NIFTY®

Non-Invasive Prenatal Testing

BGI

Local Laboratory Solutions

Trusted by 15+ million pregnancy women worldwide

Introduction

Founded in 1999, BGI is one of the world's leading life science and genomics organizations. BGI's mission is to use genomics to benefit mankind and to be a leader in the era of life sciences.

BGI follows a genomics development model of "research, production and application". With businesses in more than 100 countries and regions around the world, BGI has established cooperation and partnerships with thousands of different organizations across multi-disciplinary research areas including medical health, resource conservation and judicial services.

BGI is committed to applying its genetic and technological achievements to real world settings in order to realize the dream of trans-omics for a better life.

BGI Genomics, a subsidiary of BGI, is one of the world's leading genomics test and research service providers, providing genomics test and research services for medical institutions, research institutions, enterprises and institutions through genetic testing and other means.

BGI Genomics is committed to helping and accelerating scientific innovation, strengthening the prevention and control of cancer and other genetic diseases, improving diagnostic tools for the rapid diagnosis of infectious disease and helping the development of precision medicine.

Mission - Trans-omics for a better life.

Vision - To be a world leader in the age of life sciences.

Core Values - Curiosity, Application of Knowledge, Working for the Betterment of Mankind.

NIFTY® Introduction

NIFTY® is a world-leading non-invasive prenatal test (NIPT) product independently developed by BGI Genomics. By collecting >5 mL of maternal peripheral blood, extracting cell-free DNA, and using low-depth whole-genome sequencing technology, combined with bioinformatics analysis, the risk of fetal chromosomal abnormalities was determined.

It is a safe and accurate method for detecting T21, T18 and T13, and can expand detection for other autosomal aneuploidies, sex chromosome aneuploidies, and pathogenic chromosomal deletions/duplications (CNVs).

Items		NIFTY®	NIFTY® Pro
Screening options	T21, T18, T13	√	√
	Sex chromosome aneuploidies	Optional	√
	Rare autosomal aneuploidies	Optional	√
	Micro del/dup (CNV)	N/A	92 (22q11.2 deletion included)
	Y Chromosome	Optional	Optional
Reads		~6Mb	~25Mb
Sample volume		> 5mL peripheral blood or 2mL plasma	
TAT-Local Laboratory[1]		1-3 days	

The NIFTY® test is available for

Singleton Twins IVF

Safe
Only maternal peripheral blood is needed, no risk to mother or fetus.

Early
Screen as early as the 10th week of pregnancy.

Accurate
Over 99% sensitivity for trisomy 21, 18 and 13.

Fast
Whole experiment process takes about 14 hours at minimum[1].

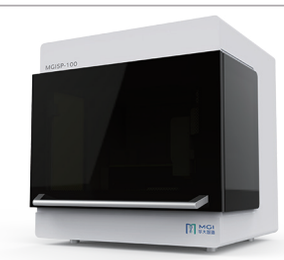
Comprehensive
NIFTY® Pro detects over 100 genetic conditions.

