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A Nonsense Mutation in *FBN1* Presented with Variable Phenotypes of Marfan Syndrome within a Chinese Family

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ABSTRACT Marfan syndrome (MFS) exhibits an irregular dominant connective tissue defect with clinical variability. It generally affects the skeletal, ocular, and cardiovascular systems. The variants in the *FBN1* gene are the main causes of this disease. In this study, Exome sequencing and Sanger sequencing were utilised to explore the pathogenic variant of a Chinese MFS family. A nonsense variant c.612C>A (C204X) of *FBN1* was identified in the both affected brothers with different clinical phenotypes and in their unaffected mother. Nevertheless, the variant was absent in 200 healthy controls. The further cell function experiment demonstrated that the FBN1 C204X mutant exhibited a lower level of mRNA and protein expression than the wild-type. The results supplemented the data of genotype-phenotype relationships in regard to *FBN1* variants, and expanded the *FBN1* variants spectrum in Asian populations. This study confirmed that MFS was a semidominant genetic disorder probably involving the special hereditary modifiers and environmental risk factors.