

# AIExome® Human Exome Panel V3

## High Coverage and Great Performance Solution for Human Exome Sequencing

### HIGHLIGHTS

#### More Complete Coverage

- Covering about 99.9% CDS region based on multiple databases
- Based on human reference genome GRCh38 and recent patch sequences

#### Effective Probe optimization

- Improve coverage of high GC regions and repeat regions to detect mutations accurately
- Probes for MAF SNP loci as internal reference to avoid sample cross-contamination

#### Reliable Performance

- Excellent enrichment performance with efficient and cost-effective workflow
- Stable data performance across different NGS platforms

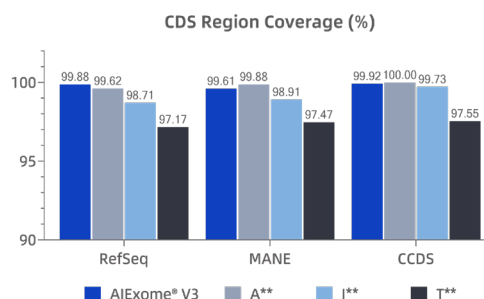
#### Flexible Panel Spike-In

- Support semi-customization of probe addition or enhancement
- Catalog product easily spike-in with good performance

Spanning 34.4 Mb target region of the human genome with efficient probe design, AIExome® Human Exome Panel V3 provides a more complete coverage of protein coding genes for the variant detection which could be widely used in clinical and academic research. The panel delivers outstanding enrichment performance of on-target rate, target coverage and uniformity, providing you a trusted sequencing result for subsequent analysis. With flexible panel customization or spike-in, AIExome® V3 could be expanded or enhanced for regions of interest that cater to your needs.

### SUPERIOR COVERAGE OF CDS REGION

AIExome® Human Exome Panel V3 covers about 34.4 Mb of CDS region (Ref:20200817) to achieve > 99% coverage of protein coding region from of RefSeq, MANE, CCDS databases. Compared to other commercial available panels, AIExome® V3 has a more complete coverage of CDS, which allows more variants detection in a single test.



**Figure 1. CDS region coverage comparison of different databases among AIExome® V3 and other products.** AIExome® V3 is compared with exome panels of A\*\*, I\*\* and T\*\* for the protein coding region coverage based on the recent releases of RefSeq, MANE, CCDS database. Sequencing was finished on NovaSeq 6000 with PE150.

## OPTIMIZED PROBE, EFFECTIVE CAPTURE AND QUALITY CONTROL

### For Hard-To-Capture Regions

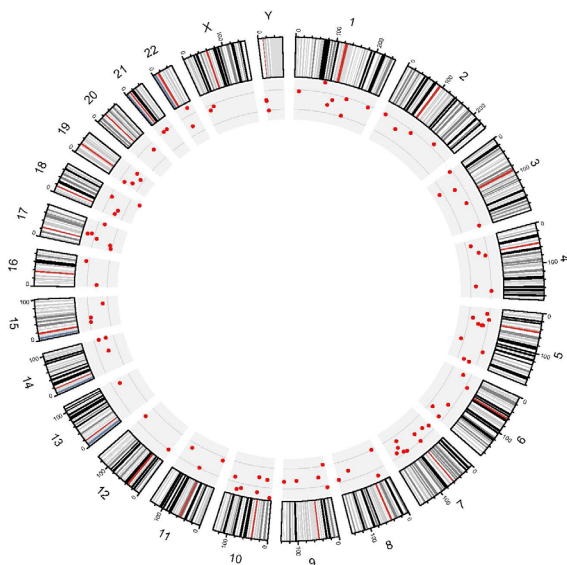
Based on iGeneTech TargetSeq® hybridization capture sequencing technology, the probe for AIExome® V3 has been optimized to have a better coverage of hard-to-capture regions (high GC content region or high repeat regions) to improve the variants detection. For example, AIExome® V3 has a more comprehensive coverage for *TERT* promoter, where the key mutation point chr5:1295113 (GRCh38) and chr5:1295135 (GRCh38) showed 7 times higher of coverage depth than similar product on the market.

