DNBSEQ™ SERVICE OVERVIEW
Whole Exome Sequencing

Service Description

For many applications, Whole Exome Sequencing is gaining popularity as a viable and cost-effective alternative for Whole Genome Sequencing. BGI Genomics has performed professional exome sequencing services for many years at several locations around the world, to support human and animal (rodents and monkeys) research and to benefit small and large-scale clinical trials and pharmaceutical drug development projects.

Besides raw sequencing data output, BGI Genomics offers standard and custom bioinformatics services to suit your specific research needs.

Sequencing Service Specification

BGI Genomics Human Exome Sequencing Service are performed with the DNBSEQ sequencing technology, featuring cPAS and DNA Nanoballs(DNB™) for superior data quality.

Sample Preparation and Services

- Agilent Sureselect exome kit for library construction and enrichment, 100/150 bp paired-end sequencing options available
- Clean data and advanced bioinformatics analysis are available in standard file formats
- Standard and custom bioinformatics data analysis
- Available data storage and bioinformatics applications

Sequencing Quality Standard

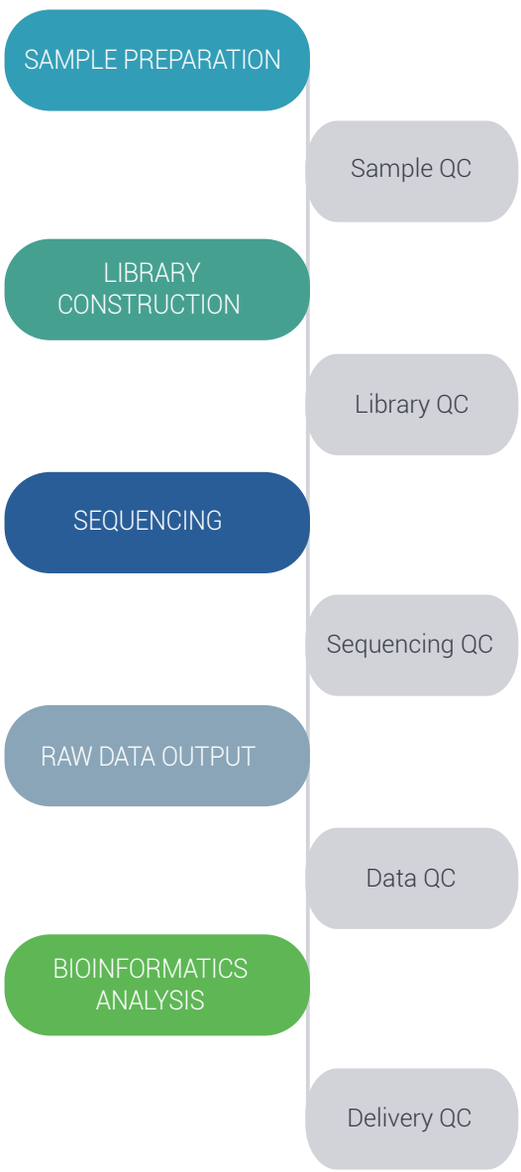
- Guaranteed ≥85% of bases with quality score of ≥Q30
- Standard sequencing coverage ≥50X; ≥100X is recommended for cancer samples

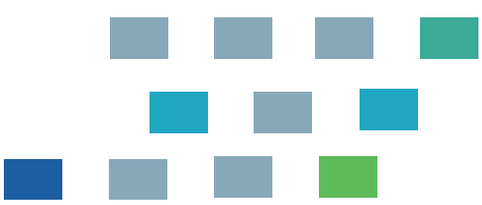
Turn Around Time

- Typical 18 days after sample acceptance for data delivery
- Rapid delivery service available

Project Workflow

We care for your samples from the start through to the result reporting. Highly experienced laboratory professionals follow strict quality procedures to ensure the integrity of your results.





Data Analysis

Besides clean data output, BGI Genomics offers a range of standard and customized bioinformatics pipelines for your whole exome sequencing project.

