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Collagen VI-related Limb-Girdle Syndrome Caused by Frequent Mutation in COL6A3 Gene With Conflicting Reports of Pathogenicity

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Abstract

Recently the scientific community has started to view Bethlem myopathy 1 and Ullrich congenital muscular dystrophy as two extremes of a collagen VI-related myopathy spectrum rather than two separate entities, as both are caused by mutations in one of the collagen VI genes. Here we report three individuals in two families who are homozygous for a COL6A3 mutation (c.7447A> G; p.Lys2483Glu), and compare their clinical features with seven previously published cases. Individuals carrying homozygous or compound heterozygous c.7447A> G, (p.Lys2483Glu) mutation exhibit mild phenotype without loss of ambulation, similar to the cases described previously as Collagen VI-related limb-girdle syndrome. The phenotype could arise due to an aberrant assembly of Von Willebrand factor A domains. Based on these data, we propose that c.7447A> G, (p.Lys2483Glu) is a common pathogenic mutation.

Keywords: COL6A3; Collagen VI-related myopathy; LGMD phenotype.

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