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Ablation of the X-linked Retinitis Pigmentosa 2 (*Rp2*) gene in mice results in opsin mistrafficking and photoreceptor degeneration

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Abstract

X-linked Retinitis Pigmentosa (XLRP) is a debilitating disorder of the eye characterized by degeneration of rod and cone photoreceptors. Mutations in the RP2 gene are the second major cause of XLRP which associated with 10-15% of XLRP cases. To elucidate the molecular mechanism of pathogenesis of RP2-mediated photoreceptor degeneration, we utilized the Cre/loxp system to delete exon2, a mutational hotspot in humans, of the Rp2 gene in mice using transgenic mice expressing the Cre under the control of the ubiquitously expressing CAG promoter. The mutant retina (Rp2-conditional knock out; Rp2CKO) exhibited undetectable RP2 protein levels, as determined by immunoblotting and immunofluorescence analyses. The Rp2CKO mice showed progressive decline in photopic (cone) and scotopic (rod) ERG, starting at 2 months of age. Histological analysis revealed progressive degeneration of the photoreceptor layer in the mutant retina. Deletion of Rp2 resulted in disorganized outer segment discs in rods and cones, while sparing outer segment development. Degeneration of both dorsal and ventral cones and mislocalization of cone opsins to the nuclear and synaptic layers prior to the onset of degeneration were detected in the mutant retina. There was no detectable defect in the expression and localization of rod and cone arrestin, cone transducin subunits or RP2-interacting protein ARL3. Our study suggested that RP2 is involved in the maintenance of photoreceptor function and that cone opsin misliocalization plays a critical role in the pathogenesis of RP2-associated disease.

Biography

Linjing Li has completed her Ph.D from Tottori University Faculty of Medicine and conducted postdoctoral studies in SUNY Downstate Medical Center. She is now a postdoctoral fellow in University of Massachusetts Medical School. Her studies have been published in reputed journals.