Reports and output data files are delivered in industry standard file formats: BAM, VCF, .xls, .png

Standard Bioinformatics Analysis

- Data filtering (removing adaptors,contamination, and low-quality reads from raw reads)
- Align reads to the human reference genome.
- SNP calling
- SNP annotation
- SNP validation and comparison
- Functionality and conservation prediction of SNPs
- Statistics of SNPs in each functional element
- InDel calling
- InDel annotation
- InDel validation and comparasion
- Statistics of InDel in each functional element

Note: The annotation databases above are only suitable for human samples.

Human-CancerAn Alysis

- Data filtering (removing adaptors,contamination, and low-quality reads from raw reads)
- Align reads to the human reference genome;
- Germline mutation calling;
- SNP detection;
- InDel detection;
- Somatic SNV detection;
- Somatic InDel detection;
- Somatic CNV detection;
- Tumor purity and ploidy analysis ;
- Verification of homology of paired samples;
- Susceptibility gene screening;
- Drug targeted annotation;
- Driving gene prediction;
- Identifying significant mutated genes;
- Deciphering mutational signature;
- CN-neutral loss of heterozy gosity;
- Hyper-mutated sample classification;
- Clone analysis.

Human-Mendelian Disorders Analysis

- Data filtering
- Align reads to the human reference genome using BWA software
- Variants calling
- VEP annotation
- Public group AF annotation
- Harmful or conservative prediction tools
- Signaling pathway annotation
- OMIM annotation
- Normal tissue protein expression annotation
- Data screening and interpretation
  1 Screening according to ACMG genetic variation classification criteria and guidelines
  2 Screening by threshold
  3 *De Novo* mutation screening
  4 Analysis of family co-separation
  5 RoH analysis

Customized Analysis

Further customization of Bioinformatics analysis to suit your unique project is available:
Please contact your BGI Genomics technical representative

Sample Requirements

We can process your gDNA, Blood, Cell line, Fresh frozen tissue samples from a variety of species, with the following general requirements:

Sample type	Mass	Concentration	Integrity (AGE)	Sample Purity
Genomic DNA	≥500 ng	≥12.5 ng/μL	The band shown on gel electrophoresis has little degradation, or of fragment size greater than 20 kb.	No contamination with RNA, protein or salt ions; colorless and transparent; non-sticky.

Tissue Type	Requirements
Fresh Cell Culture (Number of Cell or Net Weight)	≥5×10^6 cells
Fresh Animal Tissue (Net Weight)	≥50 mg
Whole Blood (Mammals)	≥0.5 mL
FFPE	≥ 10 slides, unstained, 100 mm^2 area, 5 ~ 10 μm thickness, tumor content≥70%

DNBSEQ Sequencing Technology

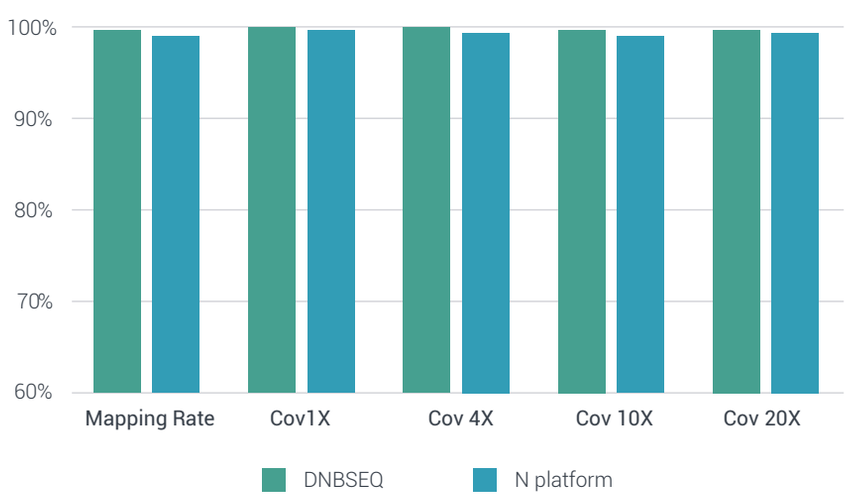
DNBSEQ system is an industry leading high-throughput sequencing solution, powered by combinatorial Probe-Anchor Synthesis (cPAS) and improved DNA Nanoballs (DNB™) technology. The cPAS chemistry works by incorporating a fluorescent probe to a DNA anchor on the DNB™, followed by high-resolution digital imaging. This combination of linear amplification and DNB™ technology reduces the error rate while enhancing the signal. In addition, the size of the DNB™ is controlled in such a way that only one DNB™ is bound per active site. This patterned array technology not only provides sequencing accuracy, but it also increases the chip utilization and sample density.

