

**DISEASE:**  
**X-linked myopathy with postural muscle atrophy**

<b>NAME:</b>	X-linked myopathy with postural muscle atrophy
<b>DESCRIPTION:</b>	X-linked myopathy with postural muscle atrophy is a rare progressive muscular dystrophy characterized by an adult-onset scapulo-axio-peroneal myopathy. Clinical presentation includes shoulder girdle atrophy, scapular winging, axial muscular atrophy of postural muscles combined with a generalized hypertrophy. Typically, neck rigidity, rigid spine, Achilles tendon shortening, and respiratory insufficiency later in disease course are present.
<b>ORPHACODE:</b>	178461
<b>SYNONYMS:</b>	XMPMA
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">OMIM</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	FHL1
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

## 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

- four and a half LIM domains 1

### Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB

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