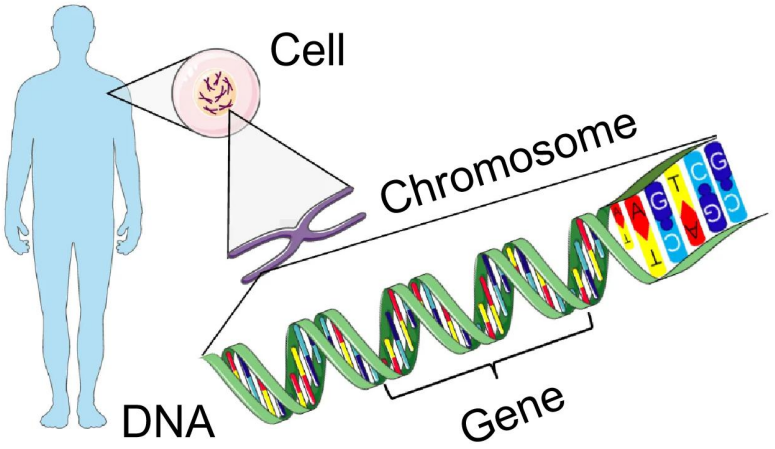
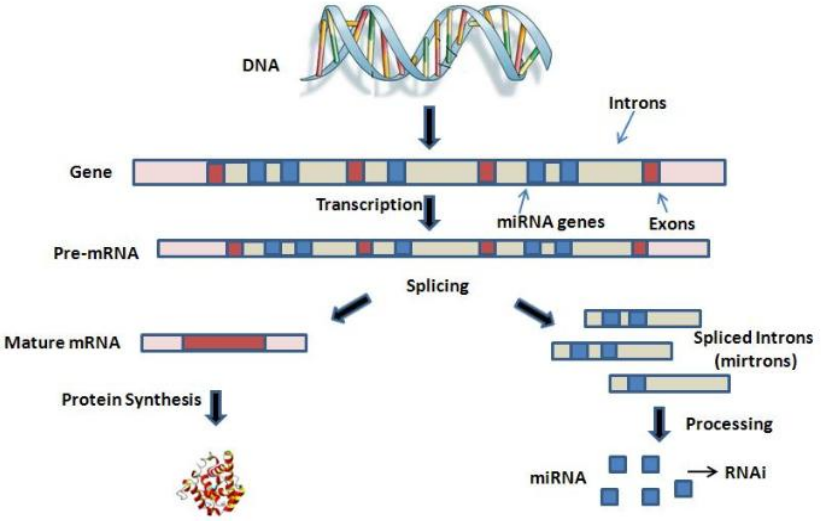
# GeneMind WES Solution

GeneMind 真迈生物

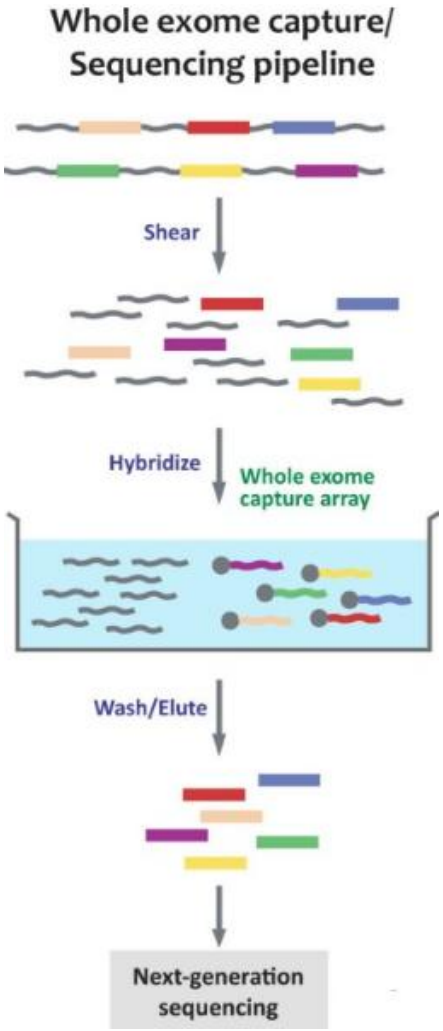
# Exons and Genetic Diseases

- **Whole-exome sequencing is a widely used next-generation sequencing (NGS) method that involves sequencing the protein-coding regions of the genome.**
- **The human exome represents less than 2% of the genome, but contains ~85% of known disease-related variants, making this method a cost-effective alternative to whole-genome sequencing.**
- **Exome sequencing using exome enrichment can efficiently identify coding variants across a broad range of applications, including population genetics, genetic disease, and cancer studies.**



# WES Principle and Advantage




## Advantage of Exome Sequencing

- Targeted view of the protein-coding regions of the genome
- Efficient Analysis of Coding Regions, provide reliable and sensitive detection results of coding variants (SNVs, Indels)
- Fast and cost effective sequencing compared with WGS(8–10 Gb of sequencing per exome compared to ~90 Gb per whole human genome)

# GeneMind WES Total Solution

| PARAMETER | | | |
|---|---|---|---|
| Sample type | gDNA / Tissue / Blood | | |
| Application | Genes and Inherited Disease | | |
| Sequencing Platform | GenoLab M* | FASTASeq 300* | SURFSeq 5000* |
| No. of samples/run[1] | 1 FCM : 6<br>1 FCH : 12<br>1 FCM+1 FCH: 18<br>2 FCM: 12<br>2 FCH: 24 | 1 FCM: 2<br>1 FCH: 6 | 1 FCM : 12<br>1 FCH : 48<br>1 FCM+1 FCH: 60<br>2 FCM: 24<br>2 FCH: 96 |
| Read length | PE150 | | |
| Effective throughput/sample | Around 10 Gb | | |
| Turn-around-time | 3-4 days | | |
| Report generation | Local analysis and report system | | |

1. Please note that the sample volume shown in this table is only for customer reference, due to the real situation, such as sample type, library quailty will be different.

*Unless otherwise informed, GeneMind sequencing platform and related sequencing reagents are not available in the USA, Canada, Australia, Japan, Singapore, Western Europe and Nordic countries yet.

# GeneMind WES solution Workflow(32 samples)





information registration, DNA extraction → Library Construction & Hybridization Capture → Incubation* → QC, pooling, Loading → Sequencing → Analysis and Report

| | | | | | | | |
|---|---|---|---|---|---|---|---|
| Automated | 2.5h | 4h | 16h | 2.5h | 24-53h | 10h | 61-88h |





| | | | | | | | |
|---|---|---|---|---|---|---|---|
| Manually | 2.5h | 4h | 16h | 2.5h | 24-53h | 10h | 61 - 88h |

*In the incubation step of hybridization capture library preparation, the hybridization mixture is incubated to allow the probes to bind specifically to their target sequences. This step does not require human intervention once the mixture is prepared.

