Capital TECHNOLOGY | CapitalBio Genomics

CapitalBio® NIPT

Non-Invasive Prenatal Testing (NIPT) for Fetal Chromosomal Aneuploidies and Microdeletions/Microduplications



HWSY-20170005-V1.0

Capital TECHNOLOGY

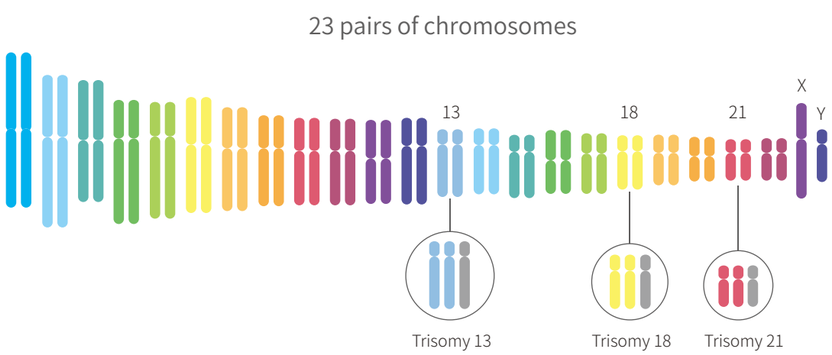
Building C, 88 Kechuang 6th Street, Yizhuang Biomedical Park
Beijing Economic-Technological Development Area
Beijing 101111
China

Tel: +86-10-69002900
Fax: +86-10-69002988
Email: globalsales@capitalbiotech.com
Website : http://www.capitalbiotech.com

CapitalBio Technology
CapitalBio Genomics

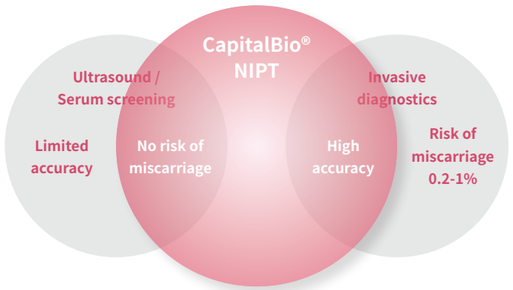
Background

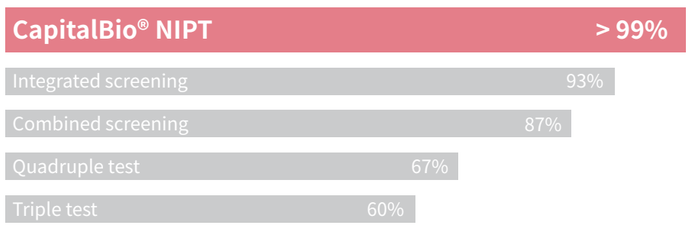
The incidence of chromosomal abnormalities is as high as 1 in 60 in China[1]. The incidence increases with maternal age and can reach 2.5% with maternal age over 35[1]. Among those with chromosomal abnormalities, fetuses with Trisomy 21 (Down Syndrome), Trisomy 18 (Edward Syndrome), and Trisomy 13 (Patau Syndrome) are most likely to survive. Therefore detection of the chromosomal abnormalities are most clinically significant.



Why CapitalBio NIPT?

High Accuracy
conventional screening methods: 60-80%
The accuracy rate of CapitalBio NIPT: 99%
Low Risk of Miscarriage
conventional screening methods: 0.2-1%
The risk of miscarriage associated with NIPT: 0
Short turnaround time
G-band Karyotyping: at least one week
Complete NIPT workflow: 4 days.







Who Should be Offered CapitalBio NIPT ?

1.Pregnant women with indicators suggesting high risk of fetal aneuploidies who wish to have an early and accurate test. The indicators include :

- High maternal age
- Positive result in serum screening
- Abnormalities discovered in ultrasonography
- History of giving birth to a baby/babies with chromosomal abnormality(ies)
- Parental abnormality(ies) involving one of the tested chromosomes

2.Pregnant women with interventional prenatal diagnosis contraindications (threatened abortion, fever, bleeding tendency, unrecovered infection, etc.)

3.Pregnant women with gestational age of above 20+6 weeks, who have missed the best time for serological screening.

What do the experts say?

