Managing Mitochondrial Dysfunction

The "powerhouse" of the cell and its implications for children with autism and other chronic conditions…

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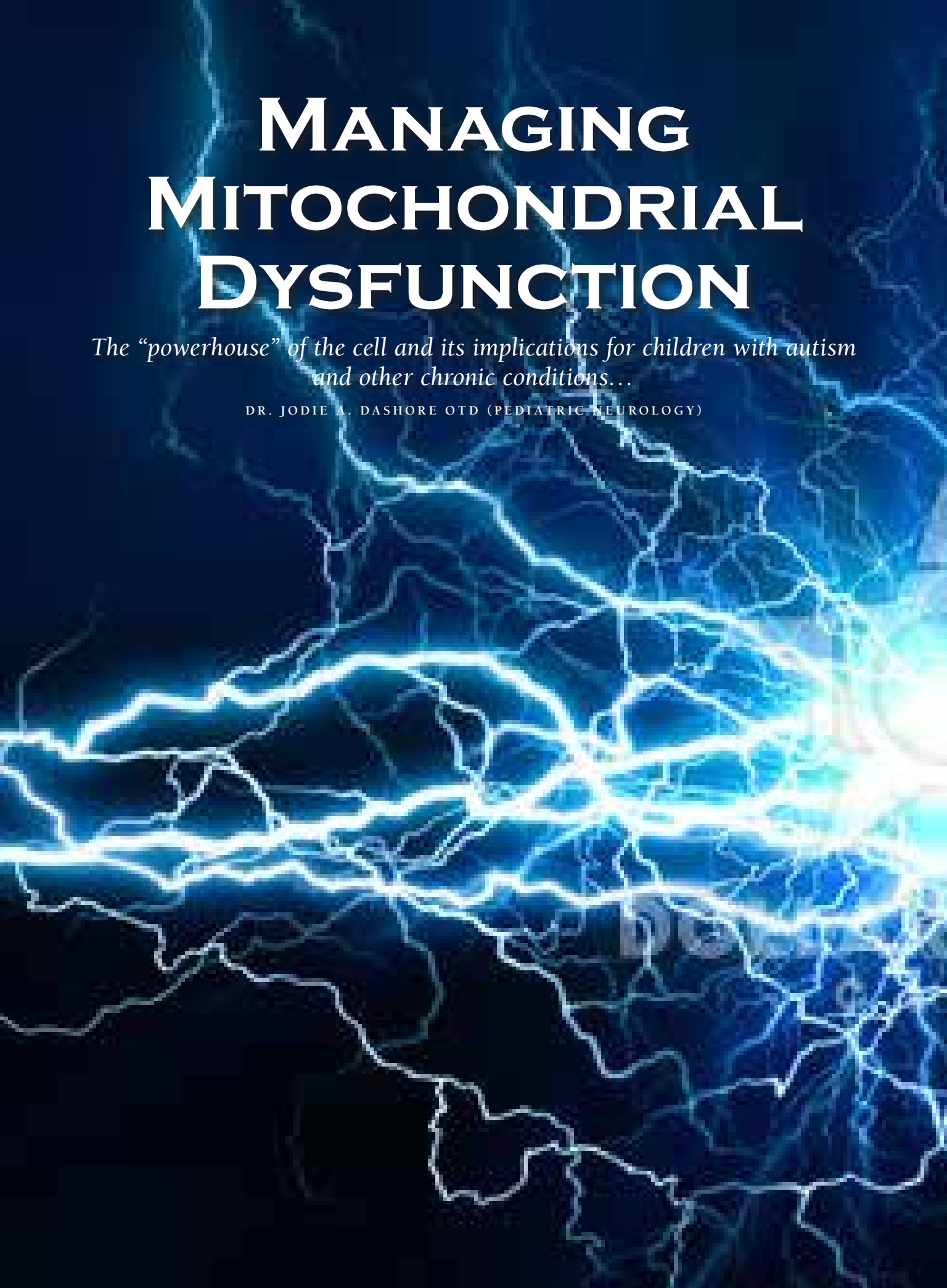

While Mitochondrial Disorders (MD) are known to be genetic in origin, over the last few years research has also looked into identifying epigenetic triggers like vaccination, emotional trauma, etc. Studies show that MD can be a predominant genetic complication in many children diagnosed with autism spectrum disorders (ASD).