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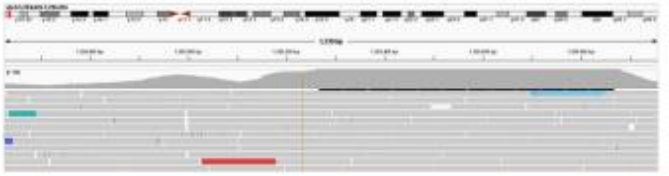
# Library Preparation and Enrichment

**Spanning 36.68 Mb target region of the human genome with efficient probe design, Genemind Universal DNA Library prep with enrichment Kit provides a more complete coverage of protein coding genes for the variant detection which could be widely used in clinical and academic research.**

| Database | CDS Region Coverage (%) |
|---|---|
| RefSeq | 99.88 |
| MANE | 99.61 |
| CCDS | 99.92 |

| Coverage Distribution from IGV | Sequencing Depth (X) | | |
|---|---|---|---|
| | Average | chr5:1295113 | chr5:1295135 |
|  | 127 | 141 | 173 |

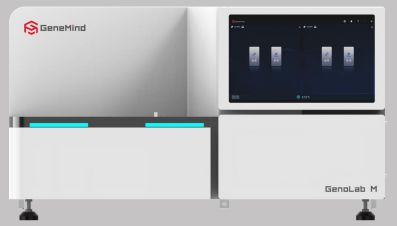
# Sequencing Platforms

## Benchtop sequencer

### FASTASeq 300*

| 100M/250M |
| --- |
| 7.5Gb-75Gb |
| mNGS/tNGS/Panel/ NIPT/PGT-A |

### GenoLab M*




| 250M/500M |
| --- |
| 18Gb-300Gb |
| NIPT/PGT-A/CNV-Seq/ mNGS/cancer panel/ WES/RNAseq |

### SURFSeq 5000*


| 500M/2000M/3600M* |
| --- |
| 50Gb-2.2Tb (available in Nov. 2024) |
| WES/WGS/cancer panel/Single cell sequencing |

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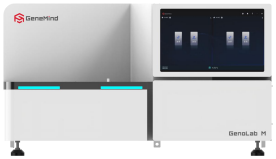
# GeneMind Sequencing Platforms



| Product | GenoLab M* | FASTASeq 300* | SURFSeq 5000* |
|---|---|---|---|
| Launch year | 2020 | 2022 | 2023 |
| Features | Comprehensive and flexible | Fast, flexible and simple-to-operate | Economical, efficient and all-round |
| Product qualification | CE IVDR | CE IVDR | CE IVDR |
| Applications | NIPT, PGT-A, Pathogen detection, cancer panel, and genetic disease diagnosis such as WES and WGS | NIPT, PGT-A, WES, cancer panel, pathogen detection,16s, forensic | Scientific and technological services, MRD, Pan-cancer analysis, agricultural breeding or species identification, WES, WGS |
| Flow cell per run | 2 Flow Cells | 1 Flow Cell | 2 Flow Cells |
| Flow cell Type | FCM: 250 M reads<br>FCH: 500 M reads | FCM: 100 M reads<br>FCH: 250 M reads<br>FCX: 100 M reads (available in July 2024) | FCM: 500 M reads<br>FCH: 2000 M reads<br>FCP: 3600 M reads (available in November 2024) |
| Read length | FCM: 75/150/300<br>FCH: 75/150/300 | FCM: 75/150/300<br>FCH: 75/150/300<br>FCX: 400/600 | FCM: 100/200/300/600<br>FCH: 50/100/200/300<br>FCP: 50/100/200/300 |

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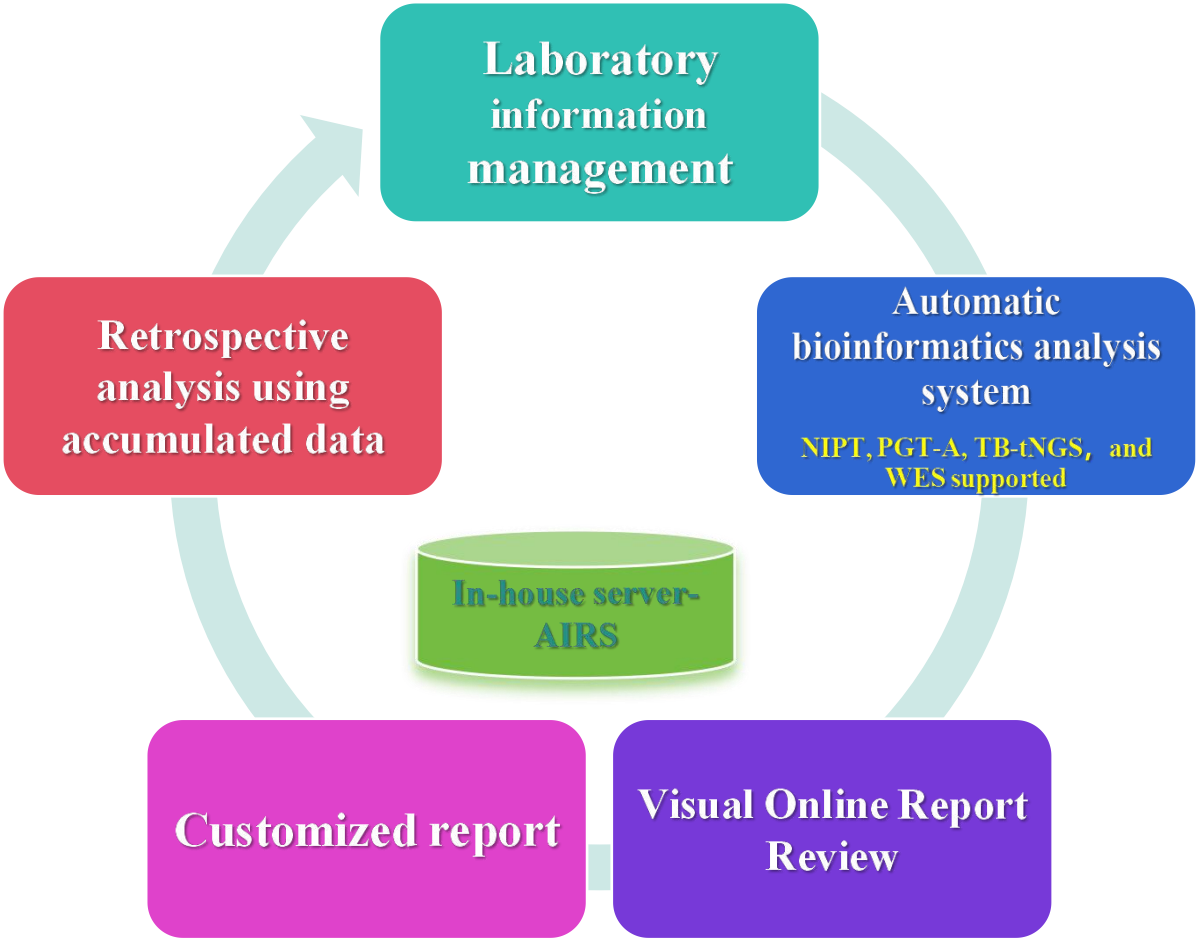
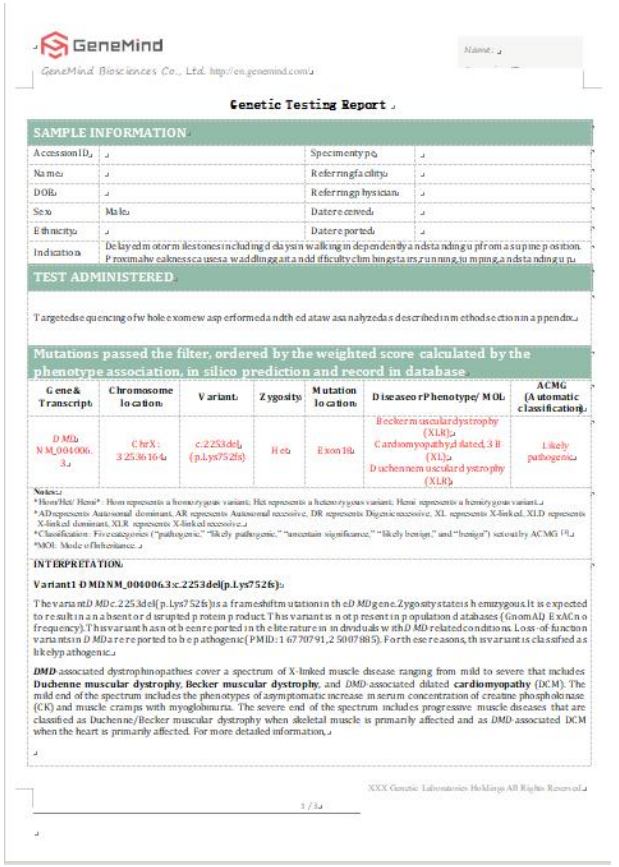
# Automatic Analysis Report System



