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## "Revolutionary" News From Medicine: 1 in 200 People Carry Mitochondrial Disease Mutation

By David Kirby - 8-11-08

**BOTH MITOCHONDRIAL "DISEASE" AND "DYSFUNCTION" APPEAR TO BE MORE COMMON THAN PREVIOUSLY THOUGHT -- IMPLICATIONS FOR AUTISM, OTHER DISORDERS ARE "EARTH SHATTERING."**

In February, when the US government conceded that vaccines had caused an autism-inducing reaction in little Hannah Poling, most experts declared that her underlying condition, a mitochondrial disorder, was exceedingly rare - so rare, in fact, that it had no bearing on other autism cases.

But on Monday, the United Mitochondrial Disease Foundation announced a "landmark research finding" showing that at least one in 200 healthy humans "harbors a pathogenic mitochondrial mutation that potentially causes disease." The finding was published in the current issue of the [American Journal of Human Genetics](#).

"This is earth shattering news," UMDF Executive Director and CEO Charles A. Mohan, Jr. told me. "Some of my colleagues are calling it 'revolutionary.' We have shown that mitochondrial disease is not rare."

Mitochondria are the little powerhouses found within most cells, and which produce most of the body's energy. Mitochondria are key for proper neurotransmission and, for obvious reasons, are highly concentrated in cells of the brain and central nervous system.

Up until now, estimates of mitochondrial disease rates have held steady at about 1-in-4000 people. But this study shows that 20 times that number have genetic mutations that could cause mitochondrial disease.

"What this says to me is that more than 1-in-4,000 people have mitochondrial disease," Mohan said. "And it tells me that 1-in-200 could develop some type of mitochondria-related disease over the course of their lifetime, depending in part on environmental triggers."

Mitochondrial disorders are found at "the core of many well known diseases and chronic illnesses, such as Alzheimer's disease, Parkinson's disease and autism spectrum disorders," a statement from the UMDF said today.

Humans have two types of DNA: nuclear, and mitochondrial. The study looked at 10 mutations in mitochondrial DNA that are known to cause disease, and identified them in the cord blood of 1 in 200 newborn children.

The study looked exclusively at classic mitochondrial "disease." In the classic form, inherited mutations of mitochondrial DNA are passed down through the mother, causing a wide variety of pathologies, including seizures, digestive problems, paralysis, blindness, heart disease, neurodevelopmental disorders and other problems.

The classic form is often quite severe, and sometimes fatal. But it is not rare.

Which brings us to Hannah Poling: She does not have "classic," maternally inherited mitochondrial disease.

Hannah does share the same single-point mutation in mitochondrial DNA as her mother, Terry. But this mutation is apparently benign (Terry Poling is just fine), is not described in the medical literature, and is not associated with any pathology at all.

Instead, Hannah seems to have had a much milder, even asymptomatic form of mitochondrial "dysfunction" - one that led to reduced cellular energy, but no obvious signs of severe mitochondrial "disease."

In April, I reported that researchers in Baltimore were studying 30 children at one autism clinic who all had nearly identical markers for mild mitochondrial dysfunction. One of them was Hannah Poling.

All 30 children were developing normally until they encountered some type of immunological stress and began showing signs of regressive autism soon afterwards.

In 28 cases, the doctors said, typical childhood fevers caused the stress, while in the other two cases, including Hannah, vaccines appeared to be the exacerbating factor.

The doctors - who spoke on a CDC conference call that included executives from the health insurance industry -- reported that mitochondrial dysfunction was found in autism "in numbers that make it not a rare occurrence."

Some estimates currently put the rate of mitochondrial dysfunction in ASD at 7-20%, while rates among regressive autism cases could climb much higher than that.

This milder form of mitochondrial disorder, the doctors said, was probably caused by a mutation found in nuclear (as opposed to mitochondrial) DNA, and inherited through the father -- rather than through the mother, as in classic mitochondrial disease.

Shockingly, the nuclear DNA mutations that bring risk of dysfunction could be as common as 1-in-400 to 1-in-50 people - though no one knows how many people have developed actual mitochondrial disorders because of it.

Even so, we can now assume that classic mitochondrial "disease" described in this study (via mutations in maternal mitochondrial DNA) and mild mitochondrial "dysfunction" found in Hannah and others (via mutations in paternal nuclear DNA) are both associated with increased risk for autism.

And we can also now assume that neither form of mitochondrial disorder is rare. Moreover, whether the low cellular energy originates in mitochondrial DNA or nuclear DNA mutations, either way it could confer increased risk for autism.

That would mean a significant number of children between the ages of 1 and 2 who are walking around right now, potentially vulnerable to autistic regression triggered by some acute immune stressor - whether vaccine related or not.

"Mitochondrial dysfunction represents a major unexplored area of human biology of vital importance to human health," the UMDF statement said, noting that it also has been implicated in autoimmune diseases such as multiple sclerosis and lupus.

"While it cannot yet be said that mitochondrial dysfunction causes these problems, it is clear that mitochondria are involved because their function is measurably disturbed," the statement said.

This new study suggests that, "mitochondrial dysfunction is a major underlying risk factor for human disease," said Dr. Douglas C. Wallace, professor of molecular medicine and director of the Center for Molecular and Mitochondrial Medicine and Genetics at the University of California-Irvine.

He should know. Dr. Wallace is one of the world's leading mitochondria researchers, and a member of the UMDF's Scientific and Medical Advisory Board. He also has a 23-year-old son with autism.

In April, Dr. Wallace told the Vaccine Safety Working Group of HHS's National Vaccine Advisory Committee that over-vaccination of people with mitochondrial disorders was a deep concern, especially in light of Hannah Poling, who got nine vaccines in one well-baby visit.

"We have always advocated spreading the immunizations out as much as possible because every time you vaccinate, you are creating a challenge for the system," Dr. Wallace testified. "And if a child has an impaired system, that could in fact trigger further clinical problems."

I take that to mean that children with impaired mitochondria might also have impaired immune systems. And children with impaired immune systems might not be able to handle, say, nine vaccines given at once.

The CDC says that multiple simultaneous vaccines are safe, "for children with normal immune systems," but makes no mention of the risk for everyone else.

But, as Dr. Wallace put it, "We do not know what is safe. We do not know what is not safe. We do not know the actual risk of a person with light mitochondrial disease has and being challenged either by vaccination or by a severe infection."

"Is there a relationship between mitochondrial disease and vaccination and mitochondrial disease and autism?" Dr. Wallace asked the HHS panel. "Would a vaccination or infection initiate an incipient mitochondrial disease, as has been suggested?"

Only major investments in scientific research will answer these questions, which have become particularly pressing now that we know that mitochondrial disorders are anything but "rare."

"This will help us educate key members of Congress to motivate and encourage NIH to appropriate more funds to focus specifically on mitochondrial dysfunction and disease," Mohan told me. "We would like to see this result in a better understanding of the links between energy metabolism and what we call the "sexy diseases."

I likewise hope our nation's researchers will jump on this particular scientific train before it leaves the station.

It would appear that far more lives are at risk for far more diseases (well beyond autism) than we ever imagined.