

Clinical Neurophysiology in Mitochondrial Disorders

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Abstract

Mitochondrial disease is a group of disorders caused by dysfunctional mitochondria, the organelles that generate energy for the cell. Mitochondria are found in every cell of the human body except red blood cells. Symptoms include poor growth, loss of muscle coordination, muscle weakness, visual problems, hearing problems, learning disabilities, heart disease, liver disease, kidney disease, gastrointestinal disorders, respiratory disorders, neurological problems, autonomic dysfunction and dementia. The effects of mitochondrial disease can be quite varied. Since the distribution of the defective mitochondrial DNA may vary from organ to organ within the body, and each mutation is modulated by other genome variants.

Although findings are not specific (e.g. generalized slowing in EEG, increased photoconvulsive response, etc.), but clinical neurophysiological studies are an important part of diagnostic workups as well as patients' follow-up. I will try to address the major changes in neurophysiological tracing according to the type of the disorder.

Keywords: Pediatric; Clinical neurophysiology; Mitochondrial disorder

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