GMR Genetics and Molecular Research
Online Journal
ISSN:1676-5680

A novel splicing site mutation of the *GPR143* gene in a Chinese X-linked ocular albinism pedigree

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Genet. Mol. Res. 12 (4): 5673-5679 (2013)
Received March 26, 2013
Accepted September 16, 2013
Published November 18, 2013
DOI http://dx.doi.org/10.4238/2013.November.18.16

ABSTRACT. Ocular albinism is an X-linked inherited disease characterized by hypopigmentation of the iris and nystagmus. To identify a new disease-causing mutation of ocular albinism, we collected a Han Chinese pedigree with 7 male congenital nystagmus patients over 3 generations. Slit-lamp photography and optical coherence tomography were performed for the proband. Genomic DNA was extracted from a whole blood sample from the proband using the high-salt method. Polymerase chain reaction (PCR) sequencing was carried out for *GPR143* and *FRMD7* genes. The three-dimensional structures of the wild-type and mutant GPR143 proteins were determined using SWISS-MODEL. The transmission of the disease in the pedigree clearly followed an X-linked pattern. The proband had significant iris and fundus hypopigmentation. Optical coherence tomography showed severe foveal hypoplasias in both eyes of the proband. A novel splicing site (G/C) mutation was found on the boundary of the 6th intron and the 7th

exon of the *GPR143* gene, resulting in a 9-amino-acid deletion (codons 257-265) in the 6th transmembrane domain of the GPR143 protein. In conclusion, a novel splicing site mutation of the *GPR143* gene was found in a Han Chinese congenital ocular albinism pedigree.

Key words: Ocular albinism; *GPR143*; Congenital nystagmus; Pedigree; Han Chinese

