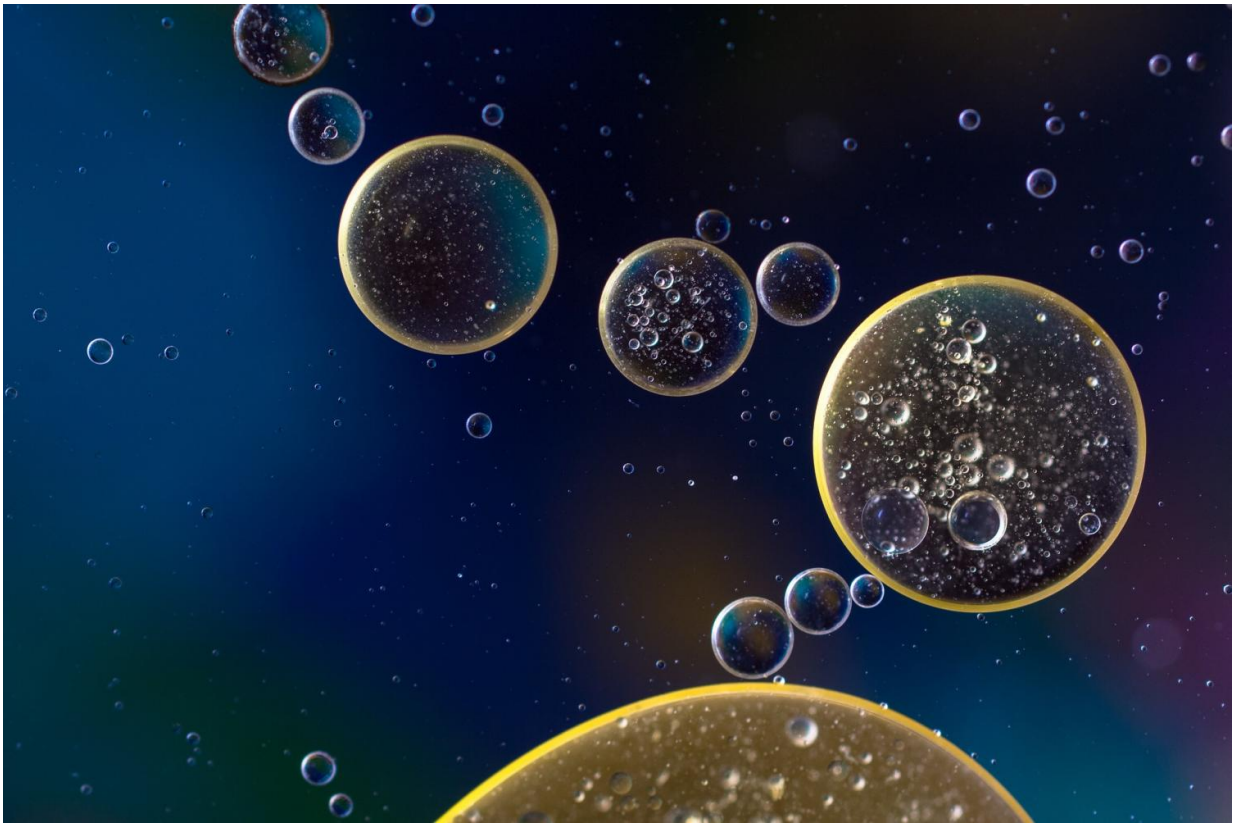
Transporter mutation alters cell energy

February 21 2020, by Leigh MacMillan



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The cotransporter NKCC1 moves sodium, potassium and chloride ions across the cell membrane and has roles regulating cell volume, epithelial transport and neuronal excitability.

Eric Delpire, Ph.D., and colleagues recently discovered that a patient

suffering from multiorgan failure had a mutation in the gene encoding NKCC1. Increased mitochondrial DNA in tissue biopsies suggested that the transporter may play a role in energy metabolism.

The researchers have now demonstrated that fibroblasts (connective tissue cells) from the patient have increased mitochondrial respiration—energy production—compared to healthy fibroblasts. They further found that fibroblasts from a mouse model of the mutated NKCC1 and cultured cells expressing the mutant cotransporter also have elevated mitochondrial respiration and increased oxidative stress.

The findings, reported in the *Journal of Cellular Physiology*, establish that the disease-associated mutation in NKCC1 affects mitochondrial respiration. Cells expressing the defective cotransporter behave as if they are in a state of starvation, the authors note.

More information: Salma Omer et al. A mutation in the Na-K-2Cl cotransporter-1 leads to changes in cellular metabolism, *Journal of Cellular Physiology* (2020). DOI: 10.1002/jcp.29623

Provided by Vanderbilt University