CENTOGENE
THE RARE DISEASE COMPANY

CentoXome®

FUTURE'S KNOWLEDGE APPLIED TODAY

When you need a medical answer

Tackling the diagnostic challenge with whole exome sequencing

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A genetic test looks into the DNA to see if there is any alteration in the genetic code or chromosome number and structure that may cause the suspected disease.

Advanced technologies, like exome sequencing allow us to sequence approximately 20.000 genes at once

WHAT IS AN EXOME?

Each cell of our body contains our DNA in the nucleus, organized as chromosomes. **DNA carries all the information we need to live** and function, coded by just four letters A, T, C, G (called nucleotides); this is the instruction manual that **determines our individual development and characteristics**. If there is a change in this letter code, or if one of the chromosomes or a part of it is missing or duplicated, the instruction manual will be altered.

Each piece of information is carried on a specific section called a gene. In total, our **DNA contains around ~20,000 genes**, each with a different combination and number of nucleotides. Genes code for proteins and, together with other molecules, compose the cells of our body.

But not all the information contained in the DNA codes for proteins (in fact most of it is not protein coding!). **Genes are composed of two types of regions** depending on their ability to produce proteins: the **exons** that induce the production of proteins; and the **introns** which themselves don´t produce proteins, but assist in selection of protein-coding exons. The entire assortment of genes is called "genome", while the whole assortment of exons is called "exome".

A CELL´S NUCLEUS CONTAINS CHROMOSOMES

CHROMOSOMES ARE MADE OF DNA

DNA CONTAINS EXONS AND INTRONS

EXONS CODE FOR PROTEINS

WHAT IS THE IMPORTANCE OF EXOME SEQUENCING IN CLINICAL PRACTICE?

The exome accounts for only 1-2% of our genome. However, **about 80% of the genetic changes** (mutations) that cause genetic disorders **occur in the exome**. We also know much more about the exome and have better means to interpret it and identify mutations, than we do for the rest of the genome.

WHAT IS A MUTATION?

A mutation is a **change in the genetic code** that modifies the gene sequence and it can affect a person's health negatively. When a disease-causing mutation is found in a gene, it generates a change in the information and therefore in the produced protein. These changes might cause dysfunctions in the cells and organs and finally **result in symptoms and disease**. Exome sequencing allows us to sequence the entire coding regions of the genome. In other words, it enables us to read a small fraction of the genome but with a high probability to identify the mutation responsible for disease.

> Exome sequencing is a recent genetic revolution that allows the sequencing of exons of thousands of genes simultaneously to identify genetic mutations

Benefits of exome sequencing:

- Provides diagnosis of a complex disease
- Patients can benefit from a better tailored therapy
- Unnecessary therapies and examinations can be avoided
- Patients can learn about the diseases that they might transmit to the next generation

SHOULD YOU UNDERGO EXOME SEQUENCING?

Our experts highly recommend exome sequencing if:

- You have an undiagnosed disease where all previous genetic testing were inconclusive
- You have, symptoms of a complex disease running in your family and inherited as a Mendelian disease (X-linked or autosomal recessive and dominant inheritance)
- You have no other way to perform genetic testing for a specific gene(s)
- You have an undiagnosed complex disease and need a pre-conception and prenatal screening for genetic markers that might be harmful for your future child
- You have a phenotype and/or a family history that strongly suggests a genetic disorder, but the genetic test targeting the responsible gene is not available

WHAT CAN YOU EXPECT FROM EXOME SEQUENCING?

There are three possible types of results we can find while analyzing your exome or genome:

Positive result:

A disease-causing change was identified in the gene/region of interest. Depending on the purpose of the test, a positive result can:

- Confirm your diagnosis or of your family members
- Identify that you or your family member is at increased risk to develop the condition in the future
- Indicate that you are a carrier of a particular genetic condition
- Allow other family members to be tested and understand their risks

Unclear result:

A change was detected in the gene/region of interest but currently, there is insufficient information in the medical literature to know if this is a disease-causing change or if this is a normal variation in the population.

Negative result:

A disease-causing change was not detected in the gene/region of interest. This might indicate that you or your family member:

- May not be affected by a particular disorder
- May not be a carrier of a specific genetic condition
- May not have an increased risk of developing a certain disease

A single test cannot always detect all possible genetic changes that cause a particular genetic condition. Hence, negative results do not completely rule out the presence of a disease. Further testing may be required to confirm a negative result. Please discuss with your doctor as each individual case is different.

Please visit our website
for more information:

www.centogene.com

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V3.1eng_December2017