NGS-based Rare Disease Tests
for Diagnosis of Genetic Disease

DES WES DGS

Comprehensive series of Rare Disease genetic test

DES
Diagnostic Exome Sequencing

5,870 genes
(including mtDNA)

A compact analysis of well-known genes related to genetic diseases.

WES
Whole Exome Sequencing

22,000 genes
(including mtDNA)

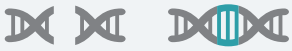
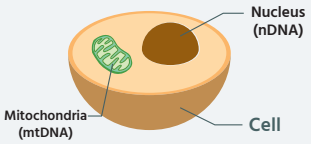
A wide analysis covering most genes related to genetic diseases.

DGS
Diagnostic Genome Sequencing

Whole Genome
(including mtDNA)

A most complete and comprehensive analysis of the human genome.

Detectable Variant types and Genes

			DES	WES	DGS
Detectable Variant types	SNV		✓	✓	✓
	InDel		✓	✓	✓
	CNV		✓	✓	✓
	GR				✓
Target Genes	nDNA		✓	✓	✓
	mtDNA		✓	✓	✓

* SNV (Single Nucleotide Variant) / InDel (Insertion & Deletion) / CNV (Copy Number Variant) / GR (Gene Rearrangement)

Test Subjects

- Patients who want to find the possible genetic cause of the diagnosed disease
- Patients whose symptoms are very broad, complex, or unspecific, not pointing towards specific disease or typical phenotype
- Patients whose prior targeted genetic testing did not provide a conclusive diagnosis so more comprehensive test is necessary
- Patients who is suspicious of mitochondrial disease

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Testable Diseases


Nervous System


Endocrine System


Otorhinolaryngology


Immune System


Opthalmology


Cardiovascular Disease


Liver and Bowels


Kidney


Mitochondria

- Result is classified into positive, inconclusive, and negative according to the clinical significance of the detected variant related to the patient's symptoms.
- Clinical interpretation from a specialized doctor is provided based on the patient's diseases and symptoms.
- Genetic counseling is available.

Service features

Test & Code	DES (ON003) / WES (OT001) / DGS (ON127)		
Specimen	EDTA WB 3 ml	**TAT**	DES (21 days) / WES (34 days) / DGS (66 days)
Method	NGS (Next Generation Sequencing)	**Sample Storage**	Room temperature (Refrigerated is recommended.)
Test description	DES analyzes more than 5,000 clinically important genes related to mendelian disorders based on the latest research data. This test can be the help of treatment decisions and patient prognosis by quick diagnosis. WES analyzes the whole exon and mitochondrial DNA of all genes encoding proteins that account for about 2% of the human genome as nucleotide sequences. If all human genes are analyzed simultaneously using a next-generation sequencing analyzer, it is possible to diagnose about 30-40% of rare diseases. DGS analyzes the whole genome consisting of approximately 30 billion base pairs, including exons and introns, from over 20,000 human genes. It is the most comprehensive rare disease test among three kinds of rare disease tests.		
Caution & Limitation	• The absence of definitive pathogenic findings does not rule out the diagnosis of a genetic disorder as some genetic abnormalities may be undetectable with this test. • It is possible that the genomic region where a disease-causing variant exists in the proband was not captured or sufficiently sequenced with low quality. • Multifactorial disorders and some types of genetic disorders due to nucleotide repeat expansion/contraction, abnormal DNA methylation, and other mechanisms may not be detectable with this test. • This test also cannot reliably detect mosaicism, chromosomal aberrations, and deletions/insertions of 20 bp or more. • Some genes have inherent sequence properties (for example repeats, homology, high GC content, and rare polymorphisms) that may result in suboptimal data, and variants in those regions may not be reliably identified.		

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