NIFTY® | NIFTY pro™ +

無創性胎兒染色體異常產前檢測

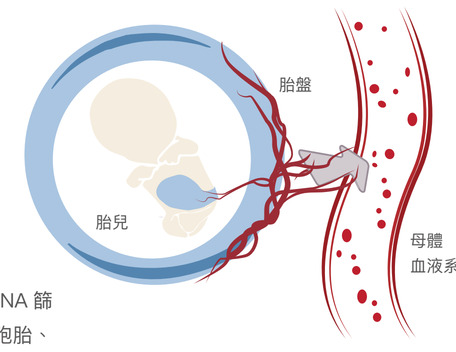
香港專利認可

簡單全面的檢測，讓各位父母安心

- 全面檢測23對染色體
- 每例樣品測序數據量達25M，進一步增加微缺失微重複檢出率
- 應用國家數量超過80個
- 超大型數據庫：全球超過1,370萬實例
- 準確率＞99%
- 專利註冊 專利編號：HK1190758
- 檢測於香港實驗室內進行

www.nifty.com.hk

Nifty / Nifty Pro+ 介紹



Nifty / Nifty Pro+ 是由華大基因（BGI）研發的一種非侵入性胎兒染色體異常檢測技術。孕婦在懷孕期間，胎兒的游離 DNA 會經過胎盤進入母體的血液系統。Nifty / Nifty Pro 檢測只需抽取媽媽靜脈血液，通過新一代 DNA 測序並結合生物信息技術，分析胎兒游離 DNA 狀況，計算出胎兒患有染色體異常（如唐氏綜合症）的風險率。

Nifty / Nifty Pro+ 是針對孕婦的無創產前基因檢測（游離 DNA 篩查），用於評估胎兒患上染色體疾病風險，適用於單胎、雙胞胎、卵子捐贈者。Nifty/ Nifty Pro+ 受到全球超過 137,000,000 孕媽媽的信賴，可以通過簡單的抽血來評估寶寶的健康狀況。Nifty / Nifty Pro+ 篩查常見的染色體異常，包括三體綜合症、性染色體非整倍體和微缺失 / 重複綜合症，並可以提供有關胎兒性別的資訊。

根據當前 ACOG 指南，不論孕婦處於任何年齡，或染色體異常風險如何，都應向孕婦提供游離 DNA 篩查。篩查是常見作為胎兒非整倍體檢測，游離 DNA 是最敏感和特異最高的篩查檢測。[1]

染色體疾病		靈敏性	特異性	參考資料
T21		99.17%	99.95%	Ultrasound Obstet Gynecol. 2015 May;45(5):530-8.
T18		98.24%	99.95%	
T13		100%	99.96%	
Del/Dup	>10Mb	88.89%	99.32%	PLoS One.2016 Jul 14; 11(7):e0159233.
	<10Mb	72.73%	99.09%	

性染色體異常	靈敏性	特異性	PPV	參考資料
XO	75%	99.9%	23.53%	BMC medical genomics vol. 5 57. 1 Dec. 2012
XXX	N/A	N/A	70%	
XXY	100%	100%	75%	Chinese medical journal vol. 133,13 (2020): 1617-1619.
XYY	100%	100%	80%	

[1] 表中數據基於歷史文獻和內部數據，僅反映過往檢測情況，不代表被測樣品的實際情況或承諾值。

- **安全無創：**不會流產風險。
- **準確可靠：**準確度高達 99% 以上。全球超過 1,370 萬實例。
- **全面篩查：**篩查胎兒 6 種常染色體三倍體綜合症、4 種性染色體異常、92 種染色體微缺失微重複綜合症、性別資訊。
- **高檢出率：**每例樣品測序數量達 25M，進一步增加微缺失微重複檢出率。
- **附加發現：**包括更罕見的染色體異常檢測 範圍覆蓋 23 對染色體。
- **信心保證：**由香港實驗室檢測和香港註冊醫療化驗師簽發報告。
- **早孕檢測：**懷孕 10 週或以上即可進行檢測（包括雙胞胎）。

NIFTY®

檢測選項：

- ✓ 三倍體綜合症(T21, T18, T13)
- ✓ 性染色體非整倍體
- ✓ 胎兒性別

NIFTY pro™ +

檢測選項：

- ✓ 三倍體綜合症（T21, T18, T13）
- ✓ 罕見常染色體非整倍體（T9, T16, T22）
- ✓ 性染色體非整倍體
- ✓ 92種染色體微缺失及微重複疾病
- ✓ 其他染色體非整倍體
- ✓ 額外發現
- ✓ 胎兒性別

NIFTY® twins

檢測選項：

- ✓ 三倍體綜合症（T21, T18, T13）
- ✓ 罕見常染色體非整倍體（T9, T16, T22）
- ✓ 92種染色體微缺失及微重複疾病
- ✓ 其他染色體非整倍體
- ✓ 額外發現
- ✓ Y染色體檢測