On the other hand, there has been increasing evidence and recognition of "acquired" mitochondrial dysfunction (not a full-blown disorder) in children with chronic and/or autoimmune conditions like autism, ADHD, ADD, SPD, Lyme Disease and PANDAS with several environmental, immunological, infectious, and inflammatory factors playing a role. Studies show that substantial percentages of these patients display several peripheral markers of mitochondrial energy metabolism dysfunction. The biochemical abnormalities are usually accompanied by highly heterogeneous clinical presentations which generally include neurological and systemic symptoms relatively unusual in an idiopathic disorder. Evidence is accumulating that these chronic disorders are characterized by certain physiological abnormalities including oxidative stress, and immune dysregulation/inflammation.

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What are Mitochondria?

Let us take a step back at look at what exactly are mitochondria and their normal role in cellular metabolism. Mitochondria are tiny organelles found in almost every cell in the body. These organelles are responsible for creating 90% of cellular energy and are commonly referred to as the "powerhouses" of the cell. They perform lots of different and important functions to keep us healthy. The most crucial role that mitochondria perform is the conversion of energy locked away in food into nutritional energy that the cell can use. In that respect, they act like miniature batteries providing power to the cell for metabolic activities when required. In fact, the main reason we breathe oxygen is so that this process of energy conversion can take place in mitochondria!

Mitochondria are essential for maintaining aspects of physiology as fundamental as cellular energy balance, the modulation of calcium signaling, defining cellular redox balance, and they house significant biosynthetic pathways. These powerhouses provide ATP, a molecule which transports chemical energy within the cell, fuelling cellular processes. This, along with other functions, means that mitochondria are essential for normal cell function including brain function. In fact, mitochondria help to recharge, cleanse, pull toxins out, and maintain cellular metabolism for optimal functioning of all organ systems in the body. They are necessary to maintain life and support growth.

What are Mitochondrial Disorders?

Genetic mitochondrial diseases usually affect tissues which are highly dependent on energy and this includes the brain, heart and muscles. Many patients have involvement of several different tissues, although in some a single tissue maybe involved like the brain tissue in ASD.

Mitochondrial disorders can affect several systemic organs, motor function, and the nervous system. Individuals can experience a wide array of symptoms and degrees of severity. Although it is commonly seen in infants and children, this chronic and genetic disease can develop at any age and diagnosis can be quite difficult.

Mitochondrial disease is when mitochondria in the cells fail to produce enough energy to sustain cell life. When enough cells cease to function properly, organs, motor functions, and the neurological system can become impaired. Research tells us that this often chronic and genetic disease affects one in every 4,000 children by the age of 10 in the United States. Mitochondrial disease is often misdiagnosed due to the fact many of the symptoms are synonymous with other, more common, diseases.

What is Oxidative Stress?

The primary role of mitochondria is the generation of ATP (energy) through oxidative phosphorylation (OXPHOS) and oxygen consumption. Central nervous system (brain) functions strongly depend on efficient mitochondrial function because brain tissue has a high energy demand.

Several studies have shown that oxidative stress and mitochondrial oxidative damage have been implicated in numerous neuro-immune and neuro-degenerative diseases. The critical mitochondrial events responsible for oxidative stress–mediated cell death have yet to be defined. Several oxidative events implicated in toxic oxidative stress include alterations in mitochondrial lipids, mitochondrial DNA, and mitochondrial proteins.

SIGNS & SYMPTOMS OF MITOCHONDRIAL DYSFUNCTION

BRAIN

- Developmental delays
- Dementia
- Neuro-psychiatric disturbances
- Migraines
- Autistic Features
- Mental retardation
- Seizures
- Atypical cerebral palsy
- Strokes

NERVES

- Weakness (may be intermittent)
- Absent reflexes
- Fainting
- Neuropathic pain
- Dysautonomia—temperature instability & other dysautonomic problems

MUSCLES

- Weakness
- Cramping
- Hypotonia
- Muscle pain

DIGESTION

- Gastrointestinal problems
- Dysmotility
- Irritable bowel syndrome
- Gastroesophogeal reflux
- Diarrhea or constipation
- Pseudo-obstruction

