



Barth Syndrome

Cardiovascular System

Barth Heart 101



Barth Syndrome
Foundation

Cardiac symptoms are not always present among those who have Barth syndrome (BTHS). There are no known methods to detect who might develop cardiac symptoms, or symptom severity even within the same family. However, individuals with BTHS do commonly have heart problems, chiefly cardiomyopathy (that may lead to heart failure and/or arrhythmias). Therefore, it is advisable for all of those who have Barth syndrome, even those who do not have any apparent cardiac symptoms, to be seen by a cardiologist for routine follow-up cardiac evaluations to monitor the heart's pumping and electrical function. Routine visits and evaluations provide baseline values for comparative change that can happen over an extended period or during a state of crisis.

It is not uncommon for those who have BTHS to experience a decline in cardiac function during periods of high growth velocity in infancy and puberty. Delayed onset of puberty is common among males. Height and weight should be closely monitored along with increased cardiac surveillance during these "growth spurts" and throughout the extended period of puberty. Due to the dangerous cardiac risks standard cardiac evaluation including EKG/ECG, Echo and Holter monitoring should be considered on an annual basis at the very least.

It is ideal to be evaluated by a cardiologist with a special interest in heart failure and/or cardiomyopathy. The cardiologist may also refer the person with Barth syndrome for an evaluation by an electrophysiologist who specializes in heart rhythm abnormalities.

The doctor should always be consulted when concerns arise and certainly consulted if the person with Barth syndrome experiences any of the following:

- Breathlessness
- Greater than usual fatigue
- Dizziness or lightheadedness
- Fainting or passing out
- Racing of the heart
- Heart palpitations
- Skipping beats of the heart
- Swelling in the abdomen or extremities
- Abnormal pallor
- Bluish tinge of the lips and nail beds
- Pain in chest
- Sudden weight gain
- Excessive sweating
- Loss of appetite
- Nausea
- Extreme thirst

Other Cardiovascular Risks

- Cardiomyopathy: Usually dilated with variable myocardial hypertrophy, left ventricular non-compaction (LVNC) and/or endocardial fibroelastosis
- Thrombotic Events
- Heart Failure
- Arrhythmia Risks (Repolarization abnormalities, ST flattening, T-wave inversion)
- Autonomic Instability
- Acute Decompensation