

## **Sparing of Myofiber Membranes in Infantile Pompe's Disease**

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Infantile Pompe's disease (type II glycogenosis), is a lysosomal disorder caused by a marked deficiency of  $\alpha$ -glucosidase. The clinical manifestations of this disorder include profound weakness, hypotonia, muscle hypertrophy, cardiomegaly, hepatomegaly, macroglossia, respiratory failure, and death during the first two years of life. The electrophysiological hallmarks are the presence of fibrillation potentials and myotonic discharges in the face of normal conduction studies.

We present two boys with this disorder. The first died untreated at 7 months. The second is alive at 19 months, receiving enzyme replacement therapy.

The thenar compound muscle action potentials (CMAPs) in both babies had normal amplitudes (7.2 mV and 6.8 mV respectively) despite the muscles being profoundly weak. Muscle biopsies showed severe vacuolization within muscle fibers with preservation of muscle membranes.

The deficiency of  $\alpha$ -glucosidase results in the lysosomal accumulation of glycogen, progressively destroying the contractile elements within myofibers. The normally large CMAPs in these markedly weak muscles showing intact membranes on biopsy indicate that the myofiber membranes are relatively spared in infantile Pompe's disease.