

Autism & Mitochondrial Disease

Mitochondrial disease can look like a number of better known diseases: Autism, Parkinson's, Alzheimer's, Lou Gehrig's disease (ALS), muscular dystrophy and chronic fatigue, among others. And it's this web of complexity and connectivity that makes mitochondrial disease research valuable to so many. Research shows that mitochondrial dysfunction is often at the crux of these more commonly recognized diseases.



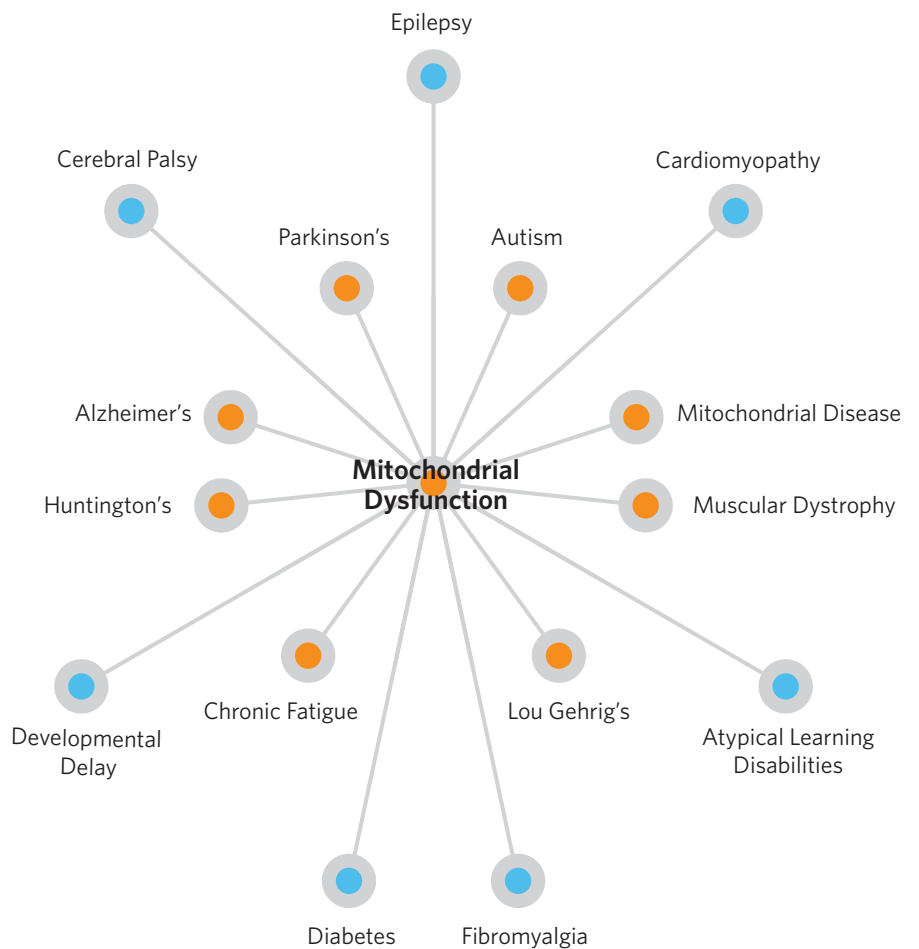
Recent reports and scientific studies linking mitochondrial disease to Autism include:

* A December 2010 study in The Journal of the American Medical Association reports impaired mitochondrial function may influence processes highly dependent on energy, such as neurodevelopment, and contribute to Autism. In this exploratory study, children with Autism were more likely to have mitochondrial dysfunction, mtDNA over-replication, and mtDNA deletions than typically developing children. *Journal of American Medical Association December 1, 2010—Vol 304, No. 21*

* Dr. Robin Morris, head of Neuropsychology at Georgia State University and Dr. John Shoffner (Medical Neurogenetics, LLC) are investigating the relationship between mitochondrial disease and Autism Spectrum Disorders. Exciting leading-edge fMRI (Functional MRI) technology from the Georgia State/Georgia Tech Joint Center for Advanced Brain Imaging will be used in this study. The study is funded
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FAMILIAR CONNECTIONS

Mitochondrial dysfunction is a central element of familiar diseases.



Research shows that mitochondrial dysfunction is often a central element of these more commonly recognized diseases. Studies and reports indicate the "orange" ones are more influenced.

