

**DISEASE:**

**Glycogen storage disease due to acid maltase deficiency, late-onset**

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| <b>NAME:</b>        | Glycogen storage disease due to acid maltase deficiency, late-onset                                                                                                                                                                                                                                                                                                                                                                                                                           |
| <b>DESCRIPTION:</b> | A form of glycogen storage disease due to acid maltase deficiency characterized by excessive accumulation of glycogen in lysosomes most notably in skeletal muscle, leading to slowly progressive muscle weakness with walking disability and reduced respiratory function. The late-onset form includes all cases in which hypertrophic cardiomyopathy did not manifest or was not diagnosed at or under the age of 1 year, as well as all cases with symptom onset above the age of 1 year. |
| <b>ORPHACODE:</b>   | 420429                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| <b>SYNONYMS:</b>    | Alpha-1,4-glucosidase acid deficiency, late-onset<br>GSD due to acid maltase deficiency, late-onset<br>GSD type 2, late-onset<br>GSD type II, late-onset<br>Glycogen storage disease type 2, late-onset<br>Glycogen storage disease type II, late-onset<br>Glycogenosis type 2, late-onset<br>Glycogenosis type II, late-onset<br>Pompe disease, late-onset                                                                                                                                   |
| <b>XREF(S):</b>     | <a href="#">Orphanet</a><br><a href="#">ICD-10</a>                                                                                                                                                                                                                                                                                                                                                                                                                                            |

|                    |                     |
|--------------------|---------------------|
| <b>ANALYTE(S):</b> | <u>GAA</u>          |
| <b>CREATED:</b>    | 13 May 2019 - 01:02 |
| <b>CHANGED:</b>    | 22 Jun 2023 - 16:14 |

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### Related Genetic Tests

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- [Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy \(with prominent contractures\) / distal arthrogyrosis \(gene panel\)](#)
- [Pompe disease, Glycogen storage disease II \(GAA gene\)](#)

### Related Laboratories

- [Centrum Medische Genetica - UZ Brussel VUB](#)

### Related Analytes

- [alpha glucosidase](#)

### Related Gene Panels

- [Neuromuscular disorders \(166 genes\) - VUB](#)

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