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Special Issue on

Combining Clinical, Genetic and Biochemical Approaches for Added Value in Understanding Congenital Stationary Night Blindness

CALL FOR PAPERS

Congenital stationary night blindness (CSNB) is a nonprogressive clinically and genetically heterogeneous retinal disorder. Using classical linkage, candidate gene approaches, and next generation sequencing, eighteen different genes have been associated with CSNB, including genes coding for proteins of the phototransduction cascade, those important for signal transmission from the photoreceptors to the bipolar cells, and genes involved in retinoid recycling in the retinal pigment epithelium. Genetics and functional analyses of the affected proteins help to decipher retinal signaling pathways and mechanisms. Conversely, functional analyses of molecules implicated in retinal signaling may identify potential gene therapy targets in patients with CSNB. Clear genotype-phenotype correlations can be drawn from gene expression and protein localization profiles associated with specific gene defects. Despite progress on gene identification and elucidation of retinal signaling, novel mutations and signaling molecules remain to be identified to explain unsolved CSNB cases and to better understand retinal physiology and pathology. These studies show how basic science can be important for patient diagnosis and vise versa and will likely prove to be important in developing future treatments for retinal disorders.

The present special issue aims to publish high-quality clinical studies on genotype-phenotype correlations, as well as research articles on functional analyses on known and candidate proteins or on novel cellular or animal models related to CSNB.

Potential topics include but are not limited to the following:

- Genotype-phenotype correlations in genes implicated in CSNB
- Novel mutations in known genes underlying CSNB
- Novel candidate genes underlying CSNB
- Cellular functional analyses of mutations leading to CSNB
- Animal models with mutations causing a CSNB phenotype
- Novel animal and cellular models for CSNB
- Investigation of pre- and postsynaptic proteins implicated in CSNB

Authors can submit their manuscripts through the Manuscript Tracking System at https://mts.hindawi.com/submit/journals/bmri/ophthalmology/csnb/.

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