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Preconception Genetic Examination Carrier screening

Modern and responsible approach to planned parenthood

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What is Preconception Genetic Examination?
The preconception examination is a set of genetic tests that are suitable for couples at the time of planning a pregnancy, i.e. before conceiving a child (conception). The aim of these tests is to reduce the risk of giving birth to a child with a severe genetic disease.

What kind of genetic tests does Preconception Genetic Examination include?
Main part of the preconception genetic examination is the "Carrier screening" test. The test examines whether an individual carries genetic predispositions (mutations) to the most common **monogenic diseases**, i.e. diseases caused by a mutation in a single gene. Examined genetic diseases include 1) autosomal recessive (AR) disorders, where the child's disability is caused by mutations in the same gene inherited from both parents (healthy carriers) and 2) X-linked diseases, where the affected son inherits from mother (a healthy carrier) the mutated gene on the X sex chromosome. To complete preconception examination, the chromosome complement (karyotype) may also be analyzed.

Is Preconception Genetic Examination suitable for a couple who has no relatives with a genetic disease?
Yes, this type of examination is especially designed for couples without known genetic burden in the family. The analyses aim to disclose those genetic predispositions that cause no symptoms in their carriers. A child, who inherits these "hidden" predispositions from both parents (or a son who inherits the mutation from mother in case of X-linked disease), will be severely affected.

Is Preconception Genetic Examination also advisable in couples whose families suffers a genetic disorder?
Prime genetic examination for couples, who have a genetic disease with known molecular cause in the family, is a test that targets this disease. However, to disclose predispositions to other genetic disorders the carrier screening can also be performed and is fully recommended.

GENETIKA PLZEŇ offers a new carrier screening for monogenic disorders

The test is designed as an "expanded" carrier screening and includes examination of **128 autosomal-recessively inherited and 23 X-linked monogenic diseases**. It targets the most common recessive diseases in all ethnic groups, including: **cystic fibrosis, spinal muscular atrophy, non-syndromic deafness (mutation in GJB2 gene), alpha and beta thallasemia, sickle cell disease, phenylketonuria, type 1 albinism** and others (for the complete list of diseases and genes see https://www. genetika-plzen.cz/). Design of the test respects all recent international professional guidelines.

The analysis is performed using the most modern available laboratory methodology - **New Generation Sequencing** (NGS), which is the first technology that allows the analysis of many genes at the same time. As entire coding sequences and losses/gains of target genes (Copy Number Variations, CNVs) are fully analyzed, the detection rate approaches 99%, significantly minimizing the residual risks of being a carrier.

In women, an additional test is performed to detect genetic predispositions to the **fragile X syndrome** (and other conditions linked to FMR1 gene).

When both partners are tested, the **reproductive risk for the couple is evaluated** and issued as a separate report.

Gene variants that may be associated with **impaired reproduction**, including inherited thrombophilia, are also part of the test. The results of this analysis are not only of diagnostic significance, but they also contribute to the individualization of potential infertility treatment by assisted reproduction methods.

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