Hot Topic in Mitochondrial Disease...

Autism and Mito
What is Autism?

- A complex neurobiological disorder that typically lasts throughout a person’s lifetime, is a part of a group of disorders known as autism spectrum disorders (ASD) and affects the ability to communicate and relate to others.
- Also associated with rigid routines and repetitive behaviors.
- 1 in 90 individuals is diagnosed with autism making it more common that pediatric cancer, diabetes and AIDS combined.
- Occurs in all racial, ethnic and social groups and is 4 times more likely to affect boys than girls.
- An underlying diagnosis is established in only 2% - 36% of cases.
Mito Disorders and Autism

- One 2005 population based study in Portugal suggested that 7.2 out of 100 patients with ASD have an underlying mito disorder.

- A 2007 study by the same group revised their population figures and noted 4.1 out of 100 patients with autism had underlying mitochondrial disease.

- Although mito appears to be a rare cause of autism, it is one of the more common definable causes of ASD.
Case Study: One

One study evaluated five patients with ASD and family histories of mitochondrial DNA diseases.

- Three patients had isolated autistic features and two had additional neurological findings.
- Two patients had the common MELAS A3243G mutation.
- One patient had mtDNA depletion.
Case Study: Two

Weissman et al 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.
Children with ASD are far more likely to have a defect in their ability to produce energy than typically developing children.

Discovered widespread reduced mitochondrial enzyme function among autistic children, affecting complex I in 60% of the patients.

Association established utilizing WBC (lymphocyte) testing.
Children on the autism spectrum also reside along a spectrum of mitochondrial dysfunction of varying severity.

Emphasized the need for ASD children to be screened for possible mitochondrial dysfunction citing improvements in children with ASD & mito dysfunction after initiation of mito disease management.
CONCLUSION OF STUDIES

- The link between mitochondrial dysfunction and autism is greater than suspected.
- It remains uncertain if this association is due to a primary defect in mitochondrial functioning due to gene mutations or dysfunction caused by other factor(s).
- Mitochondrial disease should be considered when associated with other neurological and body system complications and/or a family history of mitochondrial disease.
Recommended Evaluation for ASD Patients

Tier 1 — basic work up recommended for all patients

- Chromosome Microarray Studies
- Complete Metabolic Panel, CBC, CPK
- Ammonia Level
- Lactate and Pyruvate Levels
- Carnitine, Plasma Total and Free
- Coenzyme Q10 Level
- Plasma and Urine Amino Acids
- Urine Organic Acids
- Plasma Acylcarnitines
- Thyroid Function Tests

List is located on our website, www.virtualmdpractice.com
Recommended Evaluation

For ASD Patients

- Tier 2 — depends on clinical features & results of Tier 1 testing
  - Mitochondrial Enzyme and/or DNA Testing
  - RETT Syndrome DNA Testing
  - PTEN Mutational Analysis
  - NLGN3, NLGN4X, SHANK3, SNRPN Gene Testing
  - Lysosomal Enzyme Testing
  - Peroxisome Disease Testing (VLCFAS)
  - CSF Studies for Lactate and Pyruvate, Amino Acids and Neurotransmitters
  - Brain MRI

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Why is it important to know if an ASD patient has mito?

- For implementation of treatment & protocols
- Monitoring in affected individuals
- To determine recurrence risks for future children
- To determine risk for other family members
Mito, Autism, and Vaccines
THANK YOU!

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