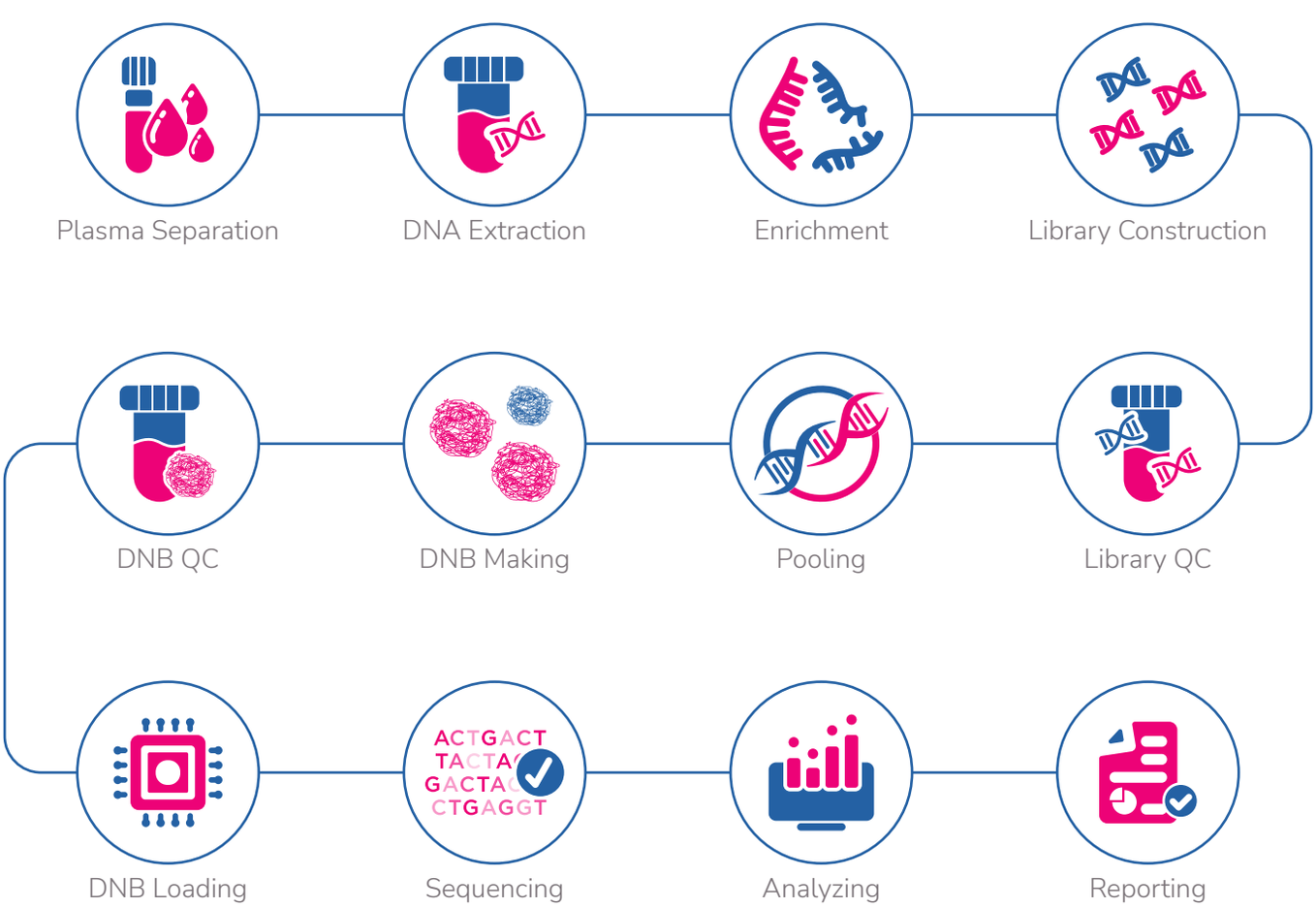
Trusted
Over 13,000,000 samples processed worldwide.

[1] Factors such as instruments, personnel operation, and scheduling can cause time differences.

Laboratory Workflow

- Completely local testing and local analysis.
- Only 1 laboratory technician is needed.
- Able to complete the test within 14 hours.

Sampling	Sample preparation	Sequencing	Bio-info & report
			
Special blood collection tube	Whole process automation	World's leading sequencer	Automated analysis and report



Solution for Localization

- Provide overall laboratory solutions.
- Provide multiple platforms for selection.
- Instruments, reagents, and software all obtained CE certification.

BGIzation of Precision Medicine Laboratory

Compliance
Instruments/reagents/
software/sampling kit
all obtained CE certification

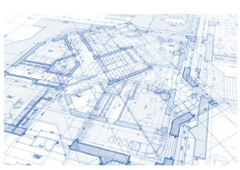
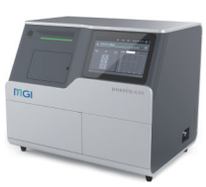
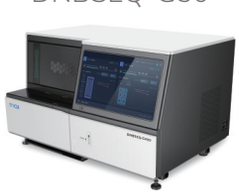
Scalability
Large throughput,
strong scalability,
meet the needs of
large-scale screening

Standardization
Laboratory construction/
operation specifications/
quality management

Intelligentization
Whole-process
intelligentization

Informatization
Whole-process informatization

Automation
Whole-process automation

Automation Platform	Sequencer	Sampling & Testing Kit	Bioinformatics	Technical Support
 MGISP-100	 DNBSEQ-G99	 Sampling Kit	 BGI HALOS	 Laboratory Design
 MGISP-960	 DNBSEQ-G50	 DNA Extraction Kit	 NIFTY®/NIFTY® Pro Software	 On-site Training
 MGISP-Smart 8	 DNBSEQ-G400	 Library Kit  Sequencing Kit		 Technical Support

Automated Sample Preparation

- 3 different automation platforms to meet laboratory needs.
- The entire process from plasma separation to DNB making can be automated[1].

MGISP-100

Specifications

Dimensions	780mm × 725mm × 777mm
Pipette Type	Fixed 8-channel pipette
Pipette Range	1 μL~200 μL

Using for NIFTY®/NIFTY® Pro

MGISP-100
DNA Extraction, Enrichment, Library Construction, Pooling, DNB Making, Reagent Preparation
~8 h/run, 1-16 samples/run

MGISP-Smart 8

Specifications

Dimensions	1410mm × 799mm × 970mm
Pipette Type	Independent 8-channel pipette
Pipette Range	1 μL~1000 μL

Using for NIFTY®/NIFTY® Pro

MGISP-Smart8 Config #4 [1]
Plasma Separation, DNA Extraction, Enrichment, Library Construction, Pooling, DNB Making, Reagent Preparation
~11 h/run, 1-48 samples/run

MGISP-960

Specifications

Dimensions	1240mm × 740mm × 1110mm
Pipette Type	Fixed 96-channel pipette
Pipette Range	1 μL~200 μL

Using for NIFTY®/NIFTY® Pro

MGISP-960 Config #2[2]
DNA Extraction, Enrichment, Library Construction
~7 h/run, 1-96 samples/run

[1] Plasma separation is only available on MGISP-Smart8
[2] Only Config #2 integrates PCR functionality

Sequencing

- NIFTY® and NIFTY® Pro can be processed on 3 different gene sequencers.
- Laboratories are free to choose sequencers based on project and sample volume.

