Novogene

Contents

Whole Exome Sequencing

Harnessing the Full Power of Whole Exome Sequencing

Novogene's comprehensive human Whole Exome Sequencing (WES) focuses on protein-coding genes to identify etiological variants in diseases such as hereditary genetic disorder and cancer. This service provides a cost-effective alternative to whole genome sequencing with a high-quality, affordable and convenient solution.

▸ Highlights

- **State-of-the-art exome capture**
  Agilent SureSelect Human All Exome V6 Kit and Agilent SureSelect $^{XT\ HS}$ Reagent Kit
- **Unsurpassed data quality**
  Guarantee of a Q30 score ⩾ 80%, exceeding Illumina's official guarantee of ⩾ 75%
- **Accurate variant calling**
  Read lengths up to 150 bp
- **Extraordinary informatics expertise**
  Cutting-edge bioinformatics pipeline with internationally recognized best-in-class software, providing customers with "publication-ready data"

Test Specifications

Workflow

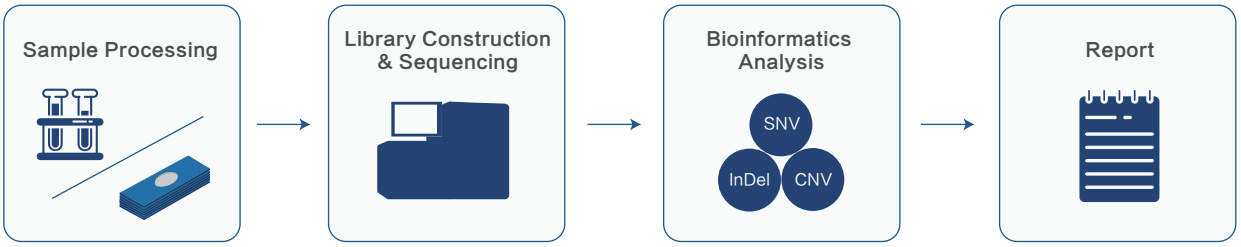

Figure 1. Whole Exome Sequencing workflow

- **Sample Requirement**

Sample Type	Sample Requirement
Whole blood	⩾2 mL
FFPE tissue	Approximately ten 4− μ m sections, each with tissue area ⩾ 25 mm^2 and tumor content ⩾ 20%
Extracted DNA	Total DNA ⩾ 500 ng; DNA concentration (quantified by Qubit) ⩾ 20 ng/ μ L, total volume ⩾ 10 μ L; Purity: OD260/280 = 1.8−2.0 without degradation or RNA contamination

- **Exome Capture**

Agilent SureSelect Human All Exon V6 Kit

Agilent SureSelect $^{XT\ HS}$ Reagent Kit

- **Sequencing Strategy**

HiSeq X Ten or NovaSeq 6000, PE150

- **Recommended Sequencing Depth**

Tumor sample: effective sequencing depth above 100 ×

Normal sample: effective sequencing depth above 50 ×

- **Data Quality Guarantee**

Guarantee of ⩾ 80% of bases having a sequencing quality score ⩾ Q30, exceeding Illumina's official guarantee of ⩾ 75%

- **Turnaround Time**

Raw data delivery within 10 calendar days for blood or DNA samples, and 12 calendar days for FFPE tissue samples

Additional 7 calendar days for standard data analysis

Bioinformatics Analysis

Novogene's bioinformatics analysis workflow includes data quality control, alignment with human reference genome, SNP/InDel/CNV calling, statistics and annotation. This service utilizes internationally recognized bioinformatics analysis tools (BWA, SAMtools, GATK, etc.) with various annotation databases (ExAC, COSMIC, 1000 Genomes, etc.) to provide a robust and comprehensive analysis of human exomes.

