abstract)*

- Khairallah RJ, Kim J, O'Shea KM, O'Connell KA, Brown BH, Galvao T, Daneault C, Des Rosiers C, Polster BM, Hoppel CL, Stanley WC. **Improved mitochondrial function with diet-induced increase in either docosahexaenoic acid or arachidonic acid in membrane phospholipids.** PLoS One. 2012;7(3):e34402. Epub 2012 Mar 30. *(PubMed Abstract)*

- Samhan-Arias AK, Ji J, Demidova OM, Sparvero LJ, Feng W, Tyurin V, Tyurina YY, Epperly MW, Shvedova AA, Greenberger JS, Bayır H, Kagan VE, Amoscato AA. **Oxidized phospholipids as biomarkers of tissue and cell damage with a focus on cardiolipin.** Biochim Biophys Acta. 2012 Mar 23. [Epub ahead of print] *(PubMed Abstract)*

- Joshi AS, Thompson MN, Fei N, Hutteman M, Greenberg ML. **Cardiolipin and mitochondrial phosphatidylethanolamine have overlapping functions in mitochondrial fusion in Saccharomyces cerevisiae.** J Biol Chem. 2012 Mar 20. [Epub ahead of print] *(PubMed – Open Access)**

- Cosson L, Toutain A, Simard G, Kulik W, G Matyas, Guichet A, Blasco H, Maakaroun-Vermesse Z, Vaillant MC, Le Caignec C, Chantepie A, Labarthe F. **Barth syndrome in a female patient.** Mol Genet Metab. 2012 May;106(1):115-20. doi: 10.1016/j.ymgme.2012.01.015. Epub 2012 Jan 24. *(PubMed Abstract)*

- Stanley WC, Khairallah RJ, Dabkowski ER. **Update on lipids and mitochondrial function: Impact of dietary n-3 polyunsaturated fatty acids.** Curr Opin Clin Nutr Metab Care. 2012 Jan 13. [Epub ahead of print] *(PubMed Abstract)*

- Kachewar SG, Sankaye SB, Kulkarni DS. **The role of radio-diagnosis in inborn errors of metabolism.** Journal of Clinical and Diagnostic Research [serial online] 2011 November [cited: 2012 Dec 5]; 5:1467-1472. *(Full Text)*

- El-Hafidi M, Meschini MC, Rizza T, Santorelli FM, Bertini E, Carrozzo R, Vázquez-Memije ME. **Cardiolipin content in mitochondria from cultured skin fibroblasts harboring mutations in the mitochondrial ATP6 gene.** J Bioenerg Biomembr. 2011 Oct 13. [Epub ahead of print] *(PubMed Abstract)*

- Spencer CT, Byrne BJ, Bryant RM, Margossian R, Maisenbacher M, Breitenger P, Benni PB, Redfearn S, Marcus E, Cade WT. **Impaired cardiac reserve and severely diminished skeletal muscle oxygen utilization mediate exercise intolerance in Barth syndrome.** Am J Physiol Heart Circ Physiol. 2011 Nov;301(5):H2122-9. Epub 2011 Aug 26. *(PubMed – Open Access)**▼

- Paradies G, Petrosillo G, Paradies V, Reiter RJ, Ruggiero FM. **Melatonin, cardiolipin and mitochondrial bioenergetics in health and disease.** J Pineal Res. 2010 May;48(4):297-310. Review. *(PubMed Abstract)*

- Swalwell H, Kirby DM, Blakely EL, Mitchell A, Salemi R, Sugiana C, Compton AG, Tucker EJ, Ke BX, Lamont PJ, Turnbull DM, McFarland R, Taylor RW, Thorburn DR. **Respiratory chain complex I deficiency caused by mitochondrial DNA mutations.** Eur J Hum Genet. 2011 Jul;19(7):769-75. doi: 10.1038/ejhg.2011.18. Epub 2011 Mar 2. *(PubMed Abstract)*

- Christie DA, Lemke CD, Elias IM, Chau LA, Kirchhof MG, Li B, Ball EH, Dunn SD, Hatch GM, Madrenas J. **Stomatin-like protein 2 binds cardiolipin and regulates mitochondrial biogenesis and function.** Mol Cell Biol. 2011 Jul 11. [Epub ahead of print] *(PubMed Abstract)*

- Sandlers Y, Baumann S. **Characterization of urine metabolites in Barth syndrome patients employing a non-targeted GC/MS screening approach.** 59th ASMS Conference on Mass Spectrometry, June 5 - 9, 2011, Denver, Colorado. (*Poster*)▼

- Wajner M, Goodman SI. **Disruption of mitochondrial homeostasis in organic acidurias: Insights from human and animal studies.** J Bioenerg Biomembr. 2011 Feb;43(1):31-8. *(PubMed Abstract)*

- Kuroda T, Tani M, Moriguchi A, Tokunaga S, Higuchi T, Kitada S, Kuge O. **FMP30 is required for the maintenance of a normal cardiolipin level and mitochondrial morphology in the absence of mitochondrial phosphatidylethanolamine synthesis.** Mol Microbiol. 2011 Apr;80(1):248-65. Epub 2011 Feb 24. *(PubMed Abstract)*

- Rosca M, Minkler P, Hoppel CL. **Cardiac mitochondria in heart failure: Normal cardiolipin profile and increased threonine phosphorylation of complex IV**. Biochim Biophys Acta. 2011 Feb 11. [Epub ahead of print] *(PubMed Abstract)*

- Jameson E, Morris AAM. **Mitochondrial disease – a review.** Paediatrics and Child Health, Volume 21, Issue 2, February 2011, Pages 80-83. *(Abstract)*

- Osman C, Voelker DR, Langer T. **Making heads or tails of phospholipids in mitochondria.** J Cell Biol. 2011 Jan 10;192(1):7-16. *(PubMed Abstract)*

- Bird SS, Marur VR, Sniatynski MJ, Greenberg HK, Kristal BS. **Lipidomics profiling by high-resolution LC-MS and high-energy collisional dissociation fragmentation: Focus on characterization of mitochondrial cardiolipins and monolysocardiolipins.** Anal Chem. 2010 Dec 30. [Epub ahead of print] *(PubMed Abstract)*

- Yoda E, Hachisu K, Taketomi Y, Yoshida K, Nakamura M, Ikeda K, Taguchi R, Nakatani Y, Kuwata H, Murakami M, Kudo I, Hara S. **Mitochondrial dysfunction and reduced prostaglandin synthesis in skeletal muscle of Group VIB Ca2+-independent phospholipase A2gamma-deficient mice.** J Lipid Res. 2010 Oct;51(10):3003-15. Epub 2010 Jul 12. *(PubMed Abstract)*

- Crimi M, Esposti MD. **Apoptosis-induced changes in mitochondrial lipids.** Biochim Biophys Acta. 2010 Sep 29. [Epub ahead of print] *(PubMed Abstract)*

- Pei W, Kratz LE, Bernardini I, Sood R, Yokogawa T, Dorward H, Ciccone C, Kelley RI, Anikster Y, Burgess HA, Huizing M, Feldman B. **A model of Costeff syndrome reveals metabolic and protective functions of mitochondrial OPA3.** Development 2010 Aug 1;137(15):2587-96. *(PubMed Abstract)*

- Rea SL, Graham BH, Nakamaru-Ogiso E, Kar A, Falk MJ. **Bacteria, yeast, worms, and flies: Exploiting simple model organisms to investigate human mitochondrial diseases.** Dev Disabil Res Rev. 2010 Jun;16(2):200-18. *(PubMed Abstract)*

- Hasson SA, Damoiseaux R, Glavin JD, Dabir DV, Walker SS, Koehler CM. **Substrate specificity of the TIM22 mitochondrial import pathway revealed with small molecule inhibitor of protein translocation.** Proc Natl Acad Sci U S A. 2010 May 25;107(21):9578-83. Epub 2010 May 10. *(PubMed Abstract)*

- Lenaz G, Genova ML. **Structure and organization of mitochondrial respiratory complexes: A new understanding of an old subject.** Antioxid Redox Signal. 2010 Apr 15;12(8):961-1008. Review. *(PubMed Abstract)*

- Nie J, Hao X, Chen D, Han X, Chang Z, Shi Y. **A novel function of the human CLS1 in phosphatidylglycerol synthesis and remodeling.** Biochim Biophys Acta. 2010 Apr;1801(4):438-45. Epub 2009 Dec 16. *(PubMed Abstract)*

- Chen S, Liu D, Finley RL Jr, Greenberg ML. **Loss of mitochondrial DNA in the yeast cardiolipin synthase crd1 mutant leads to up-regulation of the protein kinase Swe1p that regulates the G2/M transition.** J Biol Chem. 2010 Apr 2;285(14):10397-407. Epub 2010 Jan 19. *(PubMed – Open Access)**

BSF does not endorse any drugs, tests, or treatments that we may report.

- Corcelli A, Saponetti MS, Zaccagnino P, Lopalco P, Mastrodonato MG, Liquori GE, Lorusso M. **Mitochondria isolated in nearly isotonic KCl buffer: Focus on cardiolipin and organelle morphology.** Biochim Biophys Acta. 2010 Mar;1798(3):681-7. Epub 2010 Jan 20. *(PubMed Abstract)*

- Rijken PJ. **Phosphatidyl choline-protein interactions and remodeling of cardiolipin in yeast mitochondria.** Doctoral Thesis, Scheikunde Proefschriften, 2010. *(Full Text)**

- Gonzalvez F, Pariselli F, Jalmar O, Dupaigne P, Sureau F, Dellinger M, Hendrickson EA, Bernard S, Petit PX. **Mechanistic issues of the interaction of the hairpin-forming domain of tBid with mitochondrial cardiolipin.** PLoS One. 2010 Feb 22;5(2):e9342. *(PubMed Abstract)*

- Hauff KD, Choi SY, Frohman MA, Hatch GM. **Cardiolipin synthesis is required to support human cholesterol biosynthesis from palmitate upon serum removal in Hela cells.** Can J Physiol Pharmacol. 2009 Oct;87(10):813-20. *(PubMed – Open Access)**

- Taylor WA, Hatch GM. **Identification of the human mitochondrial linoleoyl-coenzyme a monolysocardiolipin acyltransferase (MLCL AT-1).** J Biol Chem. 2009 Oct 30;284(44):30360-71. Epub 2009 Sep 8. *(PubMed – Open Access)**

- Schug ZY, Gottlieb E. **Cardiolipin acts as a mitochondrial signalling platform to launch apoptosis.** Biochim Biophys Acta. 2009 Oct;1788(10):2022-31. Epub 2009 May 18. Review. *(PubMed Abstract)*

- Claypool SM. **Cardiolipin, a critical determinant of mitochondrial carrier protein assembly and function.** Biochim Biophys Acta. 2009 Oct;1788(10):2059-68. Epub 2009 May 5. Review. *(PubMed Abstract*)

- Schlattner U, Tokarska-Schlattner M, Ramirez S, Brückner A, Kay L, Polge C, Epand RF, Lee RM, Lacombe ML, Epand RM. **Mitochondrial kinases and their molecular interaction with cardiolipin.** Biochim Biophys Acta. 2009 Oct;1788(10):2032-47. Epub 2009 May 4. Review. *(PubMed – Open Access*)**

- Schlame M, Ren M. **The role of cardiolipin in the structural organization of mitochondrial membranes**. Biochim Biophys Acta. 2009 Oct;1788(10):2080-3. Epub 2009 May 4. Review. *(PubMed – Open Access)**

- Mileykovskaya E, Dowhan W. **Cardiolipin membrane domains in prokaryotes and eukaryotes.** Biochim Biophys Acta. 2009 Oct;1788(10):2084-91. Epub 2009 Apr 14. Review. *(PubMed Abstract)*

- Paradies G, Petrosillo G, Paradies V, Ruggiero FM. **Role of cardiolipin peroxidation and Ca(2+) in mitochondrial dysfunction and disease.** Cell Calcium. 2009 Jun;45(6):643-50. Epub 2009 Apr 15. Review. *(PubMed Abstract)*

- Wittig I, Schägger H. **Supramolecular organization of ATP synthas and respiratory chain in mitochondrial membranes.** Biochim Biophys Acta. 2009 Jun;1787(6):672-80. Epub 2009 Jan 8. Review. *(PubMed Abstract)*

