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Cell Dysfunction May Play Part in Autism

Mitochondria, energy center of cell, found impaired in some, researchers report

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HealthDay

TUESDAY, Nov. 30 (HealthDay News) -- Autistic children are much more likely to have defects in a cellular structure called the mitochondria, which is responsible for producing the energy used by brain cells, preliminary research finds.

These defects may help to explain the onset or the severity of autism in some children, according to the study in the Dec. 1 issue of the *Journal of the American Medical Association*.

"In this report, children with full syndrome autism were more likely to have mitochondrial dysfunction than healthy, age-matched control children," said study author Cecilia Giulivi, a professor of biochemistry at the University of California, Davis. "But we don't know if mitochondrial dysfunction is a cause of autism or a consequence of autism."

Mitochondria, sometimes called cellular "powerhouses," produce energy that's used to fuel a cell's activity -- an especially important function in the brain, Giulivi said.

When mitochondria don't function properly, the results can be devastating. Mitochondrial dysfunction been implicated in neurological conditions ranging from Parkinson's and Alzheimer's disease to schizophrenia and bipolar disorder.

Mitochondrial disease can lead to symptoms including muscle weakness, exercise intolerance (pain and muscle cramps during exercise), gastrointestinal disorders, seizures, liver disease, vision and hearing problems, developmental delays and increased susceptibility to infection.

While previous small studies have suggested some children with autism may also have mitochondrial dysfunction, measuring the function of mitochondria isn't easy because brain biopsies are out of the question, Giulivi said.

Another area of the body in which mitochondria are active is in the muscles, but biopsying muscle tissue is also invasive, she added.

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In the new study, Giulivi analyzed the mitochondria in lymphocyte cells, an immune system component found in the blood, of 10 children aged 2 to 5 with "full syndrome" autism and 10 normally developing children.

They found children with autism were far more likely to have mitochondrial dysfunction, including defects in mitochondrial DNA and abnormalities in the levels of various enzymes produced by the mitochondria.

"There has been evidence before that some children with autism have a mitochondrial disorder, but we haven't been able to do routine screening because a muscle biopsy is quite invasive," said Geraldine Dawson, chief science officer for Autism Speaks, which helped fund the research. "This study suggests we might be able to do a blood sample, which would allow us to do routine screening."

Still the findings come with caveats. While the study suggests mitochondrial dysfunction plays a role in at least come cases of autism, the researchers stressed that the results are preliminary and more studies in larger numbers of children are needed.

And there are many unknowns, including how mitochondrial dysfunction in brain cells might alter brain function in a way that leads to some of the symptoms of autism, including communication and social difficulties.

The researchers also don't know when the mitochondrial dysfunction starts -- in the womb, in infancy or later, and how that might impact the onset of autism or what other environmental or genetic factors may also contribute.

Parents should also keep in mind that autism is a "heterogenous" disorder, Dawson said. In the new study, only one child met the clinical threshold for a mitochondrial disorder, while others showed varying degrees of mitochondrial abnormalities.

"The fact that so many were showing some evidence of a mitochondrial dysfunction is remarkable because that has never been shown before," Dawson said. "This could be a subtype of autism, or could be a contributing factor to many types. It could be a cause or a consequence. We really don't know at this point, but the important thing is there really is something different about the way the mitochondria are functioning in some children with autism."

The 10 children with autism in the study all had "full syndrome" autism. Children on the less severe end of the autism spectrum were not included in the study.

Among the various mitochondrial defects, researchers found that mitochondria from children with autism consumed less oxygen than mitochondria from the children without autism, suggesting less mitochondrial activity.

The cells of children with autism also produced twice as much hydrogen peroxide, which can lead to oxidative stress, which can damage DNA.

Giulivi urged pediatricians to be on the lookout for symptoms that might indicate mitochondrial dysfunction in autistic children, including vision or hearing problems, seizures or exercise intolerance such as muscle cramps during intensive physical activity.

More information

The United Mitochondrial Disease Foundation has more on mitochondrial disease.

SOURCES: Cecilia Giulivi, Ph.D, professor, biochemistry, University of California, Davis; Geraldine Dawson, Ph.D., chief science officer, Autism Speaks, New York City; Dec. 1, 2010, *Journal of the American Medical Association*