



PRACTICAL TIPS FOR INDIVIDUALS WITH BARTH SYNDROME

MEDICAL

- **Keep thorough medical records.**
 - Request a copy of all reports and test results from each attending physician. (Records should be copied at NO cost to the patient). Some physicians have been reported to be reluctant to give them to patients and parents because these reports are not written for laymen. Most often the reason given is because the physician wants to avoid a potential misunderstanding about the contents in the reports. ⇨ Visit the medical records department at the time of the visit and sign a release to receive a copy of your medical records. This will save you time later. Often times a technician will provide you with a copy of a report at the time the study is conducted such an example would be for an ECG.
 - When requesting the medical records it may be helpful to explain that these medical records are beneficial in coordinating the care of the individual with Barth syndrome. Barth syndrome is rare and most physicians will not know much about it. You are very likely to deal with several different specialists, and it is crucial that you as the primary caregiver have all the information about your child or if you are the individual with Barth syndrome about yourself to share with them. In fact, you may be the only person who will end up with a complete file, across specialties and over time. ⇨ Keep everything; it is difficult to predict what may be important later. Three-ring binders can work well in organizing the medical records. It may be helpful to organize records in binder: by date or by test results or by specialty.
- **Share information among doctors.**
 - Since you are likely to seek the help of several different specialists (who may or may not all work at the same institution), some of the coordinating role may fall to you. Ask that each of physicians attending to the care of the individual with Barth syndrome be placed on the distribution list for reports from all physicians (sometimes this works, but often it doesn't). It is helpful to have your 3-ring binder when seeing attending physicians. Make certain the physicians have copies of pertinent documents. If they do not you may either: have multiple copies to share or ask one of the office staff to make a copy for the medical record. ⇨ Never give away your only copy or original copy of the medical document. When appropriate, particularly about highly technical or complicated issues, request that the doctors communicate directly with each other so that there is no possibility of misunderstanding or misinformation.
- **Prepare on-call physicians.**
 - One cannot predict when an emergency situation may present with Barth syndrome. Inquire how physicians within the practice are informed about Barth syndrome. It is best to be prepared for this event rather than seeing a physician who is an ill-informed, on-call sub-specialist during an emergency situation.
- **Ask questions; be knowledgeable.**
 - To the extent possible, try to understand not only about Barth syndrome itself but also about the medical status of the individual who has Barth syndrome. Do some of your own research, but be very careful to pay close attention to the sources of your information. As with all things, do not believe everything you read just because it has been printed somewhere (particularly on the Internet). ⇨ Go to reliable sources and understand where your information is coming from. At the same time, **remember that each patient is unique** and that even information specifically pertaining to Barth syndrome may or may not be relevant to every individual with Barth syndrome. It may be helpful for you to familiarize yourself with a glossary of commonly used terms. Such a glossary is available upon request from The Barth Syndrome Foundation.

