

Do You Know About Barth Syndrome?

Eli (age 2)



(Photo courtesy of Eli's family 2015)

What's Inside

- Description of Barth syndrome
- Important clinical problems
- How to diagnose
- Inheritance
- Selected highlights of clinical knowledge
- Resources for physicians and families

Our lives depend on it!



Barth Syndrome
Foundation

www.barthsyndrome.org

Saving lives through education, advances in treatment, and finding a cure for Barth syndrome.

What is Barth Syndrome?

Barth syndrome (BTHS; OMIM #302060) is a rare, life-threatening genetic disorder primarily affecting males around the world. It is caused by a mutation in the *tafazzin* gene (*TAZ*, also called G4.5), resulting in an inborn error of phospholipid metabolism.

Though not always present, cardinal characteristics of this multi-system disorder often include combinations and varying degrees of:

- **Cardiomyopathy** (usually dilated with variable myocardial hypertrophy, sometimes with left ventricular noncompaction and/or endocardial fibroelastosis)
- **Neutropenia** (can be chronic, intermittent, cyclic, or not present)
- **Low muscle mass and muscle weakness**
- **Growth delay** (short stature in the early years, followed by accelerated growth in mid- to late puberty)
- **Exercise intolerance** due to early fatigue
- **Feeding problems** (e.g., difficulty sucking, swallowing, or chewing; aversion to some food textures; selective or picky eating; frequent vomiting)
- **Cardiolipin abnormalities**
- **3-methylglutaconic aciduria** (variable but typically a 5- to 20-fold increase)

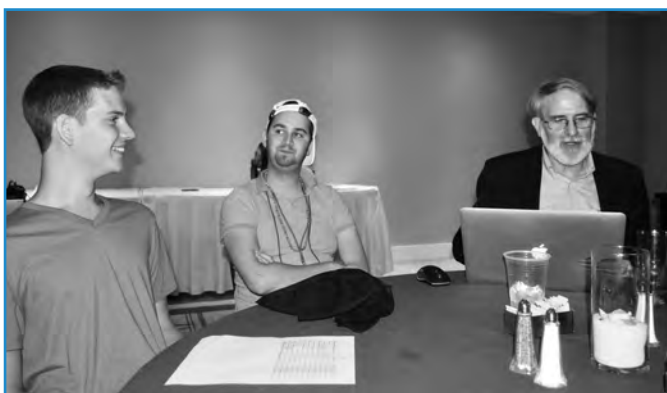
ICD-10 Code for Barth Syndrome

Barth syndrome now has a specific ICD code:

E78.71

Additional Clinical Problems May or May Not Include (in varying severity):

- Congestive heart failure
- Life-threatening bacterial infection
- Gross motor delay
- Risk of fatal arrhythmia
- Extreme fatigue
- Diarrhea and/or constipation
- Recurrent mouth ulcers
- Risk of thrombosis
- Hypoglycemia, including fasting hypoglycemia (most often in the newborn period)
- Chronic headache, abdominal pain, and/or body aches (especially during puberty)
- Osteoporosis
- Mild learning disabilities



Travis (age 21) and Oliver (age 26) consult with Dr. Richard Kelley at BSF's 2014 Conference (Photo courtesy of BSF ²⁰¹⁴)

“As a physician, I have learned more about Barth syndrome from the families’ posts on the Listserv than from any other single source — by far.” ~ Richard Kelley, MD, PhD, Genetics and Genomics, Boston Children’s Hospital, Boston, MA; Scientific & Medical Advisory Board, Barth Syndrome Foundation

A Multi-System Disorder

It is critical always to remember that Barth syndrome (BTHS) is a complex inborn error of metabolism. Because the disorder affects many systems of the body, treating a patient with BTHS often requires involvement of experts from a wide range of medical specialties.

Phases of Barth Syndrome

These general phases are often, but not always, seen in BTHS:

- Children with BTHS often are seriously ill before the age of five years.
- The ages from five to eleven years can be a “honeymoon phase,” when symptoms can appear to be few.
- This does not mean that the syndrome has been “outgrown,” as adolescence often begins another difficult period.

