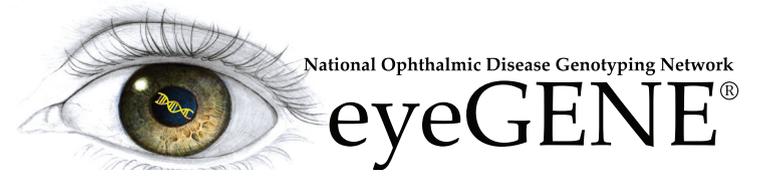




# What are the benefits and risks of eyeGENE®?



## Benefits

- **For patients:** the possibility to aid research, receive molecular diagnostic genotyping, and be contacted for future research studies.
- **For referring eye healthcare providers:** access to a secure centralized system for genetic testing and the ability to contribute to the standardization/refinement of clinical phenotypic descriptors. Providers also have the opportunity to be contacted for possible research collaborations.
- **For researchers:** a repository of linked phenotype-genotype data, and the possibility of identifying individuals with inherited eye diseases for research and/or future clinical trials.

## Risks

Genetic testing may provide information about how an eye condition is passed on within a family, which may cause stress or anxiety to some individuals and may affect family relationships. Insurance or work discrimination could occur if genetic information is disclosed, even though a federal anti-discrimination law (Genetic Information Nondiscrimination Act of 2008, or GINA) is in place. Risks are further described in the consent form and you and your patients are encouraged to ask questions.

## How do I get started?

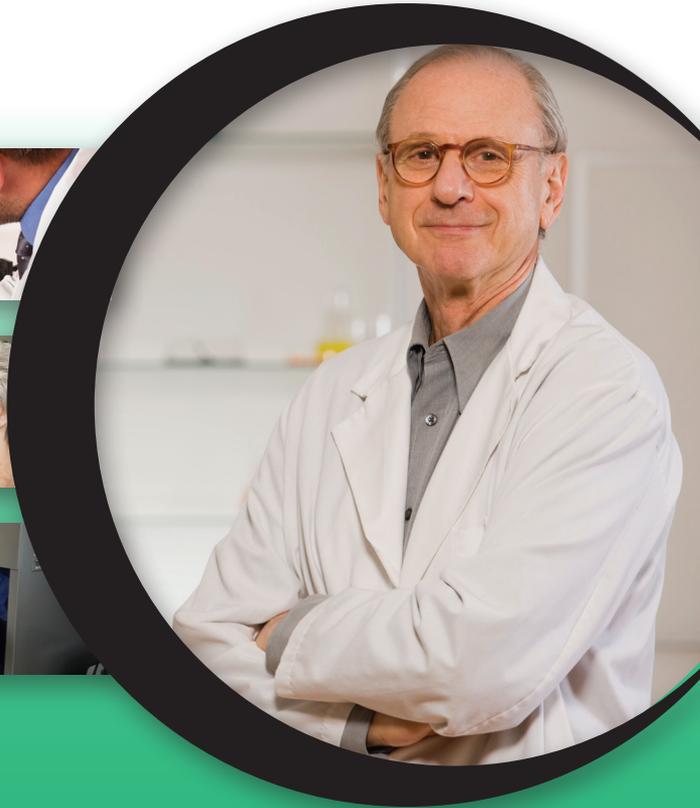
**Please contact us at:**  
 eyeGENE® Coordinating Center  
 National Eye Institute  
 National Institutes of Health

Telephone  
 301-435-3032

E-mail  
[eyeGENEinfo@nei.nih.gov](mailto:eyeGENEinfo@nei.nih.gov)

Website  
<http://www.nei.nih.gov/eyegene>

\*Clinical Laboratory Improvement Amendments (CLIA)  
<http://www.cms.gov/CLIA>



# What You Should Know

## INFORMATION FOR PHYSICIANS

*Preserving vision through genetics*





## What is eyeGENE®?



The National Ophthalmic Disease Genotyping and Phenotyping Network (eyeGENE®) was created by the National Eye Institute (NEI), part of the National Institutes of Health (NIH), in partnership with laboratories across the vision research community. One mission of the eyeGENE® Network is to facilitate research into the genetic causes of ophthalmic disease by broadening patient accessibility to diagnostic genetic testing. The eyeGENE® Network currently includes a Coordinating Center at NEI, more than 10 CLIA\*-approved molecular genetic testing laboratories around the Nation, a centralized repository for blood/DNA, and a de-identified genotype/phenotype database. The aim of this initiative is to stimulate patient and eye healthcare provider interest in genetics-based clinical care and generate involvement in ophthalmic research.



## How can I get my patients enrolled in eyeGENE®?

1. Determine if a patient has an inherited eye disease that makes the patient eligible to enroll in eyeGENE®. A list of inherited eye conditions and corresponding genes for which testing is currently available can be found at <http://www.nei.nih.gov/resources/eyegene.asp>
2. Register online at <https://nationaleyegene.nei.nih.gov> to become an eyeGENE® database user. The eyeGENE® Coordinating Center will review your information for approval. Any healthcare provider who follows patients for an eye-related condition may register (e.g., ophthalmologist, optometrist, geneticist, genetic counselor, neurologist, etc.).
3. Fill out the required family history and clinical questions online in the database. The eyeGENE® working group will review the information and determine which gene tests may be available through eyeGENE® or if more information is needed.
4. Ensure that the patient completes and signs a Research Consent Form and a DNA Diagnostic Form. Patients may choose to receive their molecular test results back and/or be re-contacted for future research or clinical treatment studies.
5. Complete the Clinician's Assurance Form and provide or arrange for genetic counseling to the patient, pre- and post-testing.



6. Draw approximately 30 ml of blood from adult participants or 7 ml of blood from children in K2 EDTA tubes.
7. Ship the samples to the Coordinating Center along with the proper forms. Forms include the Clinician's Assurance, Blood Specimen Shipping Request printed from the database, Research Consent Form (and Assent Form for minors), and DNA Diagnostic Consent Form.  
**NOTE:** There are no costs to you or the patient for molecular diagnostic testing. eyeGENE® will cover these costs. However, blood draw and shipping charges are NOT covered by eyeGENE®.

Once results are available, a molecular diagnostic report is sent to you, and results are available with your login on the secure eyeGENE® database.

**NOTE:** Turnaround time for the receipt of molecular diagnostic results varies and often depends on the gene(s) tested and availability through an eyeGENE® Network CLIA laboratory. The average turnaround time is between four months to one year.

DNA extracted from the blood samples and de-identified data for patients enrolled in eyeGENE® is available to vision researchers who have received approval for a proposal through the eyeGENE® Resource Access Subcommittee (part of the eyeGENE® Steering Committee, which oversees the eyeGENE® program).

Preserving vision through genetics