

# GeneMind Whole Exome Sequencing Total Solution

## □ Comprehensive coverage of pathogenic variants

Almost completely covers pathogenic or likely pathogenic variants in the ClinVar and LOVD databases.

## □ Supports CNVs detection

1. Single-exon level for key genes (e.g., DMD, Thalassemia);
2. 10 kb high-density probes for high-frequency pathogenic regions (e.g., 22q11.2, 1p36);
3. Genome-wide 200 kb spaced backbone probes (optional 50/100 kb densification);
4. Comprehensive CNV detection from 1 Mb down to single-exon level.

## □ Mitochondria genes is incorporated

Complete capture of the full length of the mitochondrial genome.

## □ Supports gender quality control

## □ User-friendly web system

Analysis software has integrated the latest and practical annotation tools, offering a variety of filtering options that make variants interpretation more user-friendly.



## ◎ Product Introduction

Whole-exome sequencing (WES) is a targeted next generation sequencing (NGS) approach. While the exome constitutes less than 2% of the human genome, it harbors approximately 85% of known disease-related variants, establishing WES as a highly cost-effective strategy compared to whole-genome sequencing.

GeneMind Whole Exome Sequencing solution (GMExome V2) is specifically engineered for the clinical research of hereditary diseases. Developed in accordance with the latest expert consensus and continuously updated disease and variant databases, this solution facilitates precise molecular diagnosis for patients and their families with suspected genetic disorders, thereby helping to address the diagnostic challenges posed by the heterogeneity and complexity of rare diseases.

## ◎ Automated Operation Solution and Analysis System

### Step 1 Sample Preparation



#### GenoScout EM

The Automated Nucleic Acid Extractor is a small-throughput device for nucleic acid extraction. It uses pre-packaged extraction kits (tissue/FFPE/blood/cells etc) without prior allocation.

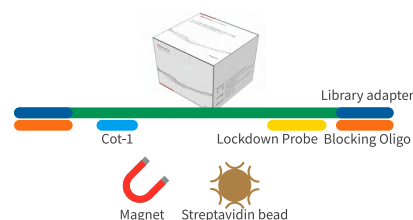
### Step 2 Library Preparation



#### MrLH-96

The Automated system enables streamlined, high-throughput NGS library prep for up to 96 samples in a single run.

### Step 3 Targeted enrichment by probe hybridization



#### Exome Enrichment kit

High efficiency capture kit, 1 reaction allow 6~12 universal libraries capture with  $\geq 80\%$  capture ratio. DNA probe coverage size: 53.55 Mb, covering 20,000+ genes

### Step 4 QC final library



#### Microplate reader

Quantification and QC for libraries using a microplate reader automatically, which can be connected to MrLH-96.

### Step 5 Sequencing



#### GenoLab™ M/FASTASeq™ 300/ SURFSeq™ 5000/SURFSeq™ Q

Provide a range of sequencers from low to high throughput, accommodating varying sample volumes. Standard PE150 sequencing model is recommended.





### Step 6 Data Analysis



#### AIRS

Data analysis and mutations reporting are performed via a local sever. Supports customized reporting.

## ◎ Product Specification

	 GenoLab™ M		 FASTASeq™ 300			 SURFSeq™ 5000			 SURFSeq™ Q	
Flow cell type	FCM	FCH	FCM	FCH	FCP	FCM	FCH	FCP	FCM	FCH
Max Output (Gb)	75	150	37.5	84	150	150	600	1100	3500	7000
Max Read Number (M)	250	500	125	280	500	500	2000	3600	11,700	23,300
Estimated Number of Samples/FC*	6	12	2	6	12	12	48	84	300	600
Estimated Number of Samples/Year**	1872	3744	312	936	1872	3744	14,976	26,208	93,600	187,200

\*Based on GMExome panel with  $\geq 100\times$  mean coverage

\*\*Theoretical throughput is calculated assuming 3 runs weekly across 52 weeks, yielding an annual total of 156 runs.

