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Establishment and rapid detection of a heterozygous missense mutation in the *CACNA1F* gene by ARMS technique with double-base mismatched primers

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ABSTRACT. Retinitis pigmentosa (RP) is a retinal degenerative disorder that often causes complete blindness. Mutations of more than 50 genes have been identified as associated with RP, including the *CACNA1F* gene. In a recent study, by employing next-generation sequencing, we identified a novel mutation in the *CACNA1F* gene. In this study, we used the amplification refractory mutation system (ARMS) and identified a single nucleotide change c.1555C>T in exon 13 of the *CACNA1F* gene, leading to the substitution of arginine by tryptophan (p.R519W) in a Chinese individual affected by RP.

This study actually confirms this novel mutation, and establishes the ARMS technique for the detection of mutations in RP.

Key words: *CACNA1F*; Missense mutation; Retinitis pigmentosa; Amplification refractory mutation system; Double-base mismatch