Regardless of the phase of Barth syndrome, the following serious risks ALWAYS exist:

Risks of Cardiac Dysfunction

- The natural history of BTHS cardiac disease has been described as “undulating.” Both the character and severity of heart dysfunction can change significantly. The cardiomyopathy can evolve from dilated to hypertrophic or vice versa, and may or may not involve left ventricular noncompaction (LVNC) and sometimes hypertrabeculation. Furthermore, sometimes a patient sick enough to be awaiting a heart transplant can improve so dramatically as to be taken off the list, especially if the underlying metabolic abnormalities have been treated and improved. Unfortunately, the reverse also can happen, and heart function can deteriorate significantly, suddenly and unexpectedly, even during otherwise simple viral or bacterial infections. Vigilant cardiac monitoring is essential.
- Life-threatening arrhythmias can occur, even when heart function is in the normal range.

Risks of Infection

- When well, a BTHS individual can have an absolute neutrophil count (ANC) approaching zero, but this can rise to normal or above during an acute infection. Thus, there are times when a normal ANC can be a sign of a serious infection.
- Many with BTHS have a normal body temperature that is substantially below 98.6°F (37°C), so even a mild fever may signify a problem.
- Taking a rectal temperature is contra-indicated due to risk of serious infection resulting from the possible introduction of fecal organisms into the bloodstream.

Risks of Nutritional and Metabolic Issues

- Because muscle is an important nutritional reserve during fasting, the intrinsically reduced muscle mass of BTHS individuals significantly limits their ability to fast. Even overnight fasting drains muscle reserves, causing relative hypoglycemia and, over time, further muscle atrophy. Eating cornstarch (*e.g.*, added to yogurt) or Extend Bars™ before bedtime can alleviate these problems.
- BTHS individuals often tolerate illnesses poorly, especially those that include diarrhea or vomiting, given their reduced muscle mass resulting in diminished body stores of electrolytes and protein. Therefore, fluid and electrolyte (particularly potassium and phosphate) balance must be monitored closely and frequently during illnesses and caution exercised to prevent hyperkalemia when giving potassium-containing IV fluids. Underlying cardiac issues also must be considered in all of this.
- Caloric requirements in someone with BTHS often are reduced because of substantially underdeveloped muscle mass. Trying to achieve standard caloric intake for age and weight often leads to vomiting and diarrhea.
- Because rare but serious hypoglycemic crises have occurred in BTHS, any symptoms of low blood sugar (weakness, pallor or sweating) must be taken seriously.
- The metabolic strategies used by BTHS cells to maintain normal energy production can cause sufficiently severe depletion of certain amino acids, most notably arginine, that cardiac and skeletal muscle protein synthesis is impaired. As a result, the use of extra dietary protein and supplements of arginine and other amino acids may be considered to raise amino acid levels to their mid-normal ranges and thereby reverse serious deterioration in cardiac function caused by cardiac muscle wasting.
- Anesthesia for BTHS patients requires special consideration because of increased risks from the cardiac, muscular and metabolic issues involved in the disorder. Dilated cardiomyopathy is frequently present, the risk of ventricular arrhythmias is increased, and lactic acid may accumulate rapidly. The much reduced muscle mass of BTHS can lead to rapid electrolyte shifts and predispose Barth syndrome patients to hypoglycemia. Thus, care should be taken to minimize pre- and post-operative fasting and to avoid use of lactated intravenous fluids. (A BTHS-specific anesthesia protocol can be found at www.barthsyndrome.org under the Living with Barth Syndrome tab entitled FACT sheets.)

