非侵入性胎兒染色體異常檢測
NON-INVASIVE PRENATAL TEST

About 1 in 150 newborn babies has chromosomal abnormalities. Pregnant women of all ages are at risk of having a baby with chromosomal abnormalities and having such test will reduce that risk.

NIFTY® provides screening for common trisomy syndromes, including trisomy 21 (Down's Syndrome), trisomy 18 (Edward's Syndrome) and trisomy 13 (Patau Syndrome), as well as testing options for sex chromosomal aneuploidies, other chromosomal aneuploidies, certain chromosomal deletions/duplications, and gender identification.

約每 150 名新生兒中就有 1 名患有染色體異常。每位孕婦都面臨生育染色體病嬰兒的風險，均應該進行此產前檢測，降低風險。

NIFTY® 可檢測常見的三倍體綜合症，包括 T21（唐氏綜合症）、T18（愛德華氏綜合症）及 T13（巴陶氏症候群），以及性染色體非整倍體及其他染色體非整倍體的檢測選項，染色體缺失 / 重複，以及性別鑑定。

NIFTY® is a non-invasive fetal chromosomal abnormality test developed by BGI, which requires only 6mL of intravenous blood from the mother. It analysis of cell-free fetal DNA by next-generation sequencing combined with bioinformatics , which calculates the risk of the fetus having a chromosomal abnormalities, such as Down syndrome.

NIFTY® 是一種非侵入性胎兒染色體異常檢測技術。NIFTY 檢測只需抽取媽媽 5–10mL 靜脈血液，通過次世代 DNA 測序並結合生物信息技術，分析胎兒遊離 DNA，計算出胎兒患有染色體異常（如唐氏綜合症）的風險率。

NIFTY Pro™ is a non-invasive prenatal test for fetal chromosomal abnormalities. It analyzes cell-free fetal DNA through high-throughput next-generation sequencing combined with bioinformatics to determine the chromosomal health of the fetus. Compared to traditional invasive prenatal testing such as serum screening and amniocentesis, Nifty Pro™ is safer and has higher coverage than traditional invasive testing, and can be performed from the 10th week of pregnancy onwards.

NIFTY Pro™ 是一種非侵入性胎兒染色體異常的產前檢測技術。通過次世代 DNA 高通量測序並結合生物信息技術，分析胎兒游離 DNA，從而得知胎兒的染色體健康情況。相比傳統的血清篩查及羊膜穿刺術等侵入性產前檢查，使用非侵入性技術的 NIFTY Pro™ 會更為安全、檢測異常覆蓋範圍更高，更可從懷孕第 10 週後開始進行檢測。

Advantages 優勢：

Safe 安全
Non-invasive sample collection with no risk of miscarriage.
無創取樣，無流產風險。

Accurate 準確
Proven 99% sensitivity for T21, T18 & T13, based on a study of more than 146,000 pregnancies.
對近 146,000 例懷孕的研究，證明對 T21、T18 和 T13 的敏感度高於 99%。

Trusted 值得信賴
Over 1,400,000 NIFTY® tests carried out to date by clinicians in more than 100 countries.
迄今為止，100 多個國家的臨床醫生已進行了超過 1,400,000 次 NIFTY®。

Simple 檢測簡單
Test from a small >5mL maternal blood sample as early as week 10 of pregnancy.
早至懷孕第 10 週就從孕婦抽取 >5 毫升的血液樣本中進行檢測。

Comprehensive 全面
Comprehensive testing of 23 pairs of chromosomes.
全面檢測 23 對染色體。

NIFTY® is not suitable for patients with the following indications:

- Maternal, fetal and/or placental mosaicism (mixtures of chromosomally normal and abnormal cells in the pregnancy).
- Balanced or unbalanced translocation and chromosomal inversion.
- Patients who have received a blood transfusion within one year before the testing date.
- Patients who have had transplant surgery.
- Patients who have had stem cell therapy.

Vanishing twin syndrome:

- If a pregnancy is more than 8 weeks along, a reduction procedure can be performed or one of the embryos can stop developing.
- If a pregnancy is less than 8 weeks along and a reduction procedure has been done or one of the embryos has stopped developing, the pregnant woman should rest for less than 8 weeks.

