Whole exome sequencing in Korean patients with retinitis pigmentosa identified mutations in *EYS*

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Retinitis pigmentosa(RP) is a rare hereditary retinal disease characterized by progressive degeneration of photoreceptors. The worldwide prevalence of RP is about 1 in 4000. The clinical features of RP are usually confined to the eyes, but some patients with RP have a syndrome affecting non-ocular organs. RP is genetically heterogeneous and can be inherited in autosomal dominant, autosomal recessive, or X-linked modes. More than 60 genes are identified as causes of RP (RetNet-Retinal Information Network). The eyes shut homolog (*EYS*) gene is located on chromosome 6q12 and is one of the largest genes expressed in the retina. The *EYS* protein is important for maintaining the normal morphology of photoreceptor cells. A high prevalence of *EYS*-related RP was reported in Asian countries (Korea, Japan, and China), but the relationship between *EYS* mutations and clinical phenotypes is still unclear. To identify RP related *EYS* variants in a Korean cohort, we performed a whole exome sequencing analysis only in non-syndromic RP patients. *EYS* mutations account for 12% of the total RP-related mutations in this study. The 4 *EYS* variants (c.2259+1G>A, c.4957dupA, c.6557G>A, c.8805C>A) are likely to be pathogenic and compound heterozygotes according to ACMG guidelines.

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