**RUNX2 mutations in cleidocranial dysplasia**

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**ABSTRACT.** The runt-related transcription factor 2 gene (RUNX2), which is also known as CBFα1, is a master regulatory gene in bone formation. Mutations in RUNX2 have been identified in cleidocranial dysplasia (CCD) patients. CCD is a rare autosomal dominant skeletal dysplasia that is characterized by delayed closure of cranial sutures, aplastic or hypoplastic clavicle formation, short stature, and dental anomalies, including malocclusion, supernumerary teeth, and delayed eruption of permanent teeth. In this study, we recruited three de novo CCD families and performed mutational analysis of the RUNX2 gene as a candidate gene approach. The mutational study revealed three disease-causing mutations: a missense mutation (c.674G>A, p.Arg225Gln), a frameshift mutation (c.1119delC, p.Arg374Glyfs*), and a nonsense mutation (c.1171C>T, p.Arg391*). Clinical examination revealed a unique dental phenotype (no typical supernumerary teeth, but duplication of anterior teeth) in one patient. We believe that this finding will broaden the understanding of the mechanism of supernumerary teeth formation and CCD-related phenotypes.

**Key words:** RUNX2; Cleidocranial dysplasia; Supernumerary teeth; Mutation