arcensus

# myLifeExome

Unlock genetic insights
at a lower cost

Whole exome sequencing (WES) is a genetic test targeting the exons of protein-coding genes in the the nuclear genome (nDNA) and includes the entire mitochondrial genome (mtDNA).

### Indicated for:

- Patients with a suspected genetic disorder as a first line genetic test
- Healthy individuals with a family history of a genetic disease
- Patients with suspected mosaicism

### Not indicated for:

- Somatic variant analysis in tumor samples
- Alzheimer's risk assessment
- Analysis of prenatal samples
- Detection of methylation patterns

## FEATURES AND PERFORMANCE

### Types of Findings

| TYPE | DEFINITION |
|---|---|
| Primary | Variants that are relevant to the indication for which the sequencing was ordered. |
| Research | Variants that are potentially relevant to the indication for testing based on current evidence from experimental, animal, or cell studies. |
| Incidental | Variants unrelated to the individual's indication that are considered actionable based on ACMG guidelines and ClinGen recommendations. |
| Carrier | Pathogenic or likely pathogenic variants that have a direct impact on reproductive risk (heterozygous variants in a gene associated with a recessive or X-linked disorder). |

↻ **Receive a semi-annual re-evaluation during the 12 months following the initial report.**

arcensus

## Sequencing Specifications

**TAT**
All samples are processed within 20 business days

**Sequencing Platform**
Illumina NovaSeq
2×100 bp

**Sample types**
Buccal swab, saliva, blood, DBS card, isolated DNA, and others upon request

**Output**
100× median coverage; 98% at >20×
mtDNA 1000× mean coverage

**Enrichment kit**
Twist Human Core + RefSeq + Mitochondrial Panel

**Raw data options**
vcf and bam files

## Types of Variants

| DNA TYPE | VARIANT TYPE | DEFINITION |
|---|---|---|
| nDNA and mtDNA | Single nucleotide variants | A DNA sequence variant affecting 1 nucleotide |
| | Insertions / Deletions | Deletions, insertions, or duplications of DNA segments less than 50bp |
| | Copy number variants | Deletions or duplications of DNA segments of at least 50bp |
| nDNA | Chromosomal abnormalities | Trisomy, uniparental disomy, monosomy, triploidy |

## Limitations

- Interpretation is dependent on the provided clinical information and family history. Misinterpretation may occur if this data is provided incorrectly or incompletely
- Variant frequencies are subject to changes due to growing variant databases and may result in reclassification of previously reported variants
- A particular genetic variant may not be recognized as the underlying cause of the genetic disorder due to incomplete scientific knowledge about the biological function of the gene and/or the impact of the variant on the expression and/or function of the gene
- This methodology detects events of mosaicism of single nucleotide variants with a minor allele fraction >5%
- This test does not detect: translocations, partial UPD, methylation, gene conversions, low level mosaicism (VAF <5%), low heteroplasmy levels (VAF <5%)

© Arcensus GmbH 05/25_03 — www.arcensus-diagnostics.com

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

**Arcensus GmbH**
Friedrich-Barnewitz-Str. 9
18119 Rostock, Germany
support@arcensus-diagnostics.com