Bethlem myopathy in a Portuguese patient - case report.

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Abstract

Mutations of the encoding genes of collagen VI (COL6A1, COL6A2 and COL6A3), are responsible for two classical phenotypes (with a wide range of severity), the Ullrich congenital muscular dystrophy (UCMD) and the Bethlem myopathy (BM). We present a male patient of 49 years old, with symptoms of muscle weakness beginning in childhood and of very slowly progression. At the age of 42, the neurological examination revealed proximal lower limb muscle weakness and contractures of fingers flexors muscles, positive Gowers manoeuvre and a waddling gait. Serum creatine kinase (CK) values were slightly elevated, electromyographic study revealed myopathic changes and muscle MRI of the lower limbs showed a specific pattern of muscle involvement, with peripheral fat infiltration in vastus lateralis and intermedius and anterocentral infiltration in rectus femoris. Respiratory and cardiac functions were unremarkable. Whole exome sequencing identified the homozygous mutation c.1970-9G>A in COL6A2 gene.

KEYWORDS: Bethlem myopathy; collagen VI; congenital muscular dystrophy

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