

Autism, Epilepsy and Mitochondrial Disease

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Autism spectrum disorders (ASD), also called pervasive developmental disorders, neurodevelopmental disorders are including a group of processes that occur as common characteristic impaired social interaction, processes verbal and nonverbal language and existence of repetitive behaviors (stereotypies), with limited activities and interests [1]. Autism frequently has partnered with other processes or symptoms of neurological dysfunction such as epilepsy, motor disorders, hyperactivity, hypotonic, mental retardation, which we think that is a disorder neurobiological heterogeneous etiology [2].

It has been estimated the incidence of autism is case in 90 children, in United States. Nevertheless in only 15-40% of cases it can be shown etiology a medical well there may definite. It is a disease or syndrome related to autism, without implying causal etiological a relationship between both problems for it can only establish the concept of autism syndrome or secondary when we establish a clear causal relationship.

There are numerous problems causing autism where we note the following: chromosomal, neurocutaneous syndromes, congenital malformations of the central nervous system, neuro-metabolic disorders, ranging from the aminoacidopathies organic acidifies, disorders neurotransmitters, The deficit cerebral glucose transporter, lysosome diseases, prematurity congenital infections, acquired infections central nervous system, to name a few.

Since in 1985 Coleman demonstrated the relationship between autism and defects oxidative phosphorylation, there has been a growing case load identified as "Mitochondrial autism," defined as alteration or dysfunction mitochondrial respiratory chain of patient recognized as autism [3].

The first occurrence estimated impact of autism in children with mitochondrial disease in 1 of 110 and mitochondrial disease in children with autism of 1 in 2000 autistic, it seems much higher than estimated currently [1,4]. This probable higher incidence suggests possible pathogenic relationship.

Frye showed that over 50% of children with autism spectrum disorders disease mitochondrial had biomarkers abnormal. In another study showed impaired role of respiratory chain mitochondrial only just over 20% of children with disorders autism spectrum and mitochondrial disease had a known mutation of the Mitochondrial DNA [5].

In 1998, Lombard, considered could be the result of autism mitochondrial dysfunction, due to impair oxidative phosphorylation of neuronal this because of the high incidence of lactic acidosis and deficiency carnitina he found in mitochondrial dysfunction autistic patients. It could be caused by environmental factors exogenous-toxic or genetic the individual himself [6].

Also autism has been linked to mutations in mitochondrial DNA point as are mutation A3243G, responsible for most cases of MELAS and mutation G8363A, who has engaged in children with autistic regression and epilepsy. It has been considered that the dysfunction or mitochondrial disease is the cause of metabolic origin more frequently found in patients with autism spectrum disorders.

HEADD Syndrome, define association hypotonic, epilepsy, autism

and psychomotor retardation associated to mitochondrial dysfunction, mainly of respiratory chain complex or mitochondrial mutations point would not seen as common type Melas, Merrf, Leigh o Kearns- Sayre [7].

The clinical features are important to know some that allow a disease mitochondrial. An important number of autistic children have regression, about 50% multiorgan involvement and present, several organ systems are affected.

The relationship of epilepsy autism is well established in 10-40% of autistic children will develop any type of epilepsy increase. We know that the incidence of epilepsy among autistic patients or has a bimodal distribution with a higher incidence among children under 5 years and a second peak in incidence after 10 years. The factors increasing the greatest risk for development of epilepsy in an autistic child is partnership with selected etiologies, as altered chromosomal neurocutaneous diseases such as tuberous sclerosis or birth defects including metabolism mitochondrial diseases [1].

Special consideration deserves the Landau-Kleffner syndrome, a syndrome characterized fundamental for display an acquired aphasia and encephalographic disorders that can reach electric slow status during sleep in children previously normal. The particular interest of this syndrome, It is the children may occur with autistic traits, like how disruptive behavior, the tendency to "kicking tantrum" stereotypies and then we may lead to think about autism, discarding other pathologies such as epilepsy and eventual association with dysfunction mitochondria. In these cases of Landau-Kleffner syndrome have demonstrated the efficacy of ACTH, the clinical aspects such as encephalographic [8].

The association between autism, epilepsy and mitochondrial disease is uncommon, but in the group of children with autism regression or which are accompanied by signs of neurological involvement such as mental retardation, hypotonia, ophthalmoplegia, dysmorphic features, this possibility should be considered.

Technical limitations and availability for the conduct of laboratory tests, biochemical, study of the mitochondrial respiratory chain and molecular genetic analysis mitochondrial DNA is a limiting progressively to be beating to achieve the greatest possible number of diagnostics to allow adequate and better forecast for autistic children attention.

The objective of trying to establish a genotype-phenotype correlation of the diseases or mitochondrial dysfunctions in children

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with autism will result, an extraordinary in order to achieve a better understanding of genes involved and its relationship with epilepsy and autism.

Finally, I would like to highlight the importance of encouraging this knowledge among neuropediatricians to achieve a greater number of cases and diagnostics, stimulate research into these diseases.

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