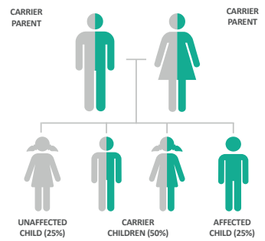
When is CarrierRef® indicated?

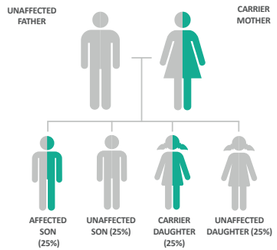
- Couples planning a pregnancy
- In assisted reproduction treatments where donated gametes are used

Risk assessment in patients with positive results



Autosomal Recessive Inheritance

Most people are asymptomatic carriers of some autosomal recessive genetic diseases. If both biological parents are carriers of a mutation in the same gene, the risk that the offspring will present the disease is 25%.



X-linked Inheritance

A woman who is a carrier and does not suffer from a genetic disease linked to the X chromosome can ignore her carrier status and transmit the mutation to 50% of her offspring. In each pregnancy, the patient will have a 25% chance of having an affected offspring.

REFERENCE LABORATORY GENETICS

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REFERENCE LABORATORY GENETICS

CarrierRef®

Carrier Screening

What is CarrierRef®?

CarrierRef® is the most comprehensive carrier screening test, combining NGS sequencing with other molecular biology techniques to study >360 diseases and >4.000 variants. The diseases studied in our test have been selected based on the following criteria:

- Clinical relevance
- Severity
- Early presentation of the disease
- Incidence
- Recommendations of the medical societies of gynaecology and genetics
- Existence of treatments or possible early intervention

The importance of offering CarrierRef® to your patients

We are all carriers of 14-18 pathogenic genetic variants

Up to 2.3% of couples carry the same pathogenic genetic variant.

Carrier couples have an **increased risk of having an affected offspring**

20% of deaths and 10% of pediatric hospitalizations in developed countries are due to hereditary diseases. **CarrierRef®** offers early and actionable information on serious genetic diseases to facilitate family planning, adequate prenatal care and access to prenatal, neonatal and/or pediatric treatments.

Why choose CarrierRef®?

PREVENTION AND PLANNING

It allows us to find out whether the parents are carriers of any pathogenic variant associated with an autosomal recessive genetic disease or linked to the X chromosome, in order to guide them in making informed decisions and prenatal/postnatal plans.

GREATER KNOWLEDGE AND SECURITY

CarrierRef® offers the highest detection rate on the market mainly due to 2 reasons:

1. It has been **designed according** to the criteria of the **most relevant health organisations** and it is constantly updated.
2. All the **genes** studied are **completely sequenced.**

GENETIC COUNSELING

- **Pre and post test genetic counselling** with our genetisists.
- We are fortunate to be able to include the experience of **Dr. Cristina Camprubí**, specialist in reproductive genetics.

ADVANTAGES OVER OTHER TESTS

- **Complete sequencing of each gene by using NGS,** guaranteeing maximum clinical sensitivity.
- **Additional molecular biology techniques** to detect pathogenic variants that cannot be determined by NGS.
- **AGG interruptions for Fragile X,** not included in most carrier screening tests, which estimate the risk only on the basis of CGG repeats (modified risk in > 83% of carriers in a premutation).
- **Silent carrier analysis for Spinal Muscular Atrophy (SMA),** improving the detection rate by 5-8%.
- Detection of **Congenital Adrenal Hyperplasia** carriers **(CYP21A2)**.
- **Detection of Thalassemias, Haemoglobinopathies and Duchenne Muscular Dystrophy (DMD).**
- Detection of 25% more carriers of **Cystic Fibrosis** than usual screenings, due to NGS technology.
- **Conclusive medical reports validated by our geneticists.**

Test code: **15048**

5 mL whole blood EDTA

Informed consent must be sent with the sample