# WES Report Template



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**Genetic Testing Report**

**SAMPLE INFORMATION**

| | | | |
|---|---|---|---|
| Accession ID | | Specimen type | |
| Name | | Referring facility | |
| DOB | | Referring physician | |
| Sex | Male | Date received | |
| Ethnicity | | Date reported | |
| Indication | Delayed motor milestones including delayed walking independently and standing up from a supine position. Proximal weakness causes a waddling gait and difficulty climbing stairs, running, jumping, and standing up. | | |

**TEST ADMINISTERED**

Targeted sequencing of whole exome was performed and the data was analyzed as described in the method section in appendix.

**Mutations passed the filter, ordered by the weighted score calculated by the phenotype association, in silico prediction and record in database**

| Gene & Transcript | Chromosome location | Variant | Zygosity | Mutation location | Disease or Phenotype/ MOI | ACMG (Automatic classification) |
|---|---|---|---|---|---|---|
| *DMD* NM_004006.3 | ChrX: 32536164 | c.2253del (p.Lys752fs) | Hem | Exon18 | Becker muscular dystrophy (XLR); Cardiomyopathy dilated, 3B (XL); Duchenne muscular dystrophy (XLR) | Likely pathogenic |

Notes:
*Hom/Het/ Hemi*: Hom represents a homozygous variant; Het represents a heterozygous variant; Hemi represents a hemizygous variant.
*AD represents Autosomal dominant, AR represents Autosomal recessive, DR represents Digenic recessive, XL represents X-linked, XLD represents X-linked dominant, XLR represents X-linked recessive.
*Classification: Five categories ("pathogenic," "likely pathogenic," "uncertain significance," "likely benign," and "benign") set out by ACMG [1].
*MOI: Mode of Inheritance.

**INTERPRETATION**

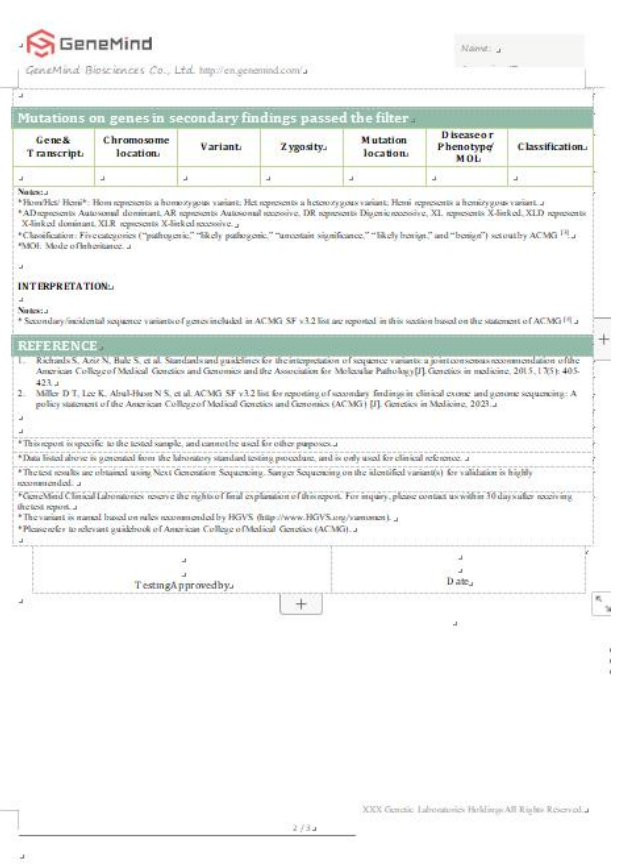
**Variant1 *DMD* NM_004006.3:c.2253del(p.Lys752fs):**

The variant *DMD* c.2253del(p.Lys752fs) is a frameshift mutation in the *DMD* gene. Zygosity status hemizygous. It is expected to result in an absent or disrupted protein product. This variant is not present in population databases (GnomAD ExAC no frequency). This variant has also been reported in the literature in individuals with *DMD*-related conditions. Loss-of-function variants in *DMD* are reported to be pathogenic (PMID: 16770791, 25007885). For these reasons, this variant is classified as likely pathogenic.