A cure for mitochondrial disease could impact cures for Autism, Parkinson's, Alzheimer's and Muscular Dystrophy

by the Department of Defense and U.S. Army Medical Research and Materiel Command (USAMRMC). In patients who have mitochondria disease expressed as an Autistic Spectrum Disorder, this study will be very important to our understanding of whether identifiable differences

exist between patients with ASD and mitochondrial disease and ASD without mitochondrial disease. This study will be essential to establishing parameters by which these patients could be followed during a treatment protocol.

* Autism Speaks recognized FMM

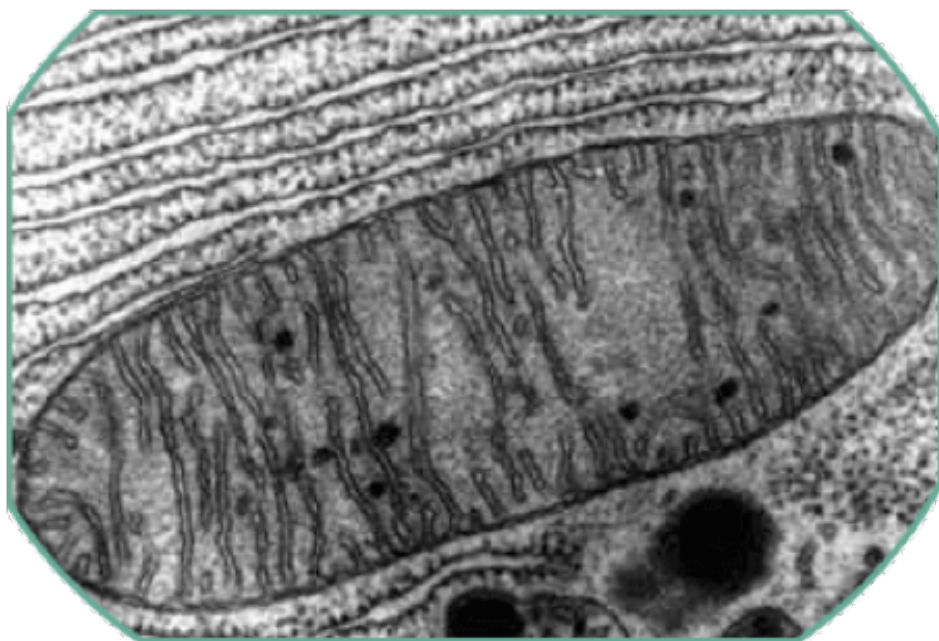
Board Member, Dr. John Shoffner, as one of the Top 10 2009 Research Achievements for finding that a subgroup of patients with mitochondrial disorders may be at risk for Autistic regression, particularly around periods of fever.

Mitochondrial Dysfunction, Autistic Regression... and Fever

Individuals with mitochondrial dysfunction and autism found to have high rate of autistic regression

Mitochondria are responsible for producing most of the energy the body uses for every day metabolic functions. More attention has recently been focused on a potential link between ASD and dysfunctional mitochondria. A study¹

published in 2009 in the Journal of Child Neurology further examined this link, finding that **a subgroup of patients with mitochondria disorders may be at increased risk for autistic regression**, especially around periods of fever.



Mitochondria are intimately tuned to the environment in which they reside and are built to respond quickly to fluctuations in the state of that environment. To characterize a relationship between mitochondria disorders and ASD, researchers from Atlanta identified a group of 28 children who had been diagnosed with both ASD and mitochondrial disease. The most common clinical observation in children with both ASD and mitochondria disorder was “hypotonia,” or muscles with low tone, followed closely by “fatigue with activity.” They also found

that approximately 60% (17 of 28) of these children experienced a regressive form of ASD, a rate of regression that is over two times greater than what is observed in ASD in general. Notably, 12 of those 17 regressions occurred in conjunction with having suffered a fever within a two week period of the regression. However, this regression did not appear to be necessarily linked to vaccinations, as two-thirds of the children that regressed with fever had not received vaccination, and of those who did receive a vaccination, none regressed without also having a fever. Finally, by showing that a subgroup of individuals with mitochondria disorders may be at risk for autistic regression, the publication highlights the continued need for enhanced awareness of the clinical signs of mitochondrial dysfunction.

1. Shoffner et al. *Fever Plus Mitochondrial Disease Could Be Risk Factors for Autistic Regression*. *J Child Neurol*. 2009 Sep 22. [PubMed link: 19773461]

2. Curran et al. *Behavioral Association with Fever in Children with Autism Spectrum Disorders*. *Pediatrics*. 2007 Dec; 120(6):e1386-92. [PubMed link: 18055656]