DISEASE:
Glycogen storage disease due to acid maltase deficiency, infantile onset

NAME:	Glycogen storage disease due to acid maltase deficiency, infantile onset
DESCRIPTION:	Glycogen storage disease due to acid maltase deficiency, infantile onset is the most severe form of glycogen storage disease due to acid maltase deficiency, characterized by cardiomegaly with respiratory distress, muscle weakness and feeding difficulties. It is often fatal.
ORPHACODE:	308552
SYNONYMS:	Alpha-1,4-glucosidase acid deficiency, infantile onset GSD due to acid maltase deficiency, infantile onset GSD type 2, infantile onset GSD type II, infantile onset Glycogen storage disease type 2, infantile onset Glycogen storage disease type II, infantile onset Glycogenosis due to acid maltase deficiency, infantile onset Glycogenosis type 2, infantile onset Glycogenosis type II, infantile onset Pompe disease, infantile onset
XREF(S):	Orphanet ICD-10
ANALYTE(S):	GAA

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RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- Enzymatic dosage Pompe disease
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)
- Pompe disease, Glycogen storage disease II (GAA gene)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- alpha glucosidase

Related Gene Panels

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- Neuromuscular disorders (166 genes) - VUB