SELECTED PUBLICATIONS SUBMITTED BY DR. WIGGS:

   This paper describes the identification of genetic variants distributed throughout the retinoblastoma gene and the use of those variants to identify individuals who are at risk for developing retinoblastoma. This was the first use of gene variants as a gene-based test capable of predicting inherited disease.

   Using a collection of sibling pairs affected with adult-onset primary open angle glaucoma we completed the first genome-wide linkage study for glaucoma and identified novel disease loci on chromosomes 14q and 15q.

   In this study we use a collection of families affected by childhood glaucoma for a genome-wide linkage study that identified two novel genomic regions likely to contain causative genes for this disease.

   Using a large case control sample from the United States, in this study we provided the first independent replication of the association of primary open angle glaucoma with CAV1/CAV2 genetic variants. The initial association was found in a population from Iceland and our results demonstrated that the association was applicable to populations with varied ethnicity such as the United States.

   Described in this article is the organization of the NEIGHBOR consortium and the clinical variables collected for over 3000 primary open angle cases and 3000 controls, the largest glaucoma case-control sample world-wide.

Haines JL. (2012) Common variants at 9p21 and 8q22 are associated with increased susceptibility to optic nerve degeneration in glaucoma. PLoS Genetics, 2012;8(4):e1002654. This paper presents the results of the NEIGHBOR primary open angle glaucoma association study which is the largest case control study for glaucoma to date. This study identified for the first time genomic regions associated with the glaucoma subgroup with increased optic nerve susceptibility.

7. Pasquale LR, Loomis SJ, Kang, JH, Yaspan BL, Abdrabou W, Budenz DL, Chen TC, Delbono E, Friedman DS, Gaasterland D, Gaasterland T, Grosskreutz CL, Lee RK, Lichter PR, Liu Y, McCarty CA, Moroi SE, Olson LM, Realini T, Rhee DJ, Schuman JS, Singh K, Vollrath D, Wollstein G, Zack DJ, Allingham RR, Pericak-Vance MA, Weinreb RN, Zhang K, Hauser MA, Richards JE, Haines JL, Wiggs JL. (2013) CDKN2B-AS1 Genotype - Glaucoma Feature Correlations in Primary Open-Angle Glaucoma Patients from the United States. Am J Ophthalmol. Feb;155(2):342-353.e5. This study from the NEIGHBOR consortium showed that genetic variants in the CDKN2BAS1 genomic region are primarily associated with glaucoma patients who have severe optic nerve disease despite normal intraocular pressures. This subgroup of glaucoma patients has an increased susceptibility to optic nerve disease compared with other glaucoma patients and this subgroup is most likely to become blind from the disease.


This study from the NEIGHBOR(HOOD) consortium shows that the genetic variants in the CAV1/CAV2 locus are primarily associated with the subgroup of glaucoma patients who have loss of central vision. These patients are more likely to become blind from the disease. These results could lead to a DNA-based diagnostic test identifying glaucoma patients who are at increased risk of losing central vision.


This study from the NEIGHBOR(HOOD) consortium reports on the first pathway analysis using genome-wide genotype data for primary open angle glaucoma (POAG) and normal-tension glaucoma (NTG). These results show that genes in the GABA metabolism pathways and Acetyl-CoA metabolism pathways are associated with POAG and NTG.


This study reports on the phenotype of the LOXL1 null mouse showing some but not all features of human pseudoexfoliation syndrome known to be significantly associated with LOXL1 gene variants.


In this study from the IGGC genome-wide genotype data was analyzed for over 35,000 individuals to find 4 new loci for intraocular pressure, a major risk factor for development of glaucoma. Dr. Wiggs is one of several senior/last authors for this manuscript.