GeneMind

# GeneMind Whole Exome Sequencing Total Solution

## ☐ Comprehensive coverage of pathogenic variants

Comprehensively 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*,*HBA1/HBA2*, *HBB*);
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 detection of CNVs ⩾1 Mb.

## ☐ Mitochondrial genome can be fully captured

Complete capture of the full length of the mitochondrial genome.

## ☐ Supports sex 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 Nucleic Acid Extraction



### 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.

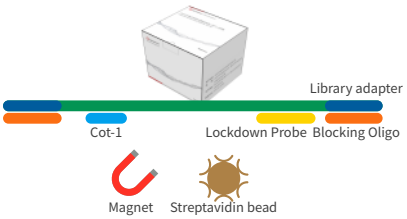
## Step 2 Library Preparation



### 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 ⩾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 server. 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 ⩾100 × mean coverage
**CNV analysis is not supported for batch sizes with fewer than 8 samples. A minimum batch size of 8 samples is required for successful CNV analysis.
***Theoretical throughput is calculated assuming 3 runs weekly across 52 weeks, yielding an annual total of 156 runs.

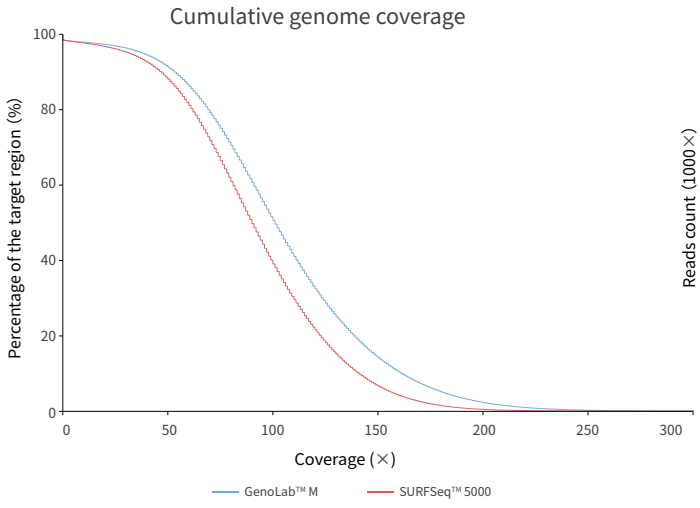
# ◎ Sequencing Data Performance of Blood Samples

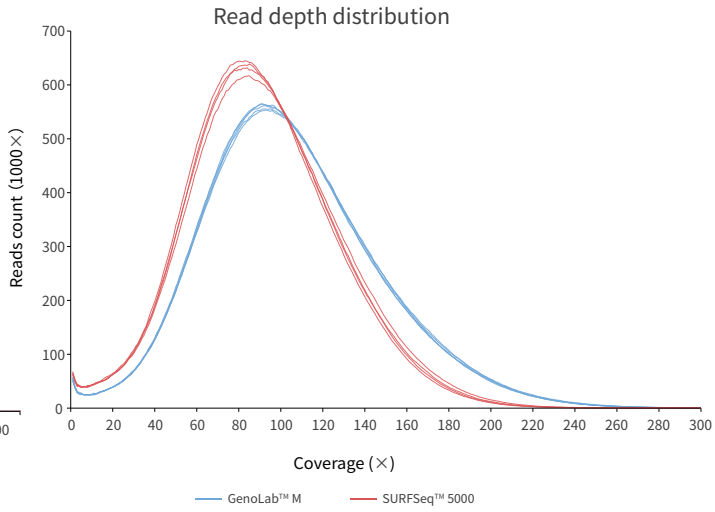
(1 RNX with 10 universal libraries, and the data volume is uniformly trimmed to 10 Gb.)

| SURFSeq™ 5000 | | | | | | | |
|---|---|---|---|---|---|---|---|
| NO. | Raw bases (Gb) | Q30 (%) | On-taget 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 |

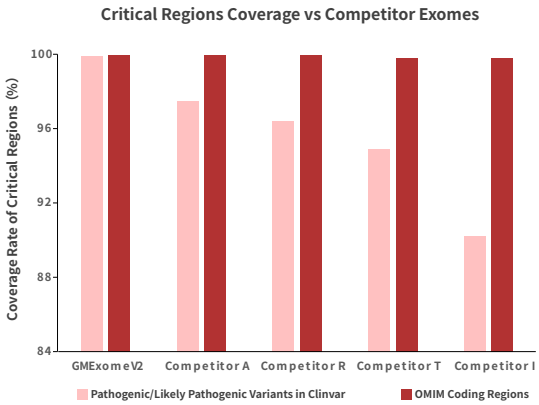
| GenoLab™ M | | | | | | | |
|---|---|---|---|---|---|---|---|
| NO. | Raw bases (Gb) | Q30 (%) | On-taget rate (%) | Average depth (×) | Coverage 20 × (%) | Fold 80 | ChrM depth |
| 1 | 10 | 93.34 | 81.67 | 117.31 | 99.1 | 1.52 | 10594 |
| 2 | 10 | 93.68 | 82.06 | 116.07 | 99.3 | 1.53 | 8680 |
| 3 | 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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