

Mitochondrial Dysfunction in Autism: Effect of Mitochondrial Supplements

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Emerging research has suggested that mitochondrial (mt) dysfunction plays a role in the pathogenesis of some patients' autism spectrum disorder (ASD). Using buccal swabs to analyze respiratory chain complexes (RCC) I (through immunocapture) and IV (with spectrophotometry), we found that 42% of 92 children with ASD had dysfunction of these RCC. We also demonstrated that RCC variations are related to ASD symptoms and that supplementation of folate or fatty acids improved mt function in ASD patients. Recently, we completed an FDA-IRB approved open-label pilot trial with a combination of carnitine, coenzyme Q10 and alpha-lipoic acid *(MitoCocktail)*. We hypothesized that patients with ASD and mt dysfunction would improve biochemically, and/or clinically, following supplementation with *MitoCocktail*. Eleven (10M/1F) ASD patients, aged 5-12 years, were studied at baseline (T1), 3 months on treatment (T2) and 3 months off treatment (T3). Mean buccal complex I/IV ratio was significantly reduced ($p<0.02$) at T2. Metabolic improvement waned at T3 in 7/11children. Neurobehavior was evaluated with Autism Diagnostic Observation Schedule (ADOS-2), Autism Spectrum Rating Scale (ASRS) and Aberrant Behavior Checklist (ABC). The 11 total or subscales scores provided showed improvement in means at T2, being statistically significant for Unusual Behavior from ASRS ($p<0.006$), and Lethargy ($p<0.01$) and Speech subscales ($p<0.02$) from ABC. At T3, these scores worsened resulting statistically significant for Lethargy ($p<0.01$) and Inappropriate Speech ($p<0.007$). We conclude that the use of mt supplements supporting mt function have therapeutic benefit, improving maladaptive behavior and speech in ASD children. Larger placebo-controlled trials are needed to demonstrate efficacy.

Biography

Agustin Legido obtained his MD deegree from Zaragoza University (Spain) in 1980. He received one PhD from The University of Bologna (Italy) in 1982 and one from the University of Zaragoza in 1985. Since 1985 he has been in the USA, where he is currently is Professor of Pediatrics and Neurology at Drexel Univesity School of Medicine (Philadelphia, PA). He is the Chief of Neurology at St. Christopher's Hospital for Children (Philadelphia, PA). He has published more than 200 papers in reputed journals, and presented more than 300 abstracts to scientifc meetings. He is the Editor-in-Chief of Seminars in Pediatric Neurology.

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