

Electrodiagnostic Studies, “Role in The Diagnosis And Follow-Up in Children With Pompe Disease”

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Abstract

Pompe disease is a neuromuscular disorder that was progressive and fatal prior to enzyme replacement therapy (ERT). The advent of treatment has made early recognition imperative. Electrodiagnostic (EDx) studies represent a valuable diagnostic tool in Pompe disease, but there has been little contemporary data.

The clinical characteristics in children and adults are very similar. Needle electromyography demonstrated spontaneous activity (SA) in 80% of children and 83% of adults. Myotonic discharges were found in 53% of children and 72% of adults, often isolated to the paraspinal muscles in adults. There could be some improvements in EDX findings after enzyme replacement therapy.

EDx studies remain a helpful tool in diagnosing Pompe disease, but do not appear to be sensitive for monitoring response to ERT.

Paraspinal examination is necessary in adults with symptoms suggestive of Pompe disease, as abnormalities may be isolated to this region. Standard EDx studies are not sufficient to monitor early response to ERT and further research on potential biomarkers is needed.

- Electrodiagnostic abnormalities are present in most patients with Pompe disease.
- Myotonic discharges and other abnormalities may be seen in this type of storage disease

Keywords: Pompe disease; Children; EDX; EMG; Myotonic discharges

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