- Malhotra A, Xu Y, Ren M, Schlame M. **Formation of molecular species of mitochondrial cardiolipin. 1. A novel transacylation mechanism to shuttle fatty acids between sn-1 and sn-2 positions of multiple phospholipid species.** Biochim Biophys Acta. 2009 Apr;1791(4):314-20. Epub 2009 Jan 21. *(PubMed – Open Access)**

- Acehan D, Khuchua Z, Houtkooper RH, Malhotra A, Kaufman J, Vaz FM, Ren M, Rockman HA, Stokes DL, Schlame M. **Distinct effects of *tafazzin* deletion in differentiated and undifferentiated mitochondria.** Mitochondrion. 2009 Apr;9(2):86-95. Epub 2008 Dec 11. *(PubMed – Open Access)**

- Rosca MG, Hoppel CL. **New aspects of impaired mitochondrial function in heart failure.** J Bioenerg Biomembr. 2009 Apr;41(2):107-12. Review. *(PubMed Abstract)*

- Schlame M. **Formation of molecular species of mitochondrial cardiolipin. 2. A mathematical model of pattern formation by phospholipid transacylation.** Biochim Biophys Acta. 2009 Apr;1791(4):321-5. Epub 2009 Jan 31. *(PubMed Abstract)*

- Rog T, Martinez-Seara H, Munck N, Karttunen M, Vattulainen I. **Role of cardiolipins in the inner mitochondrial membrane: Insight gained through atom-scale simulations.** J Phys Chem B. 2009 Mar 19;113(11):3413-22. *(PubMed Abstract)*

- Gohil VM, Greenberg ML. **Mitochondrial membrane biogenesis: Phospholipids and proteins go hand in hand.** J Cell Biol. 2009 Feb 23;184(4):469-72. *(PubMed Abstract)*

- Malhotra A, Edelman-Novemsky I, Xu Y, Plesken H, Ma J, Schlame M, Ren M. **Role of calcium-independent phospholipase A2 in the pathogenesis of Barth syndrome.** Proc Natl Acad Sci U S A. 2009 Feb 17;106(7):2337-41. Epub 2009 Jan 21. *(PubMed – Open Access)**

- Zick M, Rabl R, Reichert AS. **Cristae formation-linking ultrastructure and function of mitochondria.** Biochim Biophys Acta. 2009 Jan;1793(1):5-19. Epub 2008 Jun 20. Review. *(PubMed Abstract)*

- van Raam BJ, Kuijpers TW. **Mitochondrial defects lie at the basis of neutropenia in Barth syndrome.** Curr Opin Hematol. 2009 Jan;16(1):14-19. *(PubMed Abstract)*

- Chen S, Tarsio M, Kane PM, Greenberg ML. **Cardiolipin mediates cross-talk between mitochondria and the vacuole.** Mol Biol Cell. 2008 Dec;19(12):5047-58. Epub 2008 Sep 17. *(PubMed – Open Access)**

- Gonzalvez F, Schug ZT, Houtkooper RH, Mackenzie ED, Brooks DG, Wanders RJ, Petit PX, Vaz FM, Gottlieb E. **Cardiolipin provides an essential activating platform for caspase-8 on mitochondria.** J Cell Biol. 2008 Nov 17;183(4):681-96. Epub 2008 Nov 10. *(PubMed – Open Access)**

- Schlame M. **Cardiolipin synthesis for the assembly of bacterial and mitochondrial membranes**. J Lipid Res. 2008 Aug;49(8):1607-20. Epub 2007 Dec 12. *(PubMed Abstract)*

- **A 3D view of Barth syndrome mitochondria**. Lab Invest. (2007), 1-2.

- Acehan D, Xu Y, Stokes DL, Schlame M. **Comparison of lymphoblast mitochondria from normal subjects and patients with Barth syndrome using electron microscopic tomography.** Lab Invest. 2007 Jan;87(1):40-8. Epub 2006 Oct 16. *(PubMed – Open Access)*▼

- Chicco AJ, Sparagna GC. **Role of cardiolipin alterations in mitochondrial dysfunction and disease.** Am J Physiol Cell Physiol. 2007 Jan;292(1):C33-44. Epub 2006 Aug 9. *(PubMed Abstract)*

- DiMauro S. **Mitochondrial myopathies.** Curr Opin Rheumatol. 2006 Nov;18(6):636-41. *(PubMed Abstract)*

- Xu Y, Condell M, Plesken H, Edelman-Novemsky I, Ma J, Ren M, Schlame M. **A Drosophila model of Barth syndrome**. Proc Natl Acad Sci U S A. 2006 Aug 1;103(31):11584-8. Epub 2006 Jul 19. *(PubMed – Open Access)**

- Claypool SM, McCaffery JM, Koehler CM. **Mitochondrial mislocalization and altered assembly of a cluster of Barth syndrome mutant *tafazzins.*** J Cell Biol. 2006 Jul 31;174(3):379-90. *(PubMed Abstract)*

- Schlame M, Ren M, Xu Y, Greenberg ML, Haller I. **Molecular symmetry in mitochondrial cardiolipins.** Chem Phys Lipids. 2005 Dec;138(1-2):38-49. Epub 2005 Sep 7. *(PubMed Abstract)**

- Ellis CE, Murphy EJ, Mitchell DC, Golovko MY, Scaglia F, Barcelo´-Coblijn GC, Nussbaum RL. **Mitochondrial lipid abnormality and electron transport chain impairment in mice lacking alpha-synuclein.** Mol Cell Biol. 2005 Nov;25(22):10190-201. *(PubMed Abstract)*

- Zhong Q, Greenberg ML. **Deficiency in mitochondrial anionic phospholipid synthesis impairs cell wall biogenesis.** Biochem Soc Trans. 2005 Oct;33(Pt 5):1158-61. *(PubMed Abstract)*

- DiMauro S, Gurgel-Giannetti J. **The expanding phenotype of mitochondrial myopathy.** Curr Opin Neurol. 2005 Oct;18(5):538-42. *(PubMed Abstract)*

- Scaglia F, Towbin JA, Craigen WJ, Belmont JW, O'Brian Smith E, Neish SR, Ware SM, Hunter JV, Fernbach SD, Vladutiu GD, Wong LJ, Vogel H. **Clinical spectrum, morbidity, and mortality in 113 pediatric patients with mitochondrial disease.** Pediatrics. 2004 Oct;114(4):925-31. *(PubMed Abstract)*

- Schon EA, DiMauro S. **Medicinal and genetic approaches to the treatment of mitochondrial disease**. Curr Med Chem. 2003 Dec;10(23):2523-33. *(PubMed Abstract)*

- DiMauro S, Schon EA. **Mitochondrial respiratory-chain diseases**. N Engl J Med 2003 June 26; 348(26):2656-2668. *(PubMed Abstract)*

- DiMauro S, Eduardo B, Michaelangelo M, Massimiliano F, Sacconi S, Salviati L, Hirano M. **Mitochondrial myopathies.** Basic Appl Myol 13 (3): 145-155, 2003.

- DiMauro S, Schon EA. **Mitochondrial DNA mutations in human disease**. Am J Med Genet 2001 Spring; 106(1):18-26.*)* *(PubMed Abstract)*

- Nissenkorn A, Zeharia A, Lev D, Fatal-Valevski A, Barash V, Gutman A, Harel S, Lerman-Sagie T. **Multiple presentations of mitochondrial disorders.** Arch Dis Child. 1999 Sep;81(3):209-14. *(PubMed Abstract)*

- Schlame M, Shanske S, Doty S, Konig T, Sculco T, DiMauro S, Blanck TJ. **Microanalysis of cardiolipin in small biopsies including skeletal muscle from patients with mitochondrial disease**. J Lipid Res. 1999 Sep;40(9):1585-92. *(PubMed Abstract)*

- Varvogli L, Waisbren SE. **Personality profiles of mothers of children with mitochondrial disorders.** J Inherit Metab Dis. 1999 Jun;22(5):615-22. *(PubMed Abstract)*

- Spiro AJ, Prineas JW, Moore CL. **A new mitochondrial myopathy in a patient with salt craving**. Arch Neurol 1970 Mar; 22: 259-269. *(PubMed Abstract)*

MORPHOLOGY

- NEW Stöllberger C, Finsterer J. **Understanding left ventricular hypertrabeculation/ noncompaction: pathomorphologic findings and prognostic impact of neuromuscular comorbidities.** Expert Rev Cardiovasc Ther. 2018 Dec 20. doi: 10.1080/14779072.2019.1561280. [Epub ahead of print] *(PubMed Abstract)*

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Wortmann SB, Morava E. **3-methylglutaconic aciduria type IV: A syndrome with an evolving phenotype.** Clin Dysmorphol. 2011 Jul;20(3):168-9. No abstract available. *(PubMed)*

- Lisi EC, Cohn RD. **Genetic evaluation of the pediatric patient with hypotonia: Perspective from a hypotonia specialty clinic and review of the literature.** Dev Med Child Neurol. 2011 Jul;53(7):586-99. doi: 10.1111/j.1469-8749.2011.03918.x. Epub 2011 Mar 21. Review. *(PubMed Abstract)*

- Hastings R, Steward C, Tsai-Goodman B, Newbury-Ecob R. **Dysmorphology of Barth syndrome.** Clin Dysmorphol. 2009 Oct;18(4):185-7. *(PubMed Abstract)**▼

- Chow CW, Thorburn DR. **Morphological correlates of mitochondrial dysfunction in children**. Hum Reprod. 2000 Jul;15:Suppl 2:68-78. *(PubMed Abstract)*

- Kranzler JH, Rosenbloom AL, Proctor B, Diamond FB, Watson M. **Is short stature a handicap? A comparison of the psychosocial functioning of referred and nonreferred children with normal short stature and children with normal stature**. J Pediatr 2000 Jan; 136(1):96-102. *(PubMed Abstract)*

NEUTROPENIA

NEW Steward CG, Groves SJ, Taylor CT, Maisenbacher MK, Versluys B, Newbury-Ecob RA, Ozsahin H, Damin MK, Bowen VM, McCurdy KR, Mackey MC, Bolyard AA, Dale DC. **Neutropenia in Barth syndrome: Characteristics, risks, and management.** Curr Opin Hematol. 2018 Nov 16. doi: 10.1097/MOH.0000000000000472. [Epub ahead of print] *(PubMed Abstract)*▼

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Imai-Okazaki A, Kishita Y, Kohda M, Yatsuka Y, Hirata T, Mizuno Y, Harashima H, Hirono K, Ichida F, Noguchi A, Yoshida M, Tokorodani C, Nishiuchi R, Takeda A, Nakaya A, Sakata Y, Murayama K, Ohtake A, Okazaki Y. **Barth syndrome: Different approaches to diagnosis.** J Pediatr. 2017 Dec 15. pii: S0022-3476(17)31329-X. doi: 10.1016/j.jpeds.2017.09.075. [Epub ahead of print] *(PubMed Abstract)*

- Bakšiene M, Benušiene E, Morkuniene A, Ambrozaityte L, Utkus A, Kucinskas V. **A novel intronic splice site *tafazzin* gene mutation detected prenatally in a family with Barth syndrome.** Balkan J Med Genet. 2017 Mar 8;19(2):95-100. doi: 10.1515/bjmg-2016-0043. *(PubMed - Open Access)*

- Woiewodski L, Ezon D, Cooper J, Feingold B. **Barth syndrome with late-onset cardiomyopathy: A missed opportunity for diagnosis.** The Journal of Pediatrics, In Press, Corrected Proof, Available online 18 January 2017. *(Abstract)*

- Ferri L, Dionisi-Vici C, Taurisano R, Vaz FM, Guerrini R, Morrone A. **When silence is noise: Infantile-onset Barth syndrome caused by a synonymous substitution affecting *TAZ* gene transcription.** Clin Genet. 2016 Nov;90(5):461-465. doi: 10.1111/cge.12756. *(PubMed Abstract)*▼

