

Principal Investigator: Lucia Sobrin

Institution Affiliation: Broad

Ancillary Study Title: Genome wide association scan for diabetic retinopathy in the Jackson Heart Study

Project Overview:

The purpose of this study is to perform genome-wide association scan (GWAS) to identify single nucleotide polymorphisms (SNPs) associated with diabetic retinopathy, including macular edema and proliferative retinopathy phenotypes.

Prospective, cross-sectional, single center, GWAS of diabetic retinopathy in diabetic African-Americans.

Standard, seven-field color fundus photography will be performed on all diabetic and impaired glucose tolerance (IGT) patients in the Jackson Heart Study. Fundus photographs will be evaluated by standardized grading criteria for the presence and grade of nonproliferative diabetic retinopathy, proliferative retinopathy and macular edema by two independent graders. A third person will serve as an arbitrator in cases where the first two graders do not agree. We will take advantage of the concurrent GWAS planned in the JHS. A case-control analysis comparing diabetic subjects without retinopathy and diabetic subjects with retinopathy will be performed. In addition, severity of retinopathy will be examined as a quantitative trait in the GWAS.

If genes associated with diabetic retinopathy are identified, they will provide insights into pathophysiology of this complication and may lead to development of new therapies for retinopathy. In addition, identification of diabetic retinopathy genes may provide information with which to counsel African American patients regarding their risk of retinopathy and lead to more targeted therapy of this complication in African Americans.