KIDNEYS

- Renal tubular acidosis or wasting

HEART

- Cardiac conduction defects (heart blocks)
- Cardiomyopathy

LIVER

- Hypoglycemia (low blood sugar)
- Liver failure

EYES AND EARS

- Visual loss and blindness
- Ptosis (drooping eyelids)
- Ophthalmoplegia
- Optic atrophy
- Hearing loss and deafness
- Acquired strabismus
- Retinitis pigmentosa

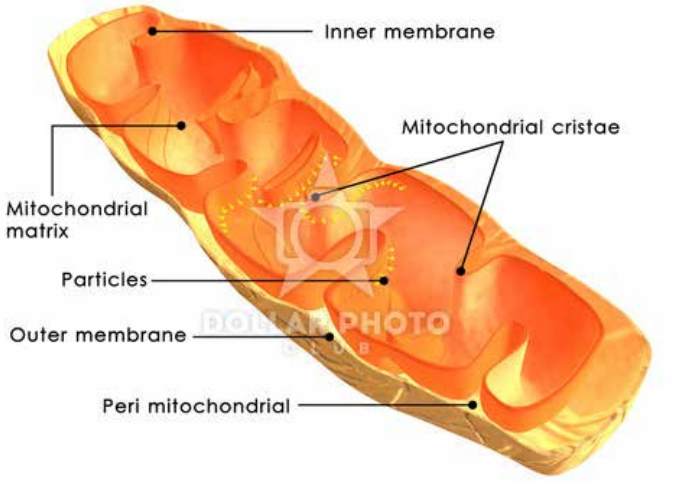
PANCREAS AND OTHER GLANDS

- Diabetes and exocrine pancreatic failure (inability to make digestive enzymes)
- Parathyroid failure (low calcium)

SYSTEMIC

- Failure to gain weight
- Fatigue
- Unexplained vomiting
- Short stature
- Respiratory problems

Source: www.umdf.org



Clinical Pearls

Genetic mitochondrial disorders are possible and certainly seen in clinical practice. However, physicians treating chronic diseases of neuro immune, autoimmune and infectious origin often report suboptimal functioning of mitochondrial functions in many patients as a result of many common triggers.

In my practice, we consider and treat autism spectrum disorders like we would other neuro immune syndromes. Most of the children with autism who I see also have other conditions like PANDAS, Lyme Disease, motor tics, etc. Interestingly, the parents and caregivers of chronically ill children experience PTSD-like symptoms themselves and often present with mitochondrial dysfunction triggered by the enormous amount of stress they have to deal with on a daily basis.

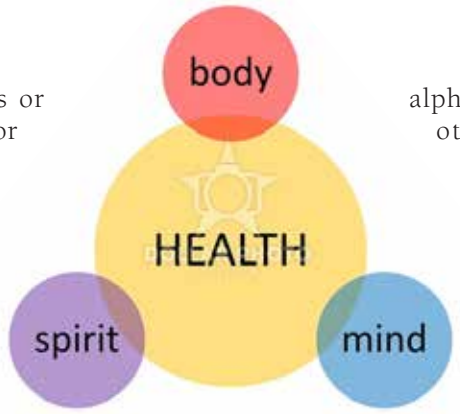
In the last few years, my office has evolved from being a full-time Pediatric Neurology specialty to a full-time all age group chronic neurological illness recovery center. We use functional medicine, European biological treatments, homotoxicology, biomedical plant medicine, preventive measures, and genetically tailored nutritional support strategies to help patients regain their health. Increasingly I find we need to address the entire family as a unit and we have seen phenomenal results when everyone from grandma to the grand child are all coping and healing together.

Biomarkers of Mitochondrial Dysfunction

Recent research has supported a role for mild mitochondrial dysfunction among ASDs. Investigators reported that levels of free and total carnitine and pyruvate were significantly reduced, while ammonia and alanine levels were considerably elevated in ASDs. These are suggestive of mild mitochondrial dysfunction. Some studies show that iodine deficiency may play an important role in the energy metabolism. Therefore, thyroid markers like TSH, anti TPO antibodies, T3, T4, T3 reverse can also contribute to the final treatment plan for a patient.

