

The slide features a decorative arrangement of six circles. Two circles are white with a light green outline, while the other four are solid light green. They are arranged in two rows: the top row has three circles and the bottom row has three circles.

Diagnostic Criteria for Barth syndrome

Pediatric Neurology Setting

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

- *A child with Barth syndrome could easily present in the pediatric neurologist's office for an array of reasons.*
- *A floppy infant or child may be referred to the neurologist without any other relevant background information, and may turn out to have:*
 - *Cardioskeletal myopathy*
 - *A lipid myopathy*
 - *3-methylglutaconic aciduria*
- *A geneticist, cardiologist or other specialist may refer a child to pediatric neurologist because they need his view to get a complete picture, either before or after the diagnosis has been made.*

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

The neuromuscular symptoms of Barth syndrome are:

- *Muscle affection mainly in a limb girdle distribution*
- *Moderate muscle wasting*
- *Moderate weakness, permitting unsupported walking*
- *Exercise intolerance*
- *Absence of contractures*
- *No involvement of bulbar musculature, with the exception of occasional mild facial muscle weakness*
- *No progression to wheelchair dependence, no involvement of extra-ocular muscles or diaphragm.*

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

- *A child with Barth syndrome showing Gowers' sign on rising.*
- *Notice moderate muscle wasting.*



Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

A boy with Barth syndrome showing

- lordotic back
- recurvated knees.....



Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting



- ...mild wasting of extremity muscles

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting



The facial appearance of Barth syndrome is not “diagnostic”, but experienced investigators point to a similarity in facial appearance in many affected boys.

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

Cardinal Characteristics besides neuromuscular involvement:

- *Cardiomyopathy (frequently dilated)*
- *Neutropenia (chronic, cyclic or intermittent)*
- *Growth retardation (can appear as failure to thrive) before puberty*
- *3 - methylgluticonic aciduria, detectable by gaschromatography and mass-spectrometry*

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

● Major Clinical Hazards:

- *Congestive heart failure*
- *Risk of serious arrhythmia including sudden cardiac death*
- *Serious bacterial infections*
- *Hypoglycemia*

● Other major problems:

- *Growth deficiency*
- *Lack of stamina*

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

● Variable Clinical Problems:

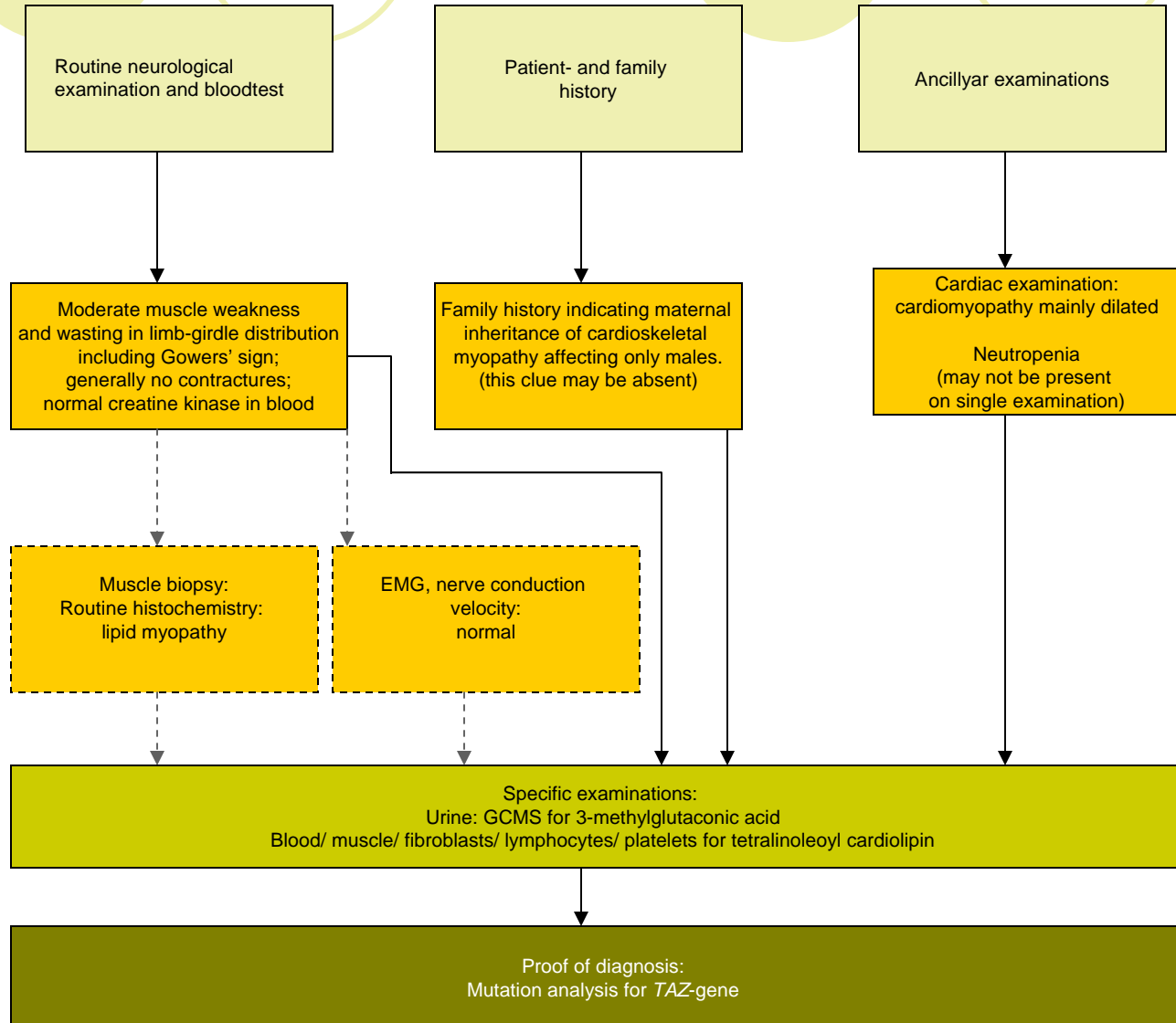
- *Frequent diarrhea*
- *Recurrent aphthous ulcers*
- *Hypoglycemia, including fasting hypoglycemia in the neonate*
- *High incidence of minor congenital malformations*
- *Low bone density*
- *Reported chronic headaches and body aches especially during puberty*
- *Feeding Problems*
- *Reports of cognitive learning difficulties*

Diagnostic Criteria for Barth Syndrome in the Pediatric Neurology Setting

● Diagnostic Testing

- *Quantitative Urine Organic Acid Analysis, including quantification of 3- methylgluticonic acid*
- *Cardiolipin analysis of muscle, platelets, lymphocytes or cultured skin fibroblasts*
- *DNA sequence analysis (Genetic testing of the tafazzin [G4.5] gene)*

Diagnostic flow-scheme for the diagnosis of Barth syndrome in an affected male patient



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