DNBSEQ-G99

Specifications

Dimensions	607mm × 680mm × 640mm
Flow Cell No.	2
Flow Cell Type	FCL
Lane No./ Flow Cell	1
Effective Reads/Flow Cell	80M

Item	Samples/Run	Run Time
NIFTY®	12 samples/FCL	~3h
NIFTY® Pro	3 samples/FCL	~3h

DNBSEQ-G50

Specifications

Dimensions	654mm × 489mm × 545 mm
Flow Cell No.	1
Flow Cell Type	FCL
Lane No./ Flow Cell	1
Effective Reads/Flow Cell	500M

Item	Samples/Run	Run Time
NIFTY®	48 samples/FCL	~9h
NIFTY® Pro	12 samples/FCL	~9h

DNBSEQ-G400

Specifications

Dimensions	1086 mm × 756 mm × 710 mm
Flow Cell No.	2
Flow Cell Type	FCL & FCS
Lane No./ Flow Cell	4
Effective Reads/Flow Cell	1500-1800M

Item	Samples/Run	Run Time
NIFTY®	192 samples/FCL	~12h
NIFTY® Pro	48 samples/FCL	~12h

Bioinformatics

- Premium and basic hardware configurations are optional to meet different needs
- Supports multiple analysis algorithms pre-installed on one HALOS
- Supports linking one HALOS to multiple sequencers
- Supports offline analysis[1]

High Integration

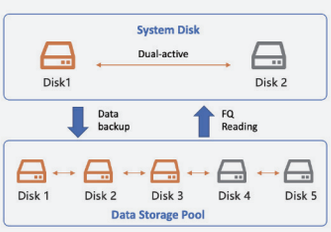
Integrated network switch and power interface inside the host

Flexible Combination

Access terminal and computing host can be separated to adapt to various laboratory environments

Native Expansion

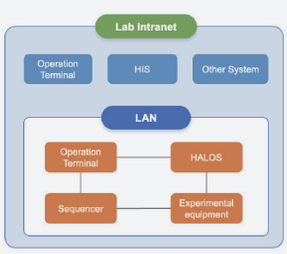
Supports expansion of computing and storage units, simple disassembly and assembly, beautiful stacking



Data Security

Built-in disaster recovery capability; Layered design of system and data disk

HALOS



Cyber Security

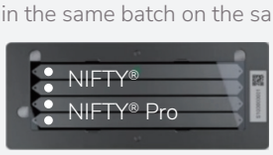
- B/S structure, high transmission efficiency
- Can operate without network
- Connect to laboratory systems or the Internet as needed

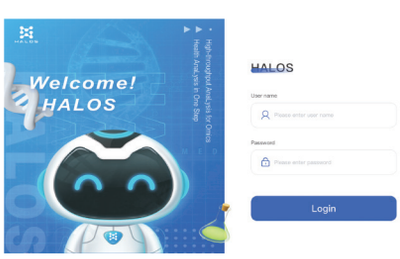
Strong Compatibility

One HALOS can be pre-installed with multiple analysis algorithms supports distributed computing, and is compatible with different analysis needs.

One HALOS can be connected to multiple sequencers and can analyze different products at the same time

One HALOS can simultaneously analyze the data of different products mixed in the same batch on the same chip





HALOS-NIFTY®

- Compatible with DNBSEQ-G400, DNBSEQ-G50 and DNBSEQ-G99
- Supports arbitrary mixed sample analysis of NIFTY® and NIFTY® Pro
- Information management of the entire experimental process
- No need to be on duty, automatic analysis, automatic report generation
- No need for data backhauling manufacturer

[1] Certain products support this mode.

Sampling & Testing Kits

- All of the sampling and testing kits have obtained CE certification.
- Used by more than 300 laboratories around the world.
- Global cold chain logistics system ensures safe supply.