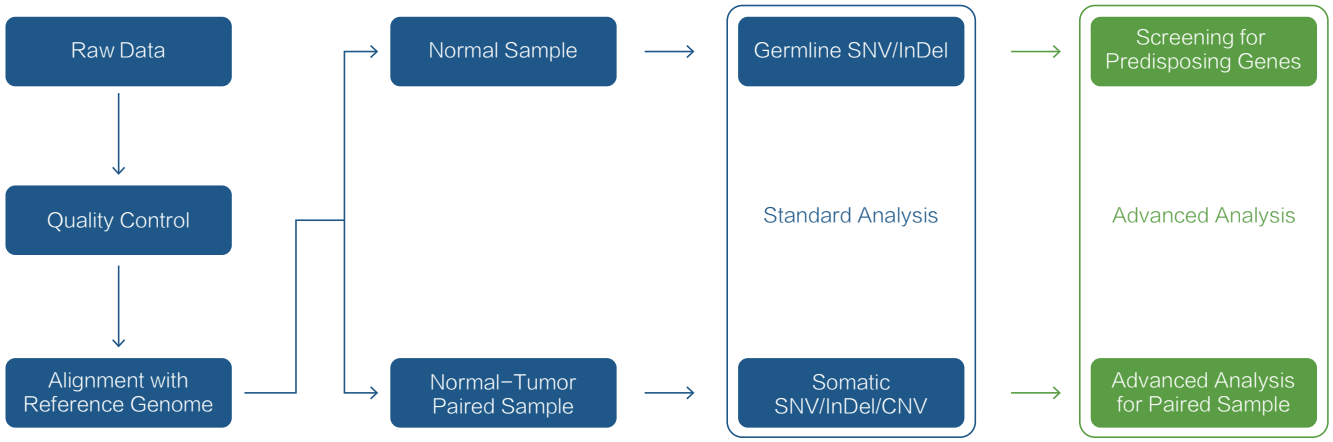

Figure 2. Workflow of standard and advanced bioinformatics analyses

Standard Analysis

- Sequencing quality and alignment
- Variants detection and annotation
- Somatic mutation calling (for normal–tumor paired samples)

Advanced Analysis

Normal Samples

- Screening for predisposing genes

Normal–Tumor Paired Samples

- Analysis of mutation spectrum and mutation signatures
- Analysis of tumor significantly mutated genes (SMG) and pathway enrichment
- Screening for known driver genes
- Display of genomic variants with Circos plots
- Analysis of tumor heterogeneity and clonal structure
- Mutation relation test (MRT) of significantly mutated genes (SMG)
- Prediction of driver genes
- Analysis of copy number variation (CNV) distribution & recurrence
- Analysis of mutation distribution
- Analysis of tumor purity & ploidy
- Analysis of loss of homozygosity (LOH)

