Adult onset distal and proximal myopathy with complete ophthalmoplegia associated with a novel de novo p.(Leu1877Pro) mutation in MYH2.


Abstract

An MYH2 mutation p.(Glu706Lys) was originally described in a family with autosomal dominant inheritance, where the affected family members presented with multiple congenital contractures and ophthalmoplegia, progressing to a proximal myopathy in adulthood. Another patient with a dominant mutation p.(Leu1870Pro) was described, presenting as a congenital myopathy with ophthalmoplegia. Here, we present a patient with symptoms beginning at age 16 years, of prominent distal but also proximal weakness, bulbar involvement and ophthalmoplegia. Initially, clinically classified as oculopharyngodistal myopathy, the patient was found to carry a novel, de novo MYH2 mutation c.5630T>C p.(Leu1877Pro). This expands the phenotype of dominant MYH2 myopathies with the clinical phenotype overlapping the oculopharyngodistal myopathy spectrum.

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KEYWORDS: MYH2; myosinopathy; oculopharyngodistal myopathy; ophthalmoplegia

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