RRH, Encoding the RPE-Expressed Opsin-Like Peropsin, Is Not Mutated in Retinitis Pigmentosa and Allied Diseases

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Ophthalmic Genetics, Volume 28, Issue 1 January 2007, pages 31 - 37

Many genes from retinoid metabolism cause retinitis pigmentosa. Peropsin, an opsin-like protein with unknown function, is specifically expressed in apical retinal pigment epithelium microvilli. Since rhodopsin and RGR, another opsin-like protein, cause retinitis pigmentosa, we used D-HPLC to screen for the peropsin gene RRH in 331 patients (288 with retinitis pigmentosa and 82 with other retinal dystrophies). We found 13 nonpathogenic variants only, among which a c.730_731delATinsG that truncates the last two transmembrane-spanning fragments and the Lys284 required for retinol binding, but does not segregate with the disease phenotype. We conclude that RRH is not a frequent gene in retinitis pigmentosa.