# Resource Summary Report

Generated by dkNET on May 19, 2025

## eyeGENE

RRID:SCR_004523
Type: Tool

### Proper Citation

eyeGENE (RRID:SCR_004523)

### Resource Information

**URL:** https://eyegene.nih.gov

**Proper Citation:** eyeGENE (RRID:SCR_004523)

**Description:** National network of research laboratories for genetic testing of eye disease. They offer testing for affected individuals coupled to registry of clinical information available through patient registry. Large data set for investigators to identify additional genetic risk factors and to explore relationship between genetic disease (genotype) and its clinical manifestation (phenotype).

**Abbreviations:** eyeGENE

**Synonyms:** eyeGENE, National Ophthalmic Disease Genotyping Network (eyeGENE), National Ophthalmic Disease Genotyping Network (eyeGENETM), National Ophthalmic Disease Genotyping Network

**Resource Type:** material resource, biomaterial supply resource

**Defining Citation:** PMID:22847030

**Keywords:** familial exudative vitreal retinopathy, fzd4, foxc1, abca4, aniridia, pax6, axenfeld - rieger syndrome, pitx2, best's disease, vmd2, bietti's crystalline corneal-retinal dystrophy, cyp4v2, c1qtnf5/ ctrp5, ca4, choroideremia, chm, cnga1, cone rod dystrophy, abca4, congenital cranial dysinnervation disease, kif21a, congenital stationary night blindness, nyx, corneal anterior stromal dystrophy, bigh3, crb1, doyne honeycomb dystrophy, efemp1, glaucoma, cyp1b1, hoxa1, impdh1, juvenile x-linked retinoschisis, xlrs1, krt12, lrp5, meesmann's epithelial dystrophy, krt3, mertk, myoc, ndp, optic atrophy, opa1, optn, pantothenate kinase-associated neuropathy, pank2, pattern dystrophy, rds, pde6a, pde6b, phox2a, prpf31, retinitis pigmentosa, retinal degeneration, abca4, rgr, rho, rlbp1, robo3, rp1,

rp2, rpe65, rpgr, sall4, sorsby fundus dystrophy, timp3, stargardt disease, elovl4, tulp1, genotype, phenotype, diagnostic, genotyping, clinical trial, genetic eye disease, blood, dna, cell line, genetic testing, treatment, genetics, ophthalmic disease, eye

**Related Condition:** Genetic eye disease, Family member

**Funding:** NEI

**Availability:** Restricted

**Resource Name:** eyeGENE

**Resource ID:** SCR_004523

**Alternate IDs:** nif-0000-00229

**Alternate URLs:** https://eyegene.nih.gov/node/38, https://eyegene.nih.gov/node/36

**Record Creation Time:** 20220129T080225+0000

**Record Last Update:** 20250517T055644+0000

## Ratings and Alerts

No rating or validation information has been found for eyeGENE.

No alerts have been found for eyeGENE.

## Data and Source Information

**Source:** SciCrunch Registry

## Usage and Citation Metrics

We found 14 mentions in open access literature.

**Listed below are recent publications.** The full list is available at dkNET.

Markovi? L, et al. (2023) Genetics in ophthalmology: molecular blueprints of retinoblastoma. Human genomics, 17(1), 82.

Mansouri V, et al. (2023) X-Linked Retinitis Pigmentosa Gene Therapy: Preclinical Aspects. Ophthalmology and therapy, 12(1), 7.

Zernant J, et al. (2022) Rare and common variants in ROM1 and PRPH2 genes trans-modify Stargardt/ABCA4 disease. PLoS genetics, 18(3), e1010129.

Xu S, et al. (2020) Mutation Screening in the miR-183/96/182 Cluster in Patients With Inherited Retinal Dystrophy. Frontiers in cell and developmental biology, 8, 619641.

Ameri H, et al. (2020) Autofluorescence of choroidal vessels in Bietti's crystalline dystrophy. BMJ open ophthalmology, 5(1), e000592.

Foote KG, et al. (2020) Comparing Cone Structure and Function in RHO- and RPGR-Associated Retinitis Pigmentosa. Investigative ophthalmology & visual science, 61(4), 42.

Gudiseva HV, et al. (2019) Next-Generation Technologies and Strategies for the Management of Retinoblastoma. Genes, 10(12).

Navale V, et al. (2019) Development of an informatics system for accelerating biomedical research. F1000Research, 8, 1430.

Gaier ED, et al. (2017) Diagnostic genetic testing for patients with bilateral optic neuropathy and comparison of clinical features according to OPA1 mutation status. Molecular vision, 23, 548.

Gao J, et al. (2017) Retrospective analysis in oculocutaneous albinism patients for the 2.7 kb deletion in the OCA2 gene revealed a co-segregation of the controversial variant, p.R305W. Cell & bioscience, 7, 22.

Arno G, et al. (2016) Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American journal of human genetics, 99(6), 1305.

Sun LW, et al. (2016) Multimodal Imaging of Photoreceptor Structure in Choroideremia. PloS one, 11(12), e0167526.

Collins DW, et al. (2013) Mitochondrial sequence variation in African-American primary open-angle glaucoma patients. PloS one, 8(10), e76627.

Gabriel LA, et al. (2011) Genetic diagnostic methods for inherited eye diseases. Middle East African journal of ophthalmology, 18(1), 24.