DNBSEQ™ SERVICE OVERVIEW
Human Whole Genome Sequencing

Service Description

Whole Genome Sequencing (WGS) determines the complete human genome sequence at one time and provides the most comprehensive collection of an individual's genetic variation based on the human reference genome.
WGS can be applied to human genetics and evolution studies to detect genome-wide genetic variations, pathogenic and susceptibility genes, and to enable genetic diversity and evolution analysis. It can also be applied in translational research to provide information on cancer and other disease-associated mutations and is one of the most important tools for precision medicine.
We offer a comprehensive range of WGS services for many sample types and coverage levels.

Sequencing Service Specification

Our Human Whole Genome Sequencing services are performed with DNBSEQ™ sequencing technology, featuring DNA Nanoballs, linear Rolling Circle Replication and cPAS technology for superior data quality.

- PCR and PCR-free library methods are available
- 100bp and 150bp Paired end sequencing options
- Choice of sequencing depth: standard (~30x), deep (~60x)and low pass (less than 10x)
- Raw data, standard and customized bioinformatics analysis available
- Available data storage and bioinformatics applications

Sequencing Quality Standard

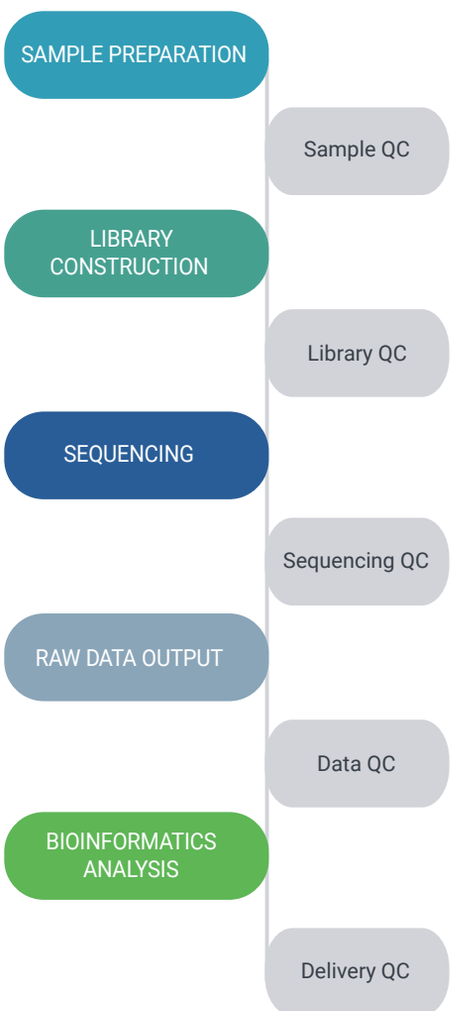
- Guaranteed ≥80% of bases with quality score of ≥Q30

Turnaround Time

- Standard Turnaround: typical 18 working days from sample QC acceptance to filtered data availability
- Rapid Turnaround: 10 working days from sample QC acceptance to filtered data availability
- Expedited services are available for all WGS sequencing options, contact our specialist for details

Project Workflow

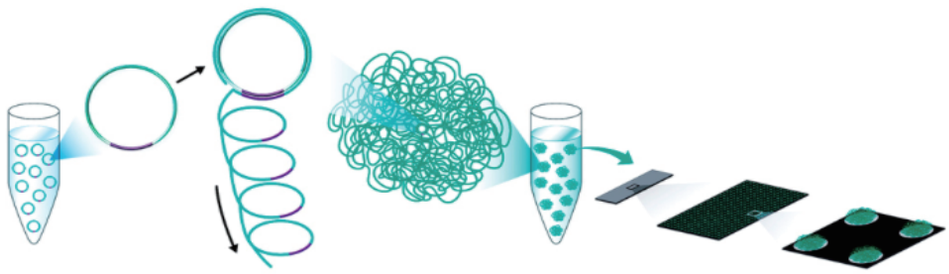
We care for your samples from the start through to the result reporting. Highly experienced laboratory professionals follow strict quality procedures to ensure the integrity of your results.



Quality Data | Fast TAT | Cost Effective

DNBSEQ™ Sequencing Technology

DNBSEQ™ is an innovative high-throughput sequencing solution. The system is powered by combinatorial Probe-Anchor Synthesis (cPAS), linear isothermal Rolling-Circle Replication and DNA Nanoballs (DNB™) echnology, followed by high-resolution digital imaging.



The combination of linear amplification and DNB technology reduces the error rate while enhancing the signal. The size of the DNB is controlled in such a way that only one DNB is bound per active site on the flow cell. This densely patterned array technology provides optimal sequencing accuracy and increases flow cell utilization.

Data Analysis

In addition to clean data output, we offer a range of standard and customized bioinformatics pipelines for your human WGS project.
Reports and output data files are delivered in industry standard FASTQ, BAM. Excel formats with publication-ready tables and figures.