Coverage Distribution from IGV	Sequencing Depth (X)		
	Average	chr5:1295113	chr5:1295135
AIExome® V3	127	141	173
A**	119	23	23

**Figure 2. The comparison of coverage on *TERT* promoter between AIExome® V3 and other product.** Compared to the similar product commercially available, AIExome® V3 had more coverage on *TERT* promoter regarding to two specific key points : chr5:1295113 (GRCh38) and chr5:1295135 (GRCh38).

### For Sample Identification

Considering possible specimen alteration or cross-contamination, AIExome® V3 includes additional 100 SNPs of high MAF as the internal reference for quality control. These SNP QC points ensure the accurate identification of samples and allow quick tracing of origins.



**Figure 3. Distribution of 100 SNPs for sample identification.** AIExome® V3 includes SNPs on autosomes and sex chromosomes for sample identification and gender assurance respectively.

## For Patch Sequences

AIExome® Human Exome Panel V3 also includes the probes for recent fixed and novel patch sequences of human reference genome GRCh38, which could effectively support the haplotype analysis and avoid missed mutation detection due to individual difference.

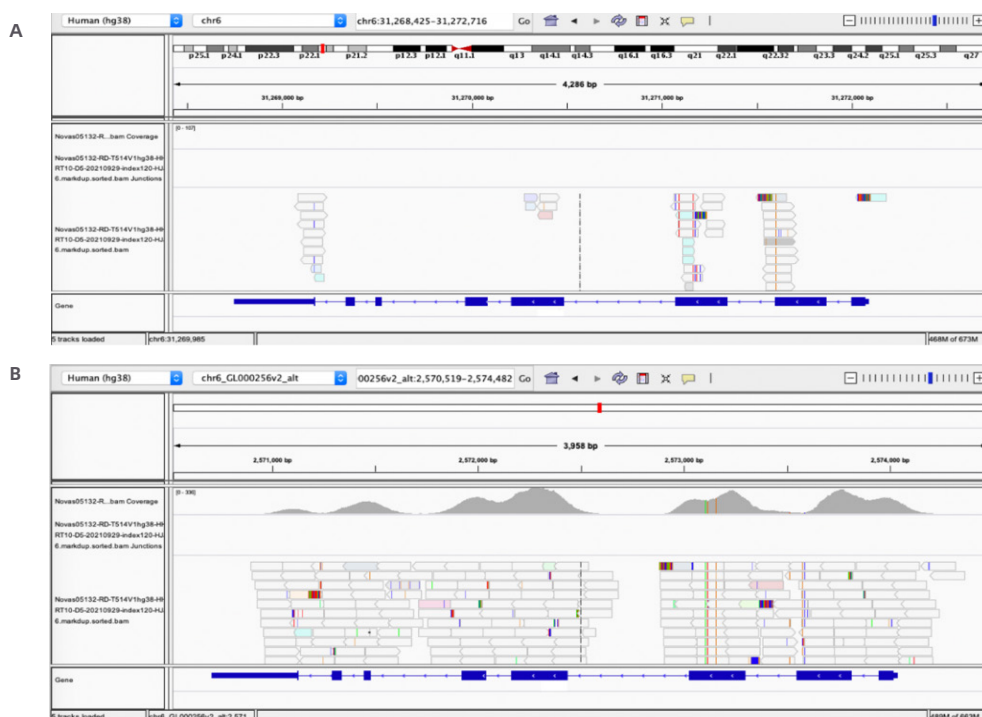


Figure 4. Improvement of coverage by additional probes for patch sequences. A. Coverage of real sample data on reference genome. B. Coverage of real sample data on patch sequence.

## EXCELLENT, STABLE AND COST-EFFECTIVE PERFORMANCE

### Better Performance Compared to Others

Together with iGeneTech simple TargetSeq One® v2.0 target enrichment workflow, AIExome® V3 had a better target enrichment performance of CDS coverage, on-target capture rate and uniformity compared to market equivalent panel. It also enables you to sequence with less raw data but higher depth per sample for cost-saving.

