

National Ophthalmic Genotyping and Phenotyping Network (eye-GENE (Registered Trademark)), Stage 3 - Expansion of DNA and Data Repositories for Rare Inherited Ophthalmic Diseases

NCT06491615

Status	RECRUITING
Sponsor	National Eye Institute (NEI)
Enrollment	1,000 participants

Key Eligibility Criteria

Inclusion (11)

- In order to be eligible to participate in this study, an individual must meet all of the following criteria:
- The participant must present with characteristics consistent with one of the following diagnoses:
 - Aniridia
 - Best disease
 - Blue-cone monochromacy
- ... and 6 more (see full listing online)

Exclusion (6)

- An individual who meets any of the following criteria will be excluded from participation in this study:
 - Those with impaired decision-making capability who do not have a legally-authorized representative.
 - Those unable to provide a saliva sample OR have any disease or condition that makes it unsafe for a subject to provide a suitable blood sample of at least 5 mL to yield more than 50 micrograms of DNA.
- An individual who meets any of the following criteria will be excluded from participation in the optional retinal imaging:
 - Those with a history of epilepsy.
- ... and 1 more (see full listing online)

Locations (2 total)

National Eye Institute (NEI), Bethesda, Maryland, United States
National Institutes of Health Clinical Center, Bethesda, Maryland, United States