

## 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 50 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:

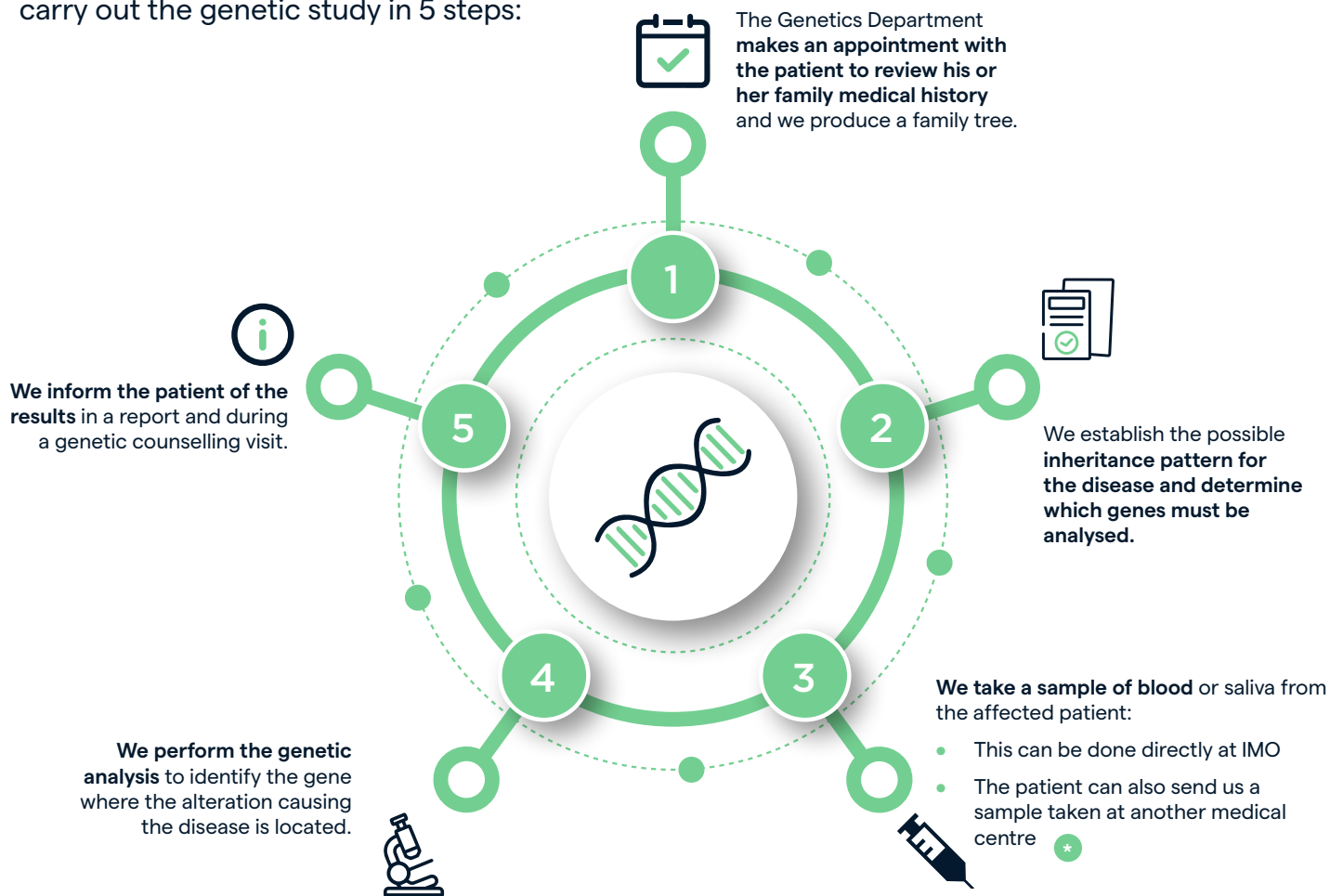
-  1 In your family medical history there are **people with hereditary or recurrent eye diseases**.
-  2 You have been diagnosed with a **hereditary eye disease**.
-  3 You want to have children and there are cases of hereditary eye disease in your family or in that of your partner.
-  4 One of your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



\* 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 50 DISEASES, WITH MORE THAN 800 GENES INVOLVED



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

A	<b>A</b> chromatopsia · Alström Syndrome · AMD (age-related macular degeneration) · Aniridia · Anophthalmia · Anophthalmia · Autosomal dominant optic atrophy · Autosomal Recessive Bestrophinopathy · Axenfeld–Rieger syndrome
B	<b>B</b> ardet–Biedl syndrome Best disease · Blepharophimosis
C	<b>C</b> entral Areolar Choroidal Dystrophy (CACD) · Choroideremia · Cone dystrophy · Cone-rod dystrophy · Congenital cataract · Congenital stationary night blindness · Congenital fibrosis of the extraocular muscles · Congenital glaucoma · Congenital hereditary endothelial dystrophy · Congenital and acquired ptosis · Congenital nystagmus
D	<b>D</b> onnai–Barrow syndrome
E	<b>F</b> amilial exudative vitreoretinopathy · Fuchs’ endothelial dystrophy
F	<b>G</b> yrate atrophy
G	<b>H</b> igh myopia
H	<b>J</b> oubert syndrome · Juvenile glaucoma
I	<b>L</b> eber congenital amaurosis · Leber’s hereditary optic neuropathy
J	<b>M</b> arshall syndrome · Meesmann corneal dystrophy · Microphthalmia
K	<b>N</b> orrie disease
L	<b>O</b> cular albinism · Oculocutaneous albinism
M	<b>P</b> rimary open-angle glaucoma (familial)
N	<b>R</b> eticular corneal dystrophy · Retinoblastoma Retinitis pigmentosa · Retinoschisis · Rothmund-Thomson syndrome
O	<b>S</b> enior Loken syndrome · Sorsby dystrophy · Stargardt disease · Stickler syndrome
P	<b>U</b> sher syndrome
Q	<b>W</b> agner syndrome



## GENETICS DEPARTMENT

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