MGZ

Medical Genetics Center

Clinician Information

CARRIER SCREENING

praeCon-Test

WHAT IS THE AIM OF THE praeCon-Test?

The **praeCon-Test**, developed by the MGZ – Medical Genetics Center, is a genetic test that can be used to identify carriers of recessive hereditary diseases in clinically healthy persons (carrier screening, heterozygote screening).

If two partners who are both carriers of the same recessively inherited disease meet at random, the couple has a 25% risk of having an affected child.

Through the praeCon-Test, an individual risk for the carrier status of certain genetic diseases can be identified in order to make an informed decision regarding personal family planning.

WHO SHOULD PURSUE THE praeCon-Test?

In contrast to traditional screening of ethnic groups, heterozygote screening can be offered to individuals independent of their ethnicity (non-directive, pan-ethnic carrier screening).

- The American College of Obstetricians and Gynecologists (ACOG) recently recognized pan-ethnic heterozygote screening as a general option for all couples who wish to have children (ACOG Committee Opinion No. 690 2017).

Heterozygote screening may also be useful if an increase in the risk of autosomal recessive diseases can be detected:

- in blood-related couples;
- in couples of the same ethnic origin for whom an increased incidence of recessive diseases is known, due to a historically isolated population;
- in couples suspected of having a genetic disease in the family that was not/could not be genetically attributed to the person concerned.

WHAT IS ANALYZED IN THE praeCon-Test?

The praeCon-Test includes the analysis of over 600 genes, whose mutations are known to be the cause of recessive diseases. It also includes an analysis of the CFTR gene to identify common carriers for cystic fibrosis (CF, mucoviscidosis). The genes for the Next-Generation Sequencing (NGS)-based screening panel were selected and evaluated according to the recommendations of the American College of Medical Genetics and Genomics (ACMG). Criteria for the selection were, for example, high penetration of the disease, a characterized clinically significant disease phenotype, sufficiently published data on clinical sensitivity/specificity of genetic variants, and a good analytical detection rate.

In addition to the NGS analysis, MLPA analysis is performed as a standard procedure in one of the two partners investigated, in order to identify a carrier for autosomal recessive spinal muscular atrophy (SMA). If the result is positive, the second partner is tested.

As an extension of the heterozygote screening, the carrier status for a Fragile X syndrome or the carrier status for alpha and beta thalassaemias can also be tested.

WHAT IS DISCLOSED IN THE REPORT?

The praeCon-Test aims to identify joint carrier status of a couple. Therefore, in the basic version of the praeCon-Test, only the joint carrier status entities are reported.

As an extension of the test (post-evaluation with additional costs), it is also possible to receive a report on carrier status entities that were found with one partner only.

IMPORTANT ASPECTS IN COUNSELING ABOUT THE praeCon-Test:

- **Many different diseases are tested, which cannot be explained in detail:** The tested diseases have different degrees of severity, some are treatable; many are not. Some diseases are so rare that their severity is not fully characterized.
- Like any genetic analysis, heterozygote screening is **not 100 % sensitive**. An inconspicuous result is equivalent to a risk reduction, but there is still a residual risk for an unidentified carrier status, the level of which cannot be stated either as a whole or for the individual diseases. It should also be pointed out that the general basic risk of 2 - 4 % exists for each expectant parent couple.
- In rare cases, the result of the test may also have **health significance for the person tested** (see Patient Consent Form in our website Downloads).
- A **conspicuous result** should be discussed individually during a genetic counseling interview, during which the couple receives comprehensive information on the type of disease, possibilities and limits of treatment, the level of risk of occurrence among the common offspring and options for family planning. This also includes the possibilities and limitations of a prenatal examination (prenatal or pre-implantation diagnostics, if necessary) as well as sperm donation.

HOW DO I REQUEST THE praeCon-Test?

- **Sample (from both partners):**
  - 2 - 4 ml EDTA blood
- **Turnaround time**
  - 3 - 6 weeks
- **Price Inquiries & Test Ordering**

  Obtain a cost statement using our convenient website contact form or submit your price inquiry via the MGZ's online portal at **www.mgz-muenchen.com**.

  To send a test request, please complete a test request form and patient consent form for both partners and submit these with the samples.
  All forms are available on our website at **www.mgz-muenchen.com**.

  For detailed price and ordering information, including shipping instructions, please see the How to Order section of our website.
  Still have questions? Contact us at **inquiry@mgz-muenchen.com**.

On our website, you will find information on other clinical topics as well as information about our company and services.

Visit us at www.mgz-muenchen.com

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