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Two Null Variants Q2417X and R3409X in *FLG* Gene are Relevant to Ichthyosis Vulgaris in a Chinese Family

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ABSTRACT Ichthyosis vulgaris (IV) is one of the most ordinary hereditary keratinising illness, characterised by scaly, dry skin, and palmoplantar hyperlinearity. The function loss of filaggrin (*FLG*) gene mutations is considered as the molecular aetiology for IV. In this study the pathogenic variants of a Chinese IV family were explored by exome sequencing and PCR sequencing. Two null heterozygous variants in *FLG* coexisted in the pedigree, including c.7249C>T (Q2417X) and c.10225C>T (R3409X). The Q2417X variant was only found in the proband and their healthy mother, whereas the R3409X variant was observed in the other affected family members. The proband with Q2417X showed a much more severe phenotype than the patients with R3409X did, but their mother with Q2417X presented no clinic symptoms. The results strongly support that IV is a semidominant keratinising disorder that involved hereditary and environmental risk aspects and expanded the *FLG* variants spectrum in Asian populations.