

**DISEASE:****Glycogen storage disease due to muscle beta-enolase deficiency**

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| <b>NAME:</b>        | Glycogen storage disease due to muscle beta-enolase deficiency   |
| <b>DESCRIPTION:</b> | A rare glycolysis disorder characterized clinically by exercise intolerance and myalgia due to severe enolase deficiency in muscle.  |
| <b>ORPHACODE:</b>   | 99849  |
| <b>SYNOMYS:</b>     | GSD due to muscle beta-enolase deficiency<br>GSDXIII<br>Glycogenosis due to muscle beta-enolase deficiency<br>Glycogenosis type 13<br>Muscle enolase deficiency<br>Muscular enolase deficiency |
| <b>XREF(S):</b>     | <a href="#">Orphanet</a><br><a href="#">ICD-10</a><br><a href="#">OMIM</a>   |
| <b>ANALYTE(S):</b>  | <a href="#">ENO3</a>   |
| <b>CREATED:</b>     | 13 May 2019 - 01:02  |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14  |

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## RELATED CONTENT

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### Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal arthrogryposis (gene panel)

### Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

### Related Analytes

- enolase 3

### Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB

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