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Barth Syndrome

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). [Barth Syndrome](#).

Barth syndrome (BT HS) is a rare, genetic disorder of lipid metabolism that primarily affects males. It is caused by a mutation in the tafazzin gene (TAZ, also called G4.5) which leads to decreased production of an enzyme required to produce cardiolipin. Cardiolipin is an essential lipid that is important in energy metabolism. BT HS, which affects multiple body systems, is considered serious. Its main characteristics often include combinations in varying degrees of heart muscle weakness (cardiomyopathy), neutropenia (low white blood cell count, which may lead to an increased risk for bacterial infections), reduced muscle tone (hypotonia), muscle weakness, undeveloped skeletal muscles, delayed growth, fatigue, varying degrees of physical disability, and methylglutaconic aciduria (an increase in an organic acid that results in abnormal mitochondria function). Although some with BT HS may have all of these characteristics, others may have only one or two and are often misdiagnosed. BT HS is an X-linked genetic condition passed from mother to son through the X chromosome. A mother who is a carrier of BT HS typically shows no signs or symptoms of the disorder herself. On average, 50 percent of children born to a carrier mother will inherit the defective gene, but only boys will develop symptoms. All daughters born to an affected male will be carriers but typically will not have symptoms.