

# What is Barth Syndrome?

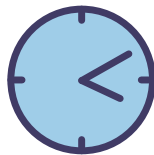
Barth syndrome is a rare, genetic, mitochondrial disorder causing metabolic abnormalities that can lead to an enlarged and weakened heart (cardiomyopathy), muscle weakness and fatigue (skeletal muscle myopathy), low levels of certain white blood cells that can lead to recurrent infections (neutropenia), growth delay that potentially can lead to short stature, and increased levels of 3-methylglutaconic acid in the urine and blood. The disease affects primarily males.

## Quick Facts



According to the limited studies available, **fewer than 10 infants** with Barth syndrome are diagnosed each year in the United States.

Individuals with Barth syndrome have **shortened life expectancy**; many die due to infections or heart failure early in life. The rate of mortality is highest during the first four years of life. Those who survive past early childhood often live only into their 40s.



Barth syndrome primarily affects the **heart, muscles, immune system** and **growth**.

Barth syndrome is inherited through a mutation on the **X chromosome**. Because women have two X chromosomes, the normal copy compensates for the defective one.

However, a boy born to a woman with the Barth syndrome mutation has a **50% chance of inheriting the mutation and developing Barth syndrome**. Girls born to a mother who is a carrier have a 50% risk of being carriers themselves.



Barth syndrome typically becomes apparent within the **first months of life**, but the onset of different symptoms can vary among affected individuals.

## Signs & Symptoms

Major clinical issues associated with Barth syndrome include:



Enlarged and weakened heart (dilated cardiomyopathy)



Heart Failure



Muscle weakness (myopathy) or low muscle mass



Serious abnormal heart rhythm (arrhythmia), resulting in sudden death



Exercise intolerance/ lack of stamina



Low levels of certain white blood cells (neutropenia)



Serious bacterial infections



Motor skills delay



Growth delay and short stature



Increased levels of 3-methylglutaconic acid in the urine and blood



Mild learning disabilities

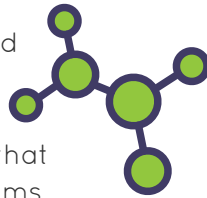
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## How Are Mitochondria Related to Barth Syndrome?



Barth syndrome is caused by **mutations in the tafazzin gene**, which provides instructions for making a protein called tafazzin. Tafazzin adds fatty acids to the cardiolipin molecule, a fat (lipid) that

is primarily found in the inner mitochondrial membrane and **helps maintain mitochondrial shape, energy production and the transport of proteins with cells**. A mutation in the tafazzin gene causes a reduced and altered cardiolipin, which **results in structurally abnormal and dysfunctional mitochondria** that contribute to the disease symptoms.



## Research & Treatments

No treatments specific for Barth syndrome have been approved. **However, there are several active clinical trials and research aim to find effective treatments.**

**Barth Syndrome Foundation** is the only international organization dedicated to saving lives through education, advances in treatment and finding a cure for Barth syndrome. Contact BSF at [www.barthsyndrome.org](http://www.barthsyndrome.org).



Barth Syndrome  
Foundation

### References

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