NIFTY pro™

NON-INVASIVE FETAL TRISOMY TEST

無創性胎兒染色體異常產前檢測

Peace of mind from a single drop

- Detect chromosomal disorders across 23 pairs of chromosomes
- Data size of each sample reaches 25M,further increase the detection rate of microdeletions/microduplications
- Available in more than 80 countries

Database: >5,000,000 cases

- Accuracy: >99%
- Obtained Patent
  Registration
  Publication No. HK1190758
- Test performed in **Hong Kong Laboratory**

HK Patented

*For further information please refer to the content of the pamphlet

BGI

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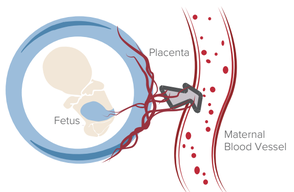
NIFTY Pro provided by BGI is a safe and easy prenatal test for detecting fetal chromosomal abnormalities. During pregnancy, the DNA of the baby will circulate into the mother's bloodstream. NIFTY Pro will require taking 6mL of the mother's peripheral blood to analyze cell free fetal DNA. Using the new generation of DNA sequencing along with bioinformatics to calculate the risk of having a fetus with chromosomal abnormalities.



Benefits of NIFTY pro

Risk Free: No miscarriage risk

Highly Accurate: 99% accurate, performed over 5,000,000 clinical samples. All Inclusive screening: Includes the screening of 6 autosomal chromosome abnormalities, 4 sex chromosome aneuploidies, 84 microdeletions/microduplications and gender determination.

High detection rate: data size of each sample reaches 25M, further increase the detection rate of microdeletions/microduplications.

Incidental findings: includes the rare chromosomal mutation apart from the above 95 test items across 23 pairs of chromosomes.

Assurance: Tests are all done in Hong Kong and reports are issued by Hong Kong MLT board registered Part I MLT.

Early Pregnancy Test: Test could be done as early as 10 week or above of pregnancy (including twin pregnancies); Nifty Pro only applicable for Singleton pregnancy.

NIFTY pro is suitable for

- Women who would like to avoid miscarriage risk following invasive prenatal testing.
- Down syndrome screening indicating an increasrd risk of abnormalities.
- Pregnancy following IVF or history of recurrent miscarriage.
- Pregnant woman who wish to have a highly accurate result on whether her baby has Down syndrome.

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NIFTY pro™ Test Content:

