



Importance of Genetic Testing

- Provides an accurate diagnosis
- Determines potential treatment options or involvement in clinical trials
- Informs patients about the potential risk of disease to other family members
- Identifies the potential risk to other organs that may be affected
- Children and infants can benefit from early diagnosis and intervention

Genetic Testing Patient Experience

Routine checkup/symptoms



Clinical evaluation by optometrist or ophthalmologist (IRD suspected)



Referred to retina specialist



Referred to genetic testing and counselling



Patient agrees to genetic testing and counselling



Pathogenic or likely pathogenic variant detected



Variant of uncertain significance detected



No variant, benign variants detected



- Genetic counselling
- Clinical management
- Test at risk family members
- Determine eligibility for available clinical studies



- Consider testing of family members
- Consider re-test



No genetic cause identified



No genetic disease



Clinical Management according to current guidelines



Periodic check-in with expert provider. (Retesting if new tests/treatments emerge)



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Genetic Testing Fact Sheet

A genetic diagnosis is a prerequisite for IRD patients to be considered for inclusion in research and clinical trials. Genetic testing and counselling can lead to greater understanding by the patient of inheritance patterns and disease progression associated with their gene. RI's Genetic Testing Landscape study shows that a best practice model on access to genetic testing and counselling services for IRDs is needed urgently.

*These data are preliminary findings from the RI Global Genetic Testing Landscape Study carried out in 2021.

Over

70%

of respondents said that there is an inadequate awareness about the benefits of genetic testing for IRDs among health care professionals.

36%

of eyecare professionals were either not aware of genetic testing for IRDs, remained neutral on patients going through the process, or did not encourage it.

Almost

90%

of respondents said that genetic testing was worth it.

Of those respondents,

32.5%

had to visit more than five physicians before receiving a genetic test.

56%

waited more than three years for a genetic diagnosis

42%

did not receive genetic counselling prior to genetic testing

34%

did not receive genetic counselling after receiving a genetic test

<https://retina-international.org/>