Neonatal fractures as a presenting feature of LMOD3-associated congenital myopathy.


Abstract

Nemaline myopathy is a rare inherited disorder characterized by weakness, hypotonia, and depressed deep tendon reflexes. It is clinically and genetically heterogeneous, with the most severe phenotype presenting as perinatal akinesia, severe muscle weakness, feeding difficulties and respiratory failure, leading to early mortality. Pathogenic variants in 12 genes, encoding components of the sarcomere or factors related to myogenesis, have been reported in patients affected with the disorder. Here, we describe an early, lethal presentation of decreased fetal movements, hypotonia, muscle weakness, and neonatal respiratory failure requiring ventilator support in three siblings from a consanguineous family. All exhibited perinatal fractures, and thus, a skeletal dysplasia was considered as possibly contributing to the phenotype. However, whole exome sequencing revealed a homozygous, loss-of-function pathogenic variant in LMOD3, which has recently been associated with nemaline myopathy and, in a subset of patients, perinatal fractures. This case demonstrates the importance of considering congenital neuromuscular disorders in the differential diagnosis of perinatal fractures.

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KEYWORDS: LMOD3; congenital fractures; congenital myopathy; nemaline myopathy; perinatal fractures

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