Nemaline Myopathy 3

National Cancer Institute

Source

National Cancer Institute. Nemaline Myopathy 3. NCI Thesaurus. Code C129870.

An inherited myopathy caused by mutations in the ACTA1 gene, encoding actin, alpha skeletal muscle. The phenotype is highly variable, and as such attempts at classification by clinical features is not optimal. Generally, affected individuals have generalized muscle weakness, typically involving proximal muscles, the face, bulbar and respiratory muscles.