arcensus

myLifeGenome

Maximise clinical utility in genetic testing

Whole Genome Sequencing (WGS) is an advanced genetic analysis that is gaining recognition as the first line genetic test. It encompasses the coding and non-coding regions of the nuclear DNA (nDNA) as well as the mitochondrial DNA (mtDNA) and maximises clinical utility as it offers a comprehensive view of the patient's genetic make-up.

Indicated for:

- Patients with a suspected genetic disorder as a first-line genetic test
- Healthy individuals with a family history of a genetic disease
- Healthy individuals interested in knowing their carrier status for family planning

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).
Pharmacogenomic	Variants associated with medication use and dosing (based on PharmCAT and CPIC Guidelines).*

* List of gene drug associations are updated frequently. Please refer to the Arcensus homepage for an up-to-date list covered by myLifeGenome.

🔁 Receive a semi-annual re-evaluation during the first 12 months following the initial report.

Sequencing specifications

TAT
All samples are processed within 20 working days

Platform
Illumina NovaSeq 6000 and/or NovaSeqX (Plus)

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

Output
90Gb +/- 10%
30× median coverage
mtDNA ≥ 1000× mean coverage

Library prep kit
Illumina TruSeq DNA Nano

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

Library
PCR Amplified, 2×150 bp

Types of Variants

DNA TYPE	VARIANT TYPE	DEFINITION
nDNA and mtDNA	Single nucleotide variants	A DNA sequence variant affecting 1 nucleotide
	Insertion / Deletion	Deletions, insertions, or duplications of DNA segments less than 500bp
	Copy number variants	Deletions or duplications of DNA segments of at least 500bp
	Structural variants	Insertions of DNA segments of at least 500bp, inversions and translocations
nDNA	Repeat expansions	Increase in the number of repeated DNA sequence motifs within a gene
	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 test does not detect the following: partial UPD, epigenetic modification, gene conversions and low levels of mosaicism (VAF <10%)
- This test may not reliably detect the following: low levels of mosaicism (VAF <30%), repeat expansion disorders, variants within pseudogene regions/duplicated segments and low levels of mtDNA heteroplasmy (<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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