qCarrier

Carrier screening testing provides essential information for family planning purposes

qCarrier is a genetic screening test that identifies if a person is **carrier of recessive diseases that could be passed on to their offspring.** Carriers generally do not manifest symptoms of the disease and in many cases, there is no family history; therefore, the only way to obtain this information is through genetic testing.

Carrier testing is an essential tool in preconceptional genetic counselling, which determines whether there is a risk of conceiving offspring with a recessive genetic disease. **qCarrier** provides relevant information to make informed decisions about:

- Family planning
- Prenatal testing options
- Preimplantation genetic diagnosis
- Egg or sperm donation

Why is it important?

Around 80% of people are carriers of at least one recessive disease. This does not have health implications for the individual, as carriers do not generally manifest symptoms of the disease. However, if both parents are carriers of the same disease, there is a high risk that their offspring may develop it.

3%

Nearly 3% of couples are carriers of the same disease, therefore in each pregnancy there is a 25% risk of having a child with the disease carried by their parents.

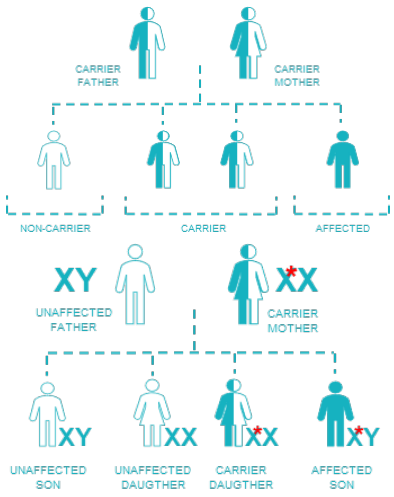
International medical guidelines emphasise the importance of carrier screening as part of reproductive healthcare, recommending specialists to inform women and their partners about the availability of this type of testing.

Conditions analysed

qCarrier analyses over 300 genes associated with more than **300 autosomal recessive and X-linked diseases.**

An autosomal recessive disease occurs when both copies of a gene are altered. If both members of a couple are carriers of the same disease, there is a 25% chance that their offspring will be affected.

X-linked diseases are conditions caused by an alteration in a gene located in the X chromosome. Females have two X chromosomes (XX) and therefore can be asymptomatic carriers. Males have one X chromosome and one Y chromosome (XY) and could present the condition. An asymptomatic female carrier will have a 50% chance of transmitting the disease to a male offspring.



Who is it for?

- ✓ Any person or couple who wants to know if they are carriers of the same disease, which implies a higher risk for their offspring.
- ✓ Gamete banks or reproduction clinics, for egg or sperm donors' testing.
- ✓ Couples who require gamete donation, to select the most appropriate donor.

Test details

Veritas provides a comprehensive service offering two testing options.

qCarrier Auto EC

Automated analysis of variants with evidence of pathogenicity, previously described as disease causing according to the ClinVar database, present in genes related to more than 300 recessive or X-linked genetic diseases with a variable impact on quality of life.

qCarrier Plus EC

Analysis of any variant present in coding and certain regulatory regions in genes related to more than 300 recessive or X-linked genetic diseases with a variable impact on quality of life.

Technical information

The test allows the detection of pathogenic/likely pathogenic variants by using several techniques and analysis software:

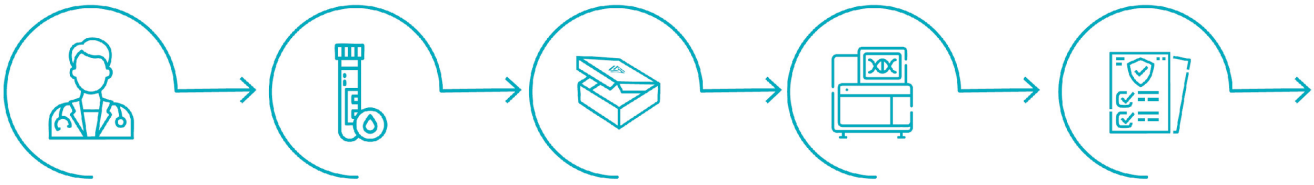
- » **Single nucleotide variants (SNVs) and deletions/duplications (CNVs):** analysed using NGS sequencing and a sophisticated bioinformatic algorithm.
- » **Fragile X syndrome and Friedreich's ataxia:** caused by triplet expansions (CGG and GAA) in the *FMR1* and *FXN* genes, respectively, and require additional analysis using specific PCR (tp-PCR).
- » **Spinal muscular atrophy:** CNVs in the *SMN1* gene are analysed by NGS and confirmed by MLPA. It is possible to identify 2+0 carriers*, but SNVs are not detected due to the high homology of the sequence.
- » **Pseudogenes:** specific molecular and bioinformatic tools are used to identify certain mutations in genes with pseudogenes such as *CYP21A2, HBA1/HBA2/HBB* for thalassaemia, and inversions in the *F8* gene.

* Carriers with two copies of the *SMN1* gene on one chromosome and a deletion of the *SMN1* gene on the other chromosome.

Sample type

The test is performed using a **blood or saliva sample** in a kit provided by Veritas.

Ordering process



The physician prescribes the test

A kit is provided to collect the sample

The sample is sent along with the requisition form

The sample is sequenced and analysed in the laboratory

The test results are sent to the ordering physician