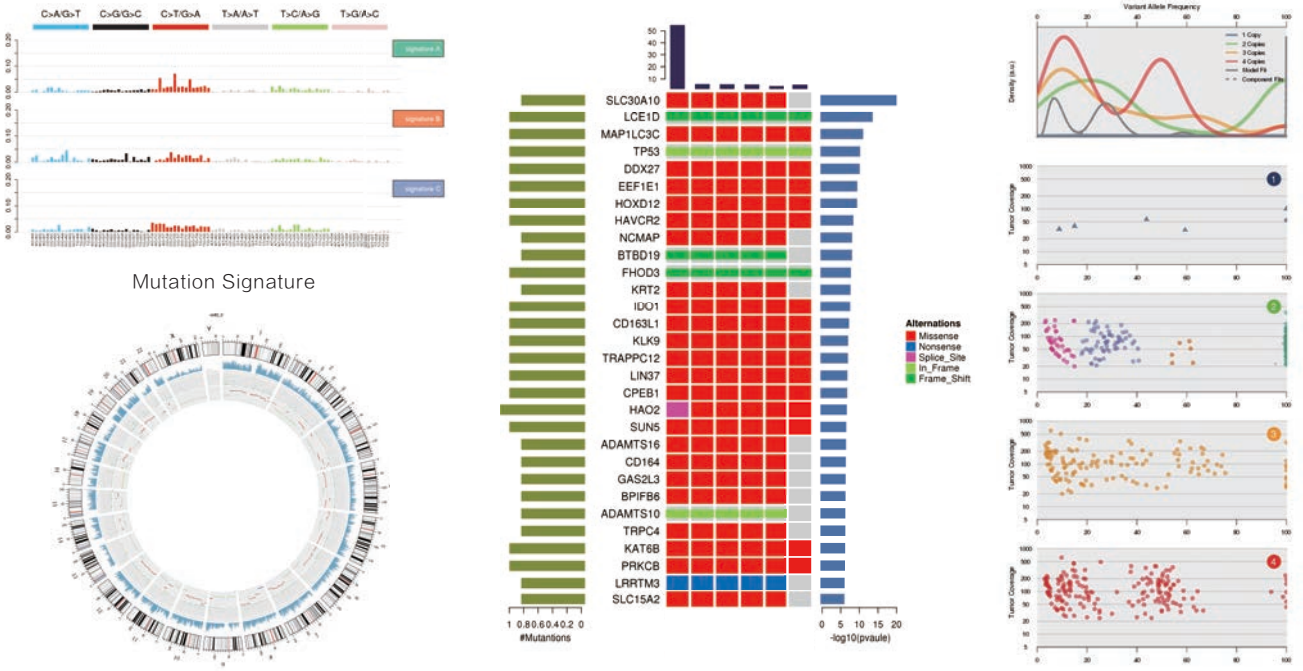

Figure 3. Examples of advanced bioinformatics analyses

Performance Validation

Unsurpassed Data Quality

Our whole exome sequencing workflow provides unsurpassed data quality with high %Q30, high mapping and capture efficiency, and low duplicate rate. This assay also performs well with low amounts of DNA from clinical samples.

Table 1. In-house data quality metrics of this WES assay (100 ng DNA input)

Data Quality Metrics	Tumor Samples	Normal Samples
Clean Data (G)	9.88 ± 2.24	6.22 ± 1.02
Effective Rate (%)	91.08 ± 2.73	96.99 ± 1.60
Q30 (%)	92.98 ± 0.32	92.11 ± 1.43
Duplicate Rate (%)	19.57 ± 3.81	21.21 ± 3.16
Mapping Rate (%)	99.53 ± 0.96	99.89 ± 0.05
Capture Rate (%)	62.06 ± 4.82	57.85 ± 6.17
Average Sequencing Depth on Target	105.44 ± 12.63	58.67 ± 8.15

Extensive Experience

We have analyzed hundreds of samples with this WES assay. Table 2. provides an example of the data quality metrics from those analyses.

Table 2. Examples of data quality metrics from tumor samples analyzed with this WES assay.

Sample Name	Clean Data (G)	Effective Rate (%)	Q30 (%)	GC (%)	Duplicate Rate (%)	Mapping Rate (%)	Capture Rate (%)	Average Sequencing Depth on Target
001	40.83	99.40	94.77	50.52	26.42	99.95	68.05	458
002	42.31	99.52	94.19	49.67	22.00	99.95	63.15	440
003	32.87	99.51	94.28	49.59	28.65	99.95	63.61	344
004	36.75	99.49	94.12	49.79	27.55	99.95	63.44	384
005	33.02	99.75	92.64	49.69	28.66	99.91	61.95	337
006	40.52	98.13	94.76	50.30	26.84	99.96	70.41	470
007	34.73	99.41	93.37	49.75	27.94	99.95	64.56	369
008	27.35	99.41	94.31	49.34	26.22	99.96	63.79	287
009	39.31	99.61	93.33	49.64	23.40	99.95	63.99	414
010	35.18	98.92	94.37	49.97	28.68	99.94	67.87	393
011	30.38	99.33	93.88	49.75	28.51	99.94	66.50	333
012	36.45	99.33	94.45	49.82	28.57	99.96	66.22	398
013	30.85	99.34	94.02	49.59	25.39	99.94	65.64	334
014	37.92	99.20	94.52	50.03	27.57	99.96	67.20	420
015	32.45	99.51	94.00	49.59	25.71	99.94	64.45	344
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Find out more at :

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