WHAT IS BARTH SYNDROME?

December 2007
Barth syndrome (BTHS; OMIM 302060) is a rare but serious, X-linked genetic disorder affecting males around the world.

It is caused by mutations in the tafazzin gene (TAZ, also called G4.5), resulting in an inborn error of lipid metabolism.
What are the Cardinal Characteristics?

Cardinal characteristics of this multi-system disorder may include combinations and degrees of:

- **Cardiomyopathy** (dilated or hypertrophic, possibly with left ventricular noncompaction and/or endocardial fibroelastosis)
- **Neutropenia** (chronic, cyclic, or intermittent)
- **Muscle hypoplasia and weakness**
- **Growth delay** (abnormal growth pattern, similar to but more severe than constitutional growth delay)
- **Exercise intolerance**
- **Cardiolipin abnormalities**
- **3-methylglutaconic aciduria**
Important clinical problems may include:

- Congestive heart failure
- Risk of life-threatening arrhythmia
- Life-threatening bacterial infection
- Feeding problems
- Frequent diarrhea
- Gross motor delay
- Short stature in the early years, followed by accelerated growth in late puberty
- Diminished capacity for exercise

(continued)
Clinical Characteristics of Barth Syndrome

Important clinical problems: (continued)

• Hypoglycemia, including fasting hypoglycemia (especially in the newborn period)
• Recurrent aphthous ulcers
• Chronic headache, abdominal pain and/or body aches (especially during puberty)
• Osteoporosis
• Extreme fatigue
• Some mild learning disabilities
Diagnostic Testing for Barth Syndrome

Diagnostic Laboratory Testing for Barth syndrome

• Quantitative urine organic acid analysis, including quantification of 3-methylglutaconic acid

• Cardiolipin analysis of muscle, platelets or cultured cells

• DNA sequence analysis (genetic testing) of the tafazzin gene (TAZ, also called G4.5)

(For more details about these tests and a listing of approved labs, please refer to www.barthsyndrome.org)
Scientific and Medical Advisory Board

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Get More Information

➢ Visit www.barthsyndrome.org

➢ Contact any member of the Scientific and Medical Advisory Board

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