Genea BIOMEDX
Genie

Genie CS kits

Product name	No. of Diseases	No. of Genes	No. of sites	Description
Genie CS lite (30)	30	25 genes	>6,000	Low cost, small panel, selected disorders, universal screening
Genie CS extended (717)	717	532 genes	>60,000	Large panel, more genes and disorders, extensive screening

* Both panels include specific disease regions such as DMD/Thalassemia/SMA etc.

Genie CS lite (30): Product Verification

· 262 samples tested

· 100% of samples passed QC, and test results were consistent with the expected results

Type of disease	Type of variation	Number of test samples	Number of samples detected
DMD	A deletion of two or more consecutive exons.	25	25
α - Thalassemia	alpha-thalassemia genotypes: $-\alpha^{3.7}$, $-\alpha^{4.2}$, $--^{SEA}$	92	92
SMA	Exon 7 deletion of the SMN1 gene	51	51
SLC25A13	IVS16ins3kb	6	6
Others	SNV/Del/Dup/Indel	79	79
CNV-negative samples	Samples without copy number variations (CNVs) in DMD, SMN1, HBA1/HBA2.	9	No copy number variations were detected in the relevant genes.

Genie CS extended (717): Product Verification

· 256 samples tested

· 100% of samples passed QC, and test results were consistent with the expected results

Type of disease	Type of variation	Number of test samples	Number of samples detected
DMD	A deletion of two or more consecutive exons.	10	10
HBA	alpha-thalassemia genotypes: $-\alpha^{3.7}$, $-\alpha^{4.2}$, $--^{SEA}$	53	53
SMA	Exon 7 deletion of the SMN1 gene	44	44
SLC25A13	IVS16ins3kb	12	12
Point mutation	SNV/Del/Dup/Indel	27	27

Sequencer: Genie Sequencer

Product	Sequencing mode	Sequencing duration	Sequencing data requirement	No. of Samples/Lane
Genie CS lite (30)	FCL-PE100	≈28h	3Mb (~0.6 Gb)	96
Genie CS extended (717)	FCL-PE100	≈28h	10Mb (~2 Gb)	40

Note:

① The sequencing duration refers only to the time from the start to the end of the sequencing process, excluding the data upload time.

② The formula for calculating data volume is: Single-end data volume * read length * 2 / 1000. For example: 1.4Mb * 150bp * 2 ends / 1000 = 0.42Gb; for example: 4Mb * 150bp * 2 ends / 1000 = 1.2Gb.

③ The number of samples that can be tested is currently limited by the number of available adapters.

Automated analysis software and genetic interpretation reporting system

Each report takes about 3 hours on average (data analysis + report generation)

Genie Carrier Screening kit for Monogenic Disorders

Two panels: 30 disorders; 717 disorders

About 3 Calendar Days

Blood、Buccal Swab、Saliva

All two carrier screening panels cover the following genes: CFTR, SMN1, DMD