***DMD*-associated dystrophinopathies cover a spectrum of X-linked muscle disease ranging from mild to severe that includes **Duchenne muscular dystrophy**, **Becker muscular dystrophy**, and *DMD*-associated dilated **cardiomyopathy** (DCM). The mild end of the spectrum includes the phenotypes of asymptomatic increase in serum concentration of creatine phosphokinase (CK) and muscle cramps with myoglobinuria. The severe end of the spectrum includes progressive muscle diseases that are classified as Duchenne/Becker muscular dystrophy when skeletal muscle is primarily affected and as *DMD*-associated DCM when the heart is primarily affected. For more detailed information,







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**Mutations on genes in secondary findings passed the filter**

| Gene & Transcript | Chromosome location | Variant | Zygosity | Mutation location | Disease or Phenotype/ MOI | Classification |
|---|---|---|---|---|---|---|
| | | | | | | |

Notes:
*Hom/Het/ Hemi*: Hom represents a homozygous variant; Het represents a heterozygous variant; Hemi represents a hemizygous variant.
*AD represents Autosomal dominant, AR represents Autosomal recessive, DR represents Digenic recessive, XL represents X-linked, XLD represents X-linked dominant, XLR represents X-linked recessive.
*Classification: Five categories ("pathogenic," "likely pathogenic," "uncertain significance," "likely benign," and "benign") set out by ACMG [1].
*MOI: Mode of Inheritance.

**INTERPRETATION**

Notes:
* Secondary/incidental sequence variants of genes included in ACMG SF v3.2 list are reported in this section based on the statement of ACMG [1].

**REFERENCE**

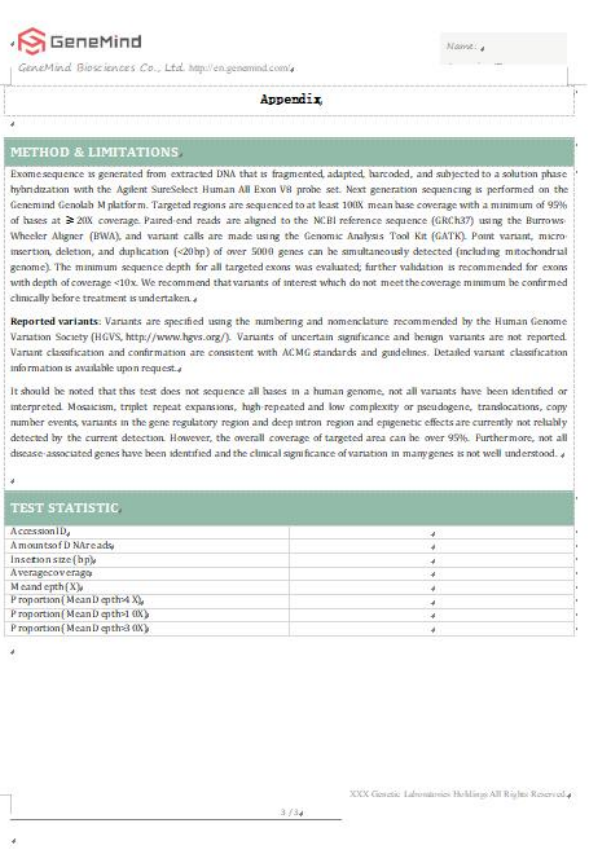

1. Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology [J]. Genetics in medicine, 2015, 17(5): 405-423.
2. Miller D T, Lee K, Abul-Husn N S, et al. ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG) [J]. Genetics in Medicine, 2023.


* This report is specific to the tested sample, and cannot be used for other purposes.
* Data listed above is generated from the laboratory standard testing procedure, and is only used for clinical reference.
* The test results are obtained using Next Generation Sequencing. Sanger Sequencing on the identified variant(s) for validation is highly recommended.
* GeneMind Clinical Laboratories reserve the rights of final explanation of this report. For inquiry, please contact us within 30 days after receiving the test report.
* The variant is named based on rules recommended by HGVS (http://www.HGVS.org/variantnam).
* Please refer to relevant guidebook of American College of Medical Genetics (ACMG).

| | |
|---|---|
| Testing Approved by | Date |







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Name:

**Appendix**

**METHOD & LIMITATIONS**

Exome sequence is generated from extracted DNA that is fragmented, adapted, barcoded, and subjected to a solution phase hybridization with the Agilent SureSelect Human All Exon V8 probe set. Next generation sequencing is performed on the Genemind Genolab M platform. Targeted regions are sequenced to at least 100X mean base coverage with a minimum of 95% of bases at ≥ 20X coverage. Paired-end reads are aligned to the NCBI reference sequence (GRCh37) using the Burrows-Wheeler Aligner (BWA), and variant calls are made using the Genomic Analysis Tool Kit (GATK). Point variant, micro-insertion, deletion, and duplication (<20bp) of over 5000 genes can be simultaneously detected (including mitochondrial genome). The minimum sequence depth for all targeted exons was evaluated; further validation is recommended for exons with depth of coverage <10x. We recommend that variants of interest which do not meet the coverage minimum be confirmed clinically before treatment is undertaken.

**Reported variants**: Variants are specified using the numbering and nomenclature recommended by the Human Genome Variation Society (HGVS, http://www.hgvs.org/). Variants of uncertain significance and benign variants are not reported. Variant classification and confirmation are consistent with ACMG standards and guidelines. Detailed variant classification information is available upon request.

It should be noted that this test does not sequence all bases in a human genome, not all variants have been identified or interpreted. Mosaicism, triplet repeat expansions, high-repeated and low complexity or pseudogene, translocations, copy number events, variants in the gene regulatory region and deep intron region and epigenetic effects are currently not reliably detected by the current detection. However, the overall coverage of targeted area can be over 95%. Furthermore, not all disease-associated genes have been identified and the clinical significance of variation in many genes is not well understood.

**TEST STATISTIC**

| | |
|---|---|
| Accession ID | |
| Amounts of DNA reads | |
| Insertion size (bp) | |
| Average coverage | |
| Mean depth (X) | |
| Proportion (Mean Depth≥4 X) | |
| Proportion (Mean Depth≥3 0X) | |
| Proportion (Mean Depth≥3 0X) | |

