Craniofacial Manifestations in Severe Nemaline Myopathy.

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Abstract
Nemaline myopathy (NM) is a rare congenital muscular disease characterized by the presence of rod (nemaline) bodies visualized on muscle biopsy. The disease is genetically and clinically heterogeneous, and the age of onset can vary from neonate to adult. Patients typically present initially with diffuse muscle weakness and hypotonia. The disease also afflicts facial musculature and can cause anomalous facial growth and development. The authors report a patient of early onset NM with significant craniofacial abnormalities. The untreated facial growth is discussed and illustrated in this article. The authors reviewed the current knowledge in the literature regarding the molecular and genetic pathogenesis of NM. The roles of both surgical and supportive management are discussed in this particular patient.

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