Exome sequencing identified *USH2A* mutations in Korean patients with retinitis pigmentosa

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Retinitis pigmentosa (RP) is a rare hereditary retinal disease. It is characterized by progressive degeneration of photoreceptors. The worldwide prevalence of RP is about 1 in 4000. The clinical manifestations of RP are usually confined to the eyes. But some patients with RP have syndrome affecting non-ocular organs. RP is genetically heterogeneous. RP can be inherited in an autosomal dominant, autosomal recessive, or X-linked. More than 60 genes are identified as causes of RP. Mutations in *USH2A* are most frequently reported as cause of non-syndromic autosomal recessive RP and Usher's syndrome type II, which is one of the most common syndromes of RP. The *USH2A* gene is located on chromosome 1q42 with 72 exons. *USH2A* gene encodes for usherin which is a basement membrane protein in the retina. It is known that this protein is crucial for the maintenance of mammalian photoreceptors. Since *USH2A* was first reported in 1998 as a cause of visual or hearing loss, many studies were done to reveal the *USH2A* mutation-phenotype correlation. To identify RP related *USH2A* variants in Korean cohort We performed a whole exome sequencing analysis to identify *USH2A* variants in 157 Korean patients with non-syndromic RP.

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