Sequencing Technology References

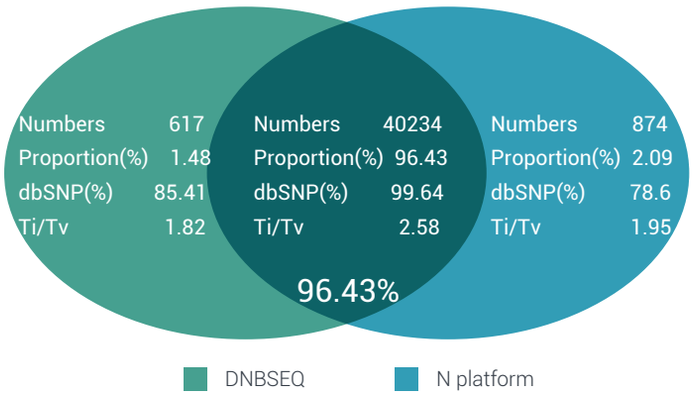
Drmanac R, Sparks AB, Callow MJ,Halpern AL, Burns NL, Kermani BG, Carnevali P et al. Human genome sequencing using unchained base reads on selfassembling DNA nanoarrays. *Science*. 2010;327(5961):78–81.

Data Performance

Following is an example of typical DNBSEQ data output for a 100X WES project with standard sample NA12878, compared with data from N platform.

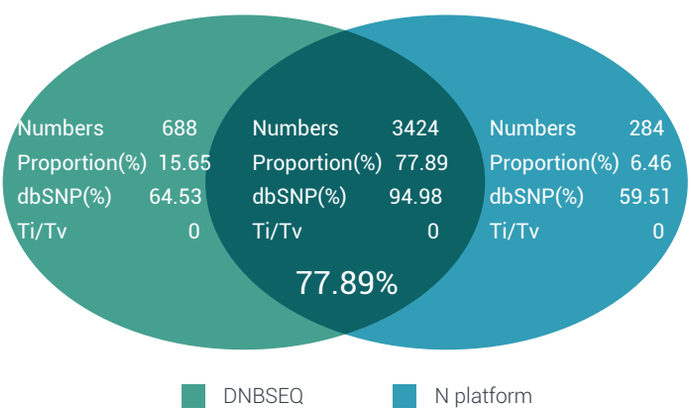


Bar-Graph showing the mapping rate and sequencing coverage of the samples using DNBSEQ and N platform of 100X WES.



SNP calling performance from the NA12878 standard sample demonstrates good concordance between platforms

SNP ANALYSIS	DNBSEQ	N platform
PRECISION	76.95%	76.44%
SENSITIVITY	95.53%	95.50%



InDel calling performance from the NA12878 standard sample demonstrates good concordance between platforms.

InDel ANALYSIS	DNBSEQ	N platform
PRECISION	74.04%	79.77%
SENSITIVITY	87.60%	84.93%

*Full demonstration data reports are available through your BGI Genomics account representative.

Request for Information or Quotation

Contact a BGI Genomics representative to discuss how we can meet your specific needs or for expert advice on experiment design, from sample to bioinformatics.

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