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Research Activities:

The research in the laboratory continued to focus on the mechanisms and genes responsible for two forms of congenital stationary night blindness (CSNB). Mice deficient in transmission between photoreceptors and second order neurons were analyzed. Mutations in the alpha-1F subunit of voltage dependent calcium channels (Cacn1af) and the metabotropic glutamate receptor 6 (Grm6) subunit were characterized. Mutations in the Cacn1af subunit result in the dendrites of post synaptic neurons extending abnormally, while the retinal morphology of the Grm6 mutant mice is normal. In a third model of CSNB, the nob mouse, studies showed that there is abnormal development resulting in spontaneous bursting of retinal ganglion cells. These mice also fail to form center surround organization of retinal ganglion cells, which is thought to be important in vision. Transgenic rescue of the phenotype of the nob mouse by expression of a EYFP:nyctalopin fusion protein, showed that the nyctalopin was only required at the first synapse to fully restore vision. The availability of this line of mice should allow the function of nyctalopin to be determined.

A second project has established an international zebrafish mutant mapping resource. This facility identified the gene responsible for 17 mutant lines of fish and provided chromosomal locations for an additional 34 to investigators world wide. This facility is greatly increasing the rate at which mutant zebrafish genes are identified.

Grants Funded:

Role: Principal Investigator

Title: Isolation of congenital stationary night blindness gene

Funding Agency: NIH

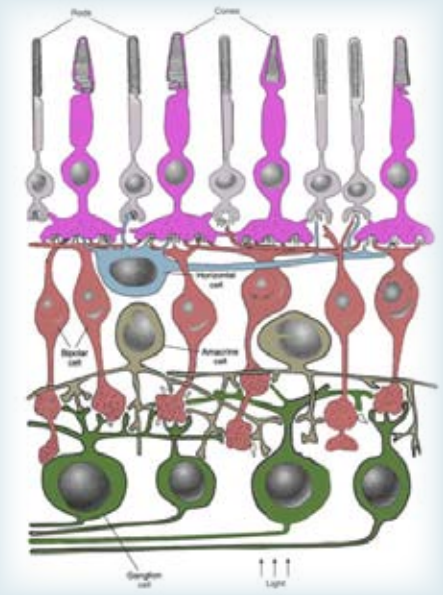
Direct Costs Funded: \$286,573

Role: Collaborator (Mervis PI)

Title: Genotype/phenotype correlations in Williams Syndrome

Funding Agency: NIH

Direct Costs Funded: \$700,000



Role: Principal Investigator

Title: Zebrafish Mutant Mapping Facility

Funding Agency: NIH

Direct Costs Funded: \$237,119

Peer-reviewed Publications:

Maddox DM, Vessey KA, Yarbrough GL, Invergo BM, Cantrell DR, Inayat S, Balannik V, Hicks WL, Hawes NL, Byers S, Smith RS, Hurd R, Howell D, **Gregg RG**, Chang B, Naggert JK, Troy JB, Pinto LH, Nishina PM, McCall MA. Allelic variance between GRM6 mutants, Grm6nob3 and Grm6nob4 results in differences in retinal ganglion cell visual responses. *J Physiol*. In press (2008).

McCall M, **Gregg RG**. Comparisons of structural and functional abnormalities in mouse b-wave mutants. *J Physiol*. In press (2008)

Marshall CR, Young EJ, Pani AM, Freckmann ML, Lacassie Y, Howald C, Fitzgerald KK, Peippo M, Morris CA, Shane K, Priolo M, Morimoto M, Kondo I, Manguoglu E, Berker-Karauzum S, Edery P, Hobart HH, Mervis CB, Zuffardi O, Reymond A, Kaplan P, Tassabehji M, **Gregg RG**, Scherer SW, Osborne LR. Infantile spasms is associated with deletion of the MAGI2 gene on chromosome 7q11.23-q21.11. *Am J Hum Genet*. 83:106-11 (2008).

Lee J, Willer JR, Willer GB, Smith K, **Gregg RG**, Gross JM. Zebrafish blowout provides genetic evidence for Patched1-mediated negative regulation of Hedgehog signaling within the proximal optic vesicle of the vertebrate eye. *Dev Biol*. 319:10-22 (2008).

Gregg RG, Kamermans M, Klooster J, Lukasiewicz PD, Peachey NS, Vessey KA, McCall MA. Nyctalopin expression in retinal bipolar cells restores visual function in a mouse model of complete X-linked congenital stationary night blindness. *J Neurophysiol*. 98:3023-33 (2007)

Palmer CA, Hollis DM, Watts RA, Houck LD, McCall MA, **Gregg RG**, Feldhoff PW, Feldhoff RC, Arnold SJ. Plethodontid modulating factor, a hypervariable salamander courtship pheromone in the three-finger protein superfamily. *FEBS J*. 274:2300-10 (2007).