

Do You Know About Barth Syndrome?



Photos courtesy of Amanda Clark ~ 2010



What's Inside

- Description of Barth syndrome (BTHS)
- Important clinical problems
- How to diagnose
- Inheritance
- Highlights of current clinical knowledge
- Resources for physicians and families

Our lives
depend on it!



Barth Syndrome
Foundation

www.barthsyndrome.org

What is Barth Syndrome?

Barth syndrome (BTHS; OMIM #302060) is a rare, serious, genetic disorder primarily affecting males across different ethnicities. It is caused by a mutation in the *tafazzin* gene (*TAZ*, also called *G4.5*), resulting in a complex inborn error of 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**
(Chronic, cyclic, or intermittent)
- **Underdeveloped skeletal musculature and muscle weakness**
- **Growth delay**
(Growth pattern similar to but often more severe than constitutional growth delay)
- **Exercise intolerance**
- **Cardiolipin abnormalities**
- **3-methylglutaconic aciduria**
(Typically a 5- to 20-fold increase)



Photo courtesy of Amanda Clark ~ 2010

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

Photo courtesy of Amanda Clark ~ 2010



Important Clinical Problems May Include (in varying severity):

- Congestive heart failure
- Life-threatening bacterial infection
- Gross motor delay
- Risk of fatal arrhythmia
- Short stature through pre-teen years, followed by accelerated growth in mid- to late puberty
- Extreme fatigue
- Diarrhea and/or constipation
- Recurrent mouth ulcers
- Feeding problems (*e.g.*, difficulty sucking, swallowing, or chewing; aversion to some food textures; selective or picky eating)
- Risk of thrombosis
- Diminished capacity for exercise
- Hypoglycemia, including fasting hypoglycemia (especially in the newborn period)
- Chronic headache, abdominal pain, and/or body aches (especially during puberty)
- Osteoporosis
- Some mild learning disabilities

A Multi-System Disorder

It is critical always to remember that Barth syndrome (BTHS) is a complex inborn error of metabolism. It affects many systems of the body, so 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 typically improve and patients tend to be crisis-free.
- This does not mean that the syndrome has been “outgrown.” Adolescence often begins another difficult period.

Despite these general phases, 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 hypertrophic to dilated or vice versa, and may or may not involve left ventricular noncompaction (LVNC). Furthermore, sometimes a patient sick enough to be awaiting a heart transplant can improve dramatically enough 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 contraindicated due to risk of serious infection.

Risks of Nutritional and Metabolic Issues

- 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 tend to tolerate illnesses poorly, especially those that include diarrhea or vomiting, given that there is reduced muscle mass and, as a result, 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.
- Rare but serious hypoglycemic crises have occurred in BTHS, so any symptoms of low blood sugar (weakness, pallor or sweating) must be taken seriously.
- There is increasing evidence that the complex strategies used by BTHS cells to maintain normal energy production can cause sufficiently severe depletion of certain amino acids to impair cardiac muscle protein synthesis. As a result, extra protein and supplements of arginine and cysteine often should be considered to reverse serious deterioration in cardiac function caused by cardiac muscle wasting.
- Anesthesia for BTHS patients requires special considerations due to increased risks from the cardiac, muscular and metabolic issues involved in the disorder. Dilated cardiomyopathy is frequently present, the risk of ventricular arrhythmias is elevated, and lactic acid may accumulate rapidly. The much reduced muscle mass of BTHS can lead to rapid electrolyte shifts and predispose BTHS patients to hypoglycemia. Thus, care should be taken to minimize fasting and to avoid use of lactated intravenous fluids.

A Multi-Disciplinary Approach

BTHS patients often have a team of specialists potentially including, but not limited to:

- Biochemical geneticist
- Cardiologist
- Clinical geneticist
- Endocrinologist
- Gastroenterologist
- General Physician
- Hematologist
- Immunologist
- Neurologist
- Nurses
- Nutritionist
- Physical Therapist
- Occupational Therapist

How to Diagnose

Barth syndrome (BTHS) is a complex, multi-system disorder. It 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 seven cardinal characteristics or in cases with family histories of multiple male deaths or fetal loss. Also note that a confirmed female BTHS case now has been reported.