Sampling Kit

Nucleic Acid Extraction Kit

Library Preparation Kit

DNBSEQ-G400
Sequencing Kit (SE50)

DNBSEQ-G50
Sequencing Kit (SE50)

DNBSEQ-G99
Sequencing Kit (SE100)

Global Logistics Network

- Global logistics network partners 80+
- Qualified by IATA (International Air Transport Association)

NIFTY® Localization Solution Options

Suggested solution 1

- Automation:
  1-16 samples/run
- TAT[1]: ~13.5 hours
- Laboratory technician: 1 person

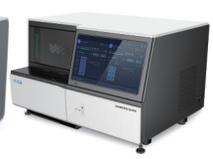


Experimental procedure	Sample preparation	Sequencing	Analysis and report
Experimental equipment	MGISP-100	DNBSEQ-G99	HALOS
Operation time	~10h (manual operation 2h)	~3h	~0.5h

Suggested solution 2

- Automation:
  1~48 samples/run
- TAT[1]: ~23.5/27.5 hours
- Laboratory technician: 1 person

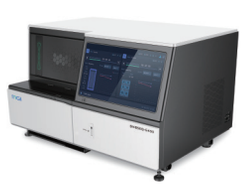





Experimental procedure	Sample preparation	Sequencing	Analysis and report
Experimental equipment	MGISP-Smart8 (Config #4)	DNBSEQ-G50 / DNBSEQ-G400	HALOS
Operation time	~12.5h (manual operation 1.5h)	~10h/12h	~1h / 3h

Suggested solution 3

- Automation:
  1~96 samples/run
- TAT[1]: ~25 hours
- Laboratory technician: 1 person


Experimental procedure	Sample preparation	Sequencing	Analysis and report
Experimental equipment	MGISP-960 (Config #2)	DNBSEQ-G400	HALOS
Operation time	~10h (manual operation 3h)	~12h	~3h

[1] Factors such as shifts and personnel proficiency in operation can affect TAT.

Our Technology

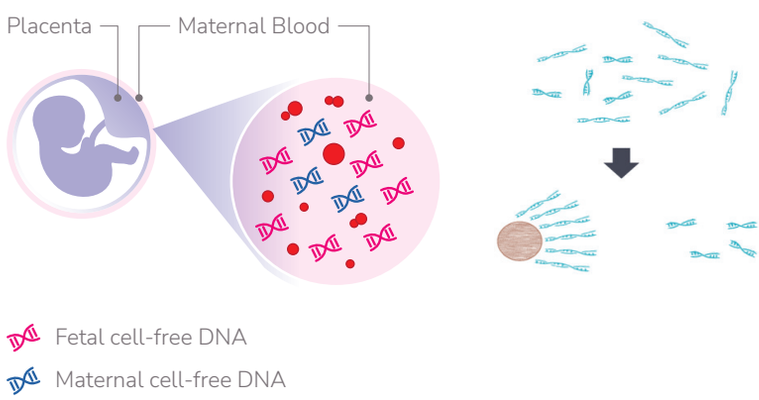
Fetal Fraction (FF%) Enrichment

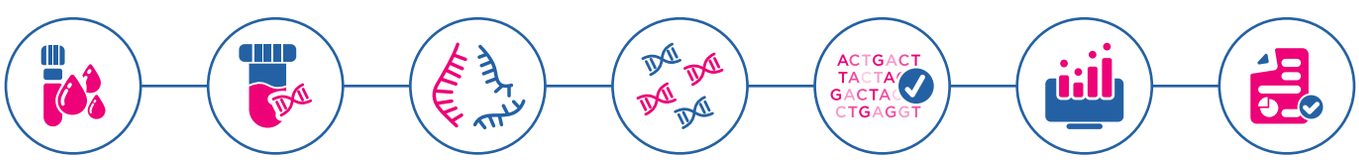
Using the enrichment process can increase the fetal cell-free DNA concentration by 1.5 to 2.0 times:

- Reduce false negatives
- Reduce re-test rate by 50%
- Improve CNV detection rate
- More significant for samples with low cffDNA concentration (≤4%)

Fetal fraction enrichment method: Magnetic beads fragment selection

Fetal cfDNA (cffDNA) fraction size is generally smaller than the maternal cfDNA. Magnetic beads will priority absorb larger fragment. The fragment selection can be achieved by using the supernatant only contain smaller fragment.





Optimized CNV Analysis Algorithms

An independent algorithm has been developed for CNV, which greatly improves the detection performance of CNV, especially DiGeorge syndrome.

Disorder	TP	FP	TN	FN	Sensitivity	Specificity	PPV
DiGeorge syndrome	9	6	16764	2	81.80%	99.96%	60.00%

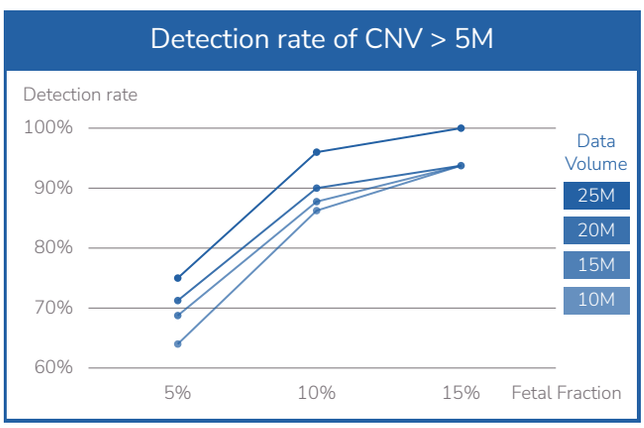
- 11 positive samples of DiGeorge syndrome, mix with 16,447 clinical samples.
- Among the 6 cases of false positives, 1 case was suspected malignant tumor, and 2 cases had ultrasound phenotype, suggesting that the actual PPV may be higher than 60%.