A Multi-Disciplinary Approach

Because Barth syndrome (BTHS) is a complex multi-system disorder, an individual who has Barth syndrome may be diagnosed by and receive care from sub-specialists involved in:

- **Cardiologist and Electrophysiologist:** Cardiomyopathy, arrhythmias, thrombosis, heart failure, ventricular noncompaction
- **Neurologist:** Low muscle tone, chronic pain, diminished capacity for exercise, muscle fatigue
- **Hematologist:** Neutropenia, frequent infections, mouth ulcers, any clotting issues
- **Metabolic specialist:** Abnormal organic acids, poor weight gain, nutrition
- **Endocrinologist:** Abnormal growth pattern, hypoglycemia, osteoporosis
- **Geneticist:** Diagnosis and family planning
- **Gastroenterologist:** Frequent vomiting, diarrhea, constipation, feeding tubes, GI motility
- **Psychologist:** Depression, anxiety, “fitting in”
- **Physical, Occupational and/or Speech Therapist:** Difficulties with gross and fine motor skills and/or speech
- **Dietician/Nutritionist:** Feeding challenges, dietary intake

Allied health professionals in the areas of occupational therapy, physical therapy, nutrition and genetic counseling are also integral to the care of those who have Barth syndrome.

How to Diagnose

Barth syndrome (BTHS) is a complicated disorder and can be difficult to recognize because all manifestations may not be simultaneously present or apparent.

The diagnosis of BTHS should be considered for a child or adult presenting with any one of its eight cardinal characteristics and in cases with family histories of multiple male deaths or fetal loss, because a diagnosis of BTHS otherwise can easily be missed.

Diagnostic Testing

- **DNA sequence analysis** (genetic testing of the *tafazzin* gene (*TAZ*, also called G4.5))
- **Cardiolipin analysis** of certain cell and tissues

Lack of family history does not exclude the diagnosis of BTHS, as there is a relatively high frequency of new mutations.

For more details about these tests, please visit GENETests™ and search for “Barth syndrome”: www.genetests.org.



Devin (age 11) and Henry (age 7)

Inheritance

Barth syndrome (BTHS) is an X-linked genetic disorder, usually transmitted from mother to son (although there is a relatively high incidence of new mutations in BTHS and are several case reports of female BTHS patients). A mother who is a carrier of a BTHS mutation (the gene is named *tafazzin* — also called *TAZ* or *G4.5*) shows no signs or symptoms of the disorder herself, probably due to skewed X-chromosome inactivation.

There is a 50% chance that a boy born to a female carrier will have BTHS, whereas girls born to a carrier have a 50% risk of being carriers themselves. All daughters of a male with BTHS will be carriers, however no sons will be affected. Because there are many non-carrier mothers, all mothers of BTHS children should be tested in order to determine the genetic risk in each family.

Any male child related through the female carrier line to a BTHS individual should be tested for the disorder, as there can be great variation in phenotype, even among affected siblings. Also note that several female BTHS cases have been confirmed and virtually asymptomatic adult male relatives of BTHS probands have been identified through extended family genetic testing.



I'm quite certain Barth syndrome is under-diagnosed. If you have never heard of the disease, you are not going to look, you are not going to find. ~ Jeffrey Towbin, MD, Co-Director, Heart Institute, Le Bonheur Children's Hospital; Chief of Cardiology, St. Jude Children's Research Hospital; Chief of Pediatric Cardiology, University of Tennessee Health Science Center, Memphis, TN

Selected Highlights of Clinical Knowledge

* Publications that acknowledge financial support contributed by Barth Syndrome Foundation (BSF) and/or BSF affiliates.

Δ Publications that acknowledge biological samples (and/or information) from Barth syndrome families, the Barth Syndrome Registry and Repository (BRR), and/or BSF affiliates.

For the most up-to-date information, including a full Barth syndrome (BTHS) bibliography and links to PubMed abstracts, please visit www.barthsyndrome.org under the Science and Medicine tab.

2016

Variable cardiac features but reduced functional exercise capacity (decreased walk times and muscle strength) in 42 BTHS individuals.

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]Δ

2015

Feeding problems found in at least 50% to 70% of BTHS individuals and often present before six months of age.

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)Δ

Pain observed to be prevalent in individuals with BTHS.

Taylor D, Brady JE, Li G, Sonty N, Saroyan JM. [Characterization of pain in patients with Barth syndrome](#). Children's Health Care, February 2015.Δ

2014

Comprehensive review article about BTHS.

