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Glycogen storage disease due to acid maltase deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Glycogen storage disease due to acid maltase deficiency. ORPHA:365*

Glycogen storage disease due to acid maltase deficiency (AMD) is an autosomal recessive trait leading to metabolic myopathy that affects cardiac and respiratory muscles in addition to skeletal muscle and other tissues. AMD represents a wide spectrum of clinical presentations caused by an accumulation of glycogen in lysosomes: Glycogen storage disease due to acid maltase deficiency, infantile onset, non-classic infantile onset and adult onset (see these terms). Early onset forms are more severe and often fatal.