[...] informing all pregnant women that NIPS is the most sensitive screening option for traditionally screened aneuploidies [...] **ACMG 2016**

[...] New evidence strongly suggests that NIPS can replace conventional screening for Patau, Edwards, and Down syndromes [...]**ACMG 2016**

[...] any patient may choose cell-free DNA analysis as a screening strategy for common aneuploidies regardless of her risk status [...] **ACOG 2015**

[...] NIPT can be used as a primary screening method for fetal trisomy 21 in pregnant women of every age and risk group. [...]**Austrian-German-Swiss Recommendations for NIPT 2016.**

CapitalBio® NIPT

Test

Safe, Rapid, Accurate and Affordable.

CapitalBio NIPT test can detected **Trisomy 21,18,13, sex chromosome aneuploidies (45,X), (47,XXY), (47,XYY) and 300 Microdeletions/ Microduplications** from maternal blood since the ninth week of pregnancy (9+0 weeks since LMP). If desired, the gender of the fetus may also be determined.

Accurate	> 99.9% Sensitive[2,3]	> 99.5% Specificity[2,3]	< 0.5% No-call rate
Safe	Take only 5-8 ml maternal peripheral blood		
Rapid	Report completed in 4 days [2]		
Affordable	The CapitalBio® NIPT offers unbeatable value for money. Contact us to learn more: tel：0086 10 6900 2900 Emal：globalsales@capitalbiotech.com		

Can be used **following assisted reproduction** – even if donor eggs are used

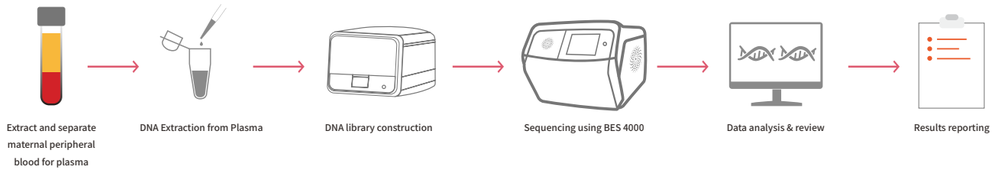
CapitalBio® NIPT

Method

The CapitalBio NIPT based on semiconductor sequencing technology offers a wide range of applications and also ensures a more cost-effective and rapid analysis.

Measures are taken in each step to ensure high quality

- The concentration of cffDNA is increased with innovative enrichment technology, improving the accuracy of z-score
- The concentration of cffDNA is calculated to discover false positive or false negative results caused by confined placental mosaicism or maternal DNA interference
- Human error is reduced to the maximum extent, since fully automated assay management and data analysis software is employed
- Samples suspected of high risk are retested



In 3 days

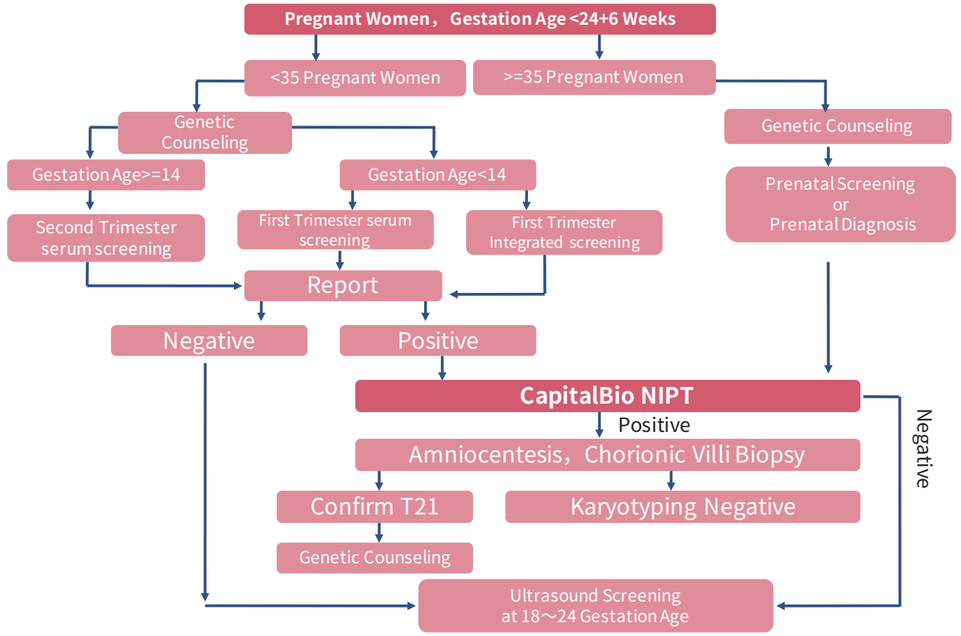
Accuracy demonstrated by clinical validation