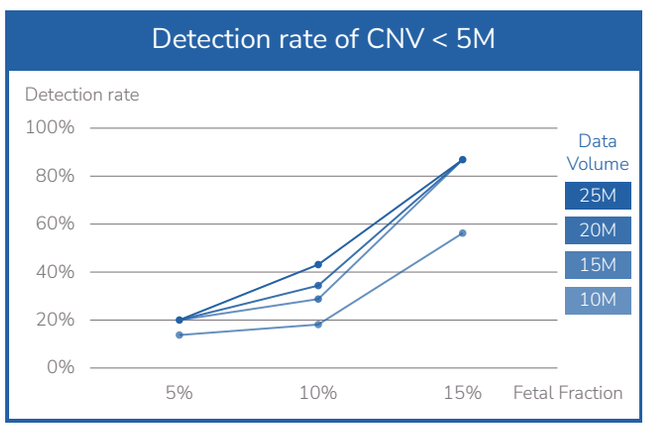
Our Technology

High Data Output Brings High Sensitivity

- Under the same FF%, as the reads increase, the detection sensitivity of CNV also increases accordingly.
- NIFTY® Pro uses a sequencing strategy of 25M reads per sample.

Higher reads improves CNV detection performance (R&D data)





As reads of NIFTY® increased to 25M:

- The positive CNV detection rate increased.
- The positive CNV detection rates for small fragments increased.
- The positivity predictive value (PPV) of CNV increased.

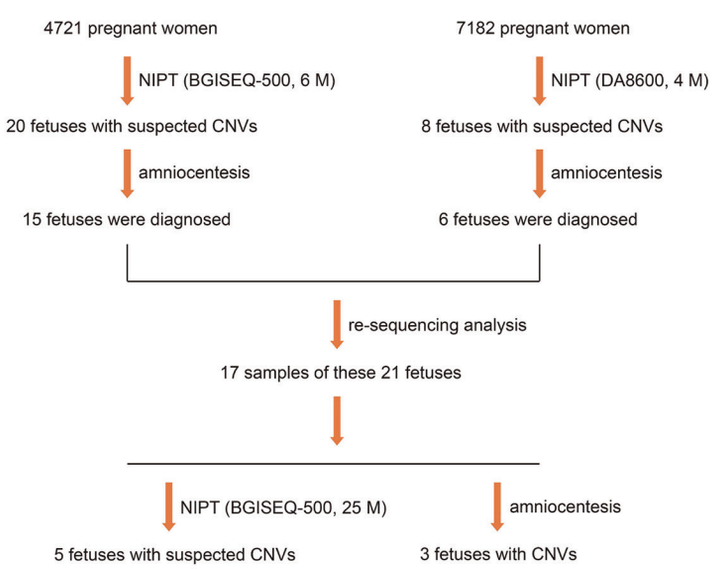
frontiers in Molecular Biosciences

ORIGINAL RESEARCH
published: 12 May 2021
doi: 10.3389/fmolb.2021.649169

Expanding the Scope of Non-invasive Prenatal Testing to Detect Fetal Chromosomal Copy Number Variations

Songchang Chen[1,2,3,4,5†], Lanlan Zhang[1,3,4,5,6†], Jiong Gao[7], Shuyuan Li[3,4,5], Chunxin Chang[3,4,5], Yiyao Chen[3,4,5], Hongjun Fei[3,4,5], Junyu Zhang[3,4,5], Yanlin Wang[3,4,5], Hefeng Huang[3,4,5], Chenming Xu[2,3,4,5] and Daru Lu[1,8*]*

CNVs					
Different fragments	Number	Detection number by NGS	Detection rate by NGS	Detection number by karyotype analysis	Detection rate by karyotype analysis
0-1 Mb	18	5	27.8%	0	0
1-3 Mb	36	30	83.3%	5	13.9%
3-10 Mb	25	19	76.0%	8	32.0%
>10 Mb	23	19	82.6%	19	82.6%
Total	102	73	71.6%	32	31.4%



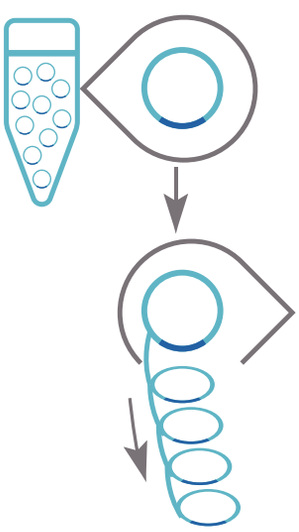
https://doi.org/10.3389/fmolb.2021.649169

