

Diagnosis

Case of the month

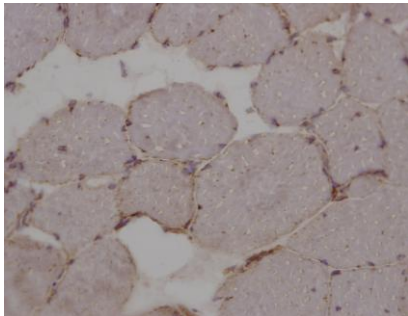
December 2011

LAMP-2 deficiency

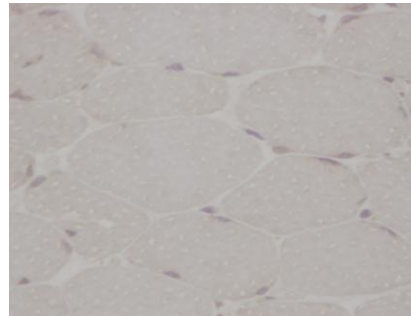
Danon's disease

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Immunostaining with antibodies against LAMP-2 was performed. No immunoreactivity was found.



Control LAMP-2



Patient LAMP-2

Genetic analysis confirmed the diagnosis. A mutation was found in the LAMP-2 gene on chromosome Xq24.

**Lysosomal-associated membrane protein 2 (LAMP-2) deficiency** is characterized by severe hypertrophic cardiomyopathy and myopathy.

Typically the disease manifests in males over 10 years of age. Onset is usually later in females.

Muscle biopsy shows intracytoplasmic vacuoles. These are autophagic vacuoles. In the myocardium there is also fibrosis .