A large-scale clinical trial of CapitalBio NIPT involved plasma samples from 2275 pregnant women have done, and the results indicated that both of the sensitivity and specificity to detection of T21,T18,T13 were greater than 99%.

Table 1. Diagnostic performance of the SSP for identifying T21, T18 and T13 in a 515 cases Retrospective Study

Type	Number of cases	Sensitivity	Specificity
Trisomy 21	55	99.94%	99.46%
Trisomy 18	16	100%	99.24%
Trisomy 13	3	100%	100%

Prenatal Screening Strategy



All positive results must be verified by chromosome karyotyping[4]

Options

Option 1

Detection/Screening of fetal trisomy 21, 18, 13

Sex chromosome aneuploidies (45,X), (47,XXY), (47,XYY), if detected
Gestation week: 9

Option 2

Detection/Screening of fetal trisomy 21, 18, 13 and other 300 common chromosomal microdeletions/microduplicaitons.*(over 1Mb[5])

Sex chromosome aneuploidies (45,X), (47,XXY), (47,XYY), if detected
Gestation week: 12

References

1.Zhang YP., et al.,(2011) Karyotype analysis of amniotic fluid cells and comparison of chromosomal abnormality rate during second trimester, Zhonghua Fu Chan Ke Za Zhi 46(9): 644-648

2.Liao C., et al. ,(2014) Noninvasive prenatal diagnosis of common aneuploidies by semiconductor sequencing, Proc Natl Acad Sci USA, 2014.111(20): p. 7415-20

3.Hu H., et al. ,(2016) Clinical Experience of Non-Invasive Prenatal Chromosomal Aneuploidy Testing in 190,277 Patient Samples. Current Molecular Medicine 2016, 16, 759-766

4.Liao C., et al.,(2014) DNA sequencing versus standard prenatal aneuploidy screening

5.Yin AH.,et al.,(2015) Noninvasive detection of fetal subchromosomal abnormalities by semiconductor sequencing of maternal plasma DNA, Proc Natl Acad Sci USA, 2015.112(47): p. 1470-1475

Product information

Introduction of BES 4000 System

CapitalBio BioelectronSeq 4000 System is a next-generation sequencing system developed and produced by CapitalBio Corporation in collaboration with Thermo Fisher Scientific Inc. in China. The system adopts Semiconductor Sequencing Technology. Working with clinical testing kits and automated data analysis & management software, it is ideal for clinical laboratories. BES 4000 system has obtained CFDA Certificate in Feb. 2015.



Specications for BioelectronSeq 4000 Sequencer

Dimensions	Width *Depth*Height (cm): 61.8*75.0*50.7
Working environment (for indoor use only)	Temperature: 68 –77° F (20 –25° C); Humidity: 40–60%, noncondensing; Altitude: <6,500 ft (2,000 m) Clearances: 12 in (30.5 cm) in rear; 4 in (10 cm) on left side; 4 in (10 cm) on right side; 4 in (10 cm) from front edge of bench to sequencer bezel; 36 in (90 cm) aisle in front of bench for operator access
Power	Voltage: 100 V (min) to 240 V (max) Current: 14 A (max) Frequency: 50/60 Hz Power draw: 1,350 W
Gas supply	Connection: 0.25 in push-to-connect fitting Pressure: 30 psi Composition: nitrogen (grade 4.8, 99.998% or better)
Other connections	Ethernet: 1 GigE USB: 2x USB 2.0
Instrument computer hardware	Processor: Dual 8-core Intel® Xeon® Sandy Bridge Memory: 128 GB RAM FPGA: Dual Altera® Stratix® V GPU processor: 1x NVIDIA® Tesla® C2075 Storage: 11 TB (SSD and HDD) Operating system: Ubuntu® 11.10