# Ordering Information

| Package code | Package name | Item | PN | Product Name | Specification | Quantity |
|---|---|---|---|---|---|---|
| A000095 | WES package (96 reactions,for GenoLab M FCM) | Extraction Kit | L000093 | GeneMind Magnetic Universal Genomic DNA Kit | 96 samples/test | 2 |
| | | Target Enrichment Kit | L000092 | GeneMind Hyb & Wash Kit v2.0 | | 1 |
| | | Library prep Kit | L000094 | GeneMind Universal DNA Library Prep Kit | | 1 |
| | | Beads | R000008 | GenoVerse DNA Selection Beads V1.0 （16ml） | | 2 |
| | | Adaptor | L000079 | GenoVerse Plate-type Single-indexed Full-Length DNAAdapters V1.0 (96 rxns) | | 1 |
| | | Sequencing Kit | S000044 | GenoLab M seqnencing Set V1.0 (FCH 300cycles) | | 8 |
| A000096 | WES package (96 reactions,for GenoLab M FCH) | Extraction Kit | L000093 | GeneMind Magnetic Universal Genomic DNA Kit | 96 samples/test | 2 |
| | | Target Enrichment Kit | L000092 | GeneMind Hyb & Wash Kit v2.0 | | 1 |
| | | Library prep Kit | L000094 | GeneMind Universal DNA Library Prep Kit | | 1 |
| | | Beads | R000008 | GenoVerse DNA Selection Beads V1.0 （16ml） | | 2 |
| | | Adaptor | L000079 | GenoVerse Plate-type Single-indexed Full-Length DNAAdapters V1.0 (96 rxns) | | 1 |
| | | Sequencing Kit | S000044 | GenoLab M seqnencing Set V1.0 (FCH 300cycles) | | 16 |
| A000097 | WES package (96 reactions,for FASTASeq 300 FCM) | Extraction Kit | L000093 | GeneMind Magnetic Universal Genomic DNA Kit | 96 samples/test | 2 |
| | | Target Enrichment Kit | L000092 | GeneMind Hyb & Wash Kit v2.0 | | 1 |
| | | Library prep Kit | L000094 | GeneMind Universal DNA Library Prep Kit | | 1 |
| | | Beads | R000008 | GenoVerse DNA Selection Beads V1.0 （16ml） | | 2 |
| | | Adaptor | L000079 | GenoVerse Plate-type Single-indexed Full-Length DNAAdapters V1.0 (96 rxns) | | 1 |
| | | Sequencing Kit | S000189 | FASTASeq 300 Sequencing Kit V1.0 （FCM 300cycles） | | 48 |
| A000098 | WES package (96 reactions,for FASTASeq 300 FCH) | Extraction Kit | L000093 | GeneMind Magnetic Universal Genomic DNA Kit | 96 samples/test | 2 |
| | | Target Enrichment Kit | L000092 | GeneMind Hyb & Wash Kit v2.0 | | 1 |
| | | Library prep Kit | L000094 | GeneMind Universal DNA Library Prep Kit | | 1 |
| | | Beads | R000008 | GenoVerse DNA Selection Beads V1.0 （16ml） | | 2 |
| | | Adaptor | L000079 | GenoVerse Plate-type Single-indexed Full-Length DNAAdapters V1.0 (96 rxns) | | 1 |
| | | Sequencing Kit | S000197 | FASTASeq 300 Sequencing Kit V1.0 （FCH 300cycles） | | 16 |
| A000099 | WES package (96 reactions, for SURFSeq 5000 FCH) | Extraction Kit | L000093 | GeneMind Magnetic Universal Genomic DNA Kit | 96 samples/test | 2 |
| | | Target Enrichment Kit | L000092 | GeneMind Hyb & Wash Kit v2.0 | | 1 |
| | | Library prep Kit | L000094 | GeneMind Universal DNA Library Prep Kit | | 1 |
| | | Beads | R000008 | GenoVerse DNA Selection Beads V1.0 （16ml） | | 2 |
| | | Adaptor | L000079 | GenoVerse Plate-type Single-indexed Full-Length DNAAdapters V1.0 (96 rxns) | | 1 |
| | | Sequencing Kit | S000242 | SURFSeq 5000 Sequencing Kit V1.0 （FCH 300cycles） | | 2 |
| A000100 | WES package (96 reactions, for SURFSeq 5000 FCM) | Extraction Kit | L000093 | GeneMind Magnetic Universal Genomic DNA Kit | 96 samples/test | 2 |
| | | Target Enrichment Kit | L000092 | GeneMind Hyb & Wash Kit v2.0 | | 1 |
| | | Library prep Kit | L000094 | GeneMind Universal DNA Library Prep Kit | | 1 |
| | | Beads | R000008 | GenoVerse DNA Selection Beads V1.0 （16ml） | | 2 |
| | | Adaptor | L000079 | GenoVerse Plate-type Single-indexed Full-Length DNAAdapters V1.0 (96 rxns) | | 1 |
| | | Sequencing Kit | S000238 | SURFSeq 5000 Sequencing Kit V1.0 （FCM 300cycles） | | 8 |

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# Paper Pubulished - GeneMind NGS Platform （38+， as of June 2024)