Another test that's useful is the patient's methylation profile and intracellular micronutrient analysis. These give us a look into enzymatic defects that may be contributing to mitochondrial issues and the intracellular

concentration of essential nutrients or the lack there of. Laboratory tests for microorganisms are widely available based on the organism suspected by a practitioner and can include bacteria, viruses, fungi, parasites, and spirochetes. Mold biotoxin illness is another piece to the puzzle and worth investigating if there is a history of exposure to water damaged buildings and mold per se.



Re-igniting the Spark

A growing body of evidence now indicates that mitochondrial dysfunction and oxidative stress have central roles in chronic disease pathogenesis. The treatment of mitochondrial dysfunction varies considerably. Most experts use a combination of vitamins, optimization of patients' nutrition and general health, and preventing worsening of symptoms during times of illness and physiologic stress.

Current research suggests that the link between these two diagnoses may very likely be greater than suspected. As a result, the paradigm for evaluation of children with autistic symptoms is changing. Mitochondrial experts have stated that some mitochondrial diseases are potentially both genetic and environmental in origin. And some believe there is a genetic predisposition with an environmental "trigger" (such as fever or illness) in some cases. (See sidebar for additional triggers.)

Appropriate identification of children with mitochondrial disease and autism may improve their overall outcome. The concept of mitochondrial therapy is a new approach, but it is being intensively tested. Coenzyme Q10 (CoQ10) is a naturally occurring antioxidant that affects mitochondrial depolarization and acts as an electron transporter for mitochondrial complexes. CoQ10 levels are low in mitochondria that have been isolated from patients, and the ratio of oxidized to reduced CoQ10 is greater in patients than in controls, suggesting increased oxidative stress in the former. A large clinical trial is now planned to test the potential of CoQ10 as a disease-modifying agent.

Clinical trials have also shown the utility of using oral replacement supplements, such as L-carnitine, alpha-lipoic acid, NADH (reduced nicotinamide adenine dinucleotide), membrane phospholipids, and other supplements. Creatine, in the form of phosphocreatine, a high-energy phosphate, buffers cellular ATP and prevents opening of the mitochondrial permeability transition pore. Combinations of these supplements can reduce significantly the fatigue and other symptoms associated with chronic disease and can naturally restore mitochondrial function, even in long-term patients with intractable fatigue.

Therapies targeting basic mitochondrial processes, such as energy metabolism or free-radical generation, or specific interactions of disease-related proteins with mitochondria, hold great promise. These aim to provide the body with the a large variety of intracellular antioxidants, sustained-release preparations of super antioxidants, minerals, and amino acids known to enhance the functional capability of the mitochondria. This assists patients who have mitochondrial dysfunction typically from methylation deficiencies, toxic exposures, and poor nutritional delivery.

Currently, these clinical treatment approaches focus on improving metabolic support and mitochondrial function through use of vitamins and supplements called the "Mito cocktail." Energy management conservation and other supportive care are equally important. Underlying infectious etiology and triggers need to be eliminated to achieve lasting relief.

In my office, the guiding principles that have worked the best are based upon the pioneering work of my mentor, Dr. Dietrich Klinghardt. This approach utilizes the best of European Biological Medicine enmeshed with ancient Eastern wisdom.

Environmental Triggers

A growing number of experts on mitochondrial diseases believe that a genetic predisposition couple with an environmental trigger precipitates dysfunction. These triggers include:

Some of these triggers are:

- Long term medications like antibiotics and chemo therapy
- Viruses like Epstein Barr
- Bacteria Like group A beta hemolytic strep, Mycoplasma species and more
- Tick borne Infections like Lyme Disease, Bartonella Henselae, Babesia Microti, etc
- Methylation blockages and key enzyme deficiencies
- Exposure to environmental toxins
- Physical or psychological trauma and stress

TOXIC WARNING

Addressing various sources of toxins is very important and often missed. Toxins can come from the earth (gases entering the house (uranium etc.), toxic building materials (particle board outgases glues), treated wood (outgases arsenic), and other wood dyes and preservatives. Railroad ties often used in playgrounds are notorious for being extremely

toxic. There may be old issues like lead in wall paint, or asbestos in roofing material which slowly leaches out. Marble and granite in your kitchen can both emit radioactivity. Non-breathing building materials can lead to mold and other problems.

Infections come inside the house in many ways such walking inside without taking your shoes off or washing your hands. Mold may be slowly growing in air duct systems, and moisture in the home can facilitate microbial growth.

