

Scholarly Activities:

The long term goal of my research is to understand the molecular events that underlie the formation and function of one of the synaptic layers, the outer plexiform layer (OPL), in the retina. This synapse connects the photoreceptors that collect light and convert it into an electrical signal, to the second order neurons, bipolar and horizontal cells. Early in development the pre- and post-synaptic neurons make contact and begin to form a synapse. This initial contact triggers a series of events that results in a synapse of extraordinary complexity. Three dendrites from post-synaptic neurons invaginate into the axon terminal of the presynaptic cell, which is the photoreceptor in this case. We have discovered a mutant mouse that fails to undergo normal maturation of the synapse and results in night blindness. This model is being used to determine what signals are involved in the maturation of this important

In a second project, we focus on the molecular components of the signal transduction cascade in the depolarizing bipolar cells. These cells utilize a metabotropic glutamate receptor, GRM6, to modulate the activity of the non-specific cation channel, TRPM1. My lab is investigating how several proteins, GRM6, TRPM1, GPR179, nyctalopin and LRIT3 interact to form a functional

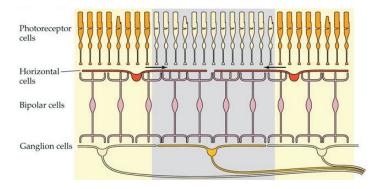
Grants:

Role: Principal Investigator

Title: Isolation of congenital stationary night blindness genes

Funding Agency: NEI

Direct Costs Funded: 2014 - R01 (\$298,059)



Publications (2013-2014):

Wilson GR, Sim JC, McLean C, Giannandrea M, Galea CA, Riseley JR, Stephenson SE, Fitzpatrick E, Haas SA, Pope K, Hogan KJ, **Gregg RG**, Bromhead CJ, Wargowski DS, Lawrence CH, James PA, Churchyard A, Gao Y, Phelan DG, Gillies G, Salce N, Stanford L, Marsh AP, Mignogna ML, Hayflick SJ, Leventer RJ, Delatycki MB, Mellick GD, Kalscheuer VM, D'Adamo P, Bahlo M, Amor DJ, Lockhart PJ. Mutations in RAB39B Cause X-Linked Intellectual Disability and Early-Onset Parkinson Disease with α-Synuclein Pathology. *Am J Hum Genet*. 95:729-35. (2014) PMID: 25434005

Ray TA, Heath KM, Hasan N, Noel JM, Samuels IS, Martemyanov KA, Peachey NS, McCall MA, **Gregg RG**. GPR179 is required for high sensitivity of the mGluR6 signaling cascade in depolarizing bipolar cells. *J Neurosci*. 34:6334-43 (2014). PMID: 24790204

Balmer J, Ji R, Ray TA, Selber F, Gassmann M, Peachey NS, **Gregg RG**, Enzmann V. Presence of the Gpr179(nob5) allele in a C3H-derived transgenic mouse. Mol Vis. 19:2615-25. (2013). PMID: 24415894

Klooster J, van Genderen MM, Yu M, Florijn RJ, Riemslag FC, Bergen AA, **Gregg RG**, Peachey NS, Kamermans M. Ultrastructural localization of GPR179 and the impact of mutant forms on retinal function in CSNB1 patients and a mouse model. *Invest Ophthalmol Vis Sci.* 54:6973-81. (2013) PMID: 24084093

External Professional Activities (2013-2014):

2013 - NIH ZRG1 F08-B (20) Fellowships: Genes, Genomes and Genetics (October meeting)

2014 - NIH ZRG1 F08-B (20) Fellowships: Genes, Genomes and Genetics (January, May and October meetings)