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myLifeGenome

Maximise clinical utility in genetic testing

Whole Genome Sequencing (WGS) 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). WGS maximises clinical utility by offering a comprehensive view of your patient's genome.

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 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.

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

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Sequencing Specifications

TAT
Reports within 20 business days

Sequencing Platform
Illumina NovaSeq

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

Output
nDNA 30× median coverage
mtDNA 4000× median coverage

Library
Illumina TruSeq DNA Nano
2×150 bp

Raw Data Options
vcf and bam files are provided

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
	Structural variants	Deletions or duplications of DNA segments of at least 50bp, inversions, repeat expansions and translocations
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 test does not detect the following: partial UPD, epigenetic modification, gene conversions and low levels of mosaicism (VAF <5%)
- This test may not reliably detect the following: low levels of mosaicism (VAF <10%), repeat expansion disorders, variants within pseudogene regions/duplicated segments and low levels of mtDNA heteroplasmy (<5%)

CAP ACCREDITED
COLLEGE of AMERICAN PATHOLOGISTS

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