Diagnosis

Case of the month

February 2012

Myophosphorylase deficiency

McArdle´s disease
Glycogenosis type V

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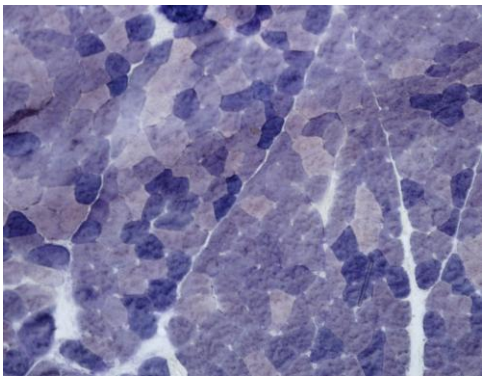
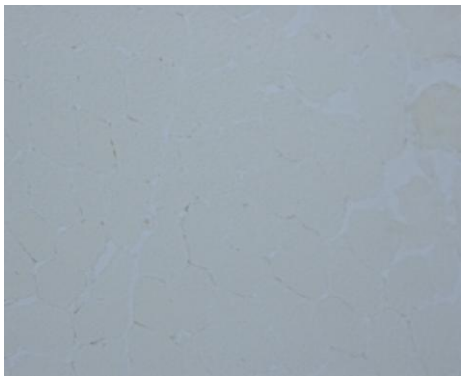
In myophospforylase staining no reactivity was found.



Control myophosphorylase



Patient myophosphorylase

Genetic analysis showed that the patient is carrier of two mutations in the **PYGM**-gene (phosporylase, glycogen, muscle). The gene codes for the enzyme myophosphorylase, which breaks down glycogen into glucose. The enzyme is only found in muscle cells. In the absence exercise will trigger pain and muscle weakness.

McArdle´s disease is an autosomal recessive disorder. The PYGM-gene is located on chromosome 11. Symtoms are usually present in childhood. Fatigue is often an early sign of the disease, followed by painful muscle contractions.
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