Mitochondrial disease has a disproportionate healthcare burden in US

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Mitochondrial diseases are a diverse group of disorders caused by mutated genes that impair energy production in a patient's cells, often with severe effects. When patients with these diseases are hospitalized, they incur high medical costs, and suffer higher-than-typical rates of comorbid diseases and in-hospital mortality. Researchers who analyzed those costs using national databases say their findings underscore the importance of developing preventive strategies and therapies for these illnesses.

"There have been few systematic investigations of the public health burden of mitochondrial disease, even as these disorders are being diagnosed more frequently than ever before," said study leader Shana E. McCormack, MD, a pediatric researcher at Children's Hospital of Philadelphia (CHOP). "We hope that our initial efforts to define the scope of this problem will lead to better medical practice in helping patients and families."

McCormack and colleagues from multiple clinical areas at CHOP published their research on April 27, 2017 in *Molecular Genetics and Metabolism*.

Tiny structures found in large numbers within cells, mitochondria function as cellular power plants, extracting energy from nutrients to drive the body. They carry their own DNA, distinct from the better-known DNA inside a cell's nucleus. Disease-causing mutations in either mitochondrial or nuclear DNA curtail cellular energy production in the

respiratory chain, an interconnected group of proteins within mitochondria. This results in a broad variety of damaging effects in potentially any organ.

Although previously considered a rare class of disorders, collectively mitochondrial disease is relatively common, with disease-causing mutations occurring in at least 1 in 4,300 Americans. Every 30 minutes, a child is born who will develop a mitochondrial disease by age 10. Patients receive supportive therapy, as there are no definitive treatments for these disorders, although clinical trials are under way in several types of mitochondrial disease.

In their analysis, the study team drew on national databases holding records of patients hospitalized in 2012 for mitochondrial disease, representing about 3,200 pediatric and 2,000 adult hospitalizations. The estimated costs associated with a typical hospitalization were twice as high for a patient with mitochondrial disease than for a patient without mitochondrial disease. In-hospital mortality rates—2.4 percent for children and 3.0 percent for adults—were also disproportionately higher. These mortality rates were six times higher than average in children and three times higher than average in adults.

The researchers also did a separate analysis of all adult and pediatric inpatients in California civilian hospitals over a five-year period, from 2007 through 2011. They found that individuals with at least one hospitalization during that time with mitochondrial disease had a median of 1.1 hospitalizations per year of follow-up. Infants under age two years had the most frequent hospitalizations. Over five years of follow-up, 9.9 percent of patients with at least one hospitalization died while in the hospital.

The study team noted that the actual costs and disease burden may well be higher than their estimates, because hospital discharge records may

fail to capture mitochondrial disease, which may be under-diagnosed. "Further studies would benefit patients by investigating better strategies for supportive care, for preventing hospitalizations and for minimizing the burden of hospitalizations on patients and families," said McCormack.

More information: Shana E. McCormack et al, Hospitalizations for mitochondrial disease across the lifespan in the U.S., *Molecular Genetics and Metabolism* (2017). DOI: 10.1016/j.ymgme.2017.04.007

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