Benefits of Genetic Testing

What is genetic testing?

Genetic testing is a medical laboratory test that looks into your genes. Genes are DNA instructions that we all inherit from our parents. Different “mistakes” in our genes, called “mutations”, can cause different diseases. Some of these diseases are rare, like cystic fibrosis or Huntington’s disease, others are more common like cancer. Genetic tests can be used to confirm the diagnosis or give you information about possible development of a disease.

What can I learn from testing?

Genetic tests can help to:
- Diagnose the disease
- Identify gene changes that may increase the risk to develop a disease
- Identify gene changes that could be passed on to children
- Give an idea about the severity of a disease
- Guide doctors in deciding on the best medicine or treatment to use

Why and when you might consider genetic testing?

- When you have a family history of a disease
- When you might have increased risk for development of disease
- If you develop signs/symptoms of a disease
- When you might pass the genetic disease (mutation) to your children
- If you are pregnant and have reason to believe that your baby is at risk of having a disease-causing mutation

What are the benefits of genetic testing?

- It can give you an accurate diagnosis
- Your physician can select the most suitable therapeutic option and treatment for you
- It can give a more accurate prognosis: how the disease may progress, will stabilize or get worse
- It can tell you about any risks of mutations being passed to your children
- You might be able to change your lifestyle to reduce your risk of getting a disease or to improve your quality of life
- It can help your doctor to give you the best possible care
- Your genetic test result might tell your doctor which medications are most effective to treat you
What does genetic testing involve?

Genetic testing involves several easy-to-follow steps:

Doctor examines patient

- Clinical diagnosis and suggestion for gene test
- Sample collection and sending to CENTOGENE
- DNA isolation from samples

Genetic analysis

- Results collection and interpretation by CENTOGENE’s experts
- Report writing

Report sent to doctor

BENEFITS of genetics testing:

- Accurate diagnosis of the patient's disease
- Selection of the most suitable therapy and support for patient
- Prediction of possible progress and severity of disease
- Prediction of possible risk of disease transmission to the patient’s children
What are the risks of genetic testing?

The results of genetic testing are not always black and white. You may get an uncertain result. Therefore, it’s very important to discuss genetic testing with your doctor and/or a genetic counsellor to be aware of the benefits and risks so that you are fully prepared. Some drawbacks of genetic testing to consider are:

- Negative results do not 100% guarantee that you will not be affected.
- A positive genetic test result can establish a correct diagnosis but often there is no treatment available.
- Waiting for the results of genetic testing can be stressful for you and other family members.
- A positive genetic test result can cause tension between family members, feelings of guilt, anxiety about the future. Genetic counsellors are trained to help you and family members cope with the result and make informed decisions.

What does a test result mean?

<table>
<thead>
<tr>
<th>Positive</th>
<th>Negative</th>
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<tr>
<td>° If you are ill, a positive test result confirms the suspicion.</td>
<td>° If you are ill, a negative test result means that you still don’t have a definite diagnosis and may need to go for further genetic testing.</td>
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<tr>
<td>° If you are healthy, a positive test result means you may be more likely to get a particular disease than other people, but it does not mean you will definitely get the disease.</td>
<td>° If you are healthy it may mean that you won’t be affected by a disease that runs in your family or it may mean that your risk of developing the disease is the same as anyone else in the general public.</td>
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## Different types of genetic testing

Genetic testing can be done for several reasons: to confirm a clinical diagnosis, to see if you are a healthy carrier of a disease, to see if you will develop a disease in the future (predictive testing), to test an unborn child (prenatal) and to test all newborn children for diseases that need to be treated immediately (newborn screening).