- Oláhová M, Thompson K, Hardy SA, Barbosa IA, Besse A, Anagnostou ME, White K, Davey T, Simpson MA, Champion M, Enns G, Schelley S, Lightowlers RN, Chrzanowska-Lightowlers ZM, McFarland R, Deshpande C, Bonnen PE, Taylor RW. **Pathogenic variants in HTRA2 cause an early-onset mitochondrial syndrome associated with 3-methylglutaconic aciduria.** J Inherit Metab Dis. 2016 Sep 30. *(PubMed Abstract)*

- Aryal B, Rao VA. **Deficiency in cardiolipin reduces doxorubicin-induced oxidative stress and mitochondrial damage in human B-lymphocytes.** PLoS One. 2016 Jul 19;11(7):e0158376. doi: 10.1371/journal.pone.0158376. eCollection 2016. *(PubMed - Open Access)*▼

- Yoo TY, Kim MR, Son JS, Lee R, Bae SH, Chung S, Kim KS, Seong MW, Park SS. **Identification of a novel de novo mutation of the *TAZ* gene in a Korean patient with Barth syndrome.** J Cardiovasc Ultrasound. 2016 Jun;24(2):153-7. doi: 10.4250/jcu.2016.24.2.153. Epub 2016 Jun 22. *(PubMed - Open Access)*

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents *TAZ* knockdown-induced apoptosis in myeloid progenitor cells.** Biochem Biophys Res Commun. 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print] *(PubMed Abstract)**

- Ye Y, Carlsson G, Karlsson-Sjöberg JM, Borregaard N, Modéer TU, Andersson ML, Pütsep KL. **The antimicrobial propeptide hCAP-18 plasma levels in neutropenia of various aetiologies: A prospective study.** Sci Rep. 2015 Jun 29;5:11685. doi: 10.1038/srep11685. *(PubMed - Open Access)*

- Saunders C, Smith L, Wibrand F, Ravn K, Bross P, Thiffault I, Christensen M, Atherton A, Farrow E, Miller N, Kingsmore, Elsebet Ostergaard. **CLPB variants associated with autosomal-recessive mitochondrial disorder with cataract, neutropenia, epilepsy, and methylglutaconic aciduria.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Wortmann SB, Zietkiewicz S, Kousi M, Szklarczyk R, Haack TB, Gersting SW, Muntau AC, Rakovic A, Renkema GH, Rodenburg RJ, Strom TM, Meitinger T, Rubio-Gozalbo ME, Chrusciel E, Distelmaier F, Golzio C, Jansen JH, van Karnebeek C, Lillquist Y, Lücke T, Õunap K, Zordania R, Yaplito-Lee J, van Bokhoven H, Spelbrink JN, Vaz FM, Pras-Raves M, Ploski R, Pronicka E, Klein C, Willemsen MAAP, de Brouwer APM, Prokisch H, Katsanis N, Wevers RA. **CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder.** The American Journal of Human Genetics, Available online 15 January 2015. *(ScienceDirect Abstract)*

- Sreeramkumar V, Adrover JM, Ballesteros I, Cuartero MI,Rossaint J, Bilbao I,Nácher M,Pitaval C, Radovanovic I,Fukui Y,McEver RP, Filippi MD, Lizasoain I,Ruiz-Cabello J, ZarbockA, Moro MA, Hidalgo A. **Neutrophils scan for activated platelets to initiate inflammation.** Science. 2014 Dec 5;346(6214):1234-8. doi: 10.1126/science.1256478. Epub 2014 Dec 4. *(PubMed Abstract)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Donadieu J, Rigaud C, Lebre AS, Touraine R, Ottolenghi C, Chabli A, Charron P, Rio M, De Lonlay P, Bonnet D. **Syndrome de Barth: le reconnaître, le traiter. Recommandations pour la prise en charge (Barth syndrome: Guidelines for diagnosis, follow-up and medical therapy).** Revue d'Oncologie Hématologie Pédiatrique, Available online 27 August 2014. *(ScienceDirect Abstract)*

- Folsi V, Miglietti N, Lombardi AM, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, Plebani A. **Cardiomyopathy in a male patient with neutropenia and growth delay.** Italian Journal of Pediatrics 2014, 40:45 doi:10.1186/1824-7288-40-45. *(PubMed – Open Access)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Jefferies JL. **Barth syndrome.** Am J Med Genet C Semin Med Genet. 2013 Jul 10. doi: 10.1002/ajmg.c.31372. [Epub ahead of print] *(PubMed – Open Access)*▼

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Rigaud C, Lebre A, Touraine R, Beaupain B, Ottolenghi C, Chabli A, Ansquer H, Ozsahin H, Di Filippo S, De Lonlay P, Borm B, Rivier F, Vaillant M, Mathieu-Dramard M, Goldenberg A, Viot G, Charron P, Rio M, Bonnet D, Donadieu J. **Natural history of Barth syndrome: A national cohort study of 22 patients.** Orphanet J Rare Dis. 2013 May 8;8:70. doi: 10.1186/1750-1172-8-70. *(PubMed – Open Access)**▼

- Dedieu N, Giardini A, Steward CG, Fenton M, Karimova A, Hsia TY, Burch M. **Successful mechanical circulatory support for 251 days in a child with intermittent severe neutropenia due to Barth syndrome.** Pediatr Transplant. 2013 Mar;17(2):E46-9. doi: 10.1111/petr.12027. Epub 2012 Nov 28. *(PubMed Abstract)*▼

- Ferri L, Donati MA, Funghini S, Malvagia S, Catarzi S, Lugli L, Ragni L, Bertini E, Vaz FM, Cooper DN, Guerrini RR, Morrone A. **New clinical and molecular insights on Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 14;8(1):27. doi: 10.1186/1750-1172-8-27. *(PubMed Abstract)*▼

- Whited K, Baile MG, Currier P, Claypool SM. **Seven functional classes of Barth syndrome mutation.** Hum Mol Genet. 2013 Feb 1;22(3):483-92. doi: 10.1093/hmg/dds447. Epub 2012 Oct 24. *(PubMed Abstract)*

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth syndrome in adulthood: A clinical case.** [Article in English, Spanish] Rev Esp Cardiol. 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. *(PubMed Abstract)*

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Sokolic R. **Neutropenia in primary immunodeficiency.** Curr Opin Hematol. 2013 Jan;20(1):55-65. doi:10.1097/MOH.0b013e32835aef1c. *(PubMed Abstract)*

- Finsterer JA, Frank M. **Haematological features in Barth syndrome.** Curr Opin Hematol. 2013 Jan;20(1):36-40. doi: 10.1097/MOH.0b013e32835a01d9. *(PubMed Abstract)*

- Sabater-Molina M, Guillén-Navarro E, García-Molina E, Ballesta-Martínez MJ, Escudero F, Ruiz-Espejo F. **Barth Syndrome in adulthood: A clinical case.** Rev Esp Cardiol (Engl Ed). 2013 Jan;66(1):68-70. doi: 10.1016/j.recesp.2012.05.015. Epub 2012 Sep 21. (*PubMed Abstract*)

- Boxer LA. **How to approach neutropenia.** Hematology Am Soc Hematol Educ Program. 2012;2012:174-82. doi: 10.1182/asheducation-2012.1.174. *(PubMed Abstract)*

- Piga A, Longo F, Musallam KM, Veltri A, Ferroni F, Chiribiri A, Bonamini R. **Left ventricular noncompaction in patients with β-thalassemia: Uncovering a previously unrecognized abnormality.** Am J Hematol. 2012 Dec;87(12):1079-83. doi: 10.1002/ajh.23323. Epub 2012 Sep 11. *(PubMed Abstract)*

- Bowron A, Frost R, Powers VE, Thomas PH, Heales SJ, Steward CG. **Diagnosis of Barth syndrome using a novel LC-MS/MS method for leukocyte cardiolipin analysis.** J Inherit Metab Dis. 2012 Oct 30. [Epub ahead of print] *(PubMed Abstract)*

- Wan C, Yu HH, Lu MY, Lee JH, Wang LC, Lin YT, Yang YH, Chiang BL. **Clinical manifestations and outcomes of pediatric chronic neutropenia.** J Formos Med Assoc. 2012 Apr;111(4):220-7. Epub 2012 Mar 16. *(PubMed Abstract)*

- Schild L, Lendeckel U, Gardemann A, Wiswedel I, Schmidt CA, Wolke C, Walther R, Grabarczyk P, Busemann C. **Composition of molecular cardiolipin species correlates with proliferation of lymphocytes.** Exp Biol Med (Maywood). 2012 Apr;237(4):372-9. doi: 10.1258/ebm.2011.011311. Epub 2012 Apr 4. *(PubMed Abstract)*

- Makaryan V, Kulik W, Vaz FM, Allen C, Dror Y, Dale DC, Aprikyan AA**. The cellular and molecular mechanisms for neutropenia in Barth syndrome.** Eur J Haematol. 2011 Oct 24. doi: 10.1111/j.1600-0609.2011.01725.x. [Epub ahead of print] *(PubMed – Open Access)**

- Dale DC**. Editorial: Serine proteases, serpins, and neutropenia.** J Leukoc Biol. 2011 Jul;90(1):3-4. *(PubMed - No abstract available.)*

- van Raam BJ, van Bruggen R, Tool AT, Jansen MH, Warris A, Jolles S, Kuijpers TW. **Nuclear factor-{kappa}B is not essential for NADPH oxidase activity in neutrophils from anhidrotic ectodermal dysplasia patients.** Blood. 2009 May 21;113(21):5362-3. *(PubMed – No Abstract Available)*

- Rivers A, Slayton WB. **Congenital cytopenias and bone marrow failure syndromes.** Semin Perinatol. 2009 Feb;33(1):20-8. Review. *(PubMed Abstract)*

- van Raam BJ, Kuijpers TW. **Mitochondrial defects lie at the basis of neutropenia in Barth syndrome.** Curr Opin Hematol. 2009 Jan;16(1):14-19. *(PubMed Abstract)*

- Boztug K, Welte K, Zeidler C, Klein C. **Congenital neutropenia syndromes.** Immunol Allergy Clin North Am 2008 May;28(2):259-75, vii-viii. Review. *(PubMed Abstract)*

- McCanta AC, Chang AC, Weiner K. **Cardiomyopathy in a child with neutropenia and motor delay.** Curr Opin Pediatr. 2008 Oct;20(5):605-7. *(PubMed Abstract)*

- Bonilla FA, Bernstein IL, Khan DA, Ballas ZK, Chinen J, Frank MM, Kobrynski LJ, Levinson AI, Mazer B, Nelson RP Jr, Orange JS, Routes JM, Shearer WT, Sorensen RU; American Academy of Allergy, Asthma and Immunology; American College of Allergy, Asthma and Immunology; Joint Council of Allergy, Asthma and Immunology. **Practice parameter for the diagnosis and management of primary immunodeficiency.** Ann Allergy Asthma Immunol. 2005 May;94(5 Suppl 1):S1-63. *(PubMed Abstract)*

- Badolato R, Fontana S, Notarangelo LD, Savoldi G. **Congenital neutropenia: Advances in diagnosis and treatment.** Curr Opin Allergy Clin Immunol. 2004 Dec;4(6):513-21. *(PubMed Abstract)*

- Dale DC, Bolyard AA, Schwinzer B, Pracht G, Bonilla MA, Boxer L, Freedman M, Donadieu J, Kannourakis G, Alter BP, Cham B, Winkelstein J, Kinsey SE, Fier C, Zeidler C, Welte K. **The Severe Chronic Neutropenia International Registry - 10 Years of follow-up.** [abstract]. American Society of Hematology Annual Meeting; Nov 2004; Blood 104: 1458.

- Maheshwari and Christensen. **Neutropenia in the neonatal intensive care unit.** Neoreviews. 2004; 5: 431-443.