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2nd Edition

Practical Tips for Individuals with Barth syndrome

www.barthsyndrome.org

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- **Enlist your physician's help, but also be somewhat self-reliant.**
 - It is important to take charge of the health care for the individual with Barth syndrome. Listen carefully and take good advice. But if something doesn't seem right, trust your instincts and follow up or seek other opinions. It is crucial for the physicians to know when a situation is beyond their capabilities or knowledge and when to call in a specialist or seek some extra help. Even with sub-specialists, if you are not comfortable with the care being offered or with something you have been told, you should not be intimidated. Trust your instincts and pursue more information on your own. ↔ Always remember that you are dealing with a rare medical condition with which most physicians are not very familiar and about which not everything is yet known.
- **Understand that there sometimes is a fine line in knowing when to follow what a physician says and when to dig in your heels and demand more attention, information, testing or help.**
 - It does NOT pay to be a pest constantly or to take up your physician's time unnecessarily, but there may well be instances in which you need to speak up and be the "squeaky wheel." You have now been placed in a position of being the patient advocate regardless of whether the patient is a loved one or yourself.
- **Be prepared for appointments.**
 - Take information that could be helpful with you, and write a list of your questions ahead of time so that you don't forget to ask anything. You may also want to consider tape recording the doctor's visit, to listen to at a later time. Some physicians may feel a bit uncomfortable with this request so just inform him/her that you are trying to better understand the disorder and process all of the information to better aid in being a partner in care. It may also be beneficial to ask a friend to accompany you on the visit to take notes. When you have an appointment with a **new doctor**, consider sending him or her some background information about Barth syndrome in advance of the appointment. This might include a one or all of these documents:
 - [Latest summary of Barth syndrome](#) from the National Institutes of Health
 - [Summary on Barth syndrome](#) - Dr. Richard I. Kelley, MD, PhD, Kennedy Krieger Institute
- **Try to avoid waiting in the pediatrician's waiting room with other sick patients.**
 - Ask to wait outside or in a room by yourselves. Sometimes there is even a separate entrance that can be used. If your child is at an age where he wants to play with toys, bring your own so that he will not play with those handled by other children who are ill. ↔ Keep a mask, hand sanitizer and antibacterial wipes with you at all times and use them.
- **Maintain a complete list of physicians who have been consulted in the care of the individual with Barth syndrome either on an ongoing or limited basis**
 - Make certain to have addresses, phone numbers and/or e-mail addresses so you may contact them should the need present.
- **Maintain a complete record of all clinical and laboratory tests**
 - One should always assume all information is critical. Even tests that may seemingly glean no apparent findings may in reality be key information later. Save everything! When saving copies of tests make certain the parameters of normal values are in the body of the document. Every testing facility has minute differences of normal values. Ask the facility to provide that information to you. This will come in handy for future reference. You may think that you will remember these results but 10 years from now you will be thankful for the time invested to obtain the additional information.

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- **Maintain detailed records of all medications**
 - When maintaining a list of medications it is important to maintain a detailed log of prescribed medications. A useful tool to assist you in keeping these records would be the BSF medication form. ([Care Plan for School Age Children with Barth syndrome, pg. 2](#))
- **You might find it helpful to purchase a medical dictionary to aid in understanding complex medical terms used during appointments with physicians.**
 - Make note of important medical terminology that you do not understand. Never be afraid to ask for clarification if you do not understand.
 - Dorland's Medical Dictionary and Tabor's Medical Dictionary are examples of popular medical dictionaries. You can review both of these at most public libraries. Look at the format of all of the medical dictionaries available and make an informed decision about the dictionary you would most like to add to your reference library at home. These are not inexpensive dictionaries. They are about 10 times more than the price of the average word dictionaries found in bookstores. These reference books are well worth your investment.

↔In summary, remember that you are a partner in care regardless if the patient is you or a loved one. Keep the lines of communication open with your professional advisor (the physician) and together you will work out a caring partnership of mutual trust and respect. Like all relationships sometimes the fit with the physician is not always a good one. The physicians are key advisors. You must be able to depend on them and trust their advice. If you don't you may wish to seek a different physician.

DAILY LIFE

- **Because most individuals with Barth syndrome are neutropenic at least sometimes, take care to protect them from unnecessary germs and be diligent about their personal hygiene and daily routines concerning germs:**
 - Avoid people you know are ill and contagious
 - Wash hands frequently with soap and water or with an antimicrobial lotion that can easily be kept on hand at all times
 - Do not allow sharing of drinks or food
 - Wash fruits and vegetables thoroughly
 - Cook meat appropriately
 - Brush teeth regularly and thoroughly and have teeth cleaned 2 – 4 times per year
 - Replace toothbrush every 6-8 weeks
 - Keep fingernails short and clean
 - **NEVER take a rectal temperature**
 - Use antibacterial ointment on cuts and scrapes (once they have been cleaned completely)
- **When dealing with young children, who have Barth syndrome, make certain friends, family and baby-sitters understand the special risks of bacterial infection and are careful around your medically compromised child.**
 - Remember that it is always best to expect the unexpected. An emergency situation is more stressful than one could predict. It is most important to be prepared. It is a good idea to have information readily accessible for first responders. This information can be placed on a form in a conspicuous place. ↔ [Child Care Provider Emergency Sheet](#)