檢測內容

NIFTY® | NIFTY pro™ + 檢測內容

	染色體疾病	發病率	徵狀	檢測準確度
NIFTY BASIC	**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]
	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常帶有三條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，即使能夠出生大多數嬰兒都會在出生後一週內夭折。陶氏綜合症嬰兒有可能有心臟缺陷，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缺陷。	靈敏性：＞99.99%[2] 特異性：＞99.96%[2]
Nifty Pro / Nifty Pro+	**三倍體綜合症：**			
	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
	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。	
NIFTY BASIC	**性染色體異常綜合症：**			
	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%
	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色體異常疾病。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	單胎孕婦：檢測靈敏率	＞99%

[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

染色體微缺失及微重複疾病列表

	疾病	染色體位置		疾病	染色體位置
1	染色體1p36缺失綜合症	1p36	47	染色體10q26缺失綜合症	10q26
2	染色體1q41–q42缺失綜合症	1q41–q42	48	染色體10p12–p11缺失綜合症	10p11.21–p12.31
3	染色體1p32–p31缺失綜合症	1p32–p31	49	染色體10p重複	10p
4	染色體2p16.1–p15缺失綜合症	2p16.1–p15	50	染色體11p13缺失綜合	11p13
5	染色體2q33.1缺失綜合症	2q33.1	51	染色體11p11.2缺失綜合症	11p11.2
6	染色體2q31.1重複綜合症	2q31.1	52	Jacobsen綜合症	11q23–25
7	染色體2q37缺失綜合症	2q37	53	染色體11q23缺失綜合症	11q23
8	染色體2q31.1微缺失綜合症	2q31.1	54	染色體12q14微缺失綜合症	12q14
9	染色體2q重複	2q	55	染色體12p12.1微缺失綜合症	12p12.1
10	染色體3pter–p25缺失綜合症	3pter–p25	56	染色體12p重複	12p
11	Dandy–Walker綜合症	3q22–q24	57	染色體13q14缺失綜合症	13q14
12	染色體3q13.31缺失綜合症	3q13.31	58	染色體13q遠端缺失	13q32–qter
13	染色體3p遠端重複	3pter–p25	59	染色體14q11–q22缺失綜合症	14q11–q22
14	染色體3q重複	3q	60	染色體14q22缺失綜合症	14q22.1–q22.3
15	染色體4p16.3缺失綜合症	4p16.3	61	染色體14q近端缺失	cen–14q22
16	染色體4q21缺失綜合症	4q21	62	染色體14q重複	14q
17	染色體4p重複	4p	63	Angelman綜合症	15q11–q13
18	染色體4q遠端重複	4q21–q35	64	Prader–Willi綜合症	15q11–q13
19	染色體4q遠端缺失	4q31–qter	65	染色體15q26–qter缺失綜合症	15q26–qter
20	Cri–du–Chat綜合症	5p15	66	Levy–Shanske綜合症	15q26–qter
21	染色體5q14.3缺失綜合症	5q14.3	67	染色體15q14缺失綜合症	15q14
22	染色體5q12缺失綜合症	5q12	68	染色體15q24微缺失綜合症	15q24
23	染色體5p13重複綜合症	5p13	69	染色體15q26過度生長綜合症	15q26
24	染色體5p重複	5p	70	染色體15q遠端缺失	15q22–q26
25	染色體6pter–p24缺失綜合症	6pter–p24	71	染色體16p12.2–p11.2缺失綜合症	16p12.2–p11.2
26	染色體6q24–q25缺失綜合症	6q24–q25	72	染色體16p12.2–p11.2重複綜合症	16p12.2–p11.2
27	染色體6q11–q14缺失綜合症	6q11–q14	73	染色體16p13.3缺失綜合症	16p13.3
28	染色體6p缺失	6p	74	染色體16p13.3重複綜合症	16p13.3
29	染色體6q15–q23缺失綜合症	6q15–q23	75	染色體16q近端重複	16q11–q13
30	染色體6q25–qter缺失綜合症	6q25–qter	76	Smith–Magenis綜合症	17p11.2
31	染色體6q26–q27缺失綜合症	6q26–q27	77	染色體17p13.3缺失綜合症	17p13.3
32	染色體7q缺失	7q	78	Potocki–Lupski綜合症	17p11.2
33	染色體7q11.23缺失綜合症	7q11.23	79	染色體17p13.3重複綜合症	17p13.3
34	染色體7q21–q32缺失	7q21–q32	80	Yuan–Harel–Lupski綜合症	17p12–p11.2
35	染色體7q31–q32缺失	7q31–q32	81	染色體17p重複	17p
36	染色體8p23.1缺失綜合症	8p23.1	82	染色體18p缺失綜合症	18p
37	染色體8p23.1重複綜合症	8p23.1	83	染色體18q遠端缺失綜合症	18q22.3–q23
38	Langer–Giedion綜合症	8q23.3–q24.11	84	Alagille綜合症 I 型	20p12
39	染色體8q22.1缺失綜合症	8q22.1	85	染色體20p重複	20p
40	染色體8q22.1重複綜合症	8q22.1	86	染色體21q22缺失	21q22
41	染色體8p重複	8p	87	染色體22q11.2缺失綜合症	22q11.2
42	染色體8q重複	8q	88	染色體Xp11.23–p11.22重複綜合症	Xp11.23–p11.22
43	染色體9p缺失綜合症	9p	89	染色體Xp21缺失綜合症	Xp21
44	染色體9p重複	9p	90	染色體Xq27.3–q28重複綜合症	Xq27.3–q28
45	DiGeorge綜合症 II 型	10p14–p13	91	染色體Xq21缺失綜合症	Xq21
46	染色體10q22.3–q23.2缺失綜合症	10q22.3–q23.2	92	染色體Xq22.3缺失綜合症	Xq22.3