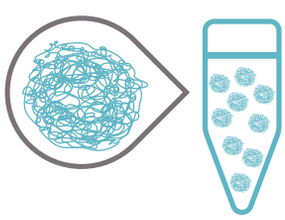
Our Technology

DNBSEQ™ Technology

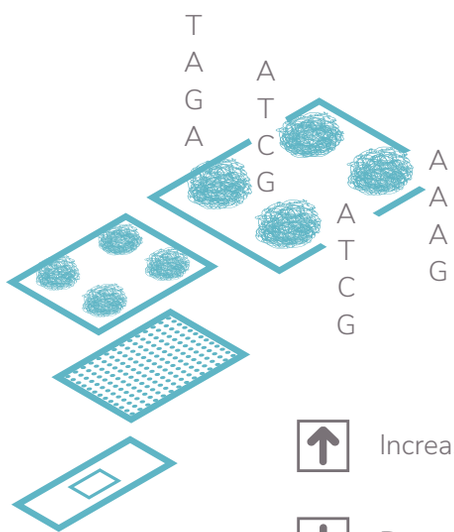
The DNA sequencing instruments utilize the state-of-the-art core technology called DNBSEQ™. DNBs (DNA nanoballs) are pumped with by the fluidics system and loaded onto a Patterned Array chip. Sequencing primer is then added and hybridized to the adaptor region of the DNB. The sequencing reaction starts by pumping sequencing reagents containing fluorescently labelled dNTP probes and DNA polymerase. Images are taken after the fluorescently labelled probes on the DNB are excited with lasers. The images are then converted into a digital signal using the propriety software. This information is then used to determine the DNA sequence of the sample.

Compared to other existing sequencing platforms, DNBSEQ™ sequencing technology combines the advantages from low amplification error rates from DNBs and high density patterned arrays. These advantages dramatically improve sequencing accuracy and have much lower duplication rates in WGS/WES applications.





DNA Nanoballs (DNB)



Patterned Array

- Increased Accuracy
- Decreased Duplicates
- Reduced Index Hopping

Our Technology

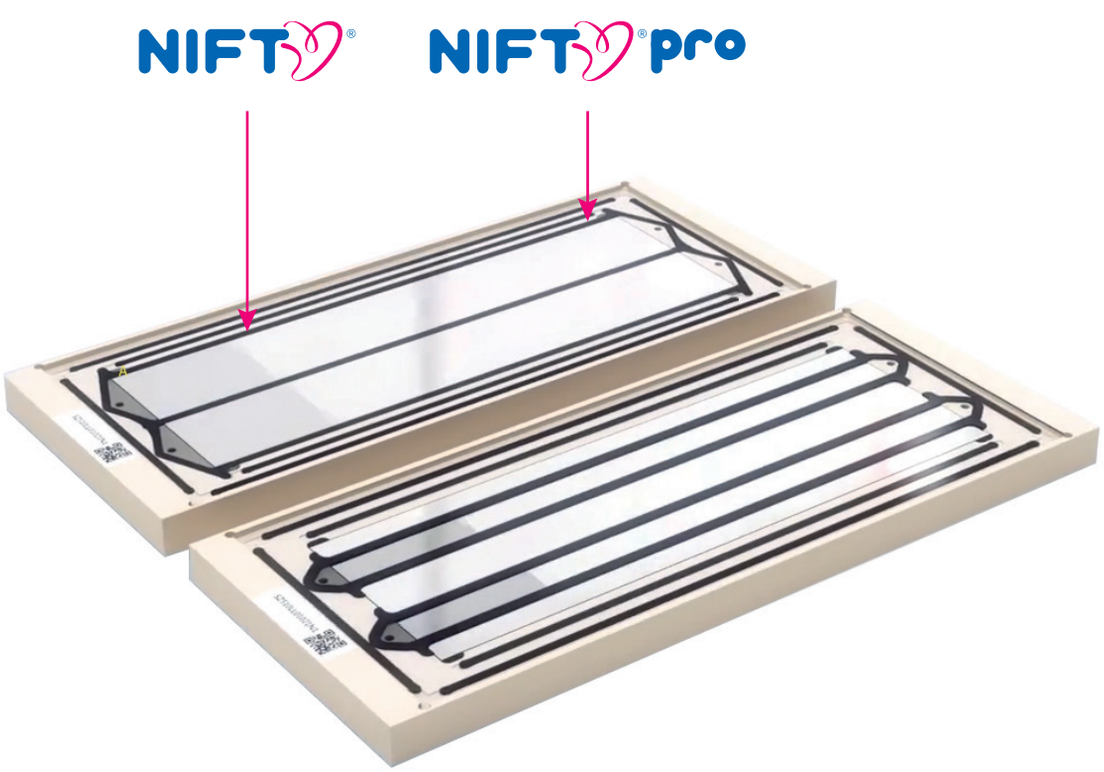
Flexible Mixed-sample Testing

HALOS can be pre-installed with NIFTY® and NIFTY® Pro analysis software, supports single sample analysis, and can automatically retrieve offline data to complete NIFTY® and NIFTY® Pro analysis simultaneously.