- Kuijpers TW, Maianski NA, Tool AT, Becker K, Plecko B, Valianpoour F, Wanders RJ, Pereira R, Van Hove J, Verhoeven AJ, Roos D, Baas F, Barth PG. **Neutrophils in Barth syndrome (BTHS) avidly bind annexin-V in the absence of apopotosis**. Blood 2004 May 15; 103(10):3915-3923. Epub 2004 Feb 5. *(PubMed – Open Access)**

- Maianski NA, Geissler J, Srinivasula SM, Alnemri ES, Roos D, Kuijpers TW. **Functional characterization of mitochondria in neutrophils: A role restricted to apoptosis**. Cell Death Differ. 2004 Feb;11(2):143-53. *(PubMed Abstract)*

- Maianski NA, Maianski AN, Kuijpers TW, Roos D. **Apoptosis of neutrophils**. Acta Haematol 2004; 111(1-2):56-66. *(PubMed Abstract)*

- Versluys B, Bowen V, McCurdy K, Mann S, Cantlay A, Newbury-Ecob R, Kern I, Goulden N, Steward C. **X-linked organic aciduria: An important cause of neutropenia in males**. Blood 2003 Nov 16; 102(11), #965. *(Abstract)*▼

- Stein SM, Dale DC. **Molecular basis and therapy of disorders associated with chronic neutropenia.** Curr Allergy Asthma Rep 2003 Sept; 3(5):385-388. *(PubMed Abstract)*

- Dale DC, Cottle TE, Fier CJ, Bolyard AA, Bonilla MA, Boxer LA, Cham B, Freedman MH, Kannourakis G, Kinsey SE, Davis R, Scarlata D, Schwinzer B, Zeidler C, Welte K. **Severe chronic neutropenia: Treatment and follow-up of patients in the Severe Chronic Neutropenia International Registry.** Am J Hematol. 2003 Feb;72(2):82-93. *(PubMed Abstract)*

- Horwitz M, Benson KF, Duan Z, Person RE, Wechsler J, Williams K, Albani D, Li FQ. **Role of neutrophil elastase in bone marrow failure syndromes: Molecular genetic revival of the chalone hypothesis.** Curr Opin Hematol 2003 Jan;10(1):49-54. *(PubMed Abstract)*

- Cottle TE, Fier CJ, Donadieu J, Kinsey SE. **Risk and benefit of treatment of severe chronic neutropenia with granulocyte colony-stimulating factor**. Semin in Hematol. 2002 Apr; 39(2):134-40. *(PubMed Abstract)*

- Freedman MH, Alter BP. **Risk of myelodysplastic syndrome and acute myeloid leukemia in congenital neutropenias**. Semin in Hematol.2002 Apr; 39(2):128-33. *(PubMed Abstract)*

- Dale DC, Bolyard AA, Aprikyan A. **Cyclic neutropenia**. Semin Hematol. 2002 Apr;39(2):89-94. *(PubMed Abstract)*

- Boxer L, Dale DC. **Neutropenia: Causes and consequences**. Semin Hematol. 2002 Apr;39(2):75-81. *(PubMed Abstract)*

- Zeidler C, Barth PG, Bonilla MA, Bolyard AA, Boxer L, Cottle T, Dale DC, Donadieu J, Fier C, Freedman M, Kannourakis G, Kinsey S, Liang B, Schwinzer B, Welte K, Cham B, for the Severe Chronic Neutropenia International Registry (SCNIR). **Neutropenia in Barth syndrome: Clinical course and treatment of neutropenia**. Blood 2001; 98(11):300a.

- Bolyard AA, Cottle T, Edwards C, Kinsey S, Schwinzer B, Zeidler C. **Understanding Severe Chronic Neutropenia – a handbook for patients and their families**. Nov 2000.

- Dale DC, Person RE, Bolyard AA, Aprikyan AG, Bos C, Bonilla MA, Boxer LA, Kannourakis G, Zeidler C, Welte K, Benson KF, Horwitz M. **Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia**. Blood 2000 Oct; 96 (7); 2317-2322. *(PubMed Abstract)*

- **Severe Chronic Neutropenia International Registry**. Update. Fall 1998; 5:1-12. *(Full text)*

- Welte K, Boxer LA. **Severe chronic neutropenia: Pathophysiology and therapy**. Semin Hematol. 1997 Oct;34(4):267-78. *(PubMed Abstract)*

- Welte K, Dale D. **Pathophysiology and treatment of severe chronic neutropenia**. Ann Hematol. 1996 Apr;72(4):158-65. *(PubMed Abstract)*

- Dong F, Brynes RK, Tidow N, Welte K, Lowenberg B, Touw IP. **Mutations in the gene for the granulocyte colony-stimulating-factor receptor in patients with acute myeloid leukemia preceded by severe congenital neutropenia**. N Engl J Med. 1995 Aug 24;333(8):487-93. *(PubMed Abstract)*

- Cox GF, Pulsipher M, Rothenberg M, Korson M, Kelley RI. **Correction of neutropenia in Barth syndrome by G-CSF**. Am J Hum Genet 1995; 57:A177. *(Full Text)*

- Bonilla MA, Dale D, Zeidler C, Last L, Reiter A, Ruggeiro M, Davis M, Koci B, Hammond W, Gillio A, Welte K. **Long-term safety of treatment with recombinant human granulocyte colony-stimulating factor (r-metHuG-CSF) in patients with severe congenital neutropenias**. Br J Haematol. 1994 Dec;88(4):723-30. *(PubMed Abstract)*

- Hammond WP, Price TH, Souza LM, Dale DC. **Treatment of cyclic neutropenia with granulocyte colony-stimulating factor**. N Engl J Med. 1989 May 18;320(20):1306-11. *(PubMed Abstract)*

- Jakubowski AA, Souza L, Kelly F, Fain K, Budman D, Clarkson B, Bonilla MA, Moore MAS, Gabrilove J. **Effects of human granulocyte colony-stimulating factor in a patient with idiopathic neutropenia**. N Engl J Med. 1989 Jan 5;320(1):38-42. *(PubMed Abstract)*

NEWBORN SCREENING

- Zhao Y, Feng Y, Ding X, Dong S, Zhang H, Ding J, Xia X. **Identification of a novel hypertrophic cardiomyopathy-associated mutation using targeted next-generation sequencing.** Int J Mol Med. 2017 Jul;40(1):121-129. doi: 10.3892/ijmm.2017.2986. Epub 2017 May 11. *(PubMed - Open Access)*

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Angelini R, Lobasso S, Gorgoglione R, Bowron A, Steward CG, Corcelli A**. Cardiolipin fingerprinting of leukocytes by MALDI-TOF/MS as screening tool for Barth syndrome.** J Lipid Res. 2015 Jul 5. pii:jlr.D059824. [Epub ahead of print] *(PubMed – Open Access)**▼

- Pass KA, Thoene J, Watson MS. **Emergency preparedness for newborn screening and genetic services.** Genet Med. 2009 Jun;11(6):455-64. *(PubMed Abstract)*

- Kulik W, van Lenthe H, Stet FS, Houtkooper RH, Kemp H, Stone JE, Steward CG, Wanders RJ, Vaz FM. **Bloodspot assay using HPLC-tandem mass spectrometry for detection of Barth syndrome.** Clin Chem. 2008 Feb;54(2):371-8. Epub 2007 Dec 10. *(PubMed – Open Access)**▼

- Leonard JV, Morris AA. **Diagnosis and early management of inborn errors of metabolism presenting around the time of birth.** Acta Paediatr. 2006 Jan;95(1):6-14. *(PubMed Abstract)*

- Wilcken B, Wiley V, Hammond J, Carpenter K. **Screening newborns for inborn errors of metabolism by tandem mass spectrometry**. N Engl J Med 2003 June 5; 348(23):2304-2312. *(PubMed Abstract)*

- Chakrapani A, Cleary MA, Wraith JE. **Detection of inborn errors of metabolism in the newborn.** Arch Dis Child Fetal Neonatal Ed. 2001 May;84(3):F205-10. *(PubMed Abstract)*

NUTRITIONAL SUPPLEMENTS

- Chen WW, Chao YJ, Chang WH, Chan JF, Hsu YHH. **Phosphatidylglycerol incorporates into cardiolipin to improve mitochondrial activity and inhibits inflammation.** Sci Rep. 2018 Mar 20;8(1):4919. doi: 10.1038/s41598-018-23190-z. *(PubMed Abstract)*

- Bradley RM, Stark KD, Duncan RE. **Influence of tissue, diet, and enzymatic remodeling on cardiolipin fatty acyl profile.** Mol Nutr Food Res. 2016 Apr 8. doi: 10.1002/mnfr.201500966. [Epub ahead of print] *(PubMed Abstract)*

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Reynolds S, Kreider CM, Meeley L, Bendixen RM. **Taste perception and sensory sensitivity: Relationship to feeding problems in boys with Barth syndrome.** J Rare Disorders. March 2015. *(Open Access)*▼

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Mulligan CM, Sparagna GC, Le CH, De Mooy AB, Routh MA, Holmes MG, Hickson-Bick DL, Zarini S, Murphy RC, Xu FY, Hatch GM, McCune SA, Moore RL, Chicco AJ. **Dietary linoleate preserves cardiolipin and attenuates mitochondrial dysfunction in the failing rat heart.** Cardiovasc Res. 2012 Jun 1;94(3):460-8. Epub 2012 Mar 12. *(PubMed Abstract)*

- Khairallah RJ, Kim J, O'Shea KM, O'Connell KA, Brown BH, Galvao T, Daneault C, Des Rosiers C, Polster BM, Hoppel CL, Stanley WC. **Improved mitochondrial function with diet-induced increase in either docosahexaenoic acid or arachidonic acid in membrane phospholipids.** PLoS One. 2012;7(3):e34402. Epub 2012 Mar 30. *(PubMed Abstract)*

- Rugolotto S, Prioli MD, Toniolo D, Pellegrino P, Catuogno S, Burlina AB. **Long-term treatment of Barth syndrome with pantothenic acid: A retrospective study**. Mol Genet Metab 2003 Dec; 80(4):408-411. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Overmars H, Vaz FM, Barth PG, van Gennip AH. **Linoleic acid supplementation of Barth syndrome fibroblasts restores cardiolipin levels: Implications for treatment**. J Lipid Res. 2003 Mar;44(3):560-6. Epub 2002 Dec 16. *(PubMed Abstract)*▼

- Kelly GS. **L-Carnitine: Therapeutic applications of a conditionally-essential amino acid**. Altern Med Rev 1998 Oct; 3(5):345-360. *(PubMed Abstract)*

- Ostman-Smith I, Brown G, Johnson A, Land JM. **Dilated cardiomyopathy due to type II X-linked 3-methylglutaconic aciduria: Successful treatment with pantothenic acid**. Br Heart J. 1994 Oct;72(4):349-53. *(PubMed Abstract)*

PATENTS AND PATENT APPLICATIONS OF INTEREST

- Wilson DT, Bamberger M. **Methods and compositions for the prevention or treatment of Barth syndrome.** Patent Application US 9,687,519 B2. June 27, 2017. *(Patent Application)*
- Chin MT, Chien WM, Dinca A. **Modified *tafazzin* proteins and methods of making and using the same.** Patent Application 20150203827. July 23, 2015. *(Patent Application)*
- Wilson D. **Methods and compositions for the prevention or treatment of Barth syndrome.** International Publication Number W0 2014/134554 A1. 2014 Sep 4. *(Patent Application)*
- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*
- Timothy Andrew Stewart. **Methods and compositions for treating Barth syndrome, cardiomyopathy, mitochondrial diseases and other conditions.** Publication Date: December 3, 2009 *(Patent Application)*
- Peter Wipf, Jingbo Xiao, Mitchell P. Fink, Valerian E. Kagan, Yulia Y Tyurina. **Selective targeting agents for mitochondria.** US Patent Issued: May 5, 2009. *(Patent Abstract)*
- Genevieve C. Sparagna, Adam J. Chicco, Russell L. Moore, Sylvia A. McCune. **Use of linoleic compounds against heart failure.** Pubication Date: Dec. 25, 2008. *(Patent Application)*

PSYCHOLOGY

- Jacob ML, Johnco C, Dane BF, Collier A, Storch EA. **Psychosocial functioning in Barth syndrome: Assessment of individual and parental adjustment.** Children's Health Care, 46:1, 66-92 (2015). *(Abstract)*▼
- Kohlmann S, Kilbert MS, Ziegler K, Schulz KH. **Supportive care needs in patients with cardiovascular disorders.** Patient Educ Couns. 2013 June;91(3):378-84. Epub 2013 Feb 4. *(PubMed Abstract)*
- Pulgaron ER, Wile D, Schneider K, Young ML, Delamater AM. **Quality of life and psychosocial functioning of children with cardiac arrhythmias.** Cardiol Young. 2012 Apr 18:1-7. [Epub ahead of print] *(PubMed Abstract)*
- Storch EA, Keeley M, Merlo LJ, St. Amant JB, Jacob M, Storch J, Spencer C, Byrne BJ. **Psychosocial functioning in youth with Barth syndrome.** Child Health Care. 2009 Apr;38(2):137-156. *(PubMed – Open Access)**▼

