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Identification of novel and recurrent mutations in the calcium binding type III repeats of cartilage oligomeric matrix protein in patients with pseudoachondroplasia

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ABSTRACT. Pseudoachondroplasia is an autosomal dominant osteochondrodysplasia characterized by disproportionate short stature, joint laxity, and early onset osteoarthrosis. Pseudoachondroplasia is caused by mutations in the gene encoding cartilage oligomeric matrix protein (*COMP*). We looked for mutations in the *COMP* gene in three sporadic Chinese pseudoachondroplasia patients and identified two novel mutations, c.1189G>T (p.D397Y) and c.1220G>A (p.C407Y), and one recurrent mutation, c.1318G>C (p.G440R), in the calcium binding type III repeats of *COMP*. This study confirms the relationship between mutations of the *COMP* gene and clinical findings of pseudoachondroplasia; it also provides evidence for the importance of the calcium binding domains to the functioning of *COMP*.

Key words: PSACH; *COMP*; Gene mutation; Skeletal dysplasia