Ferreira C, Thompson WR, Vernon H. [Barth syndrome](#). GeneReviews. October 9, 2014. (Open Access)Δ

Selected Highlights of Clinical Knowledge

2014 (cont'd)

Moderate CL deficiency associated with milder BTHS phenotype.

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] ([Open Access](#))*[Δ](#)

2013

Comprehensive review article about BTHS.

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 Journal of Rare Diseases 2013, 8:23. ([Open Access](#))*[Δ](#)

Summary of clinically important information about BTHS.

Jefferies JL. [Barth syndrome.](#) Am J Med Genet C Semin Med Genet. 2013 Aug;163(3):198-205. Epub 2013 Jul 10. ([Open Access](#))

Report from French historical experiences with BTHS individuals and discussion of how good medical practices contributed to survival.

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. ([Open Access](#))*[Δ](#)



“The cruelest irony about Barth syndrome is how deceptively healthy those who have it may appear. A casual observer would never appreciate them to have such a devastating illness.”

~ Peter Barth, MD, PhD, Pediatric Neurology (retired), Emma Children's Hospital/Academic Medical Center, Amsterdam, The Netherlands

Selected Highlights of Clinical Knowledge

2012

Review article detailing longitudinal data collected, including growth curves, from Barth Syndrome Registry and Repository.

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. Epub 2012 Oct 8. (*Open Access*)*[Δ](#)

2011

Severe exercise intolerance in BTHS due to cardiac and skeletal muscle impairments consistent with cardiac and skeletal mitochondrial myopathy.

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. (*Open Access*)*[Δ](#)

2010

First conclusive demonstration that BTHS can cause male fetal loss and stillbirth in multiple families.

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. (*Open Access*)*[Δ](#)

2009

Common childhood BTHS facial features include tall and broad forehead, round face, prominent chin, full cheeks, large ears and deep-set eyes. Gynoid stature and fat distribution often develop in late puberty.

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

Selected Highlights of Clinical Knowledge

2007

Successful cardiac transplantation in BTHS, also emphasizing the importance of accurate diagnosis for post-transplant BTHS patient care.

Mangat J, Lunnon-Wood T, Rees P, Elliott M, Burch M.
Successful cardiac transplantation in Barth syndrome:
Single-center experience of four patients. *Pediatr Transplant*
2007 May;11(3):327-31.

Our Mission is...

*Saving lives through education, advances in treatment,
and finding a cure for Barth syndrome.*



Greyson (age 8)

(Photo courtesy of Greyson's family 2016)

“BSF is truly an amazing organization. They are a lifeline to families and individuals affected by Barth syndrome. The wealth of knowledge and information, compassion and understanding, friendship and hope, that comes from being a part of an active, professional and caring foundation is beyond words.” ~ Ned, Parent of Affected Individual



Milosh & Ned

“The Family and Medical Conference is superb! Amazingly pulled off by a small, but wonderful team running BSF and done so well. I cannot say enough good things about BSF! As a parent of a son with Barth syndrome, we so value and appreciate the Foundation for so many reasons.” ~ Megan, Parent of Affected Individual



Megan & Henry

(Photos courtesy of Amanda Clark 2014)

Barth Syndrome Foundation

Saving lives through education, advances in treatment, and finding a cure for Barth syndrome.

The Barth Syndrome Foundation (BSF) and its affiliates are a group of international non-profit organizations that provide information, resources and services for healthcare professionals, families, and research scientists worldwide. Advising the group is a world-class Scientific and Medical Advisory Board (SMAB), comprised of clinicians and scientists who are leading experts in Barth syndrome (BTHS).

BSF's [website](http://www.barthsyndrome.org) (www.barthsyndrome.org) contains the most up-to-date educational materials and research findings, including a comprehensive on-line library which serves both the medical community and affected families.

BSF's **International Barth Syndrome Conference**, held every two years, is really two simultaneous meetings. One meeting brings together doctors and scientists involved in the many aspects of the disorder to discuss the latest underlying scientific developments and clinical insights; it is a unique experience that encourages collaboration and accelerates advances in understanding and treatment. The other is a family meeting in which the latest information is discussed with families. Free consultation sessions are also held, enabling families to meet with medical experts from around the world. In addition, opportunities to participate in research studies and provide important clinical data and biological samples to the Barth Syndrome Registry and Repository are offered to Barth syndrome individuals.

