

Diagnostic Criteria For Pediatric Mitochondrial Disorders

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Our body's nucleated cells contain 500-2000 mitochondria. In the liver mitochondria are specialized to detoxify ammonia in urea cycle, mitochondria are also required, for neurotransmitter metabolism. Mitochondrial encephalomyopathies are genetically, biochemically and clinically heterogeneous group of disorders associated with abnormalities of oxidative phosphorylation. The final diagnosis relied on clinical and molecular criteria.

Diagnostic criteria for pediatric mitochondrial disorders have been modified from an adult classification system. This criterion is the modified walker criteria. Because mitochondrial disease have a natural history of heterogeneous groups of disorders, the modified walker criteria is useful to evaluate the major clinical findings of infants and children with diagnosis of mitochondrial diseases.

By using the modified walker criteria, we are able to gather a large group of pediatric patients with a definite diagnosis of a mitochondrial disorder and study their clinical histories.

Keywords: Diagnostic criteria; Mitochondrial disorders; Mitochondrial encephalomyopathies

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