| List | Title | Application | ublish Tin | IF | JCR |
|---|---|---|---|---|---|
| 1 | Single molecule targeted sequencing for cancer gene mutation detection | cancer | 2016 | 4.26 | Q2 |
| 2 | Single molecule sequencing of the M13 virus genome without amplification | virus-genome | 2017 | 2.77 | Q2 |
| 3 | Fraternal twins with Phelan-McDermid syndrome not involving the SHANK3 gene: case report and literature review | disease | 2020 | 2.57 | Q2 |
| 4 | Characterizing the gene mutations associated with resistance to gatifloxacin in Mycobacterium tuberculosis through whole genome sequence | Mycobacterium-resistance | 2021 | 12.07 | Q1 |
| 5 | Comparative performance of the GenoLab M and NovaSeq 6000 sequencing platforms for transcriptome and LncRNA analysis | RNA&LncRNA | 2021 | 3.97 | Q2 |
| 6 | **Accuracy benchmark of the GeneMind GenoLab M sequencing platform for WGS and WES analysis** | WGS&WES | 2022 | 4.55 | Q2 |
| 7 | Non-invasive Prenatal Screening for Common Fetal Aneuploidies by Single Molecular Sequencing | NIPT | 2022 | 5.50 | Q1 |
| 8 | Systematic and benchmarking studies of pipelines for mammal WGBS data in the novel NGS platform | WGBS | 2023 | 3.0 | Q2 |
| 9 | Evaluation of environmental factors and microbial community structure in an important drinking-water reservoir across seasons | metagenome | 2023 | 5.2 | Q1 |
| 10 | Comparison of the Illumina NextSeq 2000 and GeneMind Genolab M sequencing platforms for spatial transcriptomics | enomics Visium spatial transcripto | 2023 | 4.4 | Q2 |
| 11 | Genetic characterization of human adenoviruses in patients using metagenomic next-generation sequencing (mNGS) in Hubei, China, from 2018 to 2019 | mNGS | 2023 | 5.2 | Q1 |
| 12 | FWAlgaeDB, an integrated genome database of freshwater algae | metagenome | 2023 | 4.6 | Q2 |
| 13 | Inhibition of Hepatitis B Virus (HBV) replication and antigen expression by Brucea javanica (L.) Merr. oil emulsion | RNA-seq | 2023 | 5.7 | Q1 |
| 14 | Ultra-high static magnetic fields cause immunosuppression through disrupting B-cell peripheral differentiation and negatively regulating BCR signaling | RNA-seq | 2023 | 9.9 | Q1 |
| 15 | Accumulation of Endogenous Adenosine Improves Cardiomyocyte Metabolism via Epigenetic Reprogramming in an Ischemia-Reperfusion Model | RNA-seq | 2023 | 11.4 | Q1 |
| 16 | Winter wheat lodging resistance characteristics as affected by nitrogen application time and the underlying mechanism | RNA-seq | 2023 | 0.9 | Q3 |
| 17 | Preparation of a Single-Cell Suspension from Tumor Biopsy Samples for Single-Cell RNA Sequencing | BD scRNA-seq | 2023 | 0.7 | Q3 |
| 18 | Development of Synbiotic Preparations That Restore the Properties of Cattle Feed Affected by Toxin-Forming Micromycetes | Microbe Genome | 2023 | 3.6 | Q1 |
| 19 | G-Protein-Coupled Receptors Mediate Modulations of Cell Viability and Drug Sensitivity by Aberrantly Expressed Recoverin 3 within A549 Cells | RNA-seq | 2023 | 5.6 | Q1 |
| 20 | 3D Spheroid Configurations Are Possible Indictors for Evaluating the Pathophysiology of Melanoma Cell Lines | RNA-seq | 2023 | 6 | Q1 |
| 21 | Comparative analysis of clinical and immunological profiles across Omicron BA.5.2 subvariants using next-generation sequencing in a Chinese cohort | Microbe Genome | 2023 | 5.7 | Q1 |
| 22 | ase Report: Four cases of SARS-CoV-2-associated Guillain-Barré Syndrome with SARS-CoV-2-positive cerebrospinal fluid detected by metagenomic next-generation sequencing: a retrospective case series from Chir | mNGS | 2023 | 7.3 | Q1 |
| 23 | Coinfection of SARS-CoV-2 and influenza A (H3N2) detected in bronchoalveolar lavage fluid of a patient with long COVID using metagenomic next−generation sequencing: a case report | mNGS | 2023 | 5.7 | Q1 |
| 24 | Systematic evaluation of multiple NGS platforms for structural variants detection | WGS | 2023 | 4.8 | Q2 |
| 25 | Draft genome sequence of perfluorinated acids C7–C9 degrading bacterium Pseudomonas mosselii 5(3), isolated from soil contaminated with pesticides | Microbe Genome | 2023 | 0.8 | Q3 |
| 26 | Biodegradation Potential of C7-C10 Perfluorocarboxylic Acids and Data from the Genome of a New Strain of Pseudomonas mosselii 5(3) | Microbe Genome | 2023 | 4.6 | Q1 |
| 27 | Systematic comparison of variant calling pipelines of target genome sequencing cross multiple next-generation sequencers | panel | 2023 | 3.7 | Q2 |
| 28 | Analysis of the transcriptional activity of model piggyBac transgenes stably integrated into different loci of the genome of CHO cells in the absence of selection pressure | RNA-seq | 2023 | 0.9 | Q4 |
| 29 | Sequencing and analysis of complete chloroplast genomes of einkorn wheats Triticum sinskajae and Triticum monococcum accession k-20970 | chloroplast genomes | 2023 | 2.0 | Q2 |
| 30 | Transcriptome analysis reveals the underlying mechanism for over-accumulation of alkaline protease in Bacillus licheniformis | RNA-seq | 2023 | 4 | Q2 |
| 31 | Expression of USP25 associates with fibrosis, inflammation and metabolism changes in IgG4-related disease | RNA-seq | 2024 | 16.6 | Q1 |
| 32 | Changes in molting frequency and expression patterns of molting-related genes in Macrobrachium rosenbergii with exogenous calcium supplement in water | RNA-seq | 2024 | 4.5 | Q1 |
| 33 | Ecophysiological responses of Phragmites australis populations to a tidal flat gradient in the Yangtze River Estuary, China | RNA-seq | 2024 | 5.6 | Q1 |
| 34 | The complete chloroplast genome sequence of Leibnitzia anandria (Linnaeus) Turczaninow | chloroplast genomes | 2024 | 0.5 | Q4 |
| 35 | Prenatal diagnosis of a trisomy 7 mosaic case: CMA, CNV-seq, karyotyping, interphase FISH, and MS-MLPA, which technique to choose? | CNV-seq | 2024 | 3.1 | Q2 |
| 36 | Modulation of Epithelial–Mesenchymal Transition Is a Possible Underlying Mechanism for Inducing Chemoresistance in MIA PaCa-2 Cells against Gemcitabine and Paclitaxel | RNA-seq | 2024 | 4.7 | Q2 |
| 37 | **Assessing the impact of sequencing platforms and analytical pipelines on whole-exome sequencing** | WES | 2024 | 3.7 | Q2 |
| 38 | Nano-bio interaction of magnetic nanoparticles with cells in a tumor at the single-cell level | tumor scRNA-seq | 2024 | 17.4 | Q1 |

# High Data Quailty (Real Clinial Results)

| | Flowcell-99 | ZM20240510QWXZ99 | ZM20240510QWXZ100 | ZM20240510QWXZ101 | ZM20240510QWXZ102 |
|---|---|---|---|---|---|
| **Raw reads** | 96,968,734 | 92,958,870 | 98,970,496 | 72,194,108 | 80,859,444 |
| **Raw bases** | 14,545,310,100 | 13,943,830,500 | 14,845,574,400 | 10,829,116,200 | 12,128,916,600 |
| **Clean reads** | 96,434,204 | 92,387,208 | 98,398,030 | 71,158,358 | 79,708,810 |
| **Clean bases** | 14,152,496,510 | 13,571,818,188 | 14,456,606,104 | 10,513,160,522 | 11,811,282,258 |
| **Q20** | 98.65% | 98.65% | 98.68% | 98.56% | 98.51% |
| **Q30** | 95.94% | 95.90% | 96.01% | 96.13% | 95.96% |
| **GC%** | 51.25% | 51.15% | 51.05% | 52.78% | 52.60% |
| **Reads with adaptors** | 11,664,012 | 10,496,078 | 11,100,306 | 6,072,120 | 5,435,740 |
| **Mapping rate** | 99.97% | 99.97% | 99.97% | 99.97% | 99.98% |
| **On target rate** | 87.14% | 86.65% | 87.11% | 86.69% | 86.43% |
| **Insert size** | 213 | 215 | 217 | 230 | 241 |
| **Coverage** | 99.60% | 99.90% | 99.90% | 99.60% | 99.60% |
| **Average depth** | 198.8X | 188.4X | 201.0X | 142.6X | 156.4X |
| **Depth > 10X** | 99.10% | 99.30% | 99.40% | 98.80% | 98.70% |
| **Depth > 20X** | 98.55% | 98.75% | 98.91% | 97.35% | 97.32% |

- **Q30>95%**
- **Mapping rate>99%**
- **On target rate>86%**
- **Average depth>140X**

