

## **Award winner-Puja Mehta Board 2022T**

Transcriptomic and Genetic regulation atlas of Glaucoma Relevant eye Tissues: Aqueous Outflow Pathways, Macula, and Optic Nerve Head.

**Mauricio Garcia:** Board 5051W:

Heterozygous Rare Loss-of-Function Variants in LTBP2 Confer Increased Risk for Adult-Onset Glaucoma: A Large-Scale Gene Burden Test in the UK Biobank

**Elizabeth Rossin:** Board 7039W:

Low population penetrance of variants associated with inherited retinal disease

### **Puja Mehta Board 2022T – Award winner**

Transcriptomic and Genetic regulation atlas of Glaucoma-Relevant eye Tissues: Aqueous Outflow Pathways, Macula, and Optic Nerve Head.

**Sudeep Mehrotra:** Board 9196T

Validation of an Unbiased Metagenomic Sequencing Approach for Diagnosis of Infectious Uveitis

### **Michelle Bartolo:** Board 9197T – Award winner

Developing a Cell-Type Specific and Multi-Ancestry Polygenic Risk Score Method with Applications to Glaucoma.

**Riccardo Sangermano:** Board 4046T:

Elucidating retinal gene function with in-vivo CRISPR-based gene perturbation assay

**Kinga Bujakowska:** Board 7040T: Uncovering Hidden Structural and Splicing Variants in Unresolved Inherited Retinal Diseases

**Songtao Xu:** Board 5026T: Developing a single-cell allele-specific expression pipeline and its application to ocular tissues and interpretation of genetic associations with glaucoma October

**Rinaldo Catta-Preta:** Board 8047T: Detecting high confidence open chromatin regions in four anterior and posterior eye tissues and their application to fine-mapping glaucoma associations.

**Swanand Koli:** Board 9200tT: Gene Supplementation Restores Retinal Ganglion Cell Function in a Mouse Model of Familial Dysautonomia

**Marianna Weener:** Board 5034F – *Award winner*  
Comparison of Empiric Data with *in silico* Prediction of Deep Intronic Pathogenicity in Inherited Retinal Disorders

**Evelyn Harper:** Board 8002F  
Advancing Genetic Diagnosis in Inherited Retinal Diseases Through Diagnostic Whole Genome Sequencing  
Friday October 17 2:30 PM - 4:30 PM

**Emily Place:** Board 2024F  
Enhancing Variant Curation Guidelines for ACO2-Related Optic Atrophy Utilizing the ClinGen Framework

**Meghana Dutta:** Board 9154F: Normal tension glaucoma genome-wide association study supports CDKN2B-AS and SIX6 as major disease loci

**Inas Aboobakar:** Board 9104F: The paradox of phenotype precision: striking the balance between statistical power and refined phenotyping reveals a novel African POAG susceptibility

locus in the All of Us Research Program

**Jyoti Lama:** Genomic Causes and Consequences in Drug Treatment Platform session

Genome-wide association study for Glucocorticoid-induced Ocular Hypertension

Saturday, October 18, 8:30 AM - 9:30 AM



