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A novel COL1A1 gene-splicing mutation (c.1875+1G>C) in a Brazilian patient with osteogenesis imperfecta

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ABSTRACT. Osteogenesis imperfecta is a heterogeneous genetic disorder characterized by bone fragility and deformity, recurrent fractures, blue sclera, short stature, and dentinogenesis imperfecta. Most cases are caused by mutations in COL1A1 and COL1A2 genes. We present a novel splicing mutation in the COL1A1 gene (c.1875+1G>C) in a 16-year-old Brazilian boy diagnosed as a type III osteogenesis imperfecta patient. This splicing mutation and its association with clinical phenotypes will be submitted to the reference database of

COL1A1 mutations, which has no other description of this mutation.

Key words: Osteogenesis imperfecta type III; Splicing mutation; Genotype/phenotype correlation

