



CENTOGENE

CentoXome[®]

Turning Years Into Days

PATIENT INFORMATION

CentoXome[®] – Whole Exome Sequencing (WES)

CENTOGENE's enhanced WES service – CentoXome[®], provides **highly uniform coverage of the entire exome and mitochondrial genome**, as well as nearly complete coverage of all known disease-causing regions throughout the genome in a single test.

The improved test design includes the **most up-to-date scientific knowledge** and unique insights based on what we believe to be the **world's largest real-world data repository** for rare and neurodegenerative diseases.

What Is an Exome?

DNA carries all the information we need to live and acts as an instruction manual that **determines our individual development and characteristics**. Each piece of information is carried on a specific section called a gene: in total, our **DNA contains around 20,000 genes**.

Genes are composed of two types of regions: the **exons** that provide the code for making proteins; and the **introns** that do not directly code for protein. The entire collection of genes is called the “genome”, while the entire collection of exons is called the “**exome**”.

What Is a Mutation?

A mutation is a **change in the genetic code** that modifies the gene sequence, and it can have a negative impact on a person’s health. When a disease-causing mutation occurs, it changes the code of the resulting protein. These changes might cause dysfunctions in the cells and organs and **result in clinical symptoms related to a rare disease**.



A human cell contains DNA in the nucleus and mitochondria



Chromosomes are made up of proteins and DNA



DNA contains exons and introns



Exons code for proteins



What Is the Importance of WES in Clinical Practice?

WES is a recognized and valuable genetic testing tool that enables the sequencing of exons of thousands of genes simultaneously to identify genetic mutations.

The exome accounts for only 1-2% of our genome. However, **about 85% of the genetic mutations** that cause genetic disorders are thought to be **found in the exome**. A lot of research has been performed on the exome, which **aids in the identification and interpretation** of mutations in this region compared to the rest of the genome.

Benefits of CentoXome Sequencing

- Provides an accurate diagnosis and avoids additional unnecessary clinical investigations
- Allows for better clinical management and prevention through precise diagnosis
- Enables tailored therapy in some cases, resulting in better outcomes for the patient
- Yields results that may be useful for future family planning

Find out more about CentoXome at
centogene.com/whole-exome-sequencing

What Can You Expect From WES?

There are three possible types of results you can expect when we analyze your exome:

Positive Results



- Indicate a causative genetic change in a gene that has been linked to your symptoms or those of your family members
- Enable your doctor to make a clear diagnosis and help decide on any treatment or other steps to safeguard your health
- Allow other family members to be tested and understand their risks

Unclear Results



Indicate a change that has been identified, and the gene of concern is known to be associated with a disorder; however, it is uncertain if the identified change is the cause of the disorder. Additional genetic testing in other family members can be considered.

Negative Results



Indicate a disease-causing change was not detected in the gene/region of interest. Either there is no genetic cause for the disorder, or a mutation exists in a region of the exome that is not covered by exome sequencing analysis.

Sequenced data of around 20,000 genes from your blood sample is carefully analyzed and interpreted by our **highly experienced medical team**, incorporating in-depth clinical information and your detailed family history. A comprehensive clinical diagnostic report will be sent to your doctor.

FOR ORDERING

www.centoportal.com

FOR MORE INFORMATION

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