CapitalBio® NIPT

Specications for BioelectronSeq 4000 Server

Product configuration	A single free-standing tower computer, included with the purchase of the BioelectronSeq 4000 System. Includes Torrent SuiteTM Software with all necessary software components to deliver signal processing, base calling, read alignment, and variant calling.
Processor	Dual 8-core 2.9 GHz CPUs
Memory	128 GB RAM
GPU processor	2x NVIDIA® Tesla ® GPUs
Storage	27 TB
Operating System	Ubuntu® 10.04
Dimensions	Width *Depth*Height (cm): 30.8*69.8*44.4 Weight: 46 kg
Power	Voltage: 100 V (min) to 240 V (max) Frequency:50/60 Hz Current: 12 A (max) Power Draw: 1,100 W

Performance specications of BioelectronSeq 4000 System

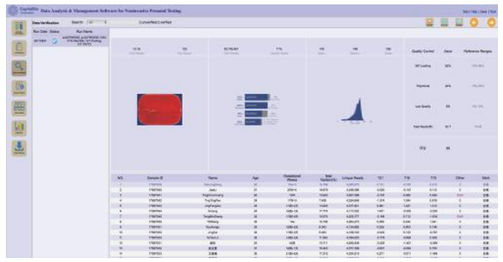
Output	PI chip: ⩾ 10Gb PII chip: ⩾ 60Gb
Read length Reads	PI chip: Up to 200 bases PII chip: Up to 100 bases
Reads	PI chip: ⩾ 80 million PII chip: ⩾ 200 million
Run time	2.5 hours
Library solutions	Ion AmpliSeqTM Library Kit Ion XpressTM Plus Fragment Library Kit Ion TargetSeqTM Exome Kit Ion Total RNA-Seq Kit
Data analysis solutions	Torrent Browser allows users to remotely access instrument status and monitor sequencing process using network connection; Variety of clinical data analysis plugins can complete clinical personalized data analysis automatedly and rapidly
Data Format	FASTQ, SFF, BAM and VCF etc.

Introduction of BES 4000 Software

CapitalBio® Data Analysis & Management Software for Noninvasive Prenatal Testing is a multifunctional software system that integrates the functions of data analysis and management, including sample information entry, experiment quality control, data analysis, data review and results reporting. The user could track, record, enter and display data, thereby achieving a comprehensive, automated and efficient data management.



Fully Automated Data Analysis



The adoption of advanced bioinformatics analysis method allows the software to automatically complete the entire process of data analysis without manual control. Each on-board sequencing results and the testing results of each sample will be clearly displayed in real time for user-friendly review.

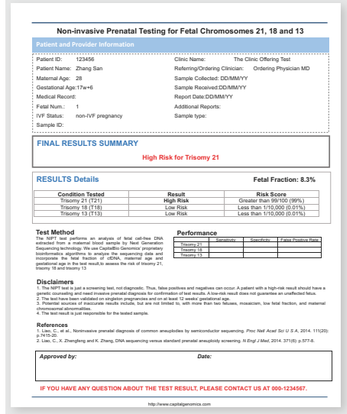
Fully Automated Results Interpretation

Based on sequencing data quality, fetal concentration, detected Z values and other QC parameters, the software can automatically calculate the testing results of all samples



CapitalBio® NIPT

One-click Report Printing



The report containing patient information, results details, fetal fraction and statement in standard layout can be printed out by clicking the button.

*Displayed only when fetal sex is requested



Ordering Information

Product	Cat. No	Size
BioelectronSeq 4000 System	180010	1 System
Data Analysis & Management Software for Noninvasive Prenatal Testing		Preinstalled in BioelectronSeq 4000 System
CapitalBio Fetal Chromosome Aneuploidy (T21,T18,T13) Testing Kit (Semiconductor Sequencing)	320020-01	120 reactions
CapitalBio Fetal Chromosome Aneuploidy (T21,T18,T13) Testing Kit (Semiconductor Sequencing)	320020-02	60 reactions
CapitalBio Fetal Chromosome Aneuploidy (T21,T18,T13) Testing Kit (Semiconductor Sequencing)	320020-03	30 reactions