arcensus

myLifeExome

Unlock genetic insights at a lower cost

Whole exome sequencing (WES) is a genetic test that sequences the exons of all protein-coding genes and the mitochondrial genome. While the exome represents only 1-2% of the human genome, it contains approximately 85% of the known disease-related variants.

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 and are in genes with an emerging disease association 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 first 12 months following the initial report.

Sequencing Specifications

TAT
All samples are processed within 20 business days

Platform
Illumina NovaSeq 6000 and/or NovaSeqX (Plus)

Sample types
Buccal swab samples, saliva, blood, DBS cards, isolated DNA (others upon request)

Output
12 Gb +/- 10% per sample
100× median coverage
20× depth of coverage for 98%
Mean 1000× depth for mitochondrial genome

Enrichment kit
Twist Human Core + RefSeq + Mitochondrial Panel
36.6 Mb CCDS coverage
99% of ClinVar variants covered

Raw data options
Vcf and bam files are available free of charge within 2 weeks after the report is issued

Library
2×100 bp

Types of Variants

DNA TYPE	VARIANT TYPE	DEFINITION
nDNA and mtDNA	Single nucleotide variants (SNV)	A DNA sequence variant affecting 1 nucleotide
	Insertion / Deletion	Deletions, insertions, or duplications of DNA segments less than 500bp
	Copy number variants (CNV)	Deletions or duplications of DNA segments of at least 500bp
nDNA	Chromosomal abnormalities	Trisomy, uniparental disomy, monosomy, triploidy

Limitations

- Interpretation is strongly 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 (SNVs) with a minor allele fraction of at least 5%
- This test does not detect: translocations, partial UPD, methylation, gene conversions, low level mosaicism (VAF <5%), low heteroplasmy levels (VAF <5%)

ISO 15189:2022 accredited, ISO 27001:2022 certified

In case you have questions about ordering the appropriate test please send an email to support@arcensus-diagnostics.com

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