Genetic testing outcomes in a nationwide cohort of **441 retinitis pigmentosa** patients

Background

- Retinitis pigmentosa (RP) is a genetically heterogeneous disease leading to progressive vision loss.
- Genetic testing is critical for RP diagnosis, prognosis, and management, especially now that gene-specific treatments and trials are available.

Aim

To describe genetic testing outcomes of patients with RP from a nationwide genetic counseling service.

Methods

Retrospective chart review of patients with a clinical diagnosis of RP seen at Genome Medical 2019-2025

RetNet database was used to characterize genes by inheritance pattern

Analysis using t-tests and Fisher's exact test

Results

n	Sex
441 Patients	**54.5%** Female (240/440)
18y or Older **95%** (416/440)	Mean Age **50.1y** SD 18.0y

441 patients with RP

→ **48.5%** (214/441) RP genotype

→ **43 different RP genes**
Most frequently:
23.8% (51/214) ***USH2A***
11.2% (24/214) ***RHO***
6.5% (14/214) ***EYS***
5.6% (12/214) ***PRPH2***
4.7% (10/214) ***RP1***

→ **50.8%** (224/441) uninformative genetic testing

→ **0.7%** (3/441) non-RP genotype

→ **non-RP genes**
33% (1/3) ***COL2A1***
33% (1/3) ***LCA5***
33% (1/3) ***OTX2***

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Results

Patients from **47** different states.
Most frequently:

- Texas: 15.5%
- New York: 8.1%
- Florida: 8.1%
- California: 7.7%
- Louisiana: 5.6%

RP Inheritance Patterns

57.5% Autosomal Recessive (123/214)
35% Autosomal Dominant (75/214)
7.5% X-Linked (16/214)

Diagnostic Yield Over Time

42.8% **2019 - 2021**
56.4% **2022 - 2025**
$p=0.006$

Conclusion

- This RP cohort is genetically heterogeneous, as seen in previous cohorts.
- Yield in testing increased over time, possibly due to changes in referrals or gene panels.
- Genetic diagnosis is increasingly important as gene-specific treatment and trials are available.