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- **Frequent meals may be required due to the possible risk of hypoglycemia symptoms.**
 - Individuals with Barth syndrome don't always have a robust appetite and have been reported to have symptoms of hypoglycemia. The risk of this symptom occurring can be lessened by encouraging frequent drinks of a nutritional beverage or encouraging the child to snack on foods between meals. ↪ 1 teaspoon per 5 kilo of weight of raw cornstarch right before bedtime has been reported to eliminate the hypoglycemic risks associated with overnight fasting with individuals who have Barth syndrome. Some parents have reported mixing the cornstarch with chocolate milk to be the most palatable for ingestion. It is best not to mix the cornstarch with a fruit juice because it will break down the cornstarch.
- **Need for frequent rest periods due to fatigue.**
 - Fatigue has been a symptom frequently reported by caregivers of those who have Barth syndrome as well as the affected individuals. It is important to know when to push and when to stop. ↪ It is not wise to push the individual to exhaustion. By allowing frequent rest periods the individual will minimize the risk of complete exhaustion. Some parents have reported a simple "time out" zone helps. Not only is there a need to educate those around you about Barth syndrome but it is also equally important to educate those who have the disorder. Give your child permission to take a rest. Encourage him to do so by making a special place for him to rest.
- **Avoid prolonged exposure to excessive heat.**
 - Individuals with Barth syndrome have been reported to decompensate during periods of prolonged exposure to heat. What is easily tolerated by a healthy individual is not tolerated well at all for someone with Barth syndrome. Avoid outings during the heat of the day on excessively warm days. Always have a beverage readily accessible to re-hydrate the individual. Plan family outings during cooler times of the year or when that is not possible select times of the day when it is cooler (morning or evening). ↪ Fatigue may be hastened during times of excessive heat so gauge your outing accordingly.
- **When traveling always be prepared for an emergency.**
 - Keep physicians contact information with you at all times in case of an emergency.
 - Take the individual's "core" three-ring binder with you so that you will have relevant information available quickly for a physician who has never been consulted in the care of you or your loved one has likely never been involved with a case of Barth syndrome.
 - In advance, do a little research on hospitals in the area in which you will be traveling. Inform the attending physicians of your travel plans and request names of physicians in the area where you will be visiting. BSF has created a **Be On the Look Out (BOLO)** document to aid families when traveling. This document was designed to be sent to area physicians and hospitals where the individual may be seen during a vacation or business travel.
 - Always take more medication than you know you need in the event of spills or unexpected travel delays. ↪ Never check medications on an airplane, train or bus; keep them with you so there is no possibility of the medications getting lost.
- **Contact with other individuals who have Barth syndrome**
 - The rarity of Barth syndrome translates to mean there are not many people who really know about or understand the condition. So, if the individual with Barth syndrome seems lonely or complains that "no one understands", sometimes putting him in contact with another individual with Barth syndrome (by phone, letter, e-mail or in person) can be very helpful. ↪ Aim to attend The Barth Syndrome Foundation conferences. Families find a deep connection with one another during these times together. The all too familiar sense of isolation is eliminated during these rare gatherings through the opportunities of exchange of information with someone else who simply "understands".

