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Bridging the Gap Between Mito & Autism

Although several studies completed by a group in Portugal in 2005 & 2007 suggested that 7.2 out of 100 autistic patients then later 4.1% of patients with autism had underlying mitochondrial disease, rendering it a rare but definable cause of ASD, a recent study out of UC Davis suggests a much stronger link between autism and mitochondrial dysfunction.

Published in the Journal of the American Medical Association (JAMA) in November 2010, the authors report that children with autism are far more likely to have defects In their bodies' ability to produce energy than typically developing children. Selecting 10 autistic children (who met stringent diagnostic criteria for ASD) ages 2 to 5 years and 10 age matched typically developing children from similar socioeconomic backgrounds as representative of the larger 1,600 participant Childhood Autism Risk from Genetics and the Environment (CHARGE) Study in Northern California, the researchers obtained blood samples from each child to complete lymphocyte (type of white blood cell) mito studies.

The study discovered widespread reduced mitochondrial enzyme function among the autistic children. Complex I was the site of the most common deficiency, found in 60% of the autistic patients, and occurred five out of six times in combination with Complex V. Other children had problems in Complexes III and IV. In addition, the autistic children showed other signs of mitochondrial impairment including elevation of blood pyruvate (a compound that is fed through the enzyme pyruvate dehydrogenase suggesting this may be compromised) and, increased hydrogen peroxide levels (sign of increased free radicals). Half of the autistic children also had higher mtDNA copy numbers in the lymphocytes, the latter a likely response to oxidative stress. When mito are functioning poorly particularly due to damage, the body produces extra copies of DNA to help promote more normal functioning. Two of the five children also had deletions in their mtDNA genes. None were found in the control children. All of these signs point to impaired mitochondrial functioning.

Despite these advances in understanding, many questions remain to be answered. Did the mitochondrial impairment occur before or after birth in these children? Does the autistic population studied have additional mitochondrial or nuclear gene defects? Ongoing testing and investigation is underway. Regardless, the study results point to a stronger link between mitochondrial dysfunction and autism than was previously believed to exist. Importantly, this association was established utilizing a cell population (lymphocytes) that is easily obtainable via a blood draw. Like buccal swab mito enzyme testing,

Biopsy Talk - MitoAction

You may listen to a MitoAction.org podcast of Dr Kendall lecturing on the ins/outs of biopsies and taking questions from monthly listeners at:

- MitoAction.org website
- ITUNES/Podcasts
- Resource Tab on www.virtualmdpractice.com

Facebook

To keep up with the latest news, "like" us on Facebook at www.facebook.com/vmpDrKendall

Links

http://mayoresearch.mayo.edu/mitochondrial-disease-biobank

www.mitoaction.org

www.umdf.org

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this non-invasive study opens the door to widespread access of mito testing to a large patient population without the risk, cost and invasiveness of traditional muscle biopsies.

MEDomics Expands Mito Gene Testing

Primary mitochondrial disease affects the intrinsic ability of the mitochondria to function properly by altering how it's many components, like pieces to a puzzle, come together to create energy. Primary mitochondrial disorders are caused by changes in mitochondrial or nuclear genes. Genes are our genetic blueprint. They are units of heredity that determine everything about us including how tall or short we are, the color of our hair and eyes and whether or not we make all of the many proteins that ultimately come together to make our energy packets known as ATP.

For about twenty years we have known a lot about the mitochondrial genes or those inherited exclusively through our mothers. These 37 mitochondrial genes are housed inside the egg cells of our moms. The remaining genes involved in energy production are inherited or passed on to us through both parents, are locating in the center of our body cells known as the nucleus, and consist of hundreds of genes. Up until very recently, technology allowed us to look at only a handful of these nuclear genes.

A number of studies suggest that 75% to 90% of mitochondrial disease in pediatric patients is due to changes in the nuclear genes inherited through both of our parents. As such, DNA testing available through most of 2010 did not allow us to find the gene causing mitochondrial disease in all patients.

All of this is about to change. In November, Medomics LLC, rolled out expanded nuclear gene testing offering the first run to several practices around the country, including VMP. This testing will look at up to 700 out of the 1500 genes involved in mitochondrial energy production. This will include the genes encoding for the proteins in Complex I, II, III, IV and V and support genes such as OXPHOS complex assembly factors, OXPHOS cofactor synthesis, and mitochondrial genome replication/transcription factors, including the 86 ribosomal protein genes. These noninvasive studies are a huge step forward towards eliminating the need for more invasive testing, specifically biopsies. This will also negate the ambiguity that can come with those often not-so-clear testing results. Ultimately, identifying the gene cause for each patient's specific mito disorder will lead to better treatment for and understanding of disease sub-types. To determine if you, your family member or other loved one may benefit from this expanded testing contact your health care provider.

Case Study: Clinical improvement due to aggressive dietary & hydration management

Dealing with teenagers can be challenging regardless of whether they have an underlying mitochondrial disease, other chronic disorder or are merely “normal” teens. However, even one of our 14 year old patients had to admit he felt much better after introduction of an aggressive dietary and hydration regimen aimed at improving his clinical symptoms. This young man had long standing myopathy (muscle weakness), ptosis (drooping eye lids) and ophthalmoplegia (inability to move his eyes) and was struggling with worsening symptoms and overwhelming fatigue.

Follow-up multisystem screening studies noted his lactate level to be increasing and his CPK (muscle enzyme) to be ten times normal. Both studies indicated that his body was struggling to function due to worsening ability to produce energy. A review of his history indicated poor daily hydration and a variable diet with intermittent food intake. We introduced multiple small frequent meals with snacks, including one at bedtime and daily hydration with Gatorade with the intent of providing his body with a more constant source of energy and less stress due to prolonged fasting and/or poor hydration status.

In follow-up several months later, his CPK & lactate levels had essentially normalized with improved stamina and less fatigue!! Several other patients have been treated with similar aggressive therapies and have also noted an improvement in their daily lives and functioning. Aggressive management by physicians who understand mitochondrial disease can and does improve the lives of affected patients!

Raising Awareness One Lecture & Event at a Time

VMP and Dr. Kendall strongly believe in education and awareness as a critical component in the fight against mitochondrial disease. Knowledge is power to patients, families and the medical community.

To support these efforts our calendar is quickly being filled with various engagements around the country to include the following:

- March/Atlanta - Mitochondrial Disease Overview for the Emory School of Nursing students;
- May/Chicago - Mito & Autism Lecture for the national Autism One Conference
- June/Chicago – Chairing panel on trends in Mitochondrial testing at the international UMDF Symposium
- October/Charlotte – Answering questions as a VIP at the 2nd Annual Carolina Foothills Chapter UMDF Walk for Life
- December/Atlanta – Guest lecturer on Mitochondrial and Genetic medicine for PCOM Medical Students.

If you have an event or lecturing opportunity and would like to request our involvement please contact us at info@virtualmdpractice.com