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<th><strong>Diagnostic testing</strong></th>
<th>if you have symptoms that suggest a certain genetic condition, testing is used to confirm or rule out the suspected disorder. Overall, diagnostic test results can help you and your doctor to make choices about your treatment and care. Results also may help family members to make healthcare decisions.</th>
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<td><strong>Carrier testing</strong></td>
<td>is used to test future parents for a previously identified familial mutation in order to determine if they are carriers and are at risk of having an affected child.</td>
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<td><strong>Prenatal genetic testing</strong></td>
<td>is used to diagnose some conditions in babies before they are even born. If an unborn child is diagnosed with a serious condition it gives the parents a chance to receive genetic counselling and consider their options, whether to terminate the pregnancy or continue to term.</td>
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<td><strong>Pre-implantation genetic testing</strong></td>
<td>is used to detect genetic changes in embryos that have been created in the laboratory by in-vitro fertilization (IVF) for parents that have difficulties conceiving naturally. During IVF, eggs are removed from the mother’s ovaries and fertilized with the father’s sperm in a laboratory dish. Once the embryo has grown for 3-6 days, either one or a small number of cells can be safely removed and tested for disease-causing genetic mutations. Only embryos who test negative are placed in the mother’s womb in the hope of delivering an unaffected, healthy child.</td>
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<tr>
<td><strong>Newborn screening</strong></td>
<td>is the most widespread form of genetic testing. In most countries newborns are screened for a number of inherited conditions such as phenylketonuria, cystic fibrosis, sickle cell disease, and other frequently inherited disorders. The advantage of testing newborn children is that some very serious disorders can be avoided if the proper treatment is started immediately.</td>
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### Predictive genetic testing

is used to detect genetic mutations associated with disorders that appear after birth, often later in life. These tests can be helpful for people without symptoms but with an affected family member. Predictive testing can tell whether a person has a high risk of developing certain genetic disorders in the future (for example, breast cancer associated with a BRCA1 or BRCA2 gene mutation).

### Whole exome sequencing (WES)

is a type of genetic testing ordered by your doctor in special situations when your medical history and physical exam strongly suggest that there is an underlying genetic etiology, but there is no strong indication for a particular genetic test. Your health care provider may recommend WES because all other previous tests have come back negative or as the very first step towards a correct diagnosis. WES is a technology in which scientists look very carefully at all of your genes at once, to try and find mutations that can explain your genetic disorder.

**WES: short explanation of the method:**

Our genes are simply small pieces of DNA. Inside each gene some of the DNA codes for a protein (exons) and some doesn't (introns). Proteins give our cells instructions on how to grow and function normally to keep us healthy. The parts of our genes that code for proteins (the exons) are the most important area for scientists to look for disease-causing mutations. So, WES looks at all the exons in all your genes (about 200,000 exons in total or 1-2% of your genome). But because WES technology isn't 100% perfect there are blind spots and a small chance that an important mutation is missed, so your doctor may need to order additional genetic tests.

### Whole genome sequencing (WGS)

is even more powerful than WES because it looks at 100% of your DNA, not just the 1-2% that codes for proteins. Your genome is the unique blueprint for your body. By sequencing your genome, scientists and health professionals are widening their search as much as possible to look for mutations in both the coding and non-coding regions of your DNA to explain your disorder. At the moment WGS is still mainly used in research. CENTOGENE as a pioneering genetic diagnostic company has the unique expertise required to offer you WGS as a fully accredited medical test. If you do choose WGS it is extremely important and helpful to review the results of your genome with a trained professional who can explain the results clearly and discuss your healthcare options. The laboratory that performs you WGS must be properly accredited. CENTOGENE has all the necessary international quality accreditations to perform WGS.
What is genetic counseling?

The information obtained from genetic testing can have a profound impact on your life. Genetic counselling is available to anyone undergoing any form of genetic testing. Genetic counselling is aimed to provide you with all the information you need to make a decision about whether you should have a genetic test or not.

Genetic counselling may include information about:

- The implications of testing positive for the condition, including the psychological impact and other consequences.
- Whether to inform relatives of your intention to test, or not to test.
- The disease you are being tested for and its potential treatments.

The information is given in a non-directive manner, as it is important that every individual reaches their own informed decision.

What about genetic testing at CENTOGENE?

At CENTOGENE, we only accept genetic tests ordered by a doctor on your behalf, to ensure that you have access to full support and back-up to make informed decisions about your future healthcare. CENTOGENE supply filtercards (CentoCard®) as a very simple way to collect your sample and send it by ordinary post to our laboratory.

Conclusion:

CENTOGENE offer genetic tests for more than 2,800 genes, specifically designed to confirm or exclude a genetic diagnosis of almost any known disorder with a genetic background. An accurate genetic diagnosis will avoid a diagnostic odyssey of perhaps several years involving many unnecessary tests. CENTOGENE offers you the most advanced in genetic testing technologies with renowned medical experts and a scientific advisory board to ensure your genetic test results are comprehensively analyzed and translated into a clinical report.

Invest wisely in your healthcare by choosing top quality expert genetic testing at CENTOGENE.

We look forward to helping you.