Diagnostic Testing

- DNA sequence analysis (genetic testing) of the *tafazzin* gene (*TAZ*, also called *G4.5*)
- Cardiolipin analysis of various cells 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:

www.barthsyndrome.org



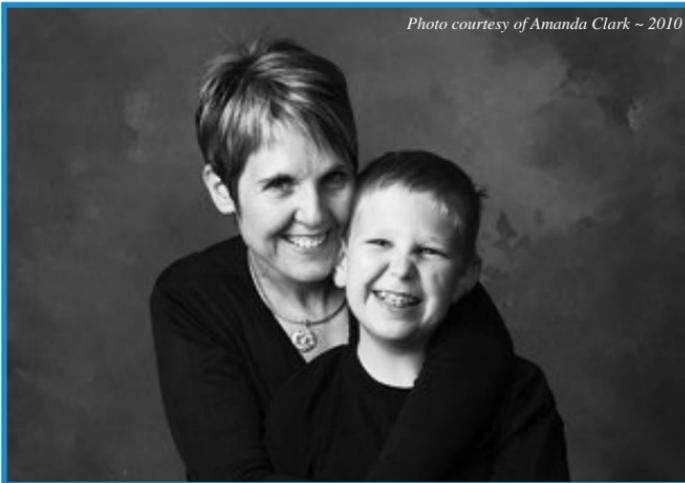
I'm quite certain it is under-diagnosed. If you have never heard of the disease, you are not going to look, you are not going to find. ~ Jeffrey Towbin, MD, FAAP, FACC, FAHA, Chief, Pediatric Cardiology, Cincinnati Children's Hospital, Cincinnati, OH

Inheritance

Barth syndrome (BTHS) is an X-linked genetic condition, usually transmitted from mother to son (although there is a relatively high incidence of new mutations in BTHS and one confirmed case report of a female BTHS patient). A mother who is a carrier of a Barth syndrome mutation (the gene is named *tafazzin* — also called *TAZ* or *G4.5*) shows no signs or symptoms of this 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 proven non-carrier mothers, all mothers of BTHS children should be tested in order to define the genetic risk in each family.

Any male child related through the female line to a BTHS individual should be tested for the disorder, as there can be great variation in phenotype even among affected siblings.



Please consider this disease in any boy with cardiomyopathy of any form, muscle weakness, neutropenia or hypoglycemia, or in any family with a history of multiple male deaths in childhood or male fetal loss and still birth. ~ Colin Steward, FRCP, FRCPCH, PhD, Pediatric Hematology, Royal Hospital for Children, Bristol, England

Highlights of Current Clinical Knowledge

†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.

For the most up-to-date information, including a full BTHS bibliography and links to PubMed abstracts, please visit:

www.barthsyndrome.org

1981 and 1983

An X-linked mitochondrial disease affecting cardiac muscle, skeletal muscle and neutrophil leucocytes, first mentioned in 1981 and then fully described in 1983 by Peter Barth (pediatric neurologist).

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 Neur Sc 1983;62:327-355. (*PubMed Abstract*)

1991

3-methylglutaconic aciduria found to be a clinical biochemical marker for BTHS.

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;119(5):738-747. (*PubMed Abstract*)

1995

G-CSF used successfully to treat neutropenia in BTHS.

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(Suppl):A177.



1996

Gene discovered on the distal arm of Xq28 (gene named *tafazzin* or *TAZ* or *G4.5*).

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; 12(4):385-389. (*PubMed Abstract*)

1998

Female carriers of BTHS asymptomatic due to X-chromosome inactivation.

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;63(5):1457-1463. (*PubMed Abstract*)

1999

Higher-than-expected unrelated BTHS cases discovered in one hospital in Bristol, UK, indicating under-diagnosis of this disease.

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): 311-315. Erratum in : J Pediatr 2000 Jun;136(1):136. (*PubMed Abstract*)

2000

***Tafazzin* involved in cardiolipin remodeling in BTHS fibroblasts.**

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



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Highlights of Current Clinical Knowledge

2001

Clinical course and treatment of neutropenia in BTHS patients presented.

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.

2003

Phospholipid abnormalities documented in children with BTHS.

