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Links

www.mitoaction.org
www.umdf.org
www.clinicaltrials.gov
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Practical Information For the School Setting: One Mito Parent's Perspective

Contributed by Stephanie Shapiro

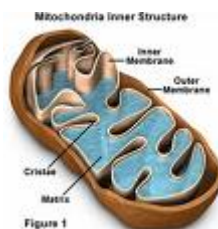
Preparing your child for a new year of school can be difficult in itself: new teachers, new materials, back to school clothes, etc, etc. Back-to-school prep for your child with a mitochondrial disorder can be especially daunting to say the least! In order to start the school year off right and to stop issues before they arise, here are a few simple suggestions for a happy and HEALTHY school year:

- 1) Educate, Educate, Educate! Be sure all the members of your child's team is knowledgeable in his/her disorder. Provide the school with multiple copies of **MITO 101** and ask for a pre-school-year meeting to answer any questions the administrative team may have about your child and mitochondrial disease. You may want to ask a mito "buddy mom" to attend this meeting with you to add additional information or suggest your school conduct a Mito 101 learning session with all faculty, staff, and nurses. Gently insist that **not all children with mito manifest the symptoms of their disease in the same way**. Just like no two children are the same, no two children with mito are the same!
- 2) Prepare a "day in the life" care plan for your child. Include best room temperature for your child, your child's baseline body temperature (which may be lower than a typical child's 98.5 degrees) the need for a water bottle and snacks to insure your child does not become dehydrated or present with low blood sugar. Also include best times of the day for educational and therapeutic lessons, fatigue issues, signs when a "time out to rest" may be in order, etc. Ask your doctor to add additional pertinent information.
- 3) Keep an open, positive, and direct line of communication with your child's teacher. Don't wait for teacher conferences, IEP or 504 meetings to discuss issues or concerns.

So, it is as easy as 1, 2, 3 ! Be proactive and hopefully there will be few times you will have to be reactive! Have a great school year and if you need support contact the UMDF at www.umdf.org

Autism and Mitochondrial Disease

Autism is a complex neurobiological disorder that typically lasts throughout a person's lifetime. It is part of a group of disorders known as autism spectrum disorders (ASD). Today, 1 in 150 individuals is diagnosed with autism, making it more common than pediatric cancer, diabetes, and AIDS combined. It occurs in all racial, ethnic, and social groups and is four times more likely to strike boys than girls. However, an underlying diagnosis is established in only 2-36% of cases.



Autism impairs a person's ability to communicate and relate to others. It is also associated with rigid routines and repetitive behaviors, such as obsessively arranging objects

Transgenomic would like to welcome Dr. Fran Kendall back into practice and best wishes on the launch of Virtual Medical Practice!

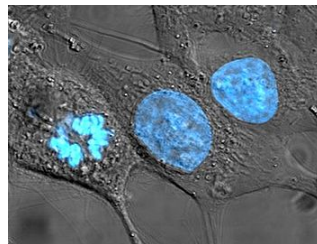
Transgenomic is a specialty genetics laboratory that excels in ultra high sensitivity genetic testing with a focus on Mitochondrial Disorders. We offer a broad range of genetic testing in this area for both mitochondrial, nuclear gene and Chromosomal Microarray testing. We also accept Medicaid in a number of states and will work with patients to get the maximum reimbursement from their insurance companies. Please contact David Keane at (404) 538-2883 with any questions.

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or following very specific routines. Symptoms can range from very mild to quite severe.

A number of genetic and metabolic disorders have been shown to be a cause of or associated with autism spectrum disorder, including mitochondrial disorders. One population based study suggested that 7.2 patients out of 100 with autistic spectrum disorder have an underlying mitochondrial disorder making it still rare but one of the more common definable causes of ASD. Several reports have shown specific mitochondrial DNA abnormalities associated with ASD including one study which evaluated five patients with ASD and family histories of mitochondrial DNA diseases. Three patients manifested isolated autistic features and two had additional neurological findings. Two of the patients were found to harbor the common MELAS A3243G mutation and another had mtDNA depletion as a cause for their difficulties. Weissman et al recently reported the association of ASD with the mtDNA A4295G mutation in a 15 year old with a number of other neurological findings including hearing loss. Overall, these various studies indicate that while likely a rare cause of ASD, mitochondrial disease can be considered when associated with other neurological complications and/or a family history of mitochondrial disease.

NIH study: Exercise benefit in mito disease?



Mitochondrial myopathies are caused by mutant mitochondrial DNA, genetic defects in parts of the mitochondrial DNA. These defects can include missing or deleted DNA that typically codes for certain proteins involved in energy production. These mutations cause individual mitochondria and the body on a whole to produce energy less efficiently. Because muscle cells require extensive energy to

function properly, they are particularly impaired by mitochondrial dysfunction. The onset of most mitochondrial myopathies occurs before the age of 20. Initially a person may experience muscle weakness and fatigue during physical activity. Other symptoms may include limited eye mobility, heart arrhythmias, slurred speech, swallowing difficulties, and impaired movement.

There is no cure yet for mitochondrial myopathies, nor is there any adequate treatment to stall disease progression. Exercise, known to boost the production and function of mitochondria in healthy people, may reduce symptoms in people with mitochondrial myopathies by increasing the number and function of normal mitochondria in an individual muscle cell. The purpose of this study is to determine the effects of exercise training versus inactivity on the expression of normal and mutant mitochondrial DNA and on mitochondrial production within muscle cells in people with mitochondrial myopathies. The study will also assess how cell function, physical endurance, heart function, and quality of life are affected by exercise training and inactivity.

Participants in this 2-year study will first undergo physiological exercise testing, magnetic resonance imaging (MRI) of heart and skeletal muscles, a needle biopsy of muscle, and a questionnaire on quality of life. Participants will then be randomly assigned to partake in regular exercise training or no training for 6 months. After 6 months, all participants will undergo repeat testing of initial evaluations. Participants who had been in the exercising group will then switch to no exercise training for 6 months, and participants who had been in the non-exercising group will switch to regular exercise training for 6 months. The second 6-month period will also be followed by repeat testing of initial evaluations. Participants will then be encouraged to continue exercise training for an additional 1 year, with retesting at the end of the second year. Each of the four evaluations will take about 15 hours over 5 days.

For more information go to www.clinicaltrials.gov