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Generation of two isogenic induced pluripotent stem cell lines from a 10-year-old typical nemaline myopathy patient with a heterozygous dominant c.541G>A (p.Asp179Asn) pathogenic variant in the ACTA1 gene

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Abstract

Nemaline myopathy (NM) is a congenital myopathy typically characterized by skeletal muscle weakness and the presence of nemaline bodies in myofibres. Approximately 25% of NM cases are caused by variants in ACTA1. We generated two induced pluripotent stem cell lines from lymphoblastoid cells of a 10-year-old female with typical NM harbouring a dominant pathogenic variant in ACTA1 (c.541C>A). The isogenic lines displayed typical iPSC morphology, expressed pluripotency markers, and could differentiate into each of the three germ layers. Although the lines have partial or complete X chromosome duplication, they may still prove useful as models of human ACTA1 disease.

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