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)

2005

Only the full-length and exon 5-deleted mRNAs of the TAZ gene appear to be functional.

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

Risk of serious arrhythmias and sudden cardiac death documented in adolescent BTHS patients.

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)

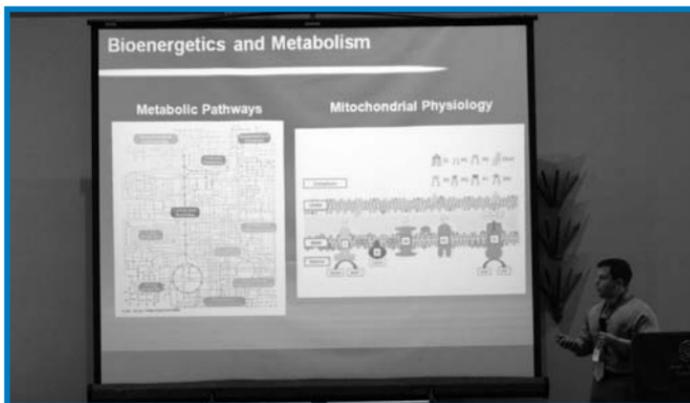


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2006

BTHS clinical phenotype described, based on data from largest cohort of BTHS patients to date.

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. † ▼ (*PubMed Abstract*)

2007

Mitochondrial size, quantity and structure abnormal in BTHS tissues and cells.

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 Abstract*)

Normal verbal but lower mathematical and visual spatial skills reported in BTHS patients indicating that educational support should be implemented during early school-age years for children with BTHS.

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

Successful cardiac transplantation in BTHS discussed, along with detailed experience with specific post-transplant medications.

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):327-31. (*PubMed Abstract*)

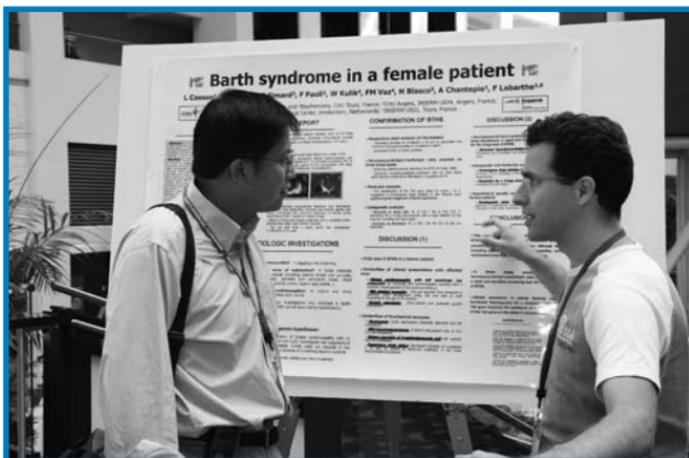


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Highlights of Current Clinical Knowledge

2008

BTHS screening using bloodspots and HPLC tandem mass spectrometry developed.

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. †
(PubMed Abstract)

Case report of a Chinese patient with BTHS presenting with acute metabolic decompensation described.

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.
(PubMed Abstract)

2009

Mitochondria-derived reactive oxygen species may be involved in the cause of neutropenia in BTHS patients.

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)

iPLA2 is a potential target for Barth syndrome therapies.

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 Abstract)



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2009 (cont'd)

Quality of life for youth with BTHS lower than that for healthy individuals and for those with cardiac disease alone.

Storch EA, Keeley M, Merlo LJ, St. Amant JB, Jacob M, Storch J, Spencer C, Byrne BJ. **Psychosocial functioning in youth with Barth syndrome.** Children's Health Care, Volume 38, Issue 2 April 2009, pp. 137-156. † ▼ (PubMed Abstract)

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 Jul 30. † ▼ (PubMed Abstract)

Cardiolipin synthesis necessary to support cholesterol biosynthesis.

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 Abstract)

Mitochondrial defects in BTHS caused not only by inner-membrane defects but also seemingly by outer-membrane protein biogenesis issues.

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 Abstract)



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Highlights of Current Clinical Knowledge

2009 (*cont'd*)

First case report of female BTHS patient confirmed by genetic analysis.

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 on Inborn Errors of Metabolism. October 2009. Molecular Genetics and Metabolism 98(2009) 89-118.