Naturally occurring problems like geographic stress (underground streams and ionizing radiation), Hartmann and Curry grid lines, places of unusually low or high magnetic fields, and natural radioactivity should all be considered.

Attention, selfless—and very tired—caregivers!

On a more spiritual level, especially when whole families are affected, I find there is a natural progression in the consciousness of self and spiritual development. I like to consider the Mind Body Spirit approach for patients of all age groups for long term positive results. A young soul is severely affected by all these issues but not aware of them and thus doesn't ask for or seek out help. A person beginning to wake up spiritually is aware of these issues but still affected in the same detrimental way. However, it is commonly seen that spiritually evolved people are aware of these issues but less affected by them.

It is important to get a support system. Explain to people around you that your child has a biological—not a behavioral—condition. Know in your heart that the child's illness is and always was the product of a society that has become too toxic for the healthy environment of a sensitive child.

Don't lose your love in the struggle, and if you are at the end of your strength, then pick yourself up and keep seeking and finding the answers. Communicate with your spouse, your child's teachers, counselors, and doctors. Envision your child improving. Try to practice positive thinking—don't give up, but find alternate strategies and implement them. Don't hand things over to someone else as you have the best healer within yourself. Accept what it is. It helps tremendously to trust that your experience is the right one at this time and that you—only you—can change it in time.

For those who like to sometimes self-treat, themselves or their children, without consulting a practitioner: please remember that anything you add to the treatment that is not necessary will dilute the treatment. Try to obtain and maintain custom protocols and regular bio-energetic retesting. Don't accept your suffering—change it! ◂

Nathan: The Disappearing Little Boy

A case study

Nathan is a seven-year-old little boy with high functioning ASD. He has been under the care of a DAN! physician since he was diagnosed at the age of two. When Nathan was five years old he went on a field trip to a local nature reserve with his special needs class from school. About six months later, his mother noticed Nathan had become increasingly fatigued and disinterested in his surroundings. Nathan then started to get thinner, developed aggressive behavioral issues and in spite of a normal even heavy appetite and a great diet full of good fats, Nathan was unable to gain weight. Nathan's nutritionist was almost ready to throw in the towel, discouraged after months of unsuccessful strategizing.

Nathan continued to decline when I first saw him. To make a long story short, we found underlying viral infections and several methylation polymorphisms including the MTHFR. Nathan's Nagalase levels were very high and his white blood cell and platelet counts were low. His eosinophils were very high indicating underlying possible parasitic infection.

We found out that Nathan's school aide had removed two ticks from Nathan's body during the field trip I mentioned earlier. Both the ticks were attached but came off easily. The aide assumed since she took them off Nathan was ok and that tick attachments were a "normal" part of a trip into the woods so she didn't report it to the teacher. As a result—unknown to anyone—Nathan had acquired Lyme Disease which is known to suppress the immune system. That also explained how Nathan's viral infections became chronic. Nathan's special labs for tick borne infections were positive.

Bio-energetic testing revealed Nathan's viral infections were intracellular and were affecting his cellular metabolism and mitochondrial function. He was put on plant medicine-based antimicrobial therapy along with mitochondrial supplements custom tailored to his methylation/genetic make-up. Biomedical detoxification and organ support for the liver, the kidneys, and the lymphatic system were also put in place to help minimize any herxheimer reaction type healing crisis.

Within four months, Nathan's activity level slowed down to a more normal level, his brain processing also slowed down, his mind was no longer racing, and he was able to better organize his thoughts and could once again communicate his needs.

Nathan has gained 10 lbs. in the last six months and is a happy, healthy and playful little boy with a twinkle in his eyes. He's now the same as he was prior to his immune and mitochondrial dysfunction. Quite a few of his food sensitivities have also decreased and he has a more varied diet to choose from. I believe many of Nathan's spectrum issues are related to underlying infections and mitochondrial dysfunction. He has a high probability of being mainstreamed in the next year or two, once his body and brain heal from the various biochemical imbalances.

In the last two years, I have treated 37 children with ASD. All 37 had mitochondrial dysfunction of varying degree, and 28 were found to have underlying chronic infections. All continue to gain higher functional capacity and are doing well with 11 of them now receiving mainstream education.