STANDARD BIOINFORMATICS ANALYSIS

- Filtering
- Alignment
- SNP calling and annotation
- SNP validation and comparison
- SNP functionality and conservation prediction
- SNP Statistics per functional element
- InDel calling and annotation
- InDel validation and comparison
- InDel statistics per functional element
- CNV calling and annotation
- SV calling and annotation

ADVANCD ANALYSIS

Advance Analysis	Contents
Cancer Analysis Paired samples are required (Normal tissue and Tumor tissue from the same person)	Somatic Mutation Calling 1 Somatic SNV calling and annotation; 2 Somatic InDel calling and annotation; 3 Somatic SV calling and annotation; 4 Somatic CNV calling and annotation; Cancer Classical Analysis; 1 Purity and Ploidy; 2 Homology Test; 3 Cancer Susceptibility Genes; 4 Drug Targeted Annotation; 5 Prediction of Driver Genes; 6 Identifying Significant Mutated Genes; 7 Deciphering Mutational Signature; 8 CN-neutral Loss of Heterozygosity; 9 Hyper-mutated Sample Classification; 10 Clone Analysis;

ADVANCD ANALYSIS

Advance Analysis	Contents
Mendelian Disorders Analysis	Public group AF annotation; Scoring 12 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 (optional, Parental samples are required) 4 Analysis of family co-separation 5 RoH analysis (optional, pedigree is consanguineous union)

CUSTOM ANALYSIS

Further customization of bioinformatics analysis to suit your unique project is available:
Please contact our technical representative.

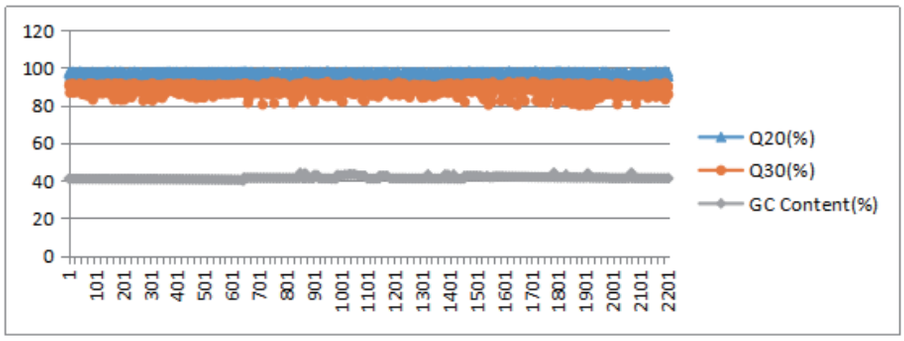
Sample Requirements

We can process your gDNA, saliva, blood, fresh frozen tissue, cell pellets and FFPE samples, with the following general requirements:

DNA Sample	Library type	Mass	Concentration	Integrity (AGE)	Sample Purity
Regular Samples	PCR	≥200ng (Recommend ≥400ng)	≥8ng/μL	The band shown on gel electrophoresis has little degradation, or of fragment size greater than 20kb.	No contamination with RNA, protein or salt ions; colorless and transparent; non-sticky
	PCR-free	≥1μg (Recommend ≥2μg)	≥12.5ng/μL		
Low Input/ FFPE Samples	PCR	≥50ng	≥2.5ng/μl	Fragment size greater than 500bp.	

Stable and High-Quality Data Performance

2,206 Samples were randomly selected from over 20,000 samples that were sequenced over a period of 6 months. The data output and data quality remained stable over that period. The average Q20 and Q30 scores were 96.78% and 88.81% respectively. The average GC content was 41.25% without obvious base bias.

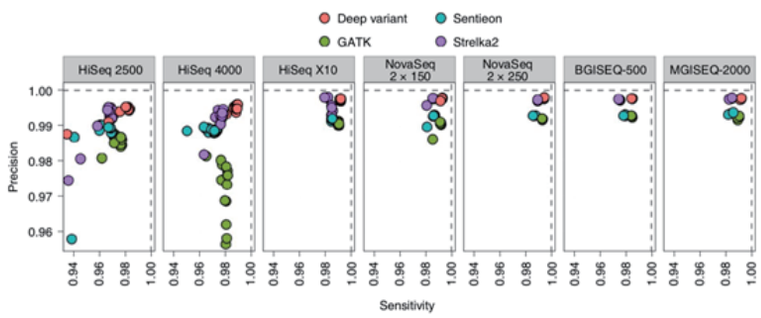


Stable Data quality scores and GC content from our DNBSEQ™ production line

Superior SNP/InDel Detection

Here the Association of Biomolecular Resource Facilities (ABRF) Next-Generation Sequencing Study[1] benchmarks the performance of a set of sequencing instruments on human reference DNA samples. It is further confirmed that the DNBSEQ™ platform has greater advantages in error rate and SNP/InDel detection.

- Among short-read instruments, the DNBSEQ™ platform provided the lowest sequencing error rates.
- For SNP/InDel, the DNBSEQ™ platform had the greatest sensitivity and precision.



Common germline haplotype variant callers were compared for each sequencing platform

References

[1].Foox, J., Tighe, S.W., Nicolet, C.M. et al. Performance assessment of DNA sequencing platforms in the ABRF Next-Generation Sequencing Study. Nat Biotechnol 39, 1129–1140 (2021).

To learn more

If you have any questions or would like to discuss how our services can help you with your research, please don't hesitate to contact us at P_contact@innomics.com. We look forward to hearing from you!

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