Assessing the Photoreceptor Expression in Retinal Organoids From a Patient With Rhodopsin Copy Number Variation

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Retinitis Pigmentosa (RP) is a common inherited retinal degeneration disease that is caused by the deterioration of rod photoreceptors, a type of retinal cell, leading to vision loss. One method to further study this disease was by the development of a lab-generated model of RP called retinal organoid (RO) using induced pluripotent stem cells (iPSC) derived from peripheral blood mononuclear cells of a male with the disease (RM). A control RO (RC) was also created using his daughter's cells because she was unaffected by RP. With this model, I wanted to find out whether there were expressed disease phenotypes with copy number variation of rhodopsin (late rod photoreceptor-specific marker) along with if there were any deleterious effects with the variation of rhodopsin copies (RHO-CNV). By obtaining images of iPSC and rod photoreceptor specific markers and Western Blot Assay images of RC and RM, I used a software called ImageJ to process images for observations and measurements of the intensities in protein production concentration. As a result, the late rod photoreceptor marker rhodopsin (RHO) was observed to be improperly localized with elevated expression. In support of the overexpression, the data analysis of the Western Blot Assay showed greater intensities rhodopsin in the father. In conclusion, the RHO-CNV expressed disease phenotypes by the mislocalization and overexpression of this late rod photoreceptor-specific marker, which showed deleterious trafficking defects in the model by rod photoreceptor degeneration, leading to vision loss.

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