# Glycogen storage disease due to muscle and heart glycogen synthase deficiency

INSERM

## Source

*INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Glycogen storage disease due to muscle and heart glycogen synthase deficiency. ORPHA:137625*

Glycogen storage disease due to muscle and heart glycogen synthase deficiency is characterised by muscle and heart glycogen deficiency. It has been described in three siblings (two brothers and their younger sister). The older brother died at 10.5 years of age as a result of sudden cardiac arrest and the younger brother presented with hypertrophic cardiomyopathy, abnormal heart rate and blood pressure during exercise, and muscle fatigability. The sister showed no symptoms but a lack of glycogen was identified through muscle biopsy. The syndrome is caused by homozygous missense mutations in the gene encoding muscle glycogen synthase.