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Genome-Wide Association Study Identify COL2A1 as a Susceptibility Gene for the Hand Development Failure of Kashin-Beck Disease

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Abstract : Kashin-Beck disease (KBD) is a chronic osteochondropathy. The mechanism of hand growth and development failure of KBD remains elusive now. In this study, we conducted a two-stage genome-wide association study (GWAS) of palmar length-width ratio (LWR) of KBD, totally involving 493 Chinese Han KBD patients. Affymetrix Genome Wide Human SNP Array 6.0 was applied for SNP genotyping. Association analysis was conducted by PLINK software. Imputation analysis was performed by IMPUTE against the reference panel of the 1000 genome project. In the GWAS, the most significant association was observed between palmar LWR and rs2071358 of COL2A1 gene (P value = 4.68×10-8). Imputation analysis identified 3 SNPs surrounding rs2071358 with significant or suggestive association signals. Replication study observed additional significant association signals at both rs2071358 (P value = 0.017) and rs4760608 (P value = 0.002) of COL2A1 gene after Bonferroni correction. Our results suggest that COL2A1 gene was a novel susceptibility gene involved in the growth and development failure of hand of KBD.

Keywords : Kashin-Beck disease, genome-wide association study, COL2A1, hand
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