Discovery of CLPB gene associated with new pediatric mitochondrial syndrome

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A new study published in the *American Journal of Human Genetics* demonstrates the continued important contributions from the Center for Pediatric Genomic Medicine at Children's Mercy Hospital in Kansas City, Mo. The study describes a new pediatric mitochondrial syndrome and discovery of the responsible gene, called CLPB.

Dr. Carol Saunders and her team partnered with collaborators in Denmark to report their collective findings based on gene mapping and exome sequencing in five children with CLPB-related disease. These patients had strikingly similar clinical findings including cataracts, severe psychomotor regression during febrile episodes, epilepsy, neutropenia with frequent infections, urinary excretion of 3-methylglutaconic aciduria, and death in early childhood.

"This research once again highlights the power of genomic medicine in the diagnosis and discovery of rare pediatric conditions," said Saunders, clinical laboratory director of the Center for Pediatric Genomic Medicine "In this case, we have identified one of the many genes, CLPB, involved in mitochondrial diseases. These findings emphasize the importance of basic research into the characterization of human CPLB gene function and will pave the way for the diagnosis of other patients."

The Center for Pediatric Genomic Medicine at Children's Mercy was the first genome center in the world inside a children's hospital, and the center's STAT-Seq test for critically ill newborns was one of TIME magazine's Top 10 Medical Breakthroughs of 2012.

More information: Learn more at http://www.childrensmercy.org/genomics.

Provided by Children's Mercy Hospital