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A novel *NF1* frame-shift mutation (c.702_703delGT) in a Chinese family with neurofibromatosis type 1

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ABSTRACT. This study aimed to characterize the clinical features of a Chinese pedigree with neurofibromatosis type 1 (NF1) and to identify mutations in the *NF1* gene. In this three-generation family containing 8 members, 5 had been diagnosed with NF1 and the others were asymptomatic. All members of the family underwent complete medical examinations. Molecular genetic analyses were performed on all subjects included in the study. All exons of *NF1* were amplified by polymerase chain reaction, sequenced, and compared with a reference database. Possible changes in function of the protein induced by amino acid variants were predicted by bioinformatic analysis. In this family, the 5 patients presented different clinical phenotypes, but all manifested typical café-au-lait macules. One novel frame-shift

mutation, c.702_703delGT, in exon 7 of *NF1* was identified in all affected family members, but not in the unaffected family members or in 102 normal controls. This mutation generates a premature stop codon at amino acid position 720. Additionally, a synonymous mutation c.702 G>A was found in 3 family members, including 2 affected and 1 normal individuals. In conclusion, our study suggests that a novel c.702_703delGT frame-shift mutation in *NF1* is likely to be responsible for the pathogenesis of NF1 in this family. To the best of our knowledge, it is the first time that a c.702_703delGT mutation has been identified in a family with neurofibromatosis type 1.

Key words: Neurofibromatosis type 1; *NF1* gene; Mutation

