A novel NF1 mutation in a Chinese patient with giant café-au-lait macule in neurofibromatosis type 1 associated with a malignant peripheral nerve sheath tumor and bone abnormality

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ABSTRACT. Neurofibromatosis type 1 (NF1; OMIM#162200) is a common neurocutaneous disorder that is characterized by multiple café-au-lait, skinfold freckling, Lisch nodules, and neurofibromas. Mutations in the NF1 gene, which encodes the neurofibromin protein, have been identified as the pathogenic gene of NF1. In this study, we present a clinical and molecular study of a Chinese patient with giant café-au-lait in NF1. The patient showed >6 café-au-lait spots on the body, axillary freckling, and multiple subcutaneous neurofibromas. He also had a malignant peripheral nerve sheath tumor and bone abnormalities. The germline mutational analysis of the NF1 gene revealed a novel missense mutation in exon 13. It is a novel heterozygous nucleotide G>A transition at position 2241 of the NF1 gene. We found no mutation in malignant peripheral nerve sheath tumor DNA from this patient. This
expands the database for \textit{NF1} gene mutations in NF1. Its absence in the normal chromosomes suggests that it is responsible for the NF1 phenotype. To our knowledge, this is the first case of giant café-au-lait macule in NF1 associated with a malignant peripheral nerve sheath tumor and bone abnormality.

\textbf{Key words}: Mutation analysis; \textit{NF1} gene; Neurofibromatosis type 1; Giant café-au-lait