The **Sci/Med Listserv** is an ongoing forum in which members of our international Scientific and Medical Advisory Board, clinicians and researchers collaborate, ask questions and exchange the latest information.

“What I think sets the BSF apart is the biennial International Scientific, Medical, and Family Conference, which brings together not only scientists, doctors, and other healthcare professionals, but families and patients too. The conference then takes on a personal quality. The science and medicine of the disease we’re discussing are intertwined with the personal side, at this truly inspiring conference.” ~ Colin Steward, PhD, FRCP, FRCPC, Pediatric Hematology, Bristol Royal Hospital for Children, Bristol, England

The **Family Listserv** is a forum in which families and selected healthcare providers engage in open discussions on the many aspects of this disorder and its treatment. It is an immediate educational resource for families.

BSF and its affiliates sponsor an annual competitive **Research Grant Program** (www.barthsyndrome.org/science--medicine/research-grant-program) to facilitate advances in Barth syndrome understanding and to encourage the discovery of new treatments. Grant applications are evaluated by BSF's international Scientific and Medical Advisory Board, with input from expert outside reviewers. Since 2002, we have awarded 95 separate grants totaling US \$4 million to 56 investigators around the world.

The **Barth Syndrome Registry & Repository** (www.barthsyndrome.org/science--medicine/-barth-syndrome-registry--repository--) was created by the Barth Syndrome Foundation to provide additional information about this rare disorder for families, physicians, and researchers. By collecting information directly from the Barth syndrome individuals or their families, we provide a resource that can help understand this disease. The data compiled in this registry will help to facilitate clinical trials and other research studies about Barth syndrome. In addition, we have select samples available. Please contact Dr. Matthew Toth, BSF Science Director, for more details. The Barth Syndrome Registry & Repository will coordinate with other rare disease registries to compare common elements and encourage research efforts to find treatments for many rare diseases including Barth syndrome.

The **Human Tafazzin Gene Mutation & Variation Database** (www.barthsyndrome.org/science--medicine/human-taz-gene-variants-database), a central, up-to-date database listing all known mutations and variations in the human *tafazzin* (*TAZ* or *G4.5*) gene was established by and is maintained by BSF. This is a very valuable resource which can be easily accessed through our main website. We strongly encourage anyone who knows of a new case (even if it involves a mutation or variation that is already listed) to contact the list master for inclusion in this database.

“A central repository for clinical data will provide a valuable resource for researchers. ... Only with a critical number of patients is it possible to know what is common and what is not, what is expected and what is not, and what works and what does not.” ~ *Gerald Cox, MD, PhD, Clinical Genetics, Boston Children’s Hospital, Boston, MA; Clinical Research, Genzyme Corporation, Cambridge, MA*

Please Join Us



Ty (age 2)



Milosh (age 6)



Mario (age 12)



Ben (age 19)



Darryl (age 35)



Bob (age 55)

- Join BSF at no cost.
- Be kept up-to-date on the latest educational materials and research findings.
- Gain access to and participate in informative Listservs to collaborate and share information.
- Receive an advance invitation to our multi-track International Scientific, Medical and Family Conference held every two years. Please visit BSF's website for additional information.
- Participate in the Barth Syndrome Registry & Repository to further research. For a direct link, please visit www.barthsyndromeregistry.org.
- Receive our newsletter which delivers relevant and timely research, medical, and organizational information.

Please contact us for more information.

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"BSF has changed our lives. When my 15 year old son was diagnosed with Barth syndrome, we were overwhelmed and lost. Through finding the Barth Syndrome Foundation, we were able to find specialists and information as to how to treat this extremely rare illness." ~ Michelle, Mother of Affected Individual, Ohio



Barth Syndrome Foundation

www.barthsyndrome.org

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Reviewed by SMAB Clinical Members
Healthcare Professional Brochure ~ May 2016
www.barthsyndrome.org