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Potassium channelopathy leading to vitreoretinal degeneration

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Abstract

Ion channels are membrane proteins which establish cellular communication and are essential for cellular existence. Their dysfunction or "channelopathy" underlies both inherited and acquired blindness that primarily affect the function of photoreceptors (PR) and retinal pigment epithelium (RPE) cells. In the apical side of the RPE cell that faces PR neurons, inwardly rectifying potassium channel (Kir7.1: that permit passage of K^+-ions into and out of cells to control cell activity) is localized. Investigation of the RPE ion channel Kir7.1 biophysical properties have demonstrated remarkable contribution of these channels to normal RPE cell function. Herein I review recent advances in RPE physiology and cell biology, and their relevance to vision most importantly the role of Kir7.1 channel in visual signal transduction. Few recent reports have confirmed the hypothesis that multiple mutations in the KCNJ13 gene that alter the function of Kir7.1 channel affect RPE cell biology and cause Snowflake Vitreoretinal Degeneration (SVD) and Leber's Congenital Amaurosis (LCA). It is puzzling how functional impairment of RPE Kir7.1 channel results in the degeneration of multiple ocular tissues as seen in SVD, an autosomal dominant disorder. LCA on the other hand is an autosomal recessive disease evidenced by severe pigmentation and reduced-amplitude of scotopic electroretinogram affecting 3 in 100,000 children. This talk will discuss on the clinical manifestations and pathophysiology of SVD and LCA. The discussion will focus more on the molecular mechanisms (structure and biology) of Kir7.1 genetic mutations.

Biography

Bikash R. Pattnaik has completed his Ph.D. from University of Delhi and postdoctoral studies in France and USA. Since 2008 as a faculty in Pediatrics, Ophthalmology and Visual Science at University of Wisconsin, Madison, Dr. Pattnaik' s research focus has been ion-channelopathy and retinal disorders. This innovative work has demonstrated how retinal cells created in a dish using STEM cell technology retain functional integrity especially visual signal transduction capabilities. He has several publications in reputed journals and serving as a guest reviewer of many Ophthalmology journals.