Depending on the type of samples arriving at the laboratory, the laboratory technician can flexibly arrange the NIFTY® and NIFTY® Pro sample sizes for sequencing in the same batch according to the formula (provided by BGI).

Sequencer	Lane No.	Max samples of NIFTY®	Max samples of NIFTY® Pro	Mixing formula
DNBSEQ-G99	1	12	3	A+4B≤12
DNBSEQ-G50	1	48	12	A+4B≤48
DNBSEQ-G400	4	192	48	A+4B≤192

*A=Samples of NIFTY®
*B=Samples of NIFTY® Pro



The flexible mixed sample detection can better meet the sample size needed for NIFTY® and NIFTY® pro, which can shorten the time to gather enough samples and save reagent cost.

Extensive Validation Data

BGI Genomics is a pioneer and a technology leader in non-invasive prenatal testing technology in China and internationally. BGI Genomics successfully developed NIPT technology in 2008 and began clinical application in 2010, serving over 13,000,000 people to date.

Accurate
High sensitivity and high specificity validated through large-scale population studies.

Insured*
Offering insurance in cases of false positive or false negative.

Comprehensive
Screen for common trisomies (T21, T18, T13), SCAs, RATs, CNVs.

Trusted
Over 13 million tests carried out to date, 70+ publications supporting performance.

NIFTY® and NIFTY® Pro has completed large-scale validation[1,2].

Chromosome abnormalities	Sensitivity (95%CI)	Specificity (95%CI)	PPV (%)
Common Trisomies	99.02% (98.38%-99.66%)	99.86% (99.84%-99.88%)	85.27%
T21	99.17% (98.52%-99.83%)	99.95% (99.93%-99.96%)	92.19%
T18	98.24% (94.93%-99.63%)	99.95% (99.93%-99.96%)	76.61%
T13	100.00% (84.56%-100.00%)	99.96% (99.95%-99.97%)	32.84%
SCAs	100.00% (90.59%-100.00%)	99.90% (99.85%-99.94%)	68.12%
RATs	100.00% (19.79%-100.00%)	99.88% (99.82%-99.92%)	6.67%
CNVs	100.00% (74.65%-100.00%)	99.94% (99.89%-99.97%)	51.72%
< 10Mb	100.00% (59.77%-100.00%)	99.86% (99.93%-99.98%)	50.00%
≥ 10Mb	100.00% (56.09%-100.00%)	99.97% (99.94%-99.99%)	53.85%

PPV, positive predictive; SCAs, sex chromosome abnormalities.
RATs, rare autosomal aneuploidies; CNVs, copy number variations.

[1]Zhang H, Gao Y, Jiang F, et al. Ultrasound Obstet Gynecol (2015), 45:530-538.
[2]Zou Y, Feng C, Qin J, et al. Front. Genet.(2023), 13:1073851.

*This service is limited to samples sent to BGI laboratory for testing only.

Services of Technology Transfer

Preparation

- Laboratory design
- Environment quality assessment
- Equipment and consumables list review
- Site inspection