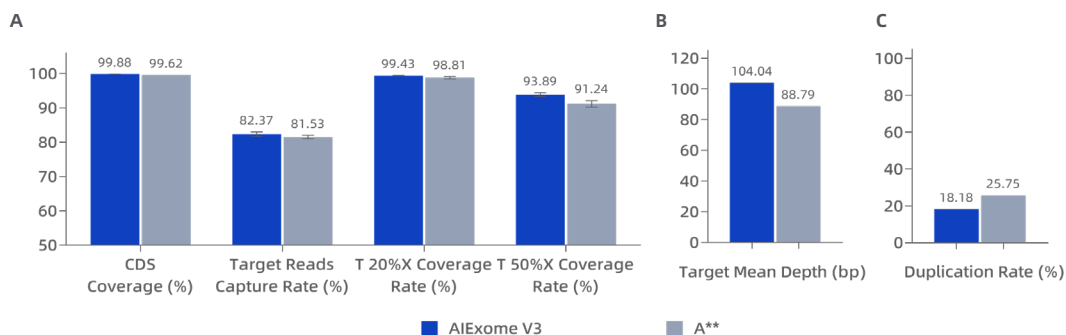
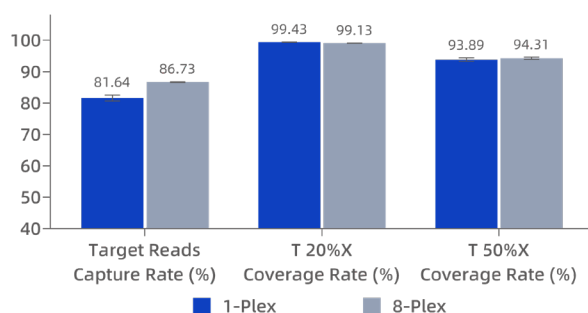


Figure 5. Data performance comparison between AIExome® V3 and other product. A. Target enrichment based on 200 ng DNA of standard reference (NA12878) with triplicates. Sequencing was performed on NovaSeq 6000 with PE150. B-C. Randomly select 8 Gb raw data for analysis of coverage depth and duplication rate. AIExome® V3 had a higher depth (~100X) and low duplication rate (~18%) than the compared commercial panel.

## Multiplex Hybridization Supported

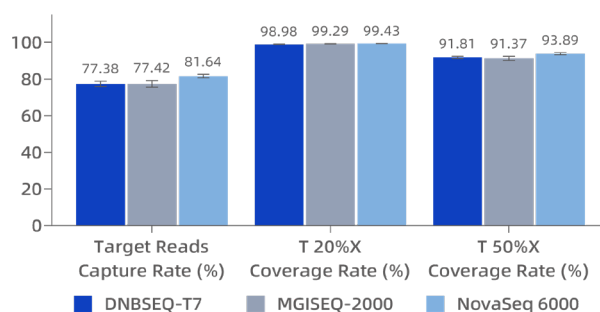
AIExome® V3 supports multiplex hybridization to capture more samples per reaction with stable and excellent performance of target coverage and uniformity.



**Figure 6. Data performance comparison between singleplex and multiplex-hybridization.** A 8-plex hybridization strategy showed over 80% target reads capture rate, 99% and more of target 20%X coverage and 94% of target 50%X coverage, which is as good as singleplex-hybridization performance. Randomly select 8 Gb raw data for analysis.

## Multiple Platforms Supported

AIExome® V3 is excellent and consistent in performance across NGS platforms. Sequencing data from DNBSEQ-T7, MGISEQ-2000 and NovaSeq 6000 all showed good performance with over 100X effective sequencing depth and 99% of target 20% X coverage.



**Figure 7. Data performance comparison between different sequencing platforms.** Enriched library was prepared with gDNA sample by using IGT® Enzyme Plus Library Prep Kit, AIExome® V3 probe and TargetSeq One® Hyb & Wash Kit v2.0. Sequencing were performed on DNBSEQ-T7, MGISEQ-2000 and NovaSeq 6000 respectively and analysis were based on randomly selected 8 Gb raw data.

## CUSTOMIZE UPON YOUR IDEA

With the increasing requirements and research purposes from genetic disorders, tumor, immunology, genotyping and other directions, our professional team supports semi-customized solution for human exome panel design. Based on our AIExome® Human Exome Panel V3, you can add additional probes for your targeted regions which are not included in our panel, or enhance the probe density for regions of interest.



### Diversiform Target Selection

Fusion, MSI, SNP backbone, mtDNA...



### Trusted Quality Assurance

Strict data QC using standard reference after probe synthesis



### Fast and Cost-Effective Panel Spike-In

Affordable cost for more coverage with short TAT



### Flexible Upgrades

Updated upon request for supplementary targeted region

According to some common research fields and purposes, we have developed mature catalog products based on AIExome® V3 with small panels spike-in for special cases, which also could be used as the base for panel customization.