- Kranzler JH, Rosenbloom AL, Proctor B, Diamond FB, Watson M. **Is short stature a handicap? A comparison of the psychosocial functioning of referred and nonreferred children with normal short stature and children with normal stature**. J Pediatr 2000 Jan; 136(1):96-102. *(PubMed Abstract)*

- The American Society of Human Genetics Board of Directors and The American College of Medical Genetics Board of Directors. ASHG/ACMG Report – **Points to consider: Ethical, legal, and psychosocial implications of genetic testing in children and adolescents**. Am J Hum Genet. 1995 Nov;57(5):1233-41. *(PubMed Abstract)*

REGISTRIES AND BIOREPOSITORIES

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Lakdawala NK. **Big data for a rare disease: Examining heart transplantation for left ventricular noncompaction in the UNOS registry.** J Heart Lung Transplant, Available online 4 April 2015. *(ScienceDirect Abstract)*

- Falk MJ, Shen L, Gonzalez M, Leipzig J, Lott MT, Stassen APM, Diroma MA, Navarro-Gomez D, Yeske P, Bai R, Boles RG, Brilhante B, Ralph D, Dare JT, Shelton R, Terry S, Zhang Z, Copeland WC, van Oven M, Prokisch H, Wallace DC, Attimonelli M, Krotoski D, Zuchner S, Gai X. **Mitochondrial Disease Sequence Data Resource (MSeqDR): A global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities.** Mol Genet Metab. 2014 Dec 4. pii: S1096-7192(14)00377-1. doi: 10.1016/j.ymgme.2014.11.016. [Epub ahead of print] *(PubMed Abstract)*

- Roberts AE, Nixon C, Steward CG, Gauvreau K, Maisenbacher M, Fletcher M, Geva J, Byrne BJ, Spencer CT. **The Barth Syndrome Registry: Distinguishing disease characteristics and growth data from a longitudinal study.** Am J Med Genet A. 2012 Nov;158A(11):2726-32. doi: 10.1002/ajmg.a.35609. Epub 2012 Oct 8. *(PubMed – Open Access)**▼

- Rubinstein YR, Groft SC, Chandros SH, Kaneshiro J, Karp B, Lockhart NC, Marshall PA, Moxley RT 3rd, Pollen GB, Miller VR, Schwartz J. **Informed consent process for patient participation in rare disease registries linked to biorepositories.** Contemp Clin Trials. 2012 Jan;33(1):5-11. Epub 2011 Oct 24. *(PubMed – No Abstract Available)*

- Alvarez JA, Orav EJ, Wilkinson JD, Fleming LE, Lee DJ, Sleeper LA, Rusconi PG, Colan SD, Hsu DT, Canter CE, Webber SA, Cox GF, Jefferies JL, Towbin JA, Lipshultz SE; for the Pediatric Cardiomyopathy Registry Investigators. **Competing risks for death and cardiac transplantation in children with dilated cardiomyopathy: Results from the Pediatric Cardiomyopathy Registry.** Circulation. 2011 Jul 25. [Epub ahead of print] *(PubMed Abstract)*

- Rubinstein YR, Groft SC, Bartek R, Brown K, Christensen RA, Collier E, Farber A, Farmer J, Ferguson JH, Forrest CB, Lockhart NC, McCurdy KR, Moore H, Pollen GB, Richesson R, Miller VR, Hull S, Vaught J. **Creating a global rare disease patient registry linked to a rare diseases biorepository database: Rare Disease-HUB (RD-HUB).** Contemp Clin Trials. 2010 Sep;31(5):394-404. Epub 2010 Jul 8. *(PubMed Abstract)*

- Rubinstein YR, Groft SC. **Driving interest in consolidating resources for the creation of a global rare disease patient registry.** Contemp Clin Trials. 2010 Sep;31(5):393. Epub 2010 Jul 7. *(PubMed - No Abstract Available)*

- Forrest CB, Bartek RJ, Rubinstein Y, Groft SC. **The case for a global rare-diseases registry.** Lancet. 2010 Jul 30. [Epub ahead of print]. *(PubMed – No Abstract Available)*

- Richesson R, Vehik K. **Patient registries: Utility, validity and inference.** Adv Exp Med Biol. 2010;686:87-104. *(PubMed Abstract)*

SKELETAL MYOPATHY

- Chabi B, Fouret G, Lecomte J, Cortade F, Pessemesse L, Baati N, Coudray C, Lin L, Tong Q, Wrutniak-Cabello C, Casas F, Feillet-Coudray C. **Skeletal muscle overexpression of short isoform Sirt3 altered mitochondrial cardiolipin content and fatty acid composition.** J Bioenerg Biomembr. 2018 Mar 27. doi: 10.1007/s10863-018-9752-1. [Epub ahead of print] *(PubMed Abstract)*

- Imai-Okazaki A, Kishita Y, Kohda M, Yatsuka Y, Hirata T, Mizuno Y, Harashima H, Hirono K, Ichida F, Noguchi A, Yoshida M, Tokorodani C, Nishiuchi R, Takeda A, Nakaya A, Sakata Y, Murayama K, Ohtake A, Okazaki Y. **Barth syndrome: Different approaches to diagnosis.** J Pediatr. 2017 Dec 15. pii: S0022-3476(17)31329-X. doi: 10.1016/j.jpeds.2017.09.075. [Epub ahead of print] *(PubMed Abstract)*

- Feingold B, Mahle WT, Auerbach S, Clemens P, Domenighetti AA, Jefferies JL, Judge DP, Lal AK, Markham LW, Parks WJ, Tsuda T, Wang PJ, Yoo SJ; American Heart Association Pediatric Heart Failure Committee of the Council on Cardiovascular Disease in the Young; Council on Clinical Cardiology; Council on Cardiovascular Radiology and Intervention; Council on Functional Genomics and Translational Biology; and Stroke Council. **Management of cardiac involvement associated with neuromuscular diseases: A scientific statement from the American Heart Association.** Circulation. 2017 Sep 26;136(13):e200-e231. doi: 10.1161/CIR.0000000000000526. Epub 2017 Aug 24. *(PubMed Abstract)*

- Fajardo VA, Mikhaeil JS, Leveille CF, Saint C, LeBlanc PJ. **Cardiolipin content, linoleic acid composition, and *tafazzin* expression in response to skeletal muscle overload and unload stimuli.** Sci Rep. 2017 May 17;7(1):2060. doi:10.1038/s41598-017-02089-1. *(PubMed - Open Access)*

- Bashir A, Bohnert KL, Reeds DN, Peterson LR, Bittel AJ, de Las Fuentes L, Pacak CA, Byrne BJ, Cade WT**. Impaired cardiac and skeletal muscle bioenergetics in children, adolescents, and young adults with Barth syndrome.** Physiol Rep. 2017 Feb;5(3). pii: e13130. doi: 10.14814/phy2.13130. *(PubMed – Open Access)*▼

- Ikon N, Ryan RO. **Barth syndrome: Connecting cardiolipin to cardiomyopathy**. Lipids. 2017 Jan 9. doi: 10.1007/s11745-016-4229-7. [Epub ahead of print] *(PubMed Abstract)*

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)**▼

- Finsterer J, Stöllberger C. **Heart disease in disorders of muscle, neuromuscular transmission, and the nerves.** Korean Circ J. 2016 Mar;46(2):117-34. doi: 10.4070/kcj.2016.46.2.117. Epub 2016 Mar 21. Review. *(PubMed Abstract)*

- Thompson WR, DeCroes B, McClellan R, Rubens J, Vaz FM, Kristaponis K, Avramopoulos D, Vernon HJ. **New targets for monitoring and therapy in Barth syndrome.** Genet Med. 2016 Feb 4. doi: 10.1038/gim.2015.204. [Epub ahead of print] *(PubMed Abstract)*▼

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S**. Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Tocchi A, Quarles EK, Basisty N, Gitari L, Rabinovitch PS. **Mitochondrial dysfunction in cardiac aging.** Biochim Biophys Acta. 2015 Jul 17. pii: S0005-2728(15)00152-8. doi: 10.1016/j.bbabio.2015.07.009. [Epub ahead of print] *(PubMed Abstract)*

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Zapala B, Platek T, Wybrańska I. **A novel *TAZ* gene mutation and mosaicism in a Polish family with Barth syndrome.** Ann Hum Genet. 2015 Mar 16. doi: 10.1111/ahg.12108. [Epub ahead of print]. *(PubMed Abstract)*

- Ferri L, Donati MA, Funghini S, Cavicchi C, Pensato V, Gellera C, Natacci F, Spaccini L, Gasperini S, Vaz FM, Cooper DN, Guerrini R, Morrone A. **Intra-individual plasticity of the *TAZ* gene leading to different heritable mutations in siblings with Barth syndrome.** Eur J Hum Genet. 2015 Mar 18. doi: 10.1038/ejhg.2015.50. [Epub ahead of print] *(PubMed Abstract)*▼

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Folsi V, Miglietti N, Lombardi A, Boccacci S, Utyatnikova T, Donati C, Squassabia L, Gazzola L, Bosio I, Borghi A, Grassi V, Notarangelo LD, Plebani A. **Cardiomyopathy in a male patient with neutropenia and growth delay.** Ital J Pediatr. 2014 May 12;40:45. doi:10.1186/1824-7288-40-45. *(PubMed – Open Access)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Finsterer J, Stöllberger C. **Unclassified cardiomyopathies in neuromuscular disorders.** Wien Med Wochenschr. 2013 Oct 24. [Epub ahead of print] *(PubMed Abstract)*

- Finsterer J, Stöllberger C. **Ultrastructural findings in noncompaction prevail with neuromuscular disorders.** Cardiology. 2013 Sep 21;126(4):219-223. [Epub ahead of print] (*PubMed Abstract)*

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Whited K, Baile MG, Currier P, Claypool SM. **Seven functional classes of Barth syndrome mutation.** Hum Mol Genet. 2013 Feb 1;22(3):483-92. doi: 10.1093/hmg/dds447. Epub 2012 Oct 24. *(PubMed Abstract)*

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Kaplan JC, Hamroun D. **The 2013 version of the gene table of neuromuscular disorders (nuclear genome).** Neuromuscular Disorders, Volume 22, Issue 12, December 2012, Pages 1108-1135. *(Abstract)*

- Cade WT, Spencer CT, Reeds DN, Waggoner AD, O'Connor R, Maisenbacher M, Crowley JR, Byrne BJ, Peterson LR. **Substrate metabolism during basal and hyperinsulinemic conditions in adolescents and young-adults with Barth syndrome.** J Inherit Metab Dis. 2012 May 12. [Epub ahead of print] *(PubMed Abstract)**▼

- Saini-Chohan HK, Mitchell RW, Vaz FM, Zelinski T, Hatch GM. **Delineating the role of alterations in lipid metabolism to the pathogenesis of inherited skeletal and cardiac muscle disorders.** J Lipid Res. 2011 Nov 7. [Epub ahead of print] *(PubMed – Open Access)**

- Spencer CT, Byrne BJ, Bryant RM, Margossian R, Maisenbacher M, Breitenger P, Benni PB, Redfearn S, Marcus E, Cade WT. **Impaired cardiac reserve and severely diminished skeletal muscle oxygen utilization mediate exercise intolerance in Barth syndrome.** Am J Physiol Heart Circ Physiol. 2011 Nov;301(5):H2122-9. Epub 2011 Aug 26. *(PubMed – Open Access)**▼

- Lisi EC, Cohn RD. **Genetic evaluation of the pediatric patient with hypotonia: Perspective from a hypotonia specialty clinic and review of the literature.** Dev Med Child Neurol. 2011 Jul;53(7):586-99. doi: 10.1111/j.1469-8749.2011.03918.x. Epub 2011 Mar 21. Review. *(PubMed Abstract)*

