eyeGENE®

Envisioning cures for rare inherited eye disorders

What is eyeGENE®?

eyeGENE®, also known as the National Ophthalmic Disease Genotyping and Phenotyping Network, was launched by the National Eye Institute (NEI) in 2006 to facilitate research into the causes and mechanisms of rare inherited eye diseases and the development of treatments and cures. A public-private partnership, the eyeGENE® Network is a collaboration among the U.S. federal government, eye health providers across the U.S. and Canada, certified molecular diagnostic laboratories, private industry, and the vision research community. eyeGENE® components include a patient registry, a curated genotype/phenotype data repository, and a DNA biorepository.

How is eyeGENE® funded?

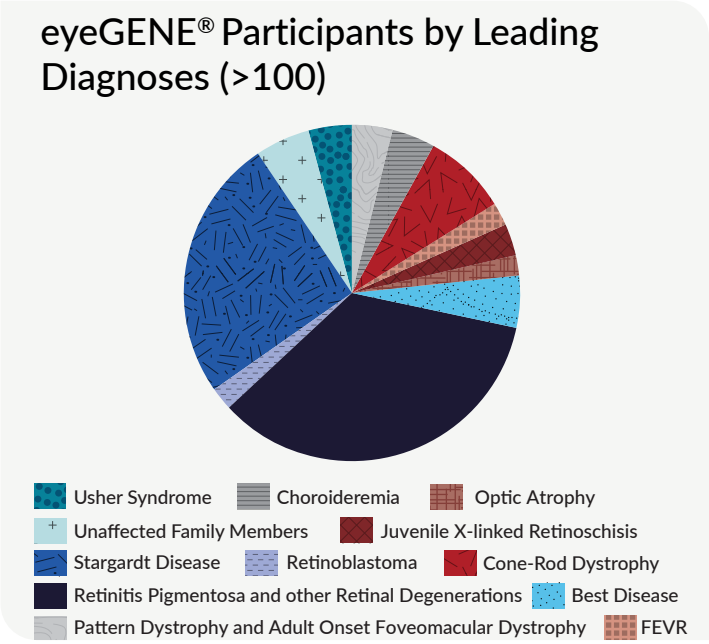
eyeGENE® is funded by federal support through the NEI, a part of the National Institutes of Health (NIH). NIH is the nation's medical research agency.

What does eyeGENE® do?

eyeGENE® connects scientists studying rare eye disease with people who have a rare inherited eye disease and want to participate in clinical research. While rare eye diseases collectively affect thousands of people, each individual disease affects relatively few people. Finding adequate numbers of people with specific mutations to study and participate in clinical trials can be challenging. Patients with rare conditions often have difficulty finding clinicians with relevant expertise.

Why study genes?

Identifying disease genes can lead to breakthrough therapies. For example, in the 1990s, researchers linked a gene called *RPE65* to the blinding eye disease Leber congenital amaurosis (LCA). In 2008, clinical trials funded by the NEI and others showed that *RPE65* gene therapy could improve the vision of people with LCA caused by this genetic mutation. Then in 2017, a gene therapy called voretigene neparvovec (Luxturna) received approval by the U.S. Food and Drug Administration (FDA) for the treatment of LCA caused by the *RPE65* mutation. Luxturna is the first FDA-approved gene-replacement therapy of any kind. Gene therapy trials for other rare eye diseases, including retinoschisis, Stargardt disease, retinitis pigmentosa, and others are underway. Their success is uncertain, which is why research to understand the genetics and disease-causing mechanisms of rare eye diseases needs continued support.



How does eyeGENE® share data?

eyeGENE® maintains a central biorepository of participant DNA samples and other clinical information, the result of partnering with more than 400 registered eye health providers and genetic testing laboratories. The biorepository is a resource open to anyone studying rare eye diseases. eyeGENE® uses the NIH Biomedical Research Informatics Computing System and has adopted Common Data Elements to enable analysis and harmonization of data across multiple sources.

What progress has eyeGENE® made?

eyeGENE® has accrued more than 6,400 participants with a variety of rare inherited eye conditions. eyeGENE® continues to identify new eye disease-causing genes, and to expand the overall scientific knowledge about genes. More than 1,600 eyeGENE® participants have received genetic test results indicating potential eligibility for current ongoing clinical trials. Eligibility for clinical trials may be based on genetics as well as other inclusion criteria.

How does eyeGENE® ensure sample quality?

eyeGENE® provided financial support to help partners meet federally mandated diagnostic testing requirements through the Clinical Laboratory Improvements Act (CLIA). In addition, the Network established quality control protocols and standard operating procedures in lab and biobank science management. eyeGENE® promotes quality assurance testing and review for each Network CLIA-certified laboratory for accurate reporting and responsible interpretations.

National Ophthalmic Disease Genotyping Network

eyeGENE®

How many genes are included in eyeGENE®?

Scientists have identified hundreds of genes related to eye disease. The eyeGENE® Network has the capacity to screen participants for up to 700 genes in over 35 disease categories.

How do eyeGENE® participants benefit?

eyeGENE® participation has the potential to empower affected individuals with knowledge about their disease. Participants can gain access to specialists who are familiar with their unique condition and can learn the latest information about clinical trials and new therapies. The results from eyeGENE® may help clinicians confirm a participant's diagnosis or may lead to an alternative diagnosis. Both scenarios lead to better clinical care. Scientists benefit from participating in eyeGENE® by gaining access to hard-to-find patient data and samples and by reaching a network of collaborators.

How can patients and researchers participate in eyeGENE®?

In 2015, eyeGENE® paused accepting new participants as many commercial diagnostic testing laboratories were making diagnostic testing available for ocular disease. The Network will soon allow additional participants to join the eyeGENE® registry so they can be matched with clinical studies. The NEI is exploring other ways for researchers and affected individuals to contribute to a newly reinvigorated eyeGENE® program.

How can I learn more about eyeGENE®?

For more information, visit: eyeGENE.nih.gov

NIH National Eye Institute
nei.nih.gov