

Diagnosis

Case of the month

October 2011

Nemaline myopathy

Mutation in ACTA1 has been found.

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**Nemaline myopathy** is a congenital myopathy. The morphologic hallmark of the disease is the formation of rods, which origin from Z bands (“nema” Gr. means thread). The rods stain red with the Gomori trichrome. Their major constituents are a-actinin and actin. They are often located in clusters at the periphery of the fibers.

Nemaline myopathy is a heterogeneous disorder, both clinically and genetically. The dominating symptoms are muscle weakness and hypotonia. The mode of inheritance may be autosomal dominant or autosomal recessive. Mutations in at least seven different genes are known to cause the disease. These are:  $\alpha$ -actin (ACTA1),  $\beta$ -tropomyosin (TPM2), a-tropomyosin slow (TPM3), nebulin (NEB), troponin T1 (TNNT1), cofilin (CFL2) and KBTBD13.