Open Peer Review on Qeios

Barth Syndrome

National Cancer Institute

Source

National Cancer Institute. <u>Barth Syndrome</u>, NCI Thesaurus. Code C84585.

A rare X-linked syndrome caused by mutations in TAZ1 gene. Signs and symptoms include cardiomyopathy, neutropenia, muscle weakness and atrophy, growth delay, cardiolipin deficiency and 3-methylglutaconic aciduria.