ReproNIM® Carrier

Genetic risk test

Inherit Life

NIMGenetics
New Integrated Medical Genetics

ReproNIM® Carrier

- **Genetic risk test** that combines massive sequencing (NGS) and other molecular genetic technologies to establish whether the parents are carriers of recessive diseases or diseases linked to the X chromosome.
- **A total of 427 genes are analysed**, resulting in a study that analyses >4,000 variants.

This study enables:

- **The assessment of genetic compatibility with most studies available on the market today.**
- **Extends the information previously obtained with other studies of carriers *.**

(*): which requires sending the previous report for evaluation

What do we study?

> 130 Diseases

including

- Muscular-spinal atrophy
- Beta thalassemia
- Becker and Duchenne muscular dystrophy
- Phenylketonuria
- Cystic fibrosis
- Haemophilia A and B
- Fragile X syndrome
- Etc...

> 4.000 Variants

ReproNIM® Carrier complete disease list available at www.nimgenetics.com

Facilitando información genética con utilidad clínica

✓ Test targeted towards the most prevalent autosomal recessive and X-linked inherited diseases.

✓ We report only the pathogenic and likely pathogenic variants.

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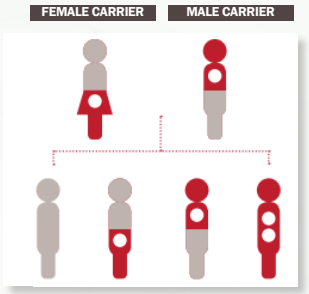
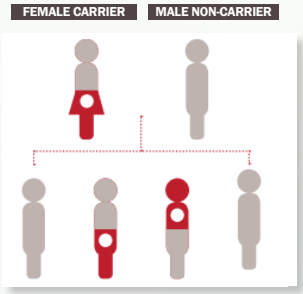
Inheritance is a determining factor

Autosomal recessive inheritance



- **50% of children** are carriers
- **25% of children** are non-carriers
- **25% of children** are affected



- **50% of children** are carriers
- **50% of children** are non-carriers

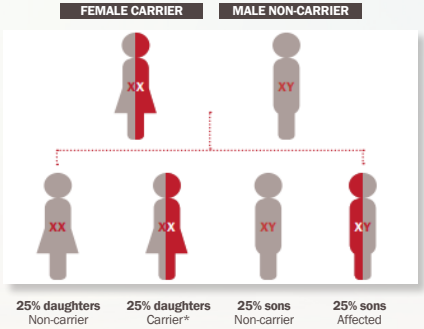
If both parents are carriers, there is a 25% chance of having an affected child

▸ EXAMPLES:

- Cystic fibrosis.
- Beta thalassemia.
- Muscular-spinal atrophy.

In exceptional circumstances, the appearance of "de novo" mutations in the child could determine the development of the disease, irrespective of the carrier status of the parents.

X-linked inheritence



▸ EXAMPLES:

- Muscular dystrophy.
- Hemophilia A and B.
- Fragile X syndrome.

(*): The presence of clinical manifestations in female carriers has been described in some diseases.

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INDICATED IN THE STUDY OF:

- Couples who want to be parents.
- Egg and sperm donor.
- Egg and sperm recipient.

DOCUMENTS NEEDED:

1. Request form*
2. Informed consent*
3. Medical report

SAMPLE CONDITIONS:

3-5 mL of blood in EDTA

RESPONSE TIME:

30 business days from the reception of the sample.

(*): If you have any questions, please, call at +34 91 037 83 54 or contact with the sales representative.

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ADVICE

Immediate accessibility to the NIMGenetics team for genetic counseling.

HAND IN HAND WITH THE MOST QUALIFIED EXPERTS

Members of the AEDP*1 and AEGH*2

(*1): Spanish Association of Prenatal Diagnosis.
(*2): Spanish Association of Human Genetics.

CERTIFIED QUALITY

Our laboratories follow the quality controls of the EMQN*3

(*3): European Molecular Genetics Quality Network.

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La Suma de Todos

Comunidad de Madrid

NIMGenetics is a Genetics Diagnosis center authorized by the Health and Consumer Department for the Community of Madrid, being duly registered under Nº CS10673

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www.nimgenetics.com