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Epub 2020 Nov 26.

Expanding the spectrum of congenital myopathies: prenatal onset with extreme hyperextension of the neck

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PMID: 33244741 DOI: [10.1007/s10072-020-04937-x](#)**Abstract**

We describe the case of a male newborn presenting with a prenatal diagnosis of persistent hyperextension of the fetal neck and severe hypotonia and respiratory insufficiency at birth. Facial weakness, increased serum creatine kinase levels, and abnormal feeding, together with other signs, such as severe contractures, also classically associated with congenital myopathies prompted to perform a muscle biopsy showing internal rods suggestive of a possible nemaline myopathy. These findings suggest that a careful neurological examination should be performed in infants with persistent hyperextension of the fetal neck to exclude weakness and a possible underlying muscle disorder.

Keywords: Nemaline myopathy; Neonatal hypotonia; Persistent hyperextension of the fetal neck.

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