# Scientists identify genetic causes of mitochondrial diseases

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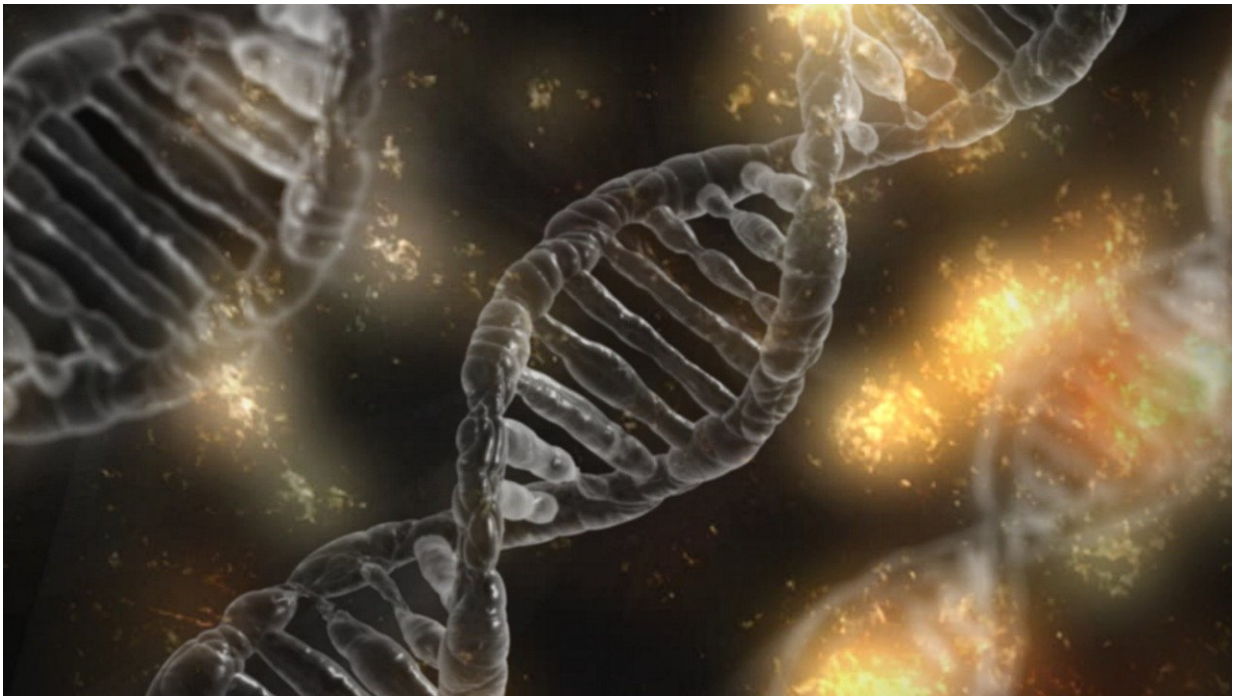

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An international team of scientists led by researchers from the University of Colorado School of Medicine have identified previously unknown genetic causes of mitochondrial diseases.

The findings are published in the October 3 edition of the journal *Nature Communications*. Johan Van Hove, MD, Ph.D., professor of pediatrics at

the CU School of Medicine, is the senior author and Marisa Friederich, Ph.D., assistant professor of pediatrics, is the first author of the article, which lists 54 co-authors from around the world.

The team studied five families with infants who had cardiomyopathy and excess acid in their blood, which appears when the cell's energy-generating system malfunctions. In these cases, the conditions appeared prenatally or when the newborn was two to five months old. In all cases, the child died before turning seven months old. One of the five families was from Colorado.

Mitochondria, which are present in almost every cell in the body, create the energy needed to sustain life and support organ function. Failure of mitochondria leads to less energy generated in the cells. Cell failure, if repeated throughout the body, can lead to the failure of whole organ systems, and can cause accumulation of lactic acid.

Mitochondrial disease is difficult to diagnose because of the complex biochemistry and because of genetic differences between individuals. In the five patient cases reviewed for this article, the scientists saw patterns that led to looking closer at a specific genes and determining that mutations in those genes caused the lethal conditions. They also identified that increasing a nutritional compound would improve the function of the cells from these patients, identifying a new opportunity for treatment.

The project involved seven research centers worldwide. In addition to Colorado, scientists from Cambridge, United Kingdom; Nijmegen, the Netherlands; Madrid, Spain; Haifa, Israel; Groningen, the Netherlands; and Adelaide, Australia, were involved in the study.

The research conducted in Colorado was supported by a local patient organization, Miracles for Mito, and by a team of Courage Classic

riders, Summits for Samantha. The Courage Classic is an annual bike ride/fundraiser sponsored by the Children's Hospital Colorado Foundation.

**More information:** Marisa W. Friederich et al, Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder, *Nature Communications* (2018). DOI: 10.1038/s41467-018-06250-w

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