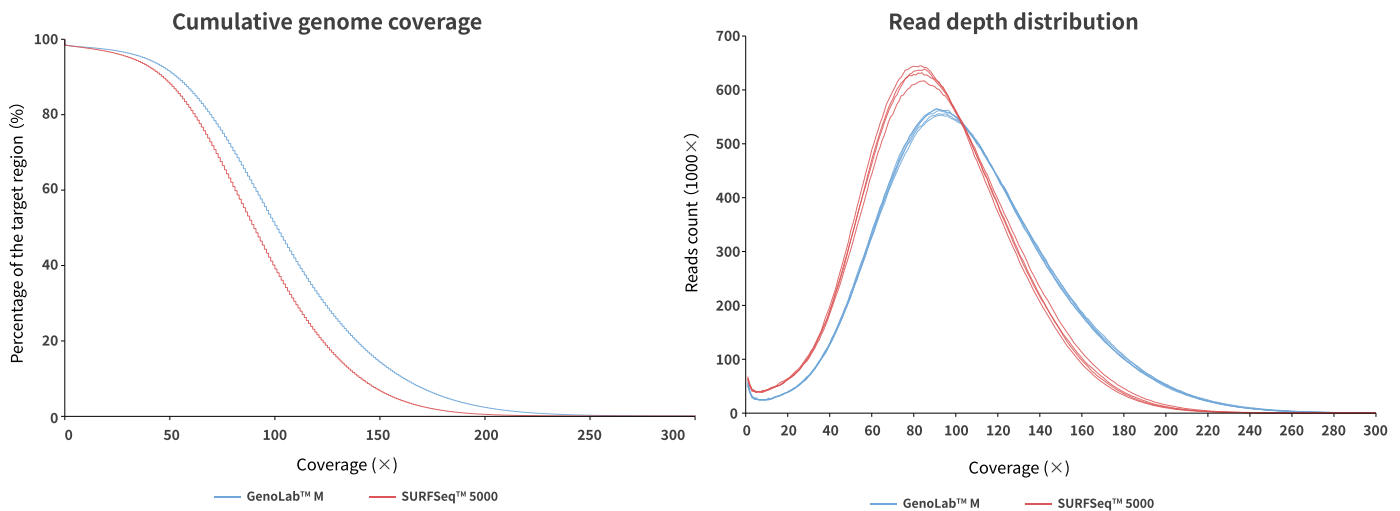
## ◎ Sequencing Data Performance of Blood Samples

(1 RNX with 10 universal libraries, and the data volume is uniformly trimmed to 10 Gb.)

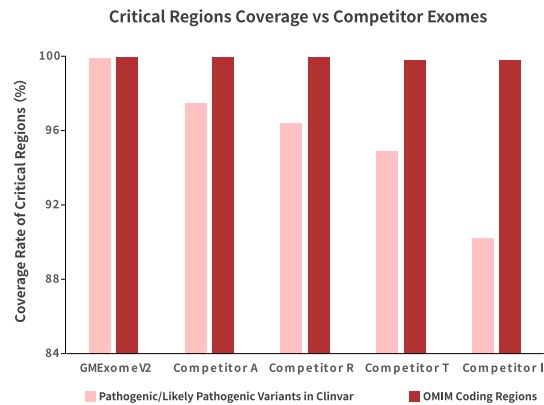
SURFSeq™ 5000							
NO.	Raw bases (Gb)	Q30 (%)	On-target rate (%)	Average depth (×)	Coverage 20 × (%)	Fold 80	ChrM depth
1	10	95.00	80.50	108.99	98.65	1.51	7307
2	10	94.95	80.47	109.24	98.62	1.51	11286
3	10	95.04	80.92	110.11	98.64	1.54	11898
4	10	95.52	82.05	112.24	98.79	1.55	16489
5	10	95.08	81.20	110.43	98.66	1.51	11277
6	10	95.34	81.19	110.28	98.81	1.54	7173
7	10	95.14	81.24	110.52	98.78	1.54	8059
8	10	95.24	82.32	114.00	98.54	1.60	41342
9	10	95.08	81.37	110.74	98.76	1.54	10074
10	10	95.21	81.07	110.25	98.68	1.52	11918

GenoLab™ M							
NO.	Raw bases (Gb)	Q30 (%)	On-target rate (%)	Average depth (×)	Coverage 20 × (%)	Fold 80	ChrM depth
1	10	93.34	81.67	117.31	99.1	1.52	10594
2	10	93.68	82.06	116.07	99.3	1.53	8680
3	10	93.87	81.54	118.58	99.32	1.54	9927
4	10	92.87	81.49	113.86	99.29	1.51	9987
5	10	93.92	82.21	117.99	99.3	1.55	9244
6	10	93.72	80.49	116.76	99.28	1.56	7733
7	10	93.37	81.84	115.68	99.09	1.49	8327
8	10	93.70	80.48	117.09	99.28	1.56	14106
9	10	93.60	82.12	119.66	99.06	1.59	40726
10	10	93.74	81.20	115.90	99.13	1.48	7972

## Percentage of the Reference Genome-NA12878



## Comprehensive coverage



Comprehensive coverage of genes associated with disease & pathogenic or likely pathogenic variants in the Clinvar and OMIM genes Coding Regions.

## ✂ Disclaimer

This document is based on company data of October, 2025, and is provided for reference only.

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