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AUTISM SPECTRUM DISORDER

Mitochondrial disease in autism

What are mitochondria and what do they do?

Mitochondria are tiny organelles present in each body cell and are known as the “powerhouses” of our cells. They burn carbohydrates, fats, and proteins to provide enough energy in the form of ATP (Adenosine TriPhosphate) for our cells to sustain life and support growth. Mitochondria are inherited directly from the mother, with each ovum or egg containing 100,000 to 1,000,000 mitochondrial DNA molecules. Each mitochondrion has its own DNA (Deoxyribonucleic acid), which is highly vulnerable to damage from free radicals, or reactive oxygen species (unstable molecules) and toxins such as heavy metals.

Body cells can have between 200 and 2,000 mitochondria per cell, but cells in the heart, brain,

muscles, lungs and the gut may have considerably higher concentrations. The human body may have 100 million billion mitochondria in total weighing approximately 10% of total body weight.

If the mitochondria do not function well and cannot convert food and oxygen to make sufficient energy, then diseases and illnesses such as autism, muscular dystrophy, atypical learning disabilities, chronic fatigue, diabetes, developmental delay, Huntington’s Disease, Alzheimer’s, cerebral palsy, bipolar, neurodegenerative disorders, metabolic syndrome, insulin resistance, obesity, Parkinson’s, epilepsy, cardio myopathy, mitochondrial disease and Lou Gehrig’s disease may prevail.

Mitochondria Structural Features

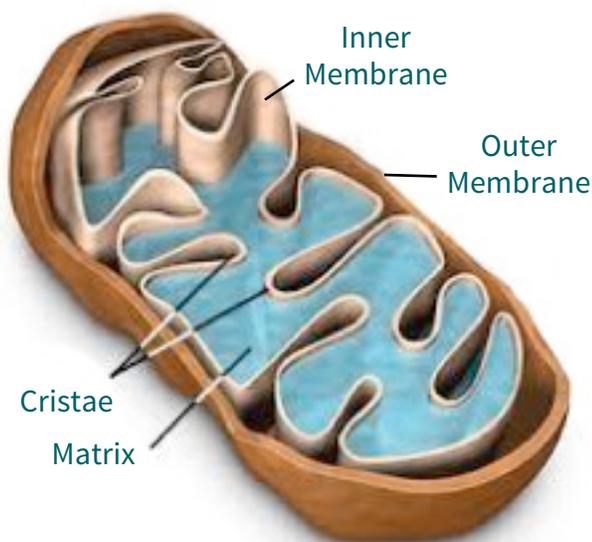


Figure 1

What causes mitochondrial dysfunction?

There are three major causes of mitochondrial dysfunction:

Oxidative Stress induced by excessive free radicals and reactive oxygen species and insufficient antioxidants, especially glutathione.

Environmental toxins including cyanide, carbon monoxide, rotenone, pyridaben, paraquat, 2, 4 dinitrophenol, atrazine, organochlorines such as dioxin, bisphenol A, organophosphates plus toxic metals including mercury, aluminum, arsenic, antimony, lead and an overload of iron and manganese. Pharmaceutical drugs also impair mitochondrial function, specifically from drugs such as acetaminophen, amino glycoside antibiotics, anti retroviral drugs, aspirin, chemotherapy agents, metformin, tamoxifen, valproic acid and statins.

Macronutrient overload especially from fructose, refined carbohydrates, alcohol and excessive saturated fat.

What are the symptoms of mitochondrial disease in autism?

Many symptoms associated with autism may be associated with mitochondrial disease. These include:

- Developmental delays
- Regression following surgery, sedation or anesthesia
- Neurological problems and seizures
- Neuropsychiatric disturbances
- Vision and/or hearing loss
- Absent reflexes
- Increased risk of infection, difficulty recovering from a minor illness such as a cold, flu or virus
- Thyroid and or/adrenal dysfunction
- Gastrointestinal problems such as reflux, constipation, diarrhea, irritable bowel syndrome
- Excessive or absent sweating
- Muscle weakness, poor muscle tone or motor incoordination
- Kidney acidosis
- Hypoglycemia (low blood sugar)
- Pancreatic dysfunction and an inability to produce sufficient digestive enzymes
- Failure to gain weight
- Difficulty handling temperature changes (heat or cold)

There is a common lack of knowledge and information within the medical community about mitochondrial disease, but it should be carefully considered when a child has three or more organ systems with problems. The United Mitochondrial Disease Foundation (UMDF) lists autistic features as a symptom of mitochondrial disease or as disorders of energy metabolism. Mitochondrial disease was thought to be rare, but it has been implicated in many diseases. Mitochondrial medicine and science is still very much in its infancy and little education is

being provided in the medical curriculum. Ironically, autism is still commonly and wrongly viewed as a psychiatric disorder and not as a biomedical disease. A detailed and thorough investigation of all underlying autism associated comorbidities including mitochondrial disease must be identified and treated wherever appropriate.

How is mitochondrial disease diagnosed?

Historically, mitochondrial disease has been difficult to diagnose as many of its symptoms are similar to those of other disorders. It is in your child's best interest to go to an expert in the field of mitochondrial medicine if you suspect he/she has this disorder. The mitochondrial medicine website provides details of qualified professionals <http://www.mitosoc.org>.





Is there a treatment for mitochondrial disease?

Treatment involves supporting the mitochondria with the correct food to provide the fuel required for energy conversion, as well as the removal of inhibiting toxins, drugs and other environmentally sourced chemicals.

The following phytochemicals are supportive of mitochondrial function. Phytochemicals are important non-nutritive compounds found in the pigments of plants that exhibit protective, disease preventing effects against environmental toxins and “bad guys” such as bacteria and fungi.

- Curcumin
- Sulforophene
- Berberine
- Quercetin
- Resveratrol
- Pterostilbene
- Green tea polyphenols

Nutrients that support mitochondrial function are:

- L- carnitine (prescription or pharmaceutical grade)
- Antioxidants including alpha lipoic acid, Vitamins C, E, A and glutathione (broccoli sprouts as a great glutathione stimulant)
- CoQ10
- Magnesium
- NADH
- N-Acetyl Cysteine
- Creatine monohydrate (prescription or pharmaceutical grade)
- Melatonin
- **B vitamins:** especially vitamin B1 (thiamine), vitamin B2 (riboflavin) and vitamin B3 (niacin). Vitamin B5 (pantothenic acid), vitamin B6 (pyridoxine), vitamin B7 (biotin) and vitamin B12 (cobalamin) may be supportive.
- **Amino acids:** especially glutamine, histidine, arginine, proline, glycine, leucine, isoleucine, valine, methionine, tyrosine and phenylalanine





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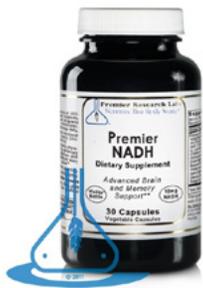
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For more information on current accepted treatments for mitochondrial disease please read “A modern Approach to the Treatment of Mitochondrial Disease” in Current Treatment Options in Neurology (2009). <http://www.ncbi.nlm.gov/pubmed/19891905>

It is important to consult a qualified healthcare provider before commencing any new treatment.