



Barth Syndrome Foundation

www.barthsyndrome.org

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**Dedicated to advancing
research, raising awareness,
and pursuing a cure
for Barth syndrome**



**Barth Syndrome
Foundation**

Barth Syndrome Foundation, Inc.

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Website: www.barthsyndrome.org

Living with Barth syndrome

Barth syndrome is a life-threatening, complex, genetic disorder that primarily affects males. It is passed from mother to son. Before recent advancements were made in the diagnosis of Barth syndrome, most boys did not survive. Today, with improved diagnosis, treatment, and management, the survival rate and future of these boys is much brighter. The main symptoms can present in a variety of ways, with differing severity, at any age.

Cardiomyopathy

A weak heart muscle usually associated with enlargement of the heart. This symptom can cause a risk of fatal arrhythmias (irregular heart beat).

Neutropenia

A reduction in “neutrophils”, a type of white blood cell that is most important for fighting bacterial infections. During the reduction of neutrophils the individual is vulnerable when exposed to infection.

Muscle weakness

All muscles, including the heart, have a cellular deficiency which limits their ability to produce energy, resulting in muscle weakness and increased fatigue. In some cases this is debilitating.

Growth delay

During childhood most affected individuals are below-average in height and weight, often substantially so. Through BSF’s registry, we have observed accelerated growth to normal height during mid- to late-teenage years.

Barth Syndrome Foundation, Inc.

“BSF” is a nonprofit, voluntary community of highly motivated and committed individuals. We provide the largest source of Barth syndrome information, resources, activities and services for this disorder worldwide.

Every year, BSF awards scientific and clinical research grants in an effort to better understand Barth syndrome. Some funded research has focused on how the cells and genes are affected, while other researchers are investigating how these cellular differences affect the person who has Barth syndrome.

All of this research will help to improve knowledge and treatment of this complex disorder. BSF has stimulated a great deal of excitement and attention on this disorder, and the prospects for further advancements are promising, but there is much crucial work that remains to be done, and we need your help in doing so.

We would like to thank our families, friends, volunteers, donors, and those in the medical community who have given their support, as both human and financial resources are necessary to pursue our goals.

“Out of isolation, we have risen up to forge a path where there once was none. Our promise is to continue to pave the way for others to follow.”

~ Shelley Bowen, President

What we do

The Barth Syndrome Foundation's (BSF) mission is to guide the search for a cure, to educate and support physicians, and to foster a caring community for affected families.

Guide the search for a cure

2005 marked BSF's 4th grant cycle, inviting researchers to apply for BSF funding to research the long-term effects and impact of Barth syndrome, cellular and genetic function and effect, and treatment of Barth syndrome. Since 2002, BSF has awarded grants to highly regarded researchers in multiple countries.

Educate and support physicians

Early diagnosis can make a tremendous difference in the quality of life ~ and in many cases, it may very well mean the difference between life and death.

- Provide educational outreach programs designed to raise public awareness.
- Participate in numerous medical and advocacy conferences.
- Provide an on-line forum for all doctors involved with BSF.
- Sponsor an international conference every two years where the world's leading experts specializing in every aspect of Barth syndrome convene to further advance the science and medicine of Barth syndrome.
- Operate BSF's website, which provides up-to-date information about its research, educational and outreach activities. Please visit www.barthsyndrome.org.



"Having already lost a son to Barth syndrome, I want to be as prepared for this baby as possible. I'm so afraid that if the doctors don't have the information they need, he won't get the care he needs. We're

providing them with everything we can. I just don't want to leave anything out." ~ **Keli**

Create a caring community

We have an open line of communication with all affected individuals and families.

- Support individuals and their loved ones in understanding this disorder and its treatment.
- Provide an up-to-date, vast library of information as it relates to Barth syndrome at www.barthsyndrome.org.
- Provide an on-line forum where families and doctors have an open discussion and immediate access to the most up-to-date information.
- Sponsor an international family conference every two years, bringing together families and world expert doctors involved in Barth syndrome.
- Sponsor clinics every two years, free of charge, where affected individuals have the opportunity to be seen by doctors from around the world who have followed the most cases of Barth syndrome.

There is power in knowledge and strength in numbers.

Your continued support will assure BSF the privilege of providing our services, at no cost, to affected families.



Today, thanks to the commitment and support of individuals, grass-roots fundraisers, corporations, and foundations, BSF is the leading independent organization dedicated to advancing Barth syndrome treatment and research throughout the world.

What is our vision?

A world in which no one will suffer or perish from Barth syndrome.

To learn more about the Barth Syndrome Foundation, Inc., please visit us at:

www.barthsyndrome.org

Barth Syndrome Foundation, Inc. Donation Form

Please help us achieve our goals by becoming a member and making a monetary contribution below, or volunteer to lend your talents to The Barth Syndrome Foundation, Inc.

- | | | |
|--------------------------|---|----------|
| <input type="checkbox"/> | Basic Member | \$50 |
| <input type="checkbox"/> | Family Member | \$100 |
| <input type="checkbox"/> | Friend of BSF | \$250 |
| <input type="checkbox"/> | Benefactor | \$500 |
| <input type="checkbox"/> | Angel | \$1,000 |
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| <input type="checkbox"/> | Company Matching Form (Please Enclose) | |
| <input type="checkbox"/> | I would prefer that my contribution NOT be acknowledged publicly | |

Please make your tax deductible contributions payable to:

The Barth Syndrome Foundation, Inc.

and mail to the address below:

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Credit Card #

Exp. date

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All gifts are tax-deductible to the fullest extent permitted by the law. The official registration and financial information of The Barth Syndrome Foundation, Inc. may be obtained from the Pennsylvania Department of State by calling toll-free, within Pennsylvania, 1-800-732-0999. Registration does not imply endorsement. The Barth Syndrome Foundation, Inc.'s Florida registration number is SC-12347. 100% of your contribution will be received by BSF, Inc.

The Barth Syndrome Foundation, Inc.
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Saving lives through education, advances in treatment and pursuit of a cure for Barth syndrome