Power outage: Research offers hint about heart weakness in Barth syndrome

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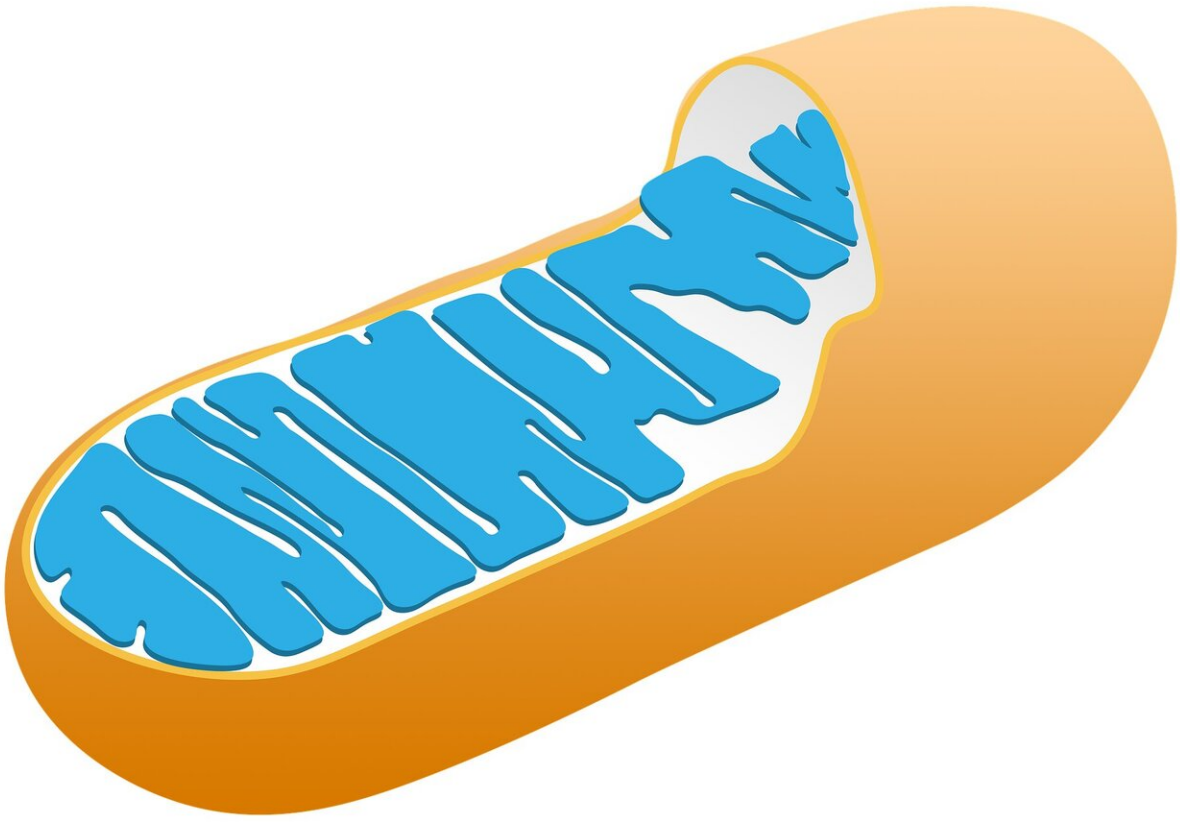

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Barth syndrome is a rare condition that occurs almost exclusively in males. Symptoms include an enlarged and weakened heart. The condition is present at birth or becomes evident early in life. Life expectancy is shortened and there is no treatment.

The laboratory of Madesh Muniswamy, Ph.D., in the Long School of Medicine at The University of Texas Health Science Center at San Antonio, found a clue about the processes that underlie this devastating condition.

The body runs on energy made by cell structures called mitochondria. The Muniswamy lab studied the interaction of a protein called MCU (mitochondrial calcium uniporter) with a phospholipid (fat) called cardiolipin. This fat is part of the mitochondria walls.

The team found that cardiolipin's binding with MCU acts as an on switch for energy production. When the two bind together, calcium ions rush into the mitochondria to produce energy needed during physiologic processes such as fasting and feeding.

The connection to Barth syndrome? Loss of energy.

"We observe reduced abundance and activity of MCU in cells of mammals that model Barth syndrome, and we also see a partial loss of cardiolipin in the cells," Dr. Muniswamy said.

"Consistently, MCU is also decreased in the cardiac tissue of human Barth syndrome patients, raising the possibility that impaired MCU function contribute to Barth syndrome pathology," he said.

The study, coauthored with colleagues at Texas A&M University, Harvard and MIT, is in *Proceedings of the National Academy of Sciences.*

More information: Sagnika Ghosh el al., "An essential role for cardiolipin in the stability and function of the mitochondrial calcium uniporter," *PNAS* (2020). www.pnas.org/cgi/doi/10.1073/pnas.2000640117

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