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No association between *FGD1* gene polymorphisms and intellectual developmental disability in the Qinba mountain area

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ABSTRACT. *FGD1* encoding a guanine nucleotide exchange factor, specifically activates Rho GTPase cell division cycle 42 (Cdc42). Dysfunction of FGD1 causes Aarskog-Scott syndrome (MIM #305400), an X-linked disorder that may affect bone and intellectual development. However, the relationship between *FGD1* and intellectual developmental disorders (IDD) remains unclear. The purpose of this study was to investigate the genetic association between the *FGD1* polymorphism and IDD. Working with families from the Qinba mountain area where the occurrence of IDD is higher than the average in China, we analyzed 456 samples from 130 nuclear families, effectively controlling for stratification and environmental factors. Five SNP loci (rs2230265, rs7881608, rs2239809, rs6614244, and

rs2284710) were selected that were well distributed within the *FGD1* gene. Genotyping was performed through single-strand conformation polymorphism and restriction fragment length polymorphism. The data were analyzed with transmission disequilibrium tests. In the Qinba mountain area, no significant association was observed between IDD and allele or genotype frequencies, or the haplotype of the 5 SNP loci of the *FGD1* gene. The results indicate that *FGD1* may not be a monogenetic X-linked factor in IDD. Further studies are required to investigate its role in intellectual development based on its specific interactions with Cdc42 or other partner proteins contributing to IDD.

Key words: *FGD1*; Restriction fragment length polymorphism; Intellectual developmental disorders; Single nucleotide polymorphism; Single-strand conformation polymorphism