- Acehan D, Vaz F, Houtkooper RH, James J, Moore V, Tokunaga C, Kulik W, Wansapura J, Toth MJ, Strauss A, Khuchua Z. **Cardiac and skeletal muscle defects in a mouse model of human Barth syndrome.** J Biol Chem. 2011 Jan 14;286(2):899-908. Epub 2010 Nov 9. *(PubMed – Open Access)**

- Finsterer J, Stollberger C. **Cardiac involvement in primary myopathies**. Cardiology 2000; 94(1):1-11. *(PubMed Abstract)*

TRANSITION FROM PEDIATRIC TO ADULT HEALTH CARE

- Shemesh E, Annunziato RA, Arnon R, Miloh T, Kerkar N. **Adherence to medical recommendations and transition to adult services in pediatric transplant recipients.** Curr Opin Organ Transplant. 2010 Jun;15(3):288-92. Review. *(PubMed Abstract)*

- Wong, LHL, Chan FWK, Wong FYY, Wong ELY, Huen KF, Yeoh EK, Fok TF. **Transition care for adolescents and families with chronic illnesses.** Journal of Adolescent Health, In Press, Corrected Proof, Available online 11 June 2010. *(Abstract)*

- Rapley P, Davidson PM. **Enough of the problem: A review of time for health care transition solutions for young adults with a chronic illness.** J Clin Nurs. 2010 Feb;19(3-4):313-23. Review. *(PubMed Abstract)*

- From the American Academy of Pediatrics: **Policy Statements-Supplemental Security Income (SSI) for children and youth with disabilities.** Council on Children with Disabilities. Pediatrics. 2009 Dec;124(6):1702-8. *(PubMed Abstract)*

- Christie D, Viner R. **Chronic illness and transition: Time for action.** Adolesc Med State Art Rev. 2009 Dec;20(3):981-7, xi. Review. *(PubMed Abstract)*

- Betz CL. **Health care transitions of youth with special health care needs: The never ending journey.** Commun Nurs Res. 2008 Spring;41:13-29. *(PubMed - No Abstract Available*)

- Lotstein DS, McPherson M, Strickland B, Newacheck PW. **Transition planning for youth with special health care needs: Results from the National Survey of Children with Special Health Care Needs.** Pediatrics. 2005 Jun;115(6):1562-8. *(PubMed Abstract)*

- Lotstein DS, Ghandour R, Cash A, McGuire E, Strickland B, Newacheck P. **Planning for health care transitions: Results from the 2005-2006 National Survey of Children with Special Health Care Needs.** Pediatrics. 2009 Jan;123(1):e145-52. *(PubMed Abstract)*

- McDonagh JE, Viner RM. **Lost in transition? Between paediatric and adult services**. BMJ. 2006 Feb 25;332(7539):435-6. *(PubMed Abstract)*

- Chamberlain MA, Kent RM. **The needs of young people with disabilities in transition from paediatric to adult services.** Eura Medicophys. 2005 Jun;41(2):111-23. *(PubMed Abstract)*

- Reiss J, Gibson R. **Health care transition: Destinations unknown**. Pediatrics. 2002 Dec;110(6 Pt 2):1307-14. *(PubMed Abstract)*

- American Academy of Pediatrics, American Academy of Family Physicians, American College of Physicians - American Society of Internal Medicine. **A consensus statement on health care transitions for young adults with special health care needs**. Pediatrics. 2002 Dec; 110(6 Pt 2):1304-6. *(PubMed Abstract)*

- Neinstein LS. **The treatment of adolescents with a chronic illness**. West J Med. 2001 Nov;175(5):293-5. *(PubMed Abstract)*

- Lewis-Gary MD. **Transitioning to adult health care facilities for young adults with chronic condition**. Pediatr Nurs 2001 Sep-Oct; 27(5):521-524. *(PubMed Abstract)*

- Callahan ST, Winitzer RF, Keenan P. **Transition from pediatric to adult-oriented health care: A challenge for patients with chronic disease**. Curr Opin Pediatr 2001 August; 13(4):310-316. *(PubMed Abstract)*

- American Academy of Pediatrics. **The role of the pediatrician in transitioning children and adolescents with developmental disabilities and chronic illness from school to work or college**. Pediatrics. 2000 Oct;106(4):854-6. *(PubMed Abstract)*

- Viner R. **Transition from paediatric to adult care. Bridging the gaps or passing the buck?** Arch Dis Child. 1999 Sep;81(3):271-5. *(PubMed Abstract)*

- American Academy of Pediatrics Committee on Children with Disabilities and Committee on Adolescence. **Transition of care provided for adolescents with special health care needs**. Pediatrics 1996 Dec; 98(6 Pt 1):1203-1206. *(PubMed Abstract)*

- Rosen DS. **Transition from pediatric to adult-oriented health care for the adolescent with chronic illness or disability**. Adolesc Med 1994 Jun; 5(2):241-248. *(PubMed Abstract)*

- Schidlow DV, Fiel SB. **Life beyond pediatrics. Transition of chronically ill adolescents from pediatric to adult health care systems**. Med Clin North Am 1990 Sep; 74(5):1113-1120. *(PubMed Abstract)*

TREATMENT OF SYMPTOMS RELEVANT TO BARTH SYNDROME PATIENTS

CARDIAC

NEW Suzuki-Hatano S, Saha M, Rizzo SA, Witko RL, Gosiker BJ, Ramanathan M, Soustek MS, Jones MD, Kang PB, Byrne BJ, Cade WT, Pacak CA. **AAV-mediated *TAZ* gene replacement restores mitochondrial and cardioskeletal function in Barth syndrome.** Hum Gene Ther. 2018 Aug 2. doi: 10.1089/hum.2018.020. [Epub ahead of print]*

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access)**

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review. *(PubMed Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)**▼

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Chanana AM, Rhee JW, Wu JC. **Human-induced pluripotent stem cell approaches to model inborn and acquired metabolic heart diseases.** Curr Opin Cardiol. 2016 Mar 26. [Epub ahead of print] *(PubMed Abstract)*

- Mathur A, Ma Z, Loskill P, Jeeawoody S, Healy KE. ***In vitro* cardiac tissue models: Current status and future prospects.** Adv Drug Deliv Rev. 2015 Sep 30. pii: S0169-409X(15)00214-8. doi: 10.1016/j.addr.2015.09.011. [Epub ahead of print] Review. *(PubMed Abstract)*▼

- Kang SL, Forsey J, Dudley D, Steward CG, Tsai-Goodman B. **Clinical characteristics and outcomes of cardiomyopathy in Barth syndrome: The UK experience.** Pediatr Cardiol. 2015 Sep 4. [Epub ahead of print] *(PubMed Abstract)*▼

- Reynolds S**. Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Wegener M, Bader A, Giri S**. How to mend a broken heart: Adult and induced pluripotent stem cell therapy for heart repair and regeneration.** Review Article. Drug Discovery Today, Available online 23 February 2015. *(ScienceDirect Abstract)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Zhao Y, Feric NT, Thavandiran N, Nunes SS, Radisic M. **The role of tissue engineering and biomaterials in cardiac regenerative medicine.** Can J Cardiol. 2014 Nov;30(11):1307-1322. doi: 10.1016/j.cjca.2014.08.027. Epub 2014 Sep 4. Review. *(PubMed Abstract)*

- Zweigerdt R, Gruh I, Martin U. **Your heart on a chip: iPSC-based modeling of Barth-syndrome-associated cardiomyopathy.** Cell Stem Cell. 2014 Jul 3;15(1):9-11. doi:10.1016/j.stem.2014.06.015. *(PubMed – Open Access)*

- Wang G, McCain ML, Yang L, He A, Pasqualini FS, Agarwal A, Yuan H, Jiang D, Zhang D, Zangi L, Geva J, Roberts AE, Ma Q, Ding J, Chen J, Wang DZ, Li K, Wang J, Wanders RJA, Kulik W, Vaz FM, Laflamme MA, Murry CE, Chien KR, Kelley RI, Church GM, Parker K, Pu WT. **Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies.** Nat Med. 2014 Jun;20(6):616-23. doi:10.1038/nm.3545 Epub. 2014 May 11. *(PubMed – Open Access)**▼

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Kohlmann S, Kilbert MS, Ziegler K, Schulz KH. **Supportive care needs in patients with cardiovascular disorders.** Patient Educ Couns. 2013June;91(3):378-84. *(PubMed Abstract)*

- Dedieu N, Giardini A, Steward CG, Fenton M, Karimova A, Hsia TY, Burch M. **Successful mechanical circulatory support for 251 days in a child with intermittent severe neutropenia due to Barth syndrome.** Pediatr Transplant. 2013 Mar;17(2):E46-9. doi: 10.1111/petr.12027. Epub 2012 Nov 28. *(PubMed Abstract)*▼

- Hanke SP, Gardner AB, Lombardi JP, Manning PB, Nelson DP, Towbin JA, Jefferies JL, Lorts A. **Left ventricular noncompaction cardiomyopathy in Barth syndrome: An example of an undulating cardiac phenotype necessitating mechanical circulatory support as a bridge to transplantation.** Pediatr Cardiol. 2012 Mar 17. [Epub ahead of print] *(PubMed Abstract)*

- Huang SC, Wu ET, Chiu SN, Hwu WL, Wu MH, Wang SS. **Mitral annuloplasty in an infant with Barth syndrome and severe mitral insufficiency: First case report and determination of annular diameter.** J Thorac Cardiovasc Surg. 2008 Oct;136(4):1095-7. *(PubMed - No Abstract Available)*

- Mangat J, Lunnon-Wood T, Rees P, Elliott M, Burch M. **Successful cardiac transplantation in Barth syndrome - single-centre experience of four patients.** Pediatr Transplant. 2007 May;11(3):327-331. *(PubMed Abstract)*

- Moffett BS, Chang AC. **Future pharmacologic agents for treatment of heart failure in children.** Pediatr Cardiol. 2006 Sep-Oct;27(5):533-51. Epub 2006 Aug 23. Review. *(PubMed Abstract)*

- Nakazawa M, Saji T, Ichida F, Oyama K, Harada K, Kusuda S. **Guidelines for the use of palivizumab in infants and young children with congenital heart disease.** Pediatr Int. 2006 Apr;48(2):190-3. *(PubMed Abstract)*

- POSITION STATEMENT (ID 2003-03). **Use of palivizumab in children with congenital heart disease.**

- DiMarco JP. **Implantable cardioverter-defibrillators**. (Review article) N Engl J Med 2003 Nov 6; 349(19):1836-1847. *(PubMed Abstract)*

- Hohnloser SH, Klingenheben T, Bloomfield D, Dabbous O, Cohen RJ. **Usefulness of microvolt t-wave alternans for prediction of ventricular tachyarrhythmic events in patients with dilated cardiomyopathy: Results from a prospective observational study**. J Am Coll Cardiol 2003 June 18; 41(12):2220-2224. *(PubMed Abstract)*

- Klingenheben T, Gronefeld G, Li Y, Hohnloser SH. **Effect of Metoprolol and *d,l*-Sotalol on microvolt-level t-wave alternans**. J Am Coll Cardiol 2001 Dec; 38(7):2013-2019. *(PubMed Abstract)*

- Bruns LA, Chrisant MK, Lamour JM, Shaddy RE, Pahl E, Blume ED, Hallowell Ss, Addonizio LJ, Canter CE. **Carvedilol as therapy in pediatric heart failure: An initial multicenter experience.** J Pediatr 2001 Apr; 138(4):505-511. *(PubMed Abstract)*

- Adwani SS, Whitehead BF, Rees PG, Morris A, Turnball DM, Elliott MJ, de Leval MR. **Heart transplantation for Barth syndrome**. Pediatr Cardiol. 1997 Mar-Apr;18(2):143-5. *(PubMed Abstract)*

- Schwartz ML, Cox GF, Lin AE, Korson MS, Perez-Atayde A, Lacro RV, Lipshultz SE. **Clinical approach to genetic cardiomyopathy in children**. Circulation. 1996 Oct 15;94(8):2021-38. *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

