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

A novel *FBN1* heterozygous mutation identified in a Chinese family with autosomal dominant Marfan syndrome

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Genet. Mol. Res. 14 (2): 4125-4132 (2015)
Received February 17, 2014
Accepted October 30, 2014
Published April 27, 2015
DOI http://dx.doi.org/10.4238/2015.April.27.27

ABSTRACT. The purpose of this study was to identify the clinical features and mutations in the fibrillin-1 gene (*FBN1*) in a large Chinese family with autosomal dominant Marfan syndrome (MFS). Seventeen members from a Chinese family of 4 generations were included in the study. All members underwent complete ophthalmic examination. Molecular genetic analysis was performed on all subjects. All exons of *FBN1* were amplified by polymerase chain reaction, sequenced, and the sequences were compared with a reference database. Variations were evaluated in family members as well as 100 normal controls. Changes in structure and function of the protein induced by amino acid variation were predicted by bioinformatic analysis. Ectopia lentis,

dolichostenomelia, arachnodactyly, and tall stature were present in all patients diagnosed with MFS. The novel heterozygous missense mutation c.2243 T>G (p.C781W) in exon 19 of *FBN1* was identified in all 5 patients, but not in other family members or 100 normal controls. This mutation caused an amino acid substitution of cysteine to tryptophan at position 781 (p.C781W) of the FBN1 protein. This mutation occurred in a highly conserved region and may cause structural and functional changes in the protein according to our bioinformatic analysis. Our results suggest that the novel mutation C781W of *FBN1* is responsible for the pathogenesis of MFS in this pedigree.

Key words: Ectopia lentis; Exon; *FBN1*; Marfan syndrome; Mutation

