Genetics and Molecular Research

NF1 frameshift mutation (c.6520_6523delGAGA) association with nervous system tumors and bone abnormalities in a Chinese patient with neurofibromatosis type 1

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Genet. Mol. Res. 15 (2): gmr.15027572
Received September 1, 2015
Accepted December 8, 2015
Published April 7, 2016
DOI http://dx.doi.org/10.4238/gmr.15027572

ABSTRACT. Neurofibromatosis type 1, also known as NF1 or von Recklinghausen's disease, is a common neurocutaneous syndrome that presents with multiple café-au-lait patches, skinfold freckling, dermatofibromas, neurofibromas, and Lisch nodules. The mutations of the gene *NF1*, encoding the protein neurofibromin, have been identified as the cause of this disease. Here, we report a clinical and molecular study of a Chinese patient with multiple café-au-lait skin freckles, dermatofibroma, central and peripheral nervous system tumors, and bone abnormalities attributed to *NF1*. The patient showed >6 café-au-lait spots on the body and multiple dermatofibromas. A brain glioma and multiple nerve sheath tumors inside and outside the vertebral canal were identified by magnetic resonance imaging, which also showed

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multiple intercostal nerve schwannomas and hydrocephalies above the cerebellar tentorium. Talipes equinus was also apparent. A mutation analysis of the *NF1* gene revealed a novel frameshift mutation in exon 43, consisting of a heterozygous deletion of four nucleotides (GAGA) between positions 6520 and 6523. No *NF1* mutations were detected in the patient's parents or younger brother. These results extend the list of known mutations in this gene. The absence of the *NF1* mutation in the healthy family members suggests that it is responsible for the NF1 phenotype. To our knowledge, this frameshift mutation represents a novel NF1 case, and may be associated with nervous system tumors and bone abnormalities.

Key words: Mutation analysis; *NF1* gene; Neurofibromatosis type 1; Multiple café-au-lait spots; Tumor; Bone abnormality

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