NIFTY® 不適用於以下人士：

- 母體、胎兒和 / 或胎盤嵌合體（妊娠期染色體正常和異常細胞的混合物）；
- 平衡或不平衡易位和染色體倒位；
- 檢測日期前一年內接受過輸血的患者；
- 接受過移植手術的患者；
- 接受過幹細胞治療的患者；

消失雙胞胎綜合症：

- 懷孕 >8 週才進行減胎手術或其中一胎停止發育；
- 懷孕 <8 週進行過減胎手術或其中一胎停止發育而休息不足 8 週的孕婦。

The test will give results of where or not the fetus has any risk of obtaining chromosomal abnormalities. This will effectively prevent giving birth to a child with chromosomal abnormalities.

預測胎兒患染色體非整倍體或微缺失微重複的風險，有效減少及預防染色體異常嬰兒的出生。

Product Specification 產品規格：

Product Name 產品名稱		NIFTY®	NIFTYpro™	NIFTYpro™+
Test Contents 檢測內容	T21, T18, T13	✓	✓	✓
	Gender Identification 性別識別	✓	✓	✓
	Sex chromosome aneuploidy 染色體非整倍體	✗	✓	✓
	Other autosome aneuploidy 其他常染色體非整倍體*	✗	✓	✓
	Chromosome deletion/ duplication (CNV) 染色體微缺失或微重複*	✗	84 (22q11.21 deletion included)	84 (22q11.21 deletion included)
	Additional Findings 額外發現	Optional 自選	✗	✓
Indication 適用人群		All pregnancies with gestation between 10–24 weeks (includes IVF pregnant) 所有懷孕 10–24 週以上的孕婦（包括 IVF 懷孕）		
Data sequencing amount 測序數據量		6M	25M	25M
Technology 技術		Low coverage WGS	Low coverage WGS	Low coverage WGS
Sample Requirements 樣本類型		5–10 mL of maternal peripheral blood 5–10 毫升孕婦的外周血		
TAT 檢測周期		5–7 working days 5–7 個工作日		

NIFTY®

10 years of dedication safeguard the health and future of babies

10 年用心，守護寶寶健康及未來

NIFTY pro™ Test Content:

	Chromosome Abnormality	Incidence Rate	Clinical Features	Accuracy Rate	
Trisomies					
NIFTY BASIC	Down Syndrome Trisomy 21	Risk will increase with the age of woman (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% Specificity: >99.97%	NIFTY PRO
NIFTY BASIC	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 who born with Edwards Syndrome will 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% Specificity: >99.97%	
NIFTY BASIC	Patau's 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% Specificity: >99.96%	
Rare Autosomal Aneuploidies(only for singleton pregnancy)					
	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.		
Sex Chromosome Aneuploidies (only for singleton pregnancy)					
NIFTY BASIC	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%	
NIFTY BASIC	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 will lack of testosterone means that the normal male sexual characteristics will not be fully developed during puberty . 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%	
NIFTY BASIC	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% of affected females.	Sensitivity: >95%	
NIFTY BASIC	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%	
NIFTY BASIC	Gender Inentification	NA	The singleton gender identification.	>99%	

