Congenital myopathy with a novel SELN missense mutation and the challenge to differentiate it from congenital muscular dystrophy.

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Abstract
Multiminicore disease is a myopathy that is pathologically characterized by the presence of multiple areas of small, short, and poorly delineated zones of sarcomeric disorganization lacking mitochondria (minicores) that can be observed in both type 1 and type 2 fibers. Most cases of multiminicore disease typically present with early-onset axial weakness, respiratory insufficiency, scoliosis, and rigid spine. There is no correlation between the frequency of minicores and clinical severity. Multiminicore disease is genetically heterogeneous and can result from recessive or dominant mutations. Genetic testing is needed to establish the precise diagnosis and provide overall prognosis. Here we report a 23-year-old woman with respiratory failure, distal joint hyperlaxity, scoliosis and rigid spine due to multiminicore disease caused by a novel compound heterozygous mutation in the selenoprotein N1-encoding gene (SELN). The preserved ambulation into adulthood and normal creatinine kinase (CK) favor the diagnosis of congenital myopathy over congenital muscular dystrophy (CMD). However, the nonspecific myopathic histopathological changes and extremely rare minicore-like structures can make it challenging to differentiate between SELN-myopathy and congenital muscular dystrophies, such as Ullrich or lamin A/C-CMD.

KEYWORDS: Multiminicore; Respiratory insufficiency; Rigid spine; SELN; Scoliosis; Selenoprotein

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