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A novel *RUNX2* mutation (T420I) in Chinese patients with cleidocranial dysplasia

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ABSTRACT. Cleidocranial dysplasia (CCD) is an autosomal-dominant heritable skeletal disease caused by heterozygous mutations in the *RUNX2* gene. We studied a Chinese family that included three affected individuals with CCD phenotypes; the clinical features of patients with CCD include delayed closure of fontanelles, frontal bossing, dysplasia of clavicles, late tooth eruption, and other skeletal anomalies. X-ray analysis showed aplasia of the clavicles. The *RUNX2* gene was studied by PCR and direct sequencing of the entire coding region and the exon-intron boundaries of the gene. A novel missense mutation (c.1259C→T[p.T420I]) in *RUNX2* gene exon 7 was identified; it was found in the affected individuals in this Chinese family, but was not present in an unaffected family member or in 100 unrelated normal controls. This is the first report that gives evidence that the T420I mutation of *RUNX2* is associated with CCD, expanding the spectrum of *RUNX2* mutations causing CCD.

Key words: Cleidocranial dysplasia; Mutation; *RUNX2*; Gene

