References.

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NEUROMETABOLIC CAUSES OF INFANTILE SPASMS

Investigators at King Abdulaziz Medical City, Riyadh, Saudi Arabia, studied the prevalence of hereditary neurometabolic causes of infantile spasms in 80 cases presenting over a 15-year period. Of 10 patients (12.5%) diagnosed with metabolic causes, 2 had a Leigh-like disorder, and 1 patient had each of the following diagnoses: ethylmalonic aciduria, nonketotic hyperglycinemia, hyperinsulinemic hypoglycemia, leukodystrophy, short-chain acyl-coenzyme A dehydrogenase deficiency, molybdenum cofactor deficiency, primary carnitine deficiency, and neonatal hypoglycemia due to panhypopituitarism. Most of the patients were born of consanguineous parents, and the hereditary group had a strong history of other family members affected. The typical hypsarrhythmia pattern in the EEG was more common in the hereditary metabolic group (P=0.003), and this group had a poor response to therapy (P=0.04). Metabolic disorders are a relatively common cause of infantile spasms in this subpopulation of Saudi patients. Early diagnosis with metabolic and genetic testing is important in selection of specific treatments and facilitating family counseling. (Alrifai MT, AlShaya MA, Abdulaban A, Alfadhel M. Hereditary neurometabolic causes of infantile spasms in 80 children presenting to a tertiary care center. **Pediatr Neurol** 2014 Sep;51(3):390-7).

COMMENTARY. In patients suspected of having a hereditary metabolic cause for infantile spasms, the authors recommend a more liberal application of advanced diagnostic techniques, such as whole exome sequencing, muscle biopsy for mitochondria biochemical and genetic studies, and newer neuroimaging techniques such as 3 Tesla MRI and PET scanning [1]. More extensive genetic testing is justified in higher risk populations where high consanguinity rates are prevalent. A review of etiology of infantile spasms in the United Kingdom where consanguinity is rare finds the common causes are hypoxic-ischemic encephalopathy (10%), chromosomal anomalies (8%), malformation (8%), perinatal stroke (8%), and tuberous sclerosis complex (7%) [2][3].

References.

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DEVELOPMENTAL DISORDERS

AAP GENETICS DIAGNOSTIC APPROACH TO INTELLECTUAL DISABILITY OR GLOBAL DEVELOPMENTAL DELAY

The American Academy of Pediatrics Committee on Genetics present a recommended clinical genetics diagnostic approach to the evaluation of intellectual disability or global developmental delays. The report addresses the advances in diagnosis and treatment of children with intellectual disabilities since the original AAP report in