

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
- Dementia, psychiatric disorders, and late-onset neurodegenerative disorders
- 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.

Sequencing Specifications



TAT

All samples are processed within 20 business days



Sequencing Platform

Illumina NovaSeq
2x100 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%)