2010

Gonadal mosaicism of TAZ (G4.5) mutation reported in obligate carrier mother of children with BTHS.

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 Mar 2. [Epub ahead of print] (*PubMed Abstract*)

First conclusive demonstration given 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. † ▼ (*PubMed Abstract*)



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2010 (cont'd)

New tafazzin knockdown mouse model showed molecular and clinical aberrations in both cardiac and skeletal muscle as well as cardiolipin abnormalities consistent with human BTHS.

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. 2010 Nov 9. [Epub ahead of print] † (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)



I have to say in all my years of practicing medicine, I have never encountered any [conference] model such as this... I learned BSF also leverages this occasion to conduct clinical research to advance the knowledge about the disease. The exceptional quality of your educational programs was made clear to me from the conversations I shared with the scientific and medical professionals attending this event. ~ Marion Burton, MD (President of the American Academy of Pediatrics)

Barth Syndrome Foundation

Resources for Healthcare Providers and Families

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 and families 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.

The BSF website (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.

Our 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 clinics are also held enabling families to consult with medical experts from around the world. In addition, the clinics offer families the opportunity to participate in research studies and provide important clinical data and biological samples to the Barth Syndrome Registry and Repository.

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

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

Our Mission is...

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

Resources for Barth Syndrome Research

Research Grant Program

BSF and its affiliates sponsor a competitive research grant program to facilitate advances in Barth syndrome (BTHS) 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. Over the past nine years, we have awarded 54 separate grants totaling US \$2.0 Million, to over 34 investigators around the world.

Barth Syndrome Registry and Repository

Through an agreement with Children's Hospital Boston and with BSF's sponsorship, the Barth Syndrome Registry and Repository (BRR) has been established to promote the collection and sharing of clinical histories and biological samples (including cell lines) from BTHS patients. The BRR is available to any qualified researcher worldwide who is interested in studying BTHS. For a direct link, please visit the homepage of BSF's website (www.barthsyndrome.org).

Human *Tafazzin* Gene Mutation & Variation Database

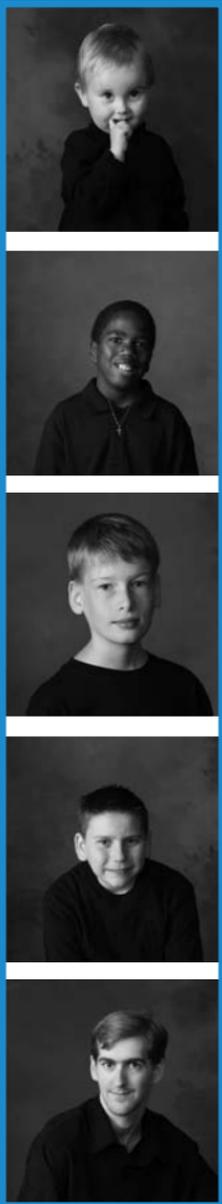
A central, up-to-date, comprehensive database listing all known mutations and variations in the human *tafazzin* (*TAZ* or *G4.5*) gene was established and is maintained by BSF. This is a very valuable resource which can be easily accessed through our main website. We strongly encourage anyone who knows 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.

For further information on our research programs, please visit our website.

www.barthsyndrome.org

A central repository for clinical data provides 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, Children's Hospital, Boston, MA; Clinical Research, Genzyme Corporation, Cambridge, MA

Please Join Us



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- 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 (with the next scheduled for late June 2012 in St. Petersburg, FL). Please visit BSF's website for additional information.
- Participate in the Barth Syndrome Registry and Repository to further research. For a direct link, please visit the homepage of BSF's website (www.barthysndrome.org).
- Receive our newsletter which delivers relevant and timely research, medical and organizational information.

Please contact us for more information.

bsfinfo@barthysndrome.org

I would rank the quality of science presented during this meeting along with the level of professionalism overall for this meeting superior in comparison to most professional meetings I have attended. I was shocked when I learned it was organized and executed by volunteers. ~ Jeffrey Harrisburg, MD, Pediatric Cardiology, Red Cross Children's Hospital, Cape Town



Barth Syndrome
Foundation

www.barthsyndrome.org

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