Lysosomal glycogen storage disease with normal acid maltase.

Danon MJ, Oh SJ, DiMauro S, Manaligod JR, Eastwood A, Naidu S, Schliselfeld LH.

Abstract

Two unrelated 16-year-old boys had mental retardation, cardiomegaly, and proximal myopathy. One also had hepatomegaly. Histochemistry and electronmicroscopy of muscle biopsies showed lysosomal glycogen storage resembling acid maltase deficiency. Biochemical studies of skeletal muscle showed increased content of glycogen of normal structure; acid alpha-glucosidase activity in both urine and muscle was normal. Other enzymes of glycogen metabolism were also normal. The cause of this apparently generalized glycogenosis with no demonstrable enzyme defect is unknown.

PMID:

6450334

[Indexed for MEDLINE]