NIFTY pro™ 檢測內容：

染色體疾病	發病率	徵狀	檢測準確度	
三倍體綜合症				
唐氏綜合症 Trisomy 21	隨著孕婦年齡增加而上升 (35歲：1/400)	21 號染色體三體症，俗稱唐氏綜合症，是由於多了一條 21 號染色體而引致的疾病。約 30% 的流產個案都是因為懷有唐氏綜合症的胎兒。根據不同的健康問題，有些患有唐氏綜合症的嬰兒需要特別的照顧及醫療護理。大多數唐氏綜合症患者會有智力障礙，程度由輕微到中度不等。早期干預已被證實能夠有效改善唐氏綜合症患者的健康及生活。	靈敏性：>99.99%[1] 特異性：>99.97%[1]	NIFTY BASIC
愛德華氏綜合症 Trisomy 18	(1/6,000)	18 號染色體三體症，又稱愛德華氏綜合症，是由於嬰兒出生時帶有三條 18 號染色體。懷有愛德華氏綜合症胎兒的孕婦會很容易流產，而大多數能夠出生的嬰兒會在出生後數星期內夭折，不足 10% 的嬰兒能夠存活一年以上。大多數愛德華氏綜合症的嬰兒會有嚴重智力障礙及出生缺陷，包括心臟、腦及腎臟不正常等；外部異常，如唇裂 / 腭裂，頭小，畸型足，手指發育不全及下腭細小等。	靈敏性：>99.99%[1] 特異性：>99.97%[1]	
巴陶氏綜合症 Trisomy 13	1/10,000–1/21,700	13 號染色體三體症，又稱巴陶氏綜合症。正常嬰兒帶有兩條 13 號染色體，巴陶氏綜合症嬰兒卻帶有三條 13 號染色體。懷有巴陶氏綜合症胎兒的孕婦會有很高的流產或死胎風險，即使能夠出生的大多數嬰兒都會在出生後一週內夭折。巴陶氏綜合症嬰兒可能有心臟缺陷，腦或脊髓的問題，額外的手指和 / 或腳趾，腭裂或免唇及肌肉張力低下。嬰兒亦會有很多其他出生器官的缺陷。	靈敏性：>99.99%[2] 特異性：>99.96%[2]	
三倍體綜合症（僅限單胎）				
9號染色體三倍體 Trisomy 9	unknown	9 號染色體三倍體是罕見的染色體疾病。完全型 9 號染色體三倍體（Full Trisomy 9）胎兒大多數於第一孕期發生自然流產，活產的嬰兒大部份活不過出生後一週。 嵌合型 9 號染色體三倍體（Mosaic Trisomy 9）表示胎兒部分細胞多出一條 9 號染色體，主要臨床症狀爲發育缺陷、先天性心臟病、智力障礙、神經系統發育遲緩及骨骼肌系統異常等。	罕見案例，檢測靈敏度未經驗證	NIFTY PRO
16號染色體三倍體 Trisomy 16	32/100,000	16 號染色體三倍體是罕見的染色體疾病。完全型 16 號染色體三倍體（Full Trisomy 16）胎兒大多數於第一孕期就自然流產。 嵌合型 16 號染色體三倍體（Mosaic Trisomy 16）表示胎兒部分細胞多出一條 16 號染色體，主要臨床症狀爲發育遲緩及認知障礙等。		
22號染色體三倍體 Trisomy 22	9/1000,000–20/100,000	22 號染色體三倍體是罕見的染色體疾病。完全型 22 號染色體三倍體（Full Trisomy 22）胎兒大多數於第一孕期發生自然流產，活產的嬰兒也無法存活長久。 嵌合型 22 號染色體三倍體（Mosaic Trisomy 22）表示胎兒部分細胞多出一條 22 號染色體，主要臨床症狀爲智力障礙、腎臟形態異常、身體兩側不對稱發育等。		
性染色體異常綜合症（僅限單胎）				
透納氏綜合症 45, X (XO) Turner Syndrome	1/2,000–1/5,000	透納氏綜合症是女性出生時 X 性染色體全部或部分缺失而引起的疾病。患有透納氏綜合症的女性有不同程度的臨床病徵及一些獨特徵狀，但絕大多數透納氏綜合症患者都有以下兩種病徵：身材比正常矮小；先天卵巢發育不良，從而導致閉經（沒有月經）及不育。	靈敏性：>95%	NIFTY BASIC
柯林菲特氏綜合症 XXY Klinefelter Syndrome	1/500	柯林菲特氏症是一種只會出現於男性身上的染身體異常疾病。受影響男性會比正常男性額外出一條 X 染色體。男性柯林菲特氏症患者的睪丸較小，在出生前及青春期不能製造足夠的男性荷爾蒙，從而導致第二性徵沒有正常發育。其他病徵包括減少鬍鬚及陰毛，乳房有輕微發育。缺少男性荷爾蒙亦都會引致其他不同徵狀，包括不育。	靈敏性：>95%	
三 X 綜合症 XXX Triple X Syndrome	1/1,000	三 X 綜合症，又稱為 X 染色體三體症，是由於女性患者多出一條 X 染色體而引致的疾病。三 X 綜合症患者的身體特徵及臨床病徵程度因人而異。有些患者並沒有任何臨床表徵，或只有輕微徵狀，有些甚致終身都未被確診。但某些患者可能表現出很多異常的情況；例如增加了學習障礙的風險，導說話和語言發展遲緩，動作技能（如坐和行走）的發育緩慢，及肌肉張力低下。這些徵狀在女性患者中有很大的差異，但 10% 受影響的女性都會出現癲癇症或腎臟異常等病徵。	靈敏性：>95%	
XYY 三體綜合症 XYY XYY Syndrome	1/1,000	XYY 三體綜合症，又稱雅各氏症，只出現於男性。患者比正常人額外多了一條 Y 染色體。XYY 綜合症患者通常身材高大，而在青春期時容易有嚴重的青春豆問題。其他徵狀包括學習障礙及一些行為上的問題，如脾氣暴躁等。	靈敏性：>95%	
胎兒性別檢測	NA	檢測單胎胎兒的性別。	>99%	