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Mutation characteristics in type I collagen genes in Chinese patients with osteogenesis imperfecta

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ABSTRACT. Osteogenesis imperfecta is normally caused by an autosomal dominant mutation in the type I collagen genes COL1A1 and COL1A2. The severity of osteogenesis imperfecta varies, ranging from perinatal lethality to a very mild phenotype. Although there have been many reports of COL1A1 and COL1A2 mutations, few cases have been reported in Chinese people. We report on five unrelated families and three sporadic cases. The mutations were detected by PCR and direct sequencing. Four mutations in COL1A1 and one in COL1A2 were found, among which three mutations were previously unreported. The mutation rates of G>C at base 128 in intron 31 of the COL1A1 gene and G>A at base 162 in intron 30 of the COL1A2 gene were higher than normal. The patients' clinical characteristics with the same mutation were variable even in the same family. We conclude that mutations in COL1A1 and COL1A2 also have an important role in osteogenesis imperfecta in the Chinese population. As the Han Chinese people account for a quarter of the world's population, these new data contribute to the type I collagen mutation map.

Key words: Osteogenesis imperfecta; COL1A1; COL1A2; Heredity; Mutation