### AIExome® Human Exome Panel V3-Inherit

AIExome® V3-Inherit, developed based on AIExome® V3, provides a more complete coverage especially for the detection of genetic disorders. AIExome® V3-Inherit covers clinically relevant non-coding pathogenic and likely pathogenic variants in ClinVar database, full length of human mitochondria genome and whole genome SNP backbone for CNV analysis to ensure a comprehensive understanding of rare and inherited diseases.

### AIExome® Human Exome Panel V3-Tumor

In spite of the CDS region from AIExome® V3, AIExome® V3-Tumor combines TargetSeq® Pan-cancer panel and HLA panel for the focus on tumor analysis. It offers a comprehensive detection of tumor related variations including SNV, InDel, Fusion and CNV, as well as some tumor-related biomarkers like MSI, TMB and HLA typing.

#### AIExome® V3-Inherit

#### AIExome® V3

#### AIExome® V3-Tumor

- ClinVar Database for variant detection of inherited diseases
- SNP Backbone for CNV detection
- Mitochondrial Genome for analysis of mitochondrial mutation and heterogeneity

- Probe enhancement of 641 tumor-related genes
- Additional probes for 38 fusion regions, 15 MSI sites, and 6 HLA genes

## ALL-IN-ONE SOLUTION

As one of the star products applying iGeneTech TargetSeq® hybridization capture sequencing technology, AIXome® Human Exome Panel V3 is compatible with our simple and cost-effective TargetSeq One® hybridization capture workflow v2.0. To enable an ultimate convenient experience, we also offer library preparation kits, adapters & indexed primers, universal blockers as well as hybridization & wash kits which are suitable for a range of capture and sequencing strategies. With this all-in-one solution for exome capture sequencing, we are confident to deliver reliable and outstanding performance that meet your requirements.

## ORDERING INFORMATION

Type	Product	Cat #
Target Probe	AIXome® Human Exome Panel V3, 16/96 rxn	PT1008091/PT1008092
	AIXome® Human Exome Panel V3 - Inherit, 16/96 rxn	PT1008101/PT1008102
	AIXome® Human Exome Panel V3 - Tumor, 16/96 rxn	PT1009181/PT1009182
Library Preparation	IGT® Fast Library Prep Kit v2.0, 16/96 rxn	C10021/C10022
	IGT® Enzyme Plus Library Prep Kit V3, 16/96/960 rxn	C11111/C11112/C11113
Adapter & Indexed Primer	IGT® Adapter & UDI Primer (for Illumina, plate), 96*1 rxn	C10042/C10052/C10062/C10072
	IGT® Adapter & UDI Primer (for MGI, plate), 96*1 rxn	C10182/C10192/C10202/C10212
	IGT® UMI Adapter & UDI Primer (for Illumina, plate), 96*1 rxn	C10092/C10102/C10112/C10122
	IGT® UMI Adapter & UDI Primer (for MGI, plate), 96*1 rxn	C10232/C10242/C10252/C10262
Hybridization & Wash	TargetSeq One® Hyb & Wash Kit v2.0 (for Illumina), 16/96 rxn	C10331/C10332
	TargetSeq One® Hyb & Wash Kit v2.0 (for MGI DI), 16/96 rxn	C10351/C10352
Universal Blocking Oligo	TargetSeq® Universal Blocking Oligo (for Illumina/MGI), 16/96 rxn	C80491/C80492/C80521/C80522
	TargetSeq® Eco Universal Blocking Oligo (for Illumina/MGI), 16/96 rxn	C80501/C80502/C80531/C80532
Magnetic Beads for Capture	TargetSeq® Cap Beads & Nuclease-Free Water	C10421/C10422/C10423
Magnetic Beads for Purification	IGT® Pure Beads	C80661/C80662

FOR MORE PRODUCT AND ORDERING INFORMATION, VISIT <https://www.igenetech.com/>

iGeneTech Bioscience Co., Ltd.

[www.igenetech.com](http://www.igenetech.com) / [sales\\_int@igenetech.com](mailto:sales_int@igenetech.com)

Add: Level 3, Block A, Building 9, No. 8 Shengmingyuan Road, Changping District, Beijing 102206, China

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