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SCHOOL

- Provide The Barth Syndrome Foundation's "*Strategies of Educational Advocacy for a child Living with Barth Syndrome – An Educator's Handbook*" to your child's teacher and school administrators.
- Enlist the assistance of your child's teachers and school administrators when sorting out educational issues in search of solutions for your child. Relevant issues will, of course, vary along with the child's age and specific situation, but they frequently may fall somewhere within the following three broad categories.
 - **Academic issues**

These may result from illness and/ or fatigue. Solutions may include such things as arranging for more time to finish tests because the physical activity of writing or calculating can be tiring and painstaking, or making sure that your child learns how to use and has access to a computer to facilitate the physical act of writing. Ask that your child have a scribe assigned to do his writing or if assistive devices are available for him to use.
 - **Social issues**

Resulting from being different ("sick," "small," "weak," "can't run as fast," "have to go to bed so early because needs more sleep than others," "has to take so much medicine," etc...). These might involve such requests as ensuring that your child has extra supervision on the playground; requesting that your child have a special buddy to help your son become more social; or depending on the age of his classmates. As children get older, sometimes it is best (assuming your son agrees) that he speak to his classmates directly about his medical situation. When representing your son's condition beware of using the phrase "compromised immune system" because there have been some incidences of people falsely leaping to the conclusion that a child has AIDS. Remember to respect your child's needs when speaking to his classmates. Some children are very private about their condition and this important to respect. Research has proven that children who suffer with chronic illness are more apt to encounter bullying in school than their healthy peers.¹ Encourage your child's teacher to be on the look out for such intolerable behavior.
 - **Practical issues**

These are of many varieties. These might include such things as requesting an extra set of textbooks to be kept at home so that book bags or back packs are not too heavy to manage, dealing with special dietary needs, requesting that your child's classes are held close to each other and also preferably near the nurse's office if walking distances under time pressure is a problem, modifying your child's physical education program, asking that your child be allowed extra time to eat his lunch, asking that those in charge at lunch time specifically encourage but not force your child to eat his lunch, or requesting a pair of Benbow scissors to facilitate your child's ability to cut. In the US many families have found an IEP (Individual Educational Plan) or a 504 plan to be invaluable tools to ensure their child's special educational requirements are being met. These tools are specially designed educational approaches to the child's education. They are in writing and provide ready access to important clues to the best approach to the child's education.

¹ Clin Pediatr (Phila). 2004 Jun;43(5):467-71

Peer victimization and psychosocial adjustment in children with type 1 diabete; Storch EA, Lewin A, Silverstein JH, Heidgerken AD, Strawser MS, Baumeister A, Geffken GR; Department of Psychiatry, University of Florida, Gainesville, FL 32610, USA.

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- **Schedule frequent team meetings**
 - Schedule a meeting of all of your child’s teachers, the school nurse, a guidance counselor and a school administrator early in the school year in order to describe your child’s condition and school situation. Sometimes it works well to do this before school even starts, and sometimes it is preferable to schedule it after your child’s teachers have had a chance to be with him for a few days so that it is a “child” you are discussing not a “case”. Set a schedule throughout the year for these meetings and encourage your son’s teachers to schedule ad hoc meetings to aid your son in his educational endeavors.
- **Make sure that the school nurse is fully armed with the information he or she needs to help keep your child safe and healthy.**
 - This probably includes more information than the standard forms. The nurse should have an understanding of your child’s specific medical condition (and this may change over the course of a year, so make sure to keep him or her up-to-date), the contact numbers of all the child’s main physicians, a list of signs and symptoms to look out for in your child, and anything else you think would be important. Some families have requested that their son’s physician or cardiologist write an order for an AED (Automatic External Defibrillator) to be placed in their son’s school, due to the possible risk of sudden arrhythmia. Some families also insist ↔every teacher who teaches the child with Barth syndrome must know CPR. BSF has created a basic [care plan](#) for children of school age with Barth syndrome.
- **Keep in close contact with the school nurse regarding what viruses and bacterial illnesses “going around” the school.**
 - If your child is neutropenic, this is particularly important information. Keeping a monthly calendar of when your child has been sick or absent may be important to record, this may determine a strong consideration for homebound schooling. Determination of a proven cycle will aid the parent and educator by anticipating “down time” the child will encounter.

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