# Scientific Paper 1 (Download Link: https://doi.org/10.1186/s12864-022-08775-3)

- Title: Accuracy benchmark of the GeneMind GenoLab M sequencing platform for WGS and WES analysis
- Sample type: HG001 and HG002
- Library type: TruSeq Nano DNA library prep kit
- Platform: NA, NT and GenoLab M*

| Samples | Library Type | Sequencing Platform | Read (M) | Bases (Gb) | Duplication rate (%) | >Q20 | >Q30 | Alignment rate (%) | Mean coverage (X) | %_bases_above_15x |
|---|---|---|---|---|---|---|---|---|---|---|
| GL_WGS_22 | WGS | GenoLab M | 442.77 | 66.42 | 1.73% | 95.35% | 88.26% | 99.88% | 22.39 | 81.30% |
| GL_WGS_33 | WGS | GenoLab M | 662.66 | 99.40 | 1.93% | 95.22% | 87.99% | 99.88% | 33.50 | 93.90% |
| NA_WGS_22 | WGS | NS | 424.9 | 63.73 | 3.57% | 95.92% | 90.05% | 99.64% | 21.37 | 87.30% |
| NA_WGS_33 | WGS | NS | 655.83 | 98.38 | 5.32% | 95.92% | 90.05% | 99.64% | 32.99 | 97.70% |
| GL_WES_100 | WES Agilent V8 | GenoLab M | 41.87 | 6.28 | 6.00% | 93.95% | 84.71% | 99.95% | 112.42 | 98.00% |
| GL_WES_raw | WES Agilent V8 | GenoLab M | 70.36 | 10.55 | 9.71% | 93.95% | 84.71% | 99.95% | 188.90 | 99.00% |
| NA_WES_100 | WES Agilent V8 | NS | 39.35 | 5.90 | 14.85% | 98.01% | 94.05% | 99.95% | 107.72 | 99.30% |
| NA_WES_raw | WES Agilent V8 | NS | 81.16 | 12.17 | 26.78% | 98.01% | 94.05% | 99.95% | 222.19 | 99.60% |
| NT_WES_100 | WES Agilent V8 | NT | 37.54 | 5.56 | 5.67% | 86.62% | 79.06% | 99.83% | 101.13 | 99.30% |
| NT_WES_raw | WES Agilent V8 | NT | 131.76 | 19.50 | 17.54% | 86.61% | 79.06% | 99.83% | 354.92 | 99.60% |

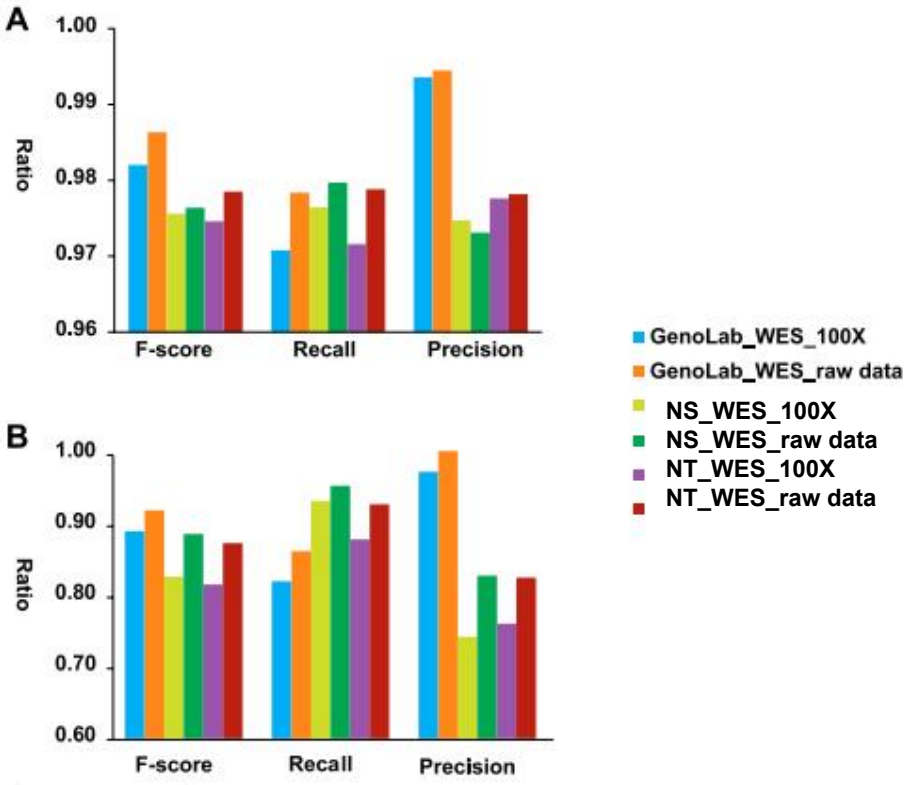
**High performance over other product:**

*Q30>84%* *Alignment rate> 99.95%* *Low duplication rate < 10%*

*Unless otherwise informed, GeneMind sequencing platform and related sequencing reagents are not available in the USA, Canada, Australia, Japan, Singapore, Western Europe and Nordic countries yet.

# Data Comparsion with Other Platforms

(Download Link: https://doi.org/10.1186/s12864-022-08775-3)




- **At 100X, the F-score and Precision in GenoLab M* were higher than NovaSeq or NextSeq, while the Recall in GenoLab M was slightly lower. GenoLab M* showed similar or superior performance to Illumina platforms at the WES application.**

*Unless otherwise informed, GeneMind sequencing platform and related sequencing reagents are not available in the USA, Canada, Australia, Japan, Singapore, Western Europe and Nordic countries yet.

# Scientific Paper 2 (Download Link: https://doi.org/10.3389/fgene.2024.1334075)

- Title: Assessing the impact of sequencing platforms and analytical pipelines on whole-exome sequencing
- Sample type: Tumor reference standard HD832 and standard HG001
- Library type: SureSelect Human All Exon V8 kit and SureSelect Human All Exon V6 kit
- Platform: NX, FASTASeq 300, NS, GenoLab M*, and SURFSeq 5000*