NEUTROPENIA

NEW Steward CG, Groves SJ, Taylor CT, Maisenbacher MK, Versluys B, Newbury-Ecob RA, Ozsahin H, Damin MK, Bowen VM, McCurdy KR, Mackey MC, Bolyard AA, Dale DC. **Neutropenia in Barth syndrome: Characteristics, risks, and management.** Curr Opin Hematol. 2018 Nov 16. doi: 10.1097/MOH.0000000000000472. [Epub ahead of print] *(PubMed Abstract)*▼

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Ikon N, Su B, Hsu FF, Forte TM, Ryan RO. **Exogenous cardiolipin localizes to mitochondria and prevents *TAZ* knockdown-induced apoptosis in myeloid progenitor cells.** Biochem Biophys Res Commun. 2015 Jul 8. pii: S0006-291X(15)30243-6. doi: 10.1016/j.bbrc.2015.07.012. [Epub ahead of print] *(PubMed Abstract)**

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Donadieu J, Rigaud C, Lebre AS, Touraine R, Ottolenghi C, Chabli A, Charron P, Rio M, De Lonlay P, Bonnet D. **Syndrome de Barth: le reconnaître, le traiter. Recommandations pour la prise en charge (Barth syndrome: Guidelines for diagnosis, follow-up and medical therapy).** Revue d'Oncologie Hématologie Pédiatrique, Available online 27 August 2014. *(ScienceDirect Abstract)*

- Aprikyan AA, Khuchua Z. **Advances in the understanding of Barth syndrome.** Br J Haematol. 2013 May;161(3):330-8. doi: 10.1111/bjh.12271. Epub 2013 Feb 25. *(PubMed Abstract)*

- Dedieu N, Giardini A, Steward CG, Fenton M, Karimova A, Hsia TY, Burch M. **Successful mechanical circulatory support for 251 days in a child with intermittent severe neutropenia due to Barth syndrome.** Pediatr Transplant. 2013 Mar;17(2):E46-9. doi: 10.1111/petr.12027. Epub 2012 Nov 28. *(PubMed Abstract)*▼

- Clarke SLN, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, Martin RP, Tsai-Goodman B, Garratt V, Ashworth M, Bowen VM, McCurdy KR, Damin MK, Spencer CT, Toth MJ, Kelley RI, Steward CG. **Barth syndrome.** Orphanet J Rare Dis. 2013 Feb 12;8:23. doi: 10.1186/1750-1172-8-23. Review. *(PubMed – Open Access)**▼

- Boxer LA. **How to approach neutropenia.** Hematology Am Soc Hematol Educ Program. 2012;2012:174-82. doi: 10.1182/asheducation-2012.1.174. *(PubMed Abstract)*

- Badolato R, Fontana S, Notarangelo LD, Savoldi G**. Congenital neutropenia: Advances in diagnosis and treatment.** Curr Opin Allergy Clin Immunol. 2004 Dec;4(6):513-21. *(PubMed Abstract)*

- Dale DC, Bolyard AA, Schwinzer B, Pracht G, Bonilla MA, Boxer L, Freedman M, Donadieu J, Kannourakis G, Alter BP, Cham B, Winkelstein J, Kinsey SE, Fier C, Zeidler C, Welte K. **The Severe Chronic Neutropenia International Registry - 10 years of follow-up** [abstract]. American Society of Hematology Annual Meeting; Blood Nov 2004; 104: 1458.

- Maheshwari and Christensen. **Neutropenia in the Neonatal Intensive Care Unit.**

- Committee on Infectious Diseases and Committee on Fetus and Newborn. **Revised indications for the use of palivizumab and respiratory syncytial virus infections immune globulin intravenous for the prevention of respiratory syncytial virus.** Pediatrics. 2003 Dec;112(6 Pt 1):1442-6. *(PubMed Abstract)*

- Dale DC, Cottle TE, Fier CJ, Bolyard AA, Bonilla MA, Boxer LA, Cham B, Freedman MH, Kannourakis G, Kinsey SE, Davis R, Scarlata D, Schwinzer B, Zeidler C, Welte K. **Severe chronic neutropenia: Treatment and follow-up of patients in the Severe Chronic Neutropenia International Registry.** Am J Hematol. 2003 Feb;72(2):82-93. *(PubMed Abstract)*

- Cottle TE, Fier CJ, Donadieu J, Kinsey SE. **Risk and benefit of treatment of severe chronic neutropenia with granulocyte colony-stimulating factor**. Semin Hematol. 2002 Apr;39(2):134-40. *(PubMed Abstract)*

- Zeidler C, Barth PG, Bonilla MA, Bolyard AA, Boxer L, Cottle T, Dale DC, Donadieu J, Fier C, Freedman M, Kannourakis G, Kinsey S, Liang B, Schwinzer B, Welte K, Cham B, for the Severe Chronic Neutropenia International Registry (SCNIR). **Neutropenia in Barth syndrome: Clinical course and treatment of neutropenia**. Blood 2001; 98(11):300a.

- Bolyard AA, Cottle T, Edwards C, Kinsey S, Schwinzer B, Zeidler C. **Understanding severe chronic neutropenia – A handbook for patients and their families**.

- Welte K, Boxer LA. **Severe chronic neutropenia: Pathophysiology and therapy**. Semin Hematol. 1997 Oct;34(4):267-78. *(PubMed Abstract)*

- Welte K, Dale D. **Pathophysiology and treatment of severe chronic neutropenia**. Ann Hematol. 1996 Apr;72(4):158-65. *(PubMed Abstract)*

- Cox GF, Pulsipher M, Rothenberg M, Korson M, Kelley RI. **Correction of neutropenia in Barth syndrome by G-CSF**. Am J Hum Genet 1995; 57:A177. *(Full Text)*

- Bonilla MA, Dale D, Zeidler C, Last L, Reiter A, Ruggeiro M, Davis M, Koci B, Hammond W, Gillio A, Welte K. **Long-term safety of treatment with recombinant human granulocyte colony-stimulating factor (r-metHuG-CSF) in patients with severe congenital neutropenias**. Br J Haematol. 1994 Dec;88(4):723-30. *(PubMed Abstract)*

- Hammond WP, Price TH, Souza LM, Dale DC. **Treatment of cyclic neutropenia with granulocyte colony-stimulating factor**. N Engl J Med. 1989 May 18;320(20):1306-11. *(PubMed Abstract)*

- Jakubowski AA, Souza L, Kelly F, Fain K, Budman D, Clarkson B, Bonilla MA, Moore MAS, Gabrilove J. **Effects of human granulocyte colony-stimulating factor in a patient with idiopathic neutropenia**. N Engl J Med. 1989 Jan 5;320(1):38-42. *(PubMed Abstract)*

METABOLIC DISORDERS

- Sudrié-Arnaud B, Marguet F, Patrier S, Martinovic J, Louillet F, Broux F, Charbonnier F, Dranguet H, Coutant S, Vezain M, Lanos R, Tebani A, Fuller M, Lamari F, Chambon P, Brehin AC, Trestard L, Tournier I, Marret S, Verspyck E, Laquerrière A, Bekri S. **Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation.** Clin Chim Acta. 2018 Jun;481:1-8. doi: 10.1016/j.cca.2018.02.023. Epub 2018 Feb 22. *(PubMed Abstract)*

- Ikon N, Hsu FF, Shearer J, Forte TM, Ryan RO. **Evaluation of cardiolipin nanodisks as lipidreplacement therapy for Barth syndrome.** J Biomed Res. 2017 Nov 1. doi: 10.7555/JBR.32.20170094. [Epub ahead of print] *(PubMed - Open Access)**

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed - Open Access)**

- Sathappa M, Mitchell W, Coscia A, Boyd K, May E, Szeto HH, Alder NN. **Investigation of the interactions of the SS-31 peptides with cardiolipin variants: A potential therapeutic for Barth syndrome.** Biophysical Journal, Volume 112, Issue 3, Supplement 1, 3 February 2017, Pages 438a. *(ScienceDirect Abstract)*

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review. *(PubMed Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)*▼

- Dinca A, Chien WM, Chin MT. **Intracellular delivery of proteins with cell-penetrating peptides for therapeutic uses in human disease.** Int J Mol Sci. 2016 Feb 22;17(2):263. doi: 10.3390/ijms17020263. Review. *(PubMed – Open Access)**

- Reynolds S. **Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Wegener M, Bader A, Giri S. **How to mend a broken heart: Adult and induced pluripotent stem cell therapy for heart repair and regeneration.** Review Article. Drug Discovery Today, Available online 23 February 2015. *(ScienceDirect Abstract)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Zweigerdt R, Gruh I, Martin U. **Your heart on a chip: iPSC-based modeling of Barth-syndrome-associated cardiomyopathy.** Cell Stem Cell. 2014 Jul 3;15(1):9-11. doi:10.1016/j.stem.2014.06.015. *(PubMed – Open Access)*

- Wang G, McCain ML, Yang L, He A, Pasqualini FS, Agarwal A, Yuan H, Jiang D, Zhang D, Zangi L, Geva J, Roberts AE, Ma Q, Ding J, Chen J, Wang DZ, Li K, Wang J, Wanders RJA, Kulik W, Vaz FM, Laflamme MA, Murry CE, Chien KR, Kelley RI, Church GM, Parker K, Pu WT. **Modeling the mitochondrial cardiomyopathy of Barth syndrome with induced pluripotent stem cell and heart-on-chip technologies.** Nat Med. 2014 Jun;20(6):616-23. doi:10.1038/nm.3545 Epub. 2014 May 11. *(PubMed – Open Access)**▼

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*▼

- Kachewar SG, Sankaye SB, Kulkarni DS. **The role of radio-diagnosis in inborn errors of metabolism.** Journal of Clinical and Diagnostic Research [serial online] 2011 November [cited: 2012 Dec 5]; 5:1467-1472. *(Full Text)*

- Rugolotto S, Prioli MD, Toniolo D, Pellegrino P, Catuogno S, Burlina AB. **Long-term treatment of Barth syndrome with pantothenic acid: A retrospective study**. Mol Genet Metab. 2003 Dec; 80(4):408-11. *(PubMed Abstract)*

- Valianpour F, Wanders RJA, Overmars H, Vaz FM, Barth PG, van Gennip AH. **Linoleic acid supplementation of Barth syndrome fibroblasts restores cardiolipin levels: Implications for treatment**. J Lipid Res. 2003 Mar;44(3):560-6. Epub 2002 Dec 16. *(PubMed Abstract)*▼

- Kelly GS. **L-Carnitine: Therapeutic applications of a conditionally-essential amino acid**. Altern Med Rev 1998 Oct; 3(5):345-360. *(PubMed Abstract)*

MITOCHONDRIAL DISEASE

- NEW Suzuki-Hatano S, Saha M, Rizzo SA, Witko RL, Gosiker BJ, Ramanathan M, Soustek MS, Jones MD, Kang PB, Byrne BJ, Cade WT, Pacak CA. **AAV-mediated *TAZ* gene replacement restores mitochondrial and cardioskeletal function in Barth syndrome.** Hum Gene Ther. 2018 Aug 2. doi: 10.1089/hum.2018.020. [Epub ahead of print]*

- Agarwal P, Cole LK, Chandrakumar A, Hauff KD, Ravandi A, Dolinsky VW, Hatch GM. **Phosphokinome analysis of Barth syndrome lymphoblasts identify novel targets in the pathophysiology of the disease.** Int J Mol Sci. 2018 Jul 12;19(7). pii: E2026. doi: 10.3390/ijms19072026. (PubMed - Open Access)*▼

- Chen WW, Chao YJ, Chang WH, Chan JF, Hsu YHH. **Phosphatidylglycerol incorporates into cardiolipin to improve mitochondrial activity and inhibits inflammation.** Sci Rep. 2018 Mar 20;8(1):4919. doi: 10.1038/s41598-018-23190-z. *(PubMed Abstract)*

- Ikon N, Hsu FF, Shearer J, Forte TM, Ryan RO. **Evaluation of cardiolipin nanodisks as lipidreplacement therapy for Barth syndrome.** J Biomed Res. 2017 Nov 1. doi: 10.7555/JBR.32.20170094. [Epub ahead of print] *(PubMed - Open Access)**