Installation

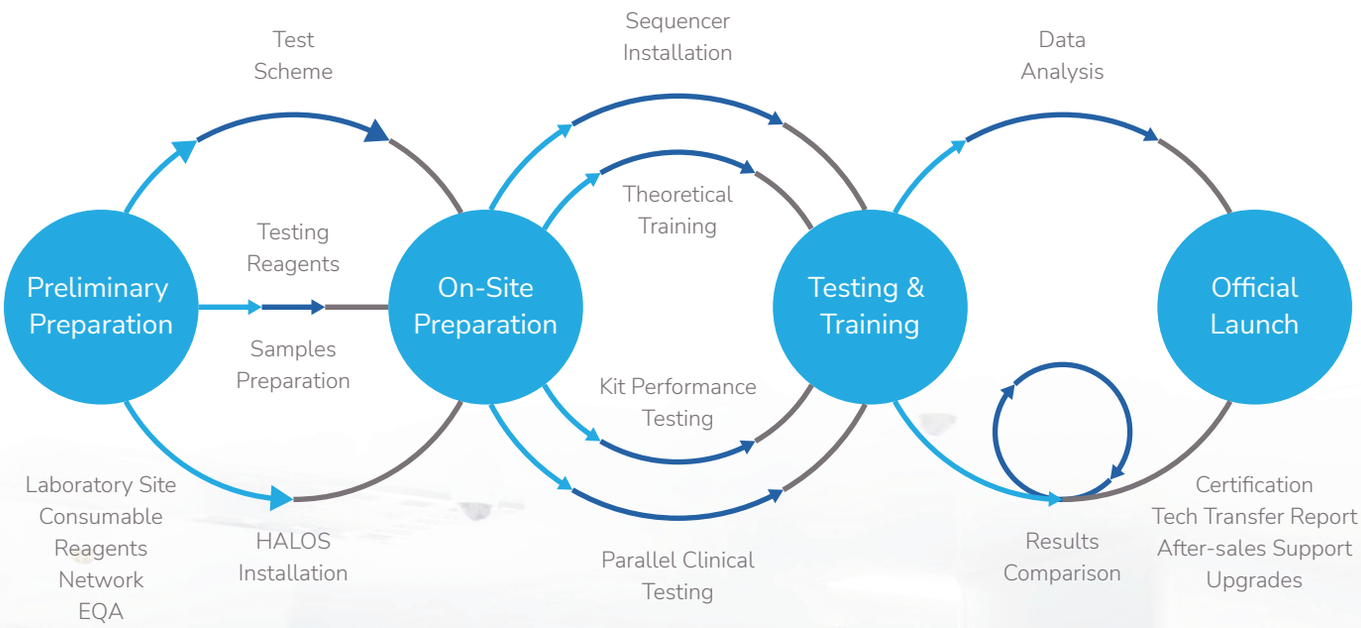
- Devices installation
- Devices performance testing

On-site Training

- Kits performance testing & training
- Parallel testing
- Results comparison
- Certification

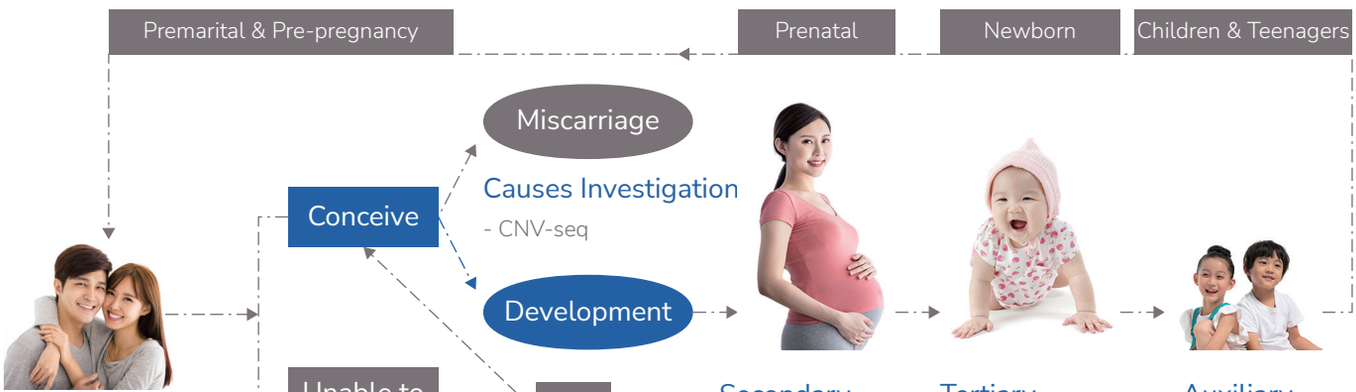
After-sale Service

- Tech transfer report
- Software upgrades
- Application support
- Field service support



Products Throughout the Life Cycle

Reproductive health



Primary Prevention

- Thalassemia genetic testing
- Carrier screening

Select healthy Embryos

- PGT-A

Secondary Prevention

- NIFTY®/NIFTY® Pro
- NIFTY® Mono
- CNV-seq

Tertiary Prevention

- Newborn metabolic screening
- Newborn genetic testing

Auxiliary Diagnosis

- WES
- WGS

Pathogenic infection precise detection

- High-throughput gene detection of pathogenic microorganisms (PMseq)
- Targeted high-throughput gene detection of pathogenic microorganisms (PTseq)

Cancer early screening and diagnosis

- Hereditary cancer gene testing
- HPV genotyping
- Colorectal cancer early screening (Colotect)
- Four-cancers genetic testing
- Cancer+discovery

Request for Information or Quotation

www.bgi.com/global/
info@bgi.com

@BGI Genomics

NIFTY® is an in vitro diagnostic test based on high-throughput sequencing technology that assesses the risk of fetal chromosomal abnormalities such as trisomy 21, 18, and 13 by detecting peripheral whole blood samples from pregnant women at ten weeks of pregnancy and above.

This product CANNOT be used as the sole basis for diagnosis or other pregnancy management decisions. Further confirmatory testing is necessary prior to making any irreversible pregnancy decision.

Unless otherwise informed, certain sequencers and sequencing reagents are not available in selected countries or regions. Please contact a representative for regional availability.The company reserves the right of final interpretation.

Published Jan 2024.