Chromosome Abnormality	Incidence Rate	Clinical Features		Accuracy Rate
Down Syndrome Trisomy 21	Risk will increase as the age of woman increases (Age 35: 1/400)	It is a condition caused by an extra copy of chromosome 21. Miscarriage occurs in about 30% of pregnancies with Down Syndrome. Those children born with Down Syndrome will need extra medical care depending on the child's specific health problems. Most children with Down Syndrome have intellectual disabilities that range from mild to moderate. Early intervention has proven to be essential in enabling individuals with Down Syndrome to lead healthy and productive lives.		Sensitivity: >99.99%[1] Specificity: >99.97%[1]
Edwards Syndrome Trisomy 18	1/6,000	It is caused when a baby has three copies of chromosome 18. Pregnancies with Edwards Syndrome are at high risk of miscarriage and most babies born with Edwards Syndrome die within the first few weeks of life while less than 10% live beyond one year. Infants with Edwards Syndrome have severe intellectual disabilities and birth defects typically involving the heart, brain, and kidneys, and external abnormalities such as cleft lip/palate, small head, club feet, underdeveloped digits, and small jaw.		Sensitivity: >99.99%[1] Specificity: >99.97%[1]
Patau Syndrome Trisomy 13	1/10,000-1/21,700	It is caused when a baby has three copies of chromosome 13. Pregnancies diagnosed with Patau Syndrome are at high risk for miscarriage or stillbirth, and most babies born with Patau Syndrome will not survive beyond the first weeks of life. Babies with Patau Syndrome may have heart defects, brain or spinal cord problems, extra fingers and/or toes, an opening in the lip (cleft lip) with or without an opening in the roof of the mouth (cleft palate), and weak muscle tone. Many babies have birth defects of other organs as well.		Sensitivity: >99.99%[2] Specificity: >99.96%[2]
Trisomy 9	unknown	Trisomy 9 is a rare chromosomal condition. Full trisomy 9 is a lethal chromosomal disorder resulting in miscarriage in the first trimester. Rare survivors usually cannot live more than a week after birth.	Mosaic Trisomy 9 is the condition that the fetus carries two entire chromosomes 9 plus part of a third copy. The Symptoms include developmental delay, dysmorphisms in the heart system, nervous systemm and skull.	Rare Sample Sensitivity rate not yet validated
Trisomy 16	32/100,000	Trisomy 16 is a rare chromosomal condition. Full trisomy 16 is a lethal chromosomal disorder resulting in miscarriage in the first trimester. Mosaic Trisomy 16 is the condition that the fetus carries two entire chromosomes 16 plus part of a third copy. The rare survivors are at increased risk for delayed growth and cognitive disorder.		
Trisomy 22	9/1000,000-20/100,000	Trisomy 22 is a rare chromosomal condition. Full trisomy 22 is a lethal chromosomal disorder resulting in miscarriage in the first trimester. The majority of live births will die during early postnatal period.	Mosaic Trisomy 22 is the condition that the fetus carries two entire chromosomes 22 plus part of a third copy. Affected individual usually experiences intellectual disability, kidney malformation and imbalanced development.	
45, X (XO) Turner Syndrome	1/2,000-1/5,000	It is caused by a completely or partially missing X sex chromosome in females. Females with Turner syndrome often have a wide range of symptoms and some distinctive characteristics. Two that occur in almost all cases of Turner syndrome are: being shorter than average. underdeveloped ovaries (female reproductive organs), resulting in a lack of monthly periods and infertility.		Sensitivity: >95%
XXY Klinefelter Syndrome	1/500	It is a genetic condition that only affects males. Affected males have an extra X chromosome. Males with Klinefelter's syndrome have small testes which do not produce enough of the male hormone testosterone before birth and during puberty. This lack of testosterone means that during puberty, the normal male sexual characteristics do not develop fully. There is reduced facial and pubic hair, and some breast tissue often develops. The lack of testosterone is also responsible for other symptoms, including infertility.		Sensitivity: >95%
XXX Triple X Syndrome	1/1,000	It is characterized by the presence of an additional X chromosome in each of a female's cells. The symptoms and physical features associated with trisomy X vary greatly from one person to another. Some females may have no symptoms (asymptomatic) or very mild symptoms and may go undiagnosed. Other women may have a wide variety of different abnormalities. Triple X syndrome is associated with an increased risk of learning disabilities and delayed development of speech and language skills. Delayed development of motor skills (such an sitting and walking), weak muscle tone (hypotonia). and behavioral and emotional difficulties are also possible, but these characteristics vary widely among affected girls and women. Seizures or kidney abnormalities occur in about 10 percent of affected females		Sensitivity: >95%
XYY XYY Syndrome	1/1,000	It affects only males and is caused by the presence of an extra Y chromosome. Affected individuals are usually very tall. Many experience severe acne during adolescence. Additional symptoms may include learning disabilities and behavioral problems such as impulsivity.		Sensitivity: >95%
Gender Inentification	NA	The singleton gender identification sensitivity rate.		>99%

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[1] 'Non-invasive prenatal testing for fetal chromosomal abnormalities by low-coverage whole-genome sequencing of maternal plasma DNA: review of 1982 consecutive cases in a single center ', Ultrasound Obstet Gynecol 2014; 43: 254-264

[2] 'Non-invasive prenatal testing for trisomies 21, 18 and 13: clinical experience from 146 958 pregnancies' , Ultrasound Obstet Gynecol 2015; 45: 530-538