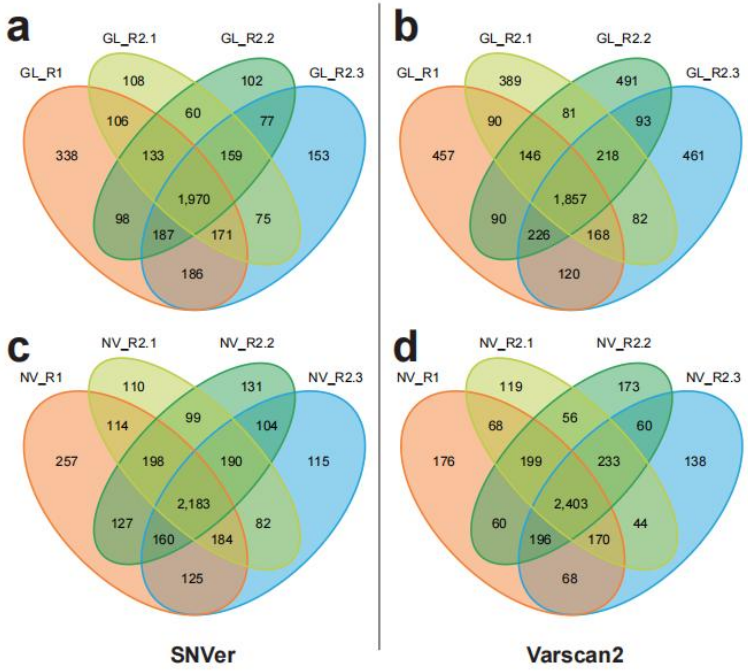
| Sample | Dataset | Reads (M) | Bases (Gb) | Q20 (%) | Q30 (%) | GC (%) | Mapped Rate (%) | Capture efficiency (%) | Duplicate Rate (%) | Average depth (×) | Coverage (≥1×) (%) | Coverage (≥4×) (%) | Coverage (≥10×) (%) | Coverage (≥30×) (%) |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| HD832 | GL_R1 | 31.38 | 8.41 | 96.79 | 92.10 | 47.50 | 99.76 | 58.68 | 8.55 | 128.92 | 99.04 | 98.87 | 98.48 | 96.46 |
| | GL_R2.1 | 29.43 | 8.61 | 97.17 | 91.48 | 45.03 | 99.91 | 51.65 | 8.17 | 123.37 | 98.98 | 98.61 | 97.61 | 92.08 |
| | GL_R2.2 | 31.50 | 9.21 | 97.85 | 92.87 | 45.01 | 99.91 | 51.57 | 8.60 | 131.87 | 98.98 | 98.60 | 97.62 | 92.41 |
| | GL_R2.3 | 31.50 | 9.21 | 97.10 | 91.40 | 46.44 | 99.90 | 51.84 | 7.78 | 132.54 | 99.03 | 98.81 | 98.33 | 95.42 |
| | NX_R1 | 32.95 | 9.75 | 95.77 | 92.26 | 49.24 | 99.93 | 56.14 | 11.29 | 143.04 | 98.92 | 98.67 | 98.23 | 96.40 |
| | NS_R1 | 32.81 | 9.06 | 98.03 | 94.85 | 50.57 | 99.70 | 57.49 | 28.07 | 118.36 | 99.07 | 98.95 | 98.77 | 97.81 |
| | NS_R2.1 | 32.95 | 9.75 | 97.45 | 93.15 | 50.19 | 99.87 | 50.75 | 21.31 | 136.42 | 99.07 | 98.94 | 98.76 | 97.75 |
| | NS_R2.2 | 32.63 | 9.65 | 97.61 | 93.25 | 50.28 | 99.87 | 51.02 | 20.28 | 138.36 | 99.07 | 98.94 | 98.77 | 97.79 |
| | NS_R2.3 | 32.20 | 9.54 | 97.73 | 93.48 | 50.26 | 99.87 | 50.67 | 23.02 | 134.45 | 99.07 | 98.94 | 98.75 | 97.73 |
| HG001 | HG001_FA | 39.14 | 11.10 | 97.32 | 92.45 | 50.65 | 99.94 | 66.52 | 3.45 | 116.68 | 99.63 | 99.41 | 98.72 | 94.01 |
| | HG001_GL | 39.20 | 11.15 | 97.59 | 91.28 | 50.03 | 99.94 | 66.16 | 5.46 | 116.53 | 99.63 | 99.41 | 98.68 | 94.14 |
| | HG001_NS | 39.23 | 11.27 | 96.87 | 92.57 | 52.03 | 99.92 | 66.14 | 20.79 | 117.91 | 99.67 | 99.52 | 99.02 | 94.09 |
| | HG001_SF | 39.05 | 11.28 | 96.30 | 90.89 | 51.32 | 99.99 | 64.61 | 24.53 | 115.28 | 99.69 | 99.44 | 98.81 | 93.81 |
| | HG001_NX | 39.23 | 11.00 | 96.80 | 93.90 | 50.47 | 99.97 | 67.25 | 6.71 | 117.12 | 99.43 | 98.98 | 97.88 | 92.47 |

- **GeneMind NGS platforms showed similar or superior data quality with Illumina platforms at WES application.**

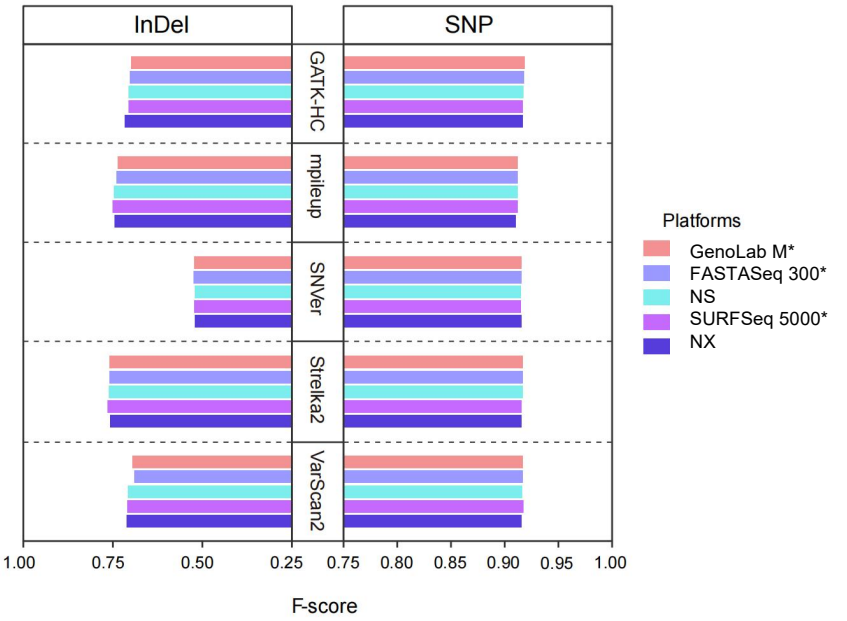
*Unless otherwise informed, GeneMind sequencing platform and related sequencing reagents are not available in the USA, Canada, Australia, Japan, Singapore, Western Europe and Nordic countries yet.

# Data Comparsion with Other Platforms

(Download Link: https://doi.org/10.3389/fgene.2024.1334075)




Venn diagram of variants calling performances in GenoLab M* and NS platforms.




Comparison of F-score for SNPs (Right) and InDels (Left) detected across five callers and five platforms.

- **The results underscore that under the identical analysis pipeline, the disparity in SNP detection among various sequencing platforms is minimal.**

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