IMO GRUPO MIRANZA | Instituto de microcirugía ocular

Genetic diagnosis and counselling at **IMO Grupo Miranza**

WHAT IS GENETIC COUNSELLING?

Genes are analysed based on a blood sample that is taken from a patient affected by a hereditary eye disease. **The aim of this study is to identify the genetic alteration** leading to the disease.

What is a hereditary disease?

Hereditary diseases often have the following characteristics:

- they are transmitted within the same family
- they are caused by an altered gene
- they may follow different inheritance patterns

Most hereditary eye diseases are minority and roughly affect less than 1 in every 2,000 people. At IMO Grupo Miranza, we provide genetic diagnosis and counselling for more than 45 hereditary diseases that affect the eyesight.

WHAT IS THE PURPOSE OF GENETIC DIAGNOSIS?

Genetic diagnosis enables us to:

1. **Confirm the clinical diagnosis** of the disease, as some diseases have similar symptoms and/or might be difficult to diagnose.
2. **Ascertain how the disease will evolve,** as its prognosis might vary depending on the type of genetic alteration.
3. **Inform the affected family.** This involves a genetic counselling visit to identify the most likely inheritance pattern, which is the way in which the disease may be transmitted within the family.
4. **Inform relatives** who might be carriers of the disease, as they could transmit it or suffer from it in the future.
5. Prepare the affected patient for **future gene and cell therapies.**
6. Find out more about hereditary diseases and **identify new genes responsible.**

SHOULD I HAVE A GENETIC STUDY?

We recommend requesting a genetic study if you meet any of these conditions:

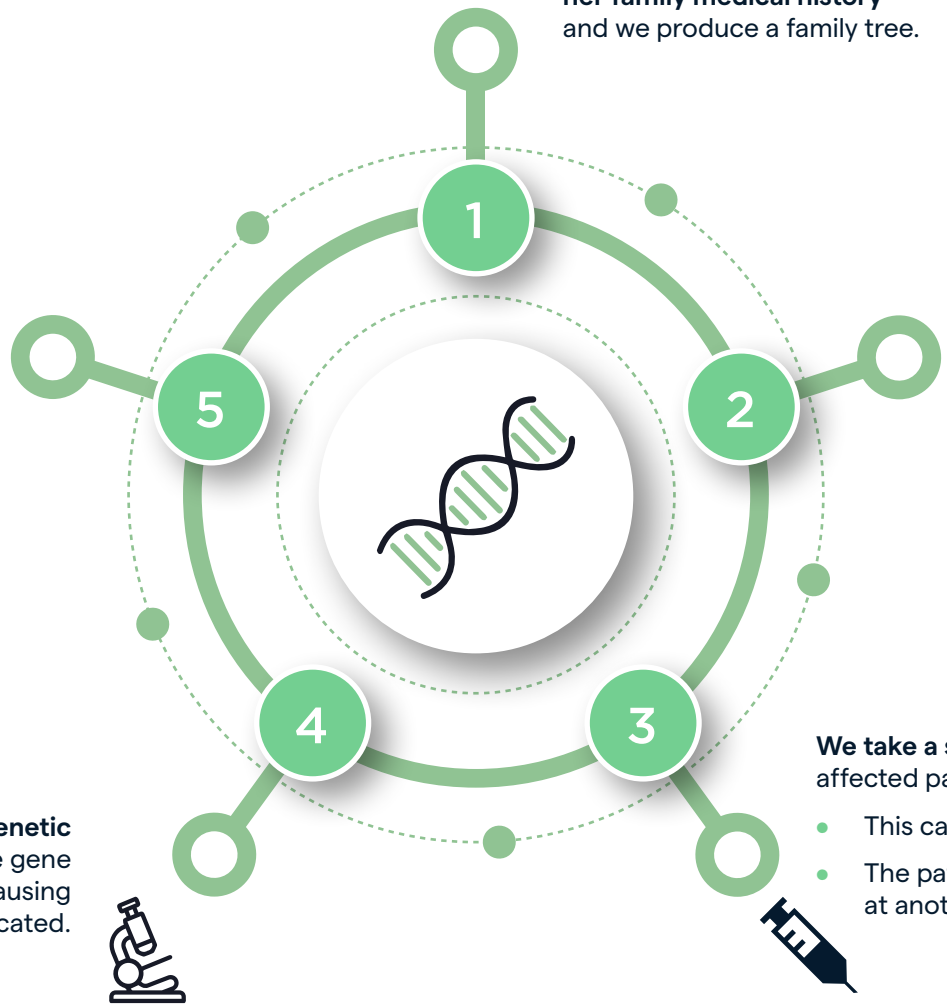
1. In your family medical history there are **people with hereditary or recurrent eye diseases.**
2. You have been diagnosed with a **hereditary eye disease.**
3. You want to have children and there are cases of hereditary eye disease in your family or in that of your partner.
4. One of your children suffers from a hereditary eye disease.

GROUND-BREAKING IN DIAGNOSIS AND RESEARCH

IMO Grupo Miranza Barcelona is the only ophthalmology centre in Spain to have its own molecular biology laboratory, which is ground-breaking in the genetic diagnosis of hereditary eye diseases. These facilities are home to ambitious basic research projects promoted by the IMO Foundation.

HOW IS A GENETIC STUDY PERFORMED?

Once the ophthalmologist has performed the clinical diagnosis, we carry out the genetic study in 5 steps:



The Genetics Department **makes an appointment with the patient to review his or her family medical history** and we produce a family tree.

We establish the possible **inheritance pattern for the disease and determine which genes must be analysed.**

We take a sample of blood or saliva from the affected patient:

- This can be done directly at IMO
- The patient can also send us a sample taken at another medical centre (*)

We perform the genetic analysis to identify the gene where the alteration causing the disease is located.

We inform the patient of the results in a report and during a genetic counselling visit.

(*) In this case, specific steps must be followed because the sample must meet a series of conditions to ensure it arrives in good condition.

OVER 30 DISEASES, WITH MORE THAN 500 GENES INVOLVED

IMO GRUPO MIRANZA

These are the hereditary eye diseases for which IMO Grupo Miranza provides a genetic diagnosis and counselling service:

A B C D E G L M N O R S U W

Achromatopsia · AMD (age-related macular degeneration) · Aniridia · Anophthalmia · Autosomal dominant optic atrophy · Autosomal Recessive Bestrophinopathy · Axenfeld–Rieger syndrome

Bardet–Biedl syndrome Best disease · Blepharophimosis

Choroideremia · Cone dystrophy · Cone-rod dystrophy · Congenital cataract · Congenital stationary night blindness · Congenital fibrosis of the extraocular muscles · Congenital glaucoma · Congenital hereditary endothelial dystrophy · Congentical and acquired ptosis

Donnai–Barrow syndrome

Familial exudative vitreoretinopathy · Fuchs' endothelial dystrophy

Gyrate atrophy

Joubert syndrome · Juvenile glaucoma

Leber congenital amaurosis · Leber's hereditary optic neuropathy

Marshall syndrome · Meesmann corneal dystrophy · Microphthalmia

Norrie disease

Ocular albinism · Oculocutaneous albinism

Primary open-angle glaucoma (familial)

Reticular corneal dystrophy · Retinoblastoma Retinitis pigmentosa · Retinoschisis · Rothmund-Thomson syndrome

Senior Loken syndrome · Sorsby dystrophy · Stargardt disease · Stickler syndrome

Usher syndrome

Wagner syndrome

GENETICS DEPARTMENT

For more information, please contact us: genetica@imo.es

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