- Huang Y, Powers C, Moore V, Schafer C, Ren M, Phoon CK, James JF, Glukhov AV, Javadov S, Vaz FM, Jefferies JL, Strauss AW, Khuchua Z. **The PPAR pan-agonist bezafibrate ameliorates cardiomyopathy in a mouse model of Barth syndrome.** Orphanet J Rare Dis. 2017 Mar 9;12(1):49. doi: 10.1186/s13023-017-0605-5. *(PubMed – Open Access)**

- Sathappa M, Mitchell W, Coscia A, Boyd K, May E, Szeto HH, Alder NN. **Investigation of the interactions of the SS-31 peptides with cardiolipin variants: A potential therapeutic for Barth syndrome.** Biophysical Journal, Volume 112, Issue 3, Supplement 1, 3 February 2017, Pages 438a. (*ScienceDirect Abstract)*

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Wu J, Ocampo A, Izpisua Belmonte JC. **Cellular metabolism and induced pluripotency.** Cell. 2016 Sep 8;166(6):1371-85. doi:10.1016/j.cell.2016.08.008. Review. *(PubMed Abstract)*

- Cai L, Fisher AL, Huang H, Xie Z. **CRISPR-mediated genome editing and human diseases.** Review Article. Genes & Diseases, Available online 30 August 2016. *(ScienceDirect – Open Access)*

- Cade WT, Reeds DN, Peterson LR, Bohnert KL, Tinius RA, Benni PB, Byrne BJ, Taylor CL. **Endurance exercise training in young adults with Barth syndrome: A pilot study.** JIMD Rep. 2016 Jun 11. [Epub ahead of print] *(PubMed Abstract)**▼

- Bradley RM, Stark KD, Duncan RE. **Influence of tissue, diet, and enzymatic remodeling on cardiolipin fatty acyl profile.** Mol Nutr Food Res. 2016 Apr 8. doi: 10.1002/mnfr.201500966. [Epub ahead of print] *(PubMed Abstract)*

- Dinca A, Chien WM, Chin MT. **Intracellular delivery of proteins with cell-penetrating peptides for therapeutic uses in human disease.** Int J Mol Sci. 2016 Feb 22;17(2):263. doi: 10.3390/ijms17020263. Review. *(PubMed – Open Access)**

- Reynolds S**. Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Healthc. 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*

- Hsu P, Liu X, Zhang J, Wang HG, Ye JM, Shi Y. **Cardiolipin remodeling by *TAZ/Tafazzin* is selectively required for the initiation of mitophagy.** Autophagy. 2015 Apr 28:0. [Epub ahead of print] *(PubMed Abstract)**

- Soustek MS, Baligand C, Falk DJ, Walter GA, Lewin AS, Byrne BJ. **Endurance training ameliorates complex 3 deficiency in a mouse model of Barth syndrome.** J Inherit Metab Dis. 2015 Apr 10. [Epub ahead of print] *(PubMed Abstract)*

- Wegener M, Bader A, Giri S**. How to mend a broken heart: Adult and induced pluripotent stem cell therapy for heart repair and regeneration.** Review Article. Drug Discovery Today, Available online 23 February 2015. *(ScienceDirect Abstract)*

- Ferreira C, Thompson WR, Vernon H. **Barth syndrome.** GeneReviews. October 9, 2014. *(PubMed – Open Access)*▼

- Malhotra A, Kahlon P, Donoho T, Doyle IC. **Pharmacogenomic considerations in the treatment of the pediatric cardiomyopathy called Barth syndrome.** Recent Pat Biotechnol. 2014 Sep 4. [Epub ahead of print] *(PubMed Abstract)*

- Chi C-S. **Diagnostic approach in infants and children with mitochondrial diseases.** Pediatr Neonatol. 2014 Aug 20. pii: S1875-9572(14)00099-0. doi: 10.1016/j.pedneo.2014.03.009. [Epub ahead of print] Review. *(PubMed Abstract)*

- Mayr JA. **Lipid metabolism in mitochondrial membranes.** J Inherit Metab Dis. 2014 Aug 1. [Epub ahead of print] *(PubMed Abstract)*

- Aires V, Delmas D, Le Bachelier C, Latruffe N, Schlemmer D, Benoist JF, Djouadi F, Bastin J. **Stilbenes and resveratrol metabolites improve mitochondrial fatty acid oxidation defects in human fibroblasts.** Orphanet J Rare Dis. 2014 Jun 5;9:79. doi: 10.1186/1750-1172-9-79. *(PubMed Abstract)*

- Vernon HJ, Sandlers Y, McClellan R, Kelley RI. **Clinical laboratory studies in Barth syndrome.** Mol Genet Metab. 2014 Jun;112(2):143-7. doi: 10.1016/j.ymgme.2014.03.007. Epub 2014 Mar 30. *(PubMed Abstract)*

- Birk AV, Chao WM, Bracken C, Warren JD, Szeto HH. **Targeting mitochondrial cardiolipin and the cytochrome c/cardiolipin complex to promote electron transport and optimize mitochondrial ATP synthesis.** Br J Pharmacol. 2014 Apr;171(8):2017-28. doi: 10.1111/bph.12468. *(PubMed Abstract)*

- Szeto HH. **First-in-class cardiolipin-protective compound as a therapeutic agent to restore mitochondrial bioenergetics.** Br J Pharmacol. 2014 Apr;171(8):2029-50. doi: 10.1111/bph.12461. Review. *(PubMed Abstract)*

- Kachewar SG, Sankaye SB, Kulkarni DS. **The role of radio-diagnosis in inborn errors of metabolism.** Journal of Clinical and Diagnostic Research [serial online] 2011 November [cited: 2012 Dec 5]; 5:1467-1472. *(Full Text)*

- Khairallah RJ, Kim J, O'Shea KM, O'Connell KA, Brown BH, Galvao T, Daneault C, Des Rosiers C, Polster BM, Hoppel CL, Stanley WC. **Improved mitochondrial function with diet-induced increase in either docosahexaenoic acid or arachidonic acid in membrane phospholipids.** PLoS One. 2012;7(3):e34402. Epub 2012 Mar 30. *(PubMed Abstract)*

- Mulligan CM, Sparagna GC, Le CH, De Mooy AB, Routh MA, Holmes MG, Hickson-Bick DL, Zarini S, Murphy RC, Xu FY, Hatch GM, McCune SA, Moore RL, Chicco AJ. **Dietary linoleate preserves cardiolipin and attenuates mitochondrial dysfunction in the failing rat heart.** Cardiovasc Res. 2012 Jun 1;94(3):460-8. Epub 2012 Mar 12. *(PubMed Abstract)*

- Parikh S, Saneto R, Falk MJ, Anselm I, Cohen BH, Haas R, Medicine Society TM. **A modern approach to the treatment of mitochondrial disease.** Curr Treat Options Neurol. 2009 Nov;11(6):414-30. *(PubMed Abstract)*

- Jackson SK, Abate W, Tonks AJ. **Lysophospholipid acyltransferases: Novel potential regulators of the inflammatory response and target for new drug discovery.** Pharmacol Ther. 2008 Jul;119(1):104-14. Epub 2008 Apr 23. Review. *(PubMed Abstract)*

- Schon EA, DiMauro S. **Medicinal and genetic approaches to the treatment of mitochondrial disease**. Curr Med Chem. 2003 Dec;10(23):2523-33. *(PubMed Abstract)*

- Ruitenbeck W, Wendel U, Hamel BC, Trijbels JM. **Genetic counselling and prenatal diagnosis in disorders of the mitochondrial energy metabolism**. J Inherit Metab Dis. 1996;19(4):581-7. *(PubMed Abstract)*

BSF does not endorse any drugs, tests, or treatments that we may report.

NEWBORN SCREENING

- Sudrié-Arnaud B, Marguet F, Patrier S, Martinovic J, Louillet F, Broux F, Charbonnier F, Dranguet H, Coutant S, Vezain M, Lanos R, Tebani A, Fuller M, Lamari F, Chambon P, Brehin AC, Trestard L, Tournier I, Marret S, Verspyck E, Laquerrière A, Bekri S. **Metabolic causes of nonimmune hydrops fetalis: A next-generation sequencing panel as a first-line investigation.** Clin Chim Acta. 2018 Jun;481:1-8. doi: 10.1016/j.cca.2018.02.023. Epub 2018 Feb 22. *(PubMed Abstract)*

- Brión M, de Castro López MJ, Santori M, Pérez Muñuzuri A, López Abel B, Baña Souto AM, Martínez Soto MI, Couce ML. **Prospective and retrospective diagnosis of Barth syndrome aided by next-generation sequencing.** Am J Clin Pathol. 2016 Apr 22. pii: aqw025. [Epub ahead of print] *(PubMed – Open Access)*

- Kachewar SG, Sankaye SB, Kulkarni DS. **The role of radio-diagnosis in inborn errors of metabolism.** Journal of Clinical and Diagnostic Research [serial online] 2011 November [cited: 2012 Dec 5]; 5:1467-1472. *(Full Text)*

- Garratt V, Riddiford D, Steward C, Tsai-Goodman B, Newbury-Ecob R. **What is Barth syndrome?** Midwives Magazine: Issue 4: 2011. *(Not peer reviewed) (Abstract)*▼

- Pass KA, Thoene J, Watson MS. **Emergency preparedness for newborn screening and genetic services.** Genet Med. 2009 Jun;11(6):455-64. *(PubMed Abstract)*

- Kulik W, van Lenthe H, Stet FS, Houtkooper RH, Kemp H, Stone JE, Steward CG, Wanders RJ, Vaz FM. **Bloodspot assay using HPLC-tandem mass spectrometry for detection of Barth syndrome.** Clin Chem. 2008 Feb;54(2):371-8. Epub 2007 Dec 10. *(PubMed – Open Access)**▼

- Leonard JV, Morris AA. **Diagnosis and early management of inborn errors of metabolism presenting around the time of birth.** Acta Paediatr. 2006 Jan;95(1):6-14. *(PubMed Abstract)*

- Wilcken B, Wiley V, Hammond J, Carpenter K. **Screening newborns for inborn errors of metabolism by tandem mass spectrometry**. N Engl J Med 2003 June 5; 348(23):2304-2312. *(PubMed Abstract)*

- Chakrapani A, Cleary MA, Wraith JE. **Detection of inborn errors of metabolism in the newborn.** Arch Dis Child Fetal Neonatal Ed. 2001 May;84(3):F205-10. *(PubMed Abstract)*

- Cardonick EH, Kuhlman K, Ganz E, Pagotto LT. **Prenatal clinical expression of 3-methylglutaconic aciduria: Barth syndrome**. Prenat Diagn. 1997 Oct;17(10):983-8. *(PubMed Abstract)*

- Ruitenbeck W, Wendel U, Hamel BC, Trijbels JM. **Genetic counselling and prenatal diagnosis in disorders of the mitochondrial energy metabolism**. J Inherit Metab Dis. 1996;19(4):581-7. *(PubMed Abstract)*

NUTRITION

- Camp KM, Krotoski D, Parisi MA, Gwinn KA, Cohen BH, Cox CS, Enns GM, Falk MJ, Goldstein AC, Gopal-Srivastava R, Gorman GS, Hersh SP, Hirano M, Hoffman FA, Karaa A, MacLeod EL, McFarland R, Mohan C, Mulberg AE, Odenkirchen JC, Parikh S, Rutherford PJ, Suggs-Anderson SK, Tang WH, Vockley J, Wolfe LA, Yannicelli S, Yeske PE, Coates PM. **Nutritional interventions in primary mitochondrial disorders: Developing an evidence base.** Mol Genet Metab. 2016 Sep 20. pii: S1096-7192(16)30191-3. doi: 10.1016/j.ymgme.2016.09.002. [Epub ahead of print] Review. *(PubMed Abstract)*

- Bradley RM, Stark KD, Duncan RE. **Influence of tissue, diet, and enzymatic remodeling on cardiolipin fatty acyl profile.** Mol Nutr Food Res. 2016 Apr 8. doi: 10.1002/mnfr.201500966. [Epub ahead of print] *(PubMed Abstract)*

- Reynolds S**. Successful management of Barth syndrome: A systematic review highlighting the importance of a flexible and multidisciplinary approach.** J Multidiscip Health 2015 Jul 29;8:345-58. doi: 10.2147/JMDH.S54802. eCollection 2015. Review. *(PubMed – Open Access)*