MEDICOVER GENETICS

# Empowering people to use genetic testing for their health and well-being

## Whole Exome Sequencing Decode&Discover

## WHAT IS WHOLE EXOME SEQUENCING?

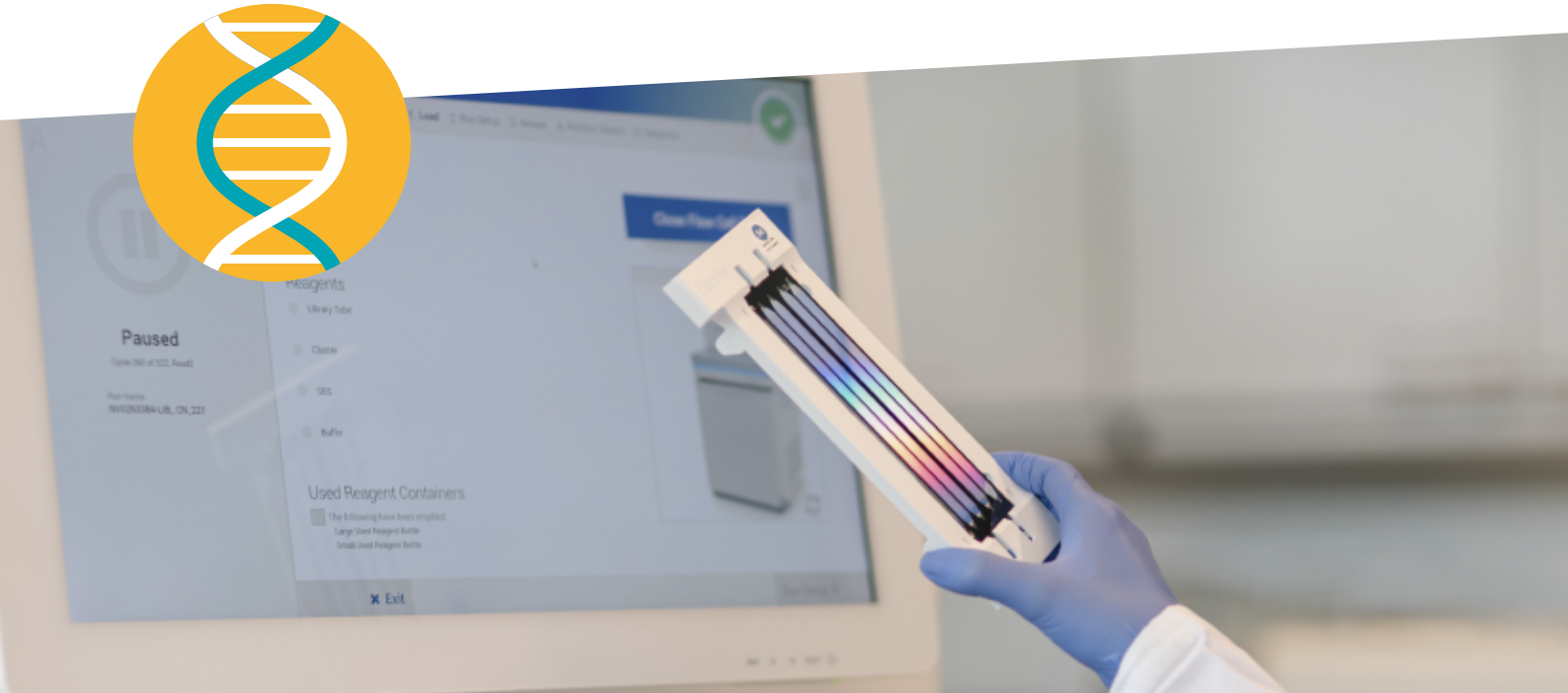
Whole exome sequencing (WES) is a **comprehensive genetic test which analyzes the protein-coding regions (exons) of all human genes (~20,000 genes)**. The sum of all the exons is called an exome. The human exome contains ~85% of known disease-related variants. The remaining 15% of disease-causing variants are located within introns (non-coding regions). **Decode&Discover** is designed to detect the exome variants, as well as the intronic variants. All of these variants can change the function of the underlying DNA sequence, thereby causing disease.

## WHY PERFORM WHOLE EXOME SEQUENCING?

WES offers a simultaneous analysis of a large number of genes. **It substantially increases the chances of finding the genetic cause of diseases with complex and non-specific symptoms**. WES reduces the time and cost from symptom presentation to diagnosis, and has an increased diagnostic yield. The results may provide information about the recurrence risk (chance of having another child with a similar condition) and may also be useful for other family members.

## WHO CAN BENEFIT FROM WHOLE EXOME DECODE&DISCOVER?

- Patients with a complicated medical history that affects many organs
- Patients who have undergone previous genetic testing with inconclusive results
- Patients with complex, non-specific symptoms with multiple differential diagnoses
- Patients with a severe presentation in the neonatal or childhood period



## OUR TEST OPTIONS

| Single WES | Duo WES | Trio WES |
|---|---|---|
| Only the patient is tested | Patient and one biological parent are tested | Patient and both biological parents are tested |

Trio WES is the most commonly performed analysis providing comprehensive results with the highest diagnostic yield. In cases where only one or no biological parent is available for testing, we can perform Duo WES or Single WES respectively. Trio WES allows simultaneous analysis for all modes of inheritance and can ascertain the significance of clinically relevant variants.

## HOW TO ORDER DECODE&DISCOVER

**Pre-Test**
GENETIC COUNSELLING

**STEP 1**
TEST ORDERED

**STEP 2**
SAMPLE COLLECTED

**STEP 3**
SAMPLE PROCESSED

**STEP 4**
RESULTS DELIVERED

**Post-Test**
GENETIC COUNSELLING

## POSSIBLE OUTCOMES OF THE TEST

**Pathogenic and likely pathogenic variants**: the genetic cause of the observed symptoms has been identified and may help determine the right treatment and management plan.

**Variants of unknown significance (VUS)**: there was not enough evidence to classify the variant as either pathogenic or neutral. However, future information may connect the variant with the patient's symptoms. If new information becomes available, the meaning of the results may change for the patient and their relatives (and they will be notified). Annual variant reclassification and testing family members is recommended.

**Incidental findings**: genetic changes that are not associated with the patient's current condition may be discovered which may be medically important and will be automatically included in the report unless they are associated with adult-onset disorders for which there are no interventions available.

**Secondary findings**: genetic changes that are not associated with the patient's current condition, which have been defined by the American College of Medical Genetics as medically important. The patient or legal guardian must consent to the reporting of these results.

***It is important to note that a negative result does not guarantee the absence of a disorder or that the disorder does not have a genetic cause. Additional knowledge may become available that could explain the underlying cause of the symptoms.***

## ABOUT US

**Medicover Genetics** supports you throughout your diagnostic journey with reliable testing and genetic counselling.

**German leader in genetic testing**

**>20 years of experience**

**Up-to-date diagnostic algorithm**

**Strong clinical team of scientists and physicians**

**Accredited laboratories**

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