A novel single-base deletion mutation of the *RUNX2* gene in a Chinese family with cleidocranial dysplasia


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ABSTRACT. We identified a disease-causing mutation of the *RUNX2* gene in a four-generation Chinese family affected with cleidocranial dysplasia (CCD). For mutation analysis, the coding region of *RUNX2* was sequenced with DNA from two patients and three unaffected family members. The *RUNX2* mutation was investigated in 50 normal controls by denaturing high pressure liquid chromatography. A heterozygous single-base deletion (c.549delC) of *RUNX2*, which predicts a termination site at the 185th codon and leads to a stop in the runt domain of *RUNX2* protein, was detected in both patients but not in the three unaffected members of the family. This mutation was also not found in 50 controls and has not been reported previously. We demonstrated that a novel mutation (c.549delC) of *RUNX2* is associated with CCD in a Chinese family, adding to the repertoire of *RUNX2* mutations related to CCD.

Key words: CCD; *RUNX2*; c.549delC; Mutation analysis; DHPLC