Condition List for Micro-deletion & Micro-duplication Syndromes

1	Chromosome 1p31 duplication syndrome	43	Jacobsen syndrome
2	Chromosome 1p32-p31 deletion syndrome	44	Chromosome 13q14 microdeletion syndrome
3	Chromosome 1p36 deletion syndrome	45	Chromosome 13q14 deletion syndrome
4	Chromosome 1q41-q42 deletion syndrome	46	Chromosome 14q11-q22 deletion syndrome
5	Chromosome 2p12-p11.2 deletion syndrome	47	Fries syndrome
6	Chromosome 2p16.1-p15 deletion syndrome	48	Chromosome 15q11-q13 duplication syndrome
7	Split-hand/foot malformation 5	49	Angelman syndrome
8	Chromosome 2q31.1 duplication syndrome	50	Prader-Willi syndrome
9	Chromosome 2q31.1 deletion syndrome	51	Chromosome 15q14 deletion syndrome
10	Chromosome 2q35 duplication syndrome	52	Chromosome 15q25 deletion syndrome
11	Holoprosencephaly 6	53	Congenital Diaphragmatic Hernia, CDH
12	Chromosome 3pter-p25 deletion syndrome	54	Chromosome 15q26-qter deletion syndrome
13	Chromosome 3q13.31 deletion syndrome	55	Levy-Shanske syndrome
14	dandy-walker syndrome	56	Chromosome 16p deletion syndrome
15	Chromosome 3q29 deletion syndrome	57	Chromosome 16p11.2-p12.2 microduplication syndrome
16	Chromosome 3q29 duplication syndrome	58	Chromosome 16p12.2-p11.2 deletion syndrome
17	Wolf-Hir schonn syndrome	59	Chromosome 16p13.3 deletion syndrome
18	Chromosome 4q21 deletion syndrome	60	Chromosome 16q22 deletion syndrome
19	Chromosome 4q32.1-q32.2 triplication syndrome	61	Potocki-Lupski syndrome
20	Cri du Chat syndrome	62	Smith-Magenis syndrome
21	Chromosome 5q32 deletion syndrome	63	Yuan-Harel-Lupski syndrome
22	Chromosome 5q34.3 deletion syndrome	64	Chromosome 17p13.3 duplication syndrome
23	Chromosome 6pter-p24 deletion syndrome	65	Chromosome 17p13.3 deletion syndrome
24	Chromosome 6q11-q14 deletion syndrome	66	Chromosome 17p12 deletion syndrome
25	Chromosome 6q24-q25 deletion syndrome	67	Chromosome 17p12 duplication syndrome
26	CHDM	68	Chromosome 17q21.31 duplication syndrome
27	Chromosome 7q deletion syndrome	69	Chromosome 17q23.1-q23.2 deletion syndrome
28	Chromosome 7q11.23 deletion syndrome	70	Chromosome 18p deletion syndrome
29	Chromosome 7q11.23 duplication syndrome	71	Chromosome 18q deletion syndrome
30	Chromosome 8p23.1 deletion syndrome	72	Chromosome 19p13.11 deletion syndrome
31	Chromosome 8p23.1 duplication syndrome	73	Holoprosencephaly 1
32	Chromosome 8q12.1-q21.2 deletion syndrome	74	Cat-Eye syndrome
33	Chromosome 8q22.1 duplication syndrome	75	Chromosome 22q11.2 deletion syndrome
34	Chromosome 8q22.1 deletion syndrome	76	Chromosome 22q11.2 duplication syndrome
35	Langer-Giedion syndrome	77	DiGeorge syndrome
36	Chromosome 9p deletion syndrome	78	Chromosome Xp11.23-p11.22 duplication syndrome
37	Digeorge syndrome 2	79	Chromosome Xp11.3 deletion syndrome
38	Chromosome 10q22.3-q23.2 deletion syndrome	80	Chromosome Xp21 deletion syndrome
39	Chromosome 10q26 deletion syndrome	81	Chromosome Xq21 deletion syndrome
40	Potocki-Shaffer syndrome	82	Chromosome Xq22.3 telomeric deletion syndrome
41	WAGR syndrome	83	Chromosome Xq27.3-q28 duplication syndrome
42	WAGRO syndrome	84	Chromosome Xq28 deletion syndrome

Niftyhk

For further inquiries about NIFTY Pro, please contact us at:
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Comparison of Prenatal Screening Tests:

Test	Accuracy	Gestation	Miscarriage Risk	False Positive Rate	TAT (Working Days)
Maternal Serum Screening -1st trimester	80-90%	11-13[+6]	0%	5%	1-2
Maternal Serum Screening -2nd trimester	60-90%	16-19[+6]	0%	5%	1-2
Fetal Nuchal Translucency	60-80%	11-13[+6]	0%	5%	1-2
Amniocentesis	>99.9%	16-21	0.5-1%	<1%	14-21
Chorionic Villus Sampling	>99.9%	11-13	1-2%	<1%	14-21
Fetal Blood Sampling	>99.9%	>20	1-2%	0%	5-7
NIFTY pro	>99%	≥10	0%	<1% (Trisomy 21)	5

NIFTY pro™ Reassures You:

Being pregnant is something each and every woman would be extremely excited for. Aside from being excited you may also want to ensure the healthiness of your baby. All mothers should go through prenatal tests to ensure that the baby has not inherited genetic disease.

NIFTY Pro test technology can accurately test the risk of your baby having down syndrome and other chromosomal disorders using peripheral blood of the mother.

Due to technological restriction, listed below are women who cannot perform this test:

1. Multiple pregnancy (even fetal reduction occurs at later stage)
2. Twin pregnancy undergone fetal reduction after 8 weeks of gestation
3. Has undergone fetal reduction within the past 8 weeks
4. Pregnant women or her spouse with chromosomal disorders
5. Pregnancy with placental mosaicisms
6. Pregnant woman with Robertsonian translocatian fetus
7. Pregnancy less than 10 weeks

Please contact you doctor if you have conditions listed as below:

1. The receival of allogenic blood transfusion within one year
2. The received of transplant surgery or stem cell therapy
3. The receival of cellular immunotherapy where exogenous DNA is introduced within 4 weeks
4. The abnormal paternal karyotype, maternal abnormal karyotype with q9+/-, pss+/-, pstk+/-, pss
5. Pregnant woman with BMI>40
6. Being affected or had history of malignant tumor or benign tumor
7. The receival of heparin therapy or heparin analogue therapy

If you may require further assistance towards the report of NIFTY Pro, please contact your doctor.
If the test result is " **high risk** ", it is recommended to perform prenatal diagnosis.