產前篩查技術比較

檢測	準確度	孕週	流產風險	假陽性率	報告週期（工作日）
血清學篩查ー早孕期篩查	80～90%	$11～13^{+6}$	0%	5%	1～2
血清學篩查ー中孕期篩查	60～90%	$16～19^{+6}$	0%	5%	1～2
胎兒頸部透明層檢查(NT)	60～80%	$11～13^{+6}$	0%	5%	1～2
羊膜穿刺	>99.9%	16～21	0.5～1%	＜1%	14～21
絨毛膜穿刺	>99.9%	11～13	1～2%	＜1%	14～21
臍帶靜脈穿刺	>99.9%	>20	1～2%	0%	5～7
NIFTY® NIFTY pro™ +	>99%	≥10	0%	＜1%（21號染色體三體症）	5

Nifty / Nifty Pro+ 讓您安心

懷孕對於每一位女士來說都是最激動人心的事情。當您在感受這份幸福的同時請為寶寶的健康做好準備。

Nifty / Nifty Pro+ 無創性胎兒染色體異常產前檢測技術，能夠為準媽媽們提供一種準確度高且無流產風險的唐氏綜合症及其他染色體疾病篩查方法。Nifty / Nifty Pro 只需要簡單抽血，即可有效地檢測寶寶的健康。

由於技術局限，以下孕婦並不能進行檢測：

1. 懷有三胞胎或以上的孕婦，不論之後是否減胎
2. 懷有雙胎，但超過八週後才減胎的孕婦
3. 距上次減胎時間不足8週的孕婦
4. 自身或配偶有染色體異常的孕婦
5. 懷有胎盤嵌合體寶寶的孕婦
6. 懷有羅氏易拉寶寶的孕婦
7. 實際孕週<10週

如受檢者有以下情況，請於進行檢測前與您的醫生相討有關安排：

1. 一年內接受過異體輸血的孕婦
2. 曾經接受過幹細胞治療或器官移植的孕婦
3. 最後一次接受抗體免疫治療注射後休息不足4週的孕婦
4. 父母任何一方有染色體異常qh+/–, ps+/–, pstk+/–, pss
5. 孕婦體重指數(BMI)>40
6. 懷孕期間或曾患有惡性腫瘤
7. 懷孕期間服用過特定藥物如抗凝血藥

*如孕婦對Nifty / Nifty Pro+的檢測內容及報告有任何問題，請向醫生諮詢詳情。
*當檢測結果為「高風險」時，孕婦應進行產前診斷。

Niftyhk
BGI www.hkbgi.com
如對Nifty / Nifty Pro+感興趣或有任何疑問，歡迎查詢：
Web: www.nifty.com.hk Tel: 3610 3525 Email: p_hkhealth@bgi.com

NIFTY® | NIFTY pro™+

NON-INVASIVE FETAL TRISOMY TEST

Peace of mind from a single drop

HK Patent Approval

- Detect chromosomal disorders across **23 pairs** of chromosomes
- Data size of each sample reaches **25M**, further increase thedetection rate of microdeletions/microduplications
- Available in more than **80 countries**
- Database: **Over 13.7 million cases worldwide**
- Accuracy: **>99%**
- Obtained Patent Registration Publication No. **HK1190758**
- Test performed in **Hong Kong Laboratory**

ISO 27001 SGS | bsi. BS 10012 Data Protection

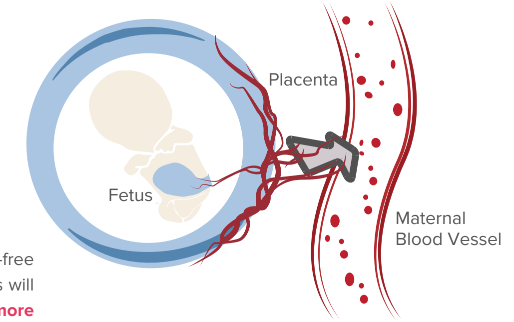
www.nifty.com.hk

Overview of Nifty / Nifty Pro+

Nifty / 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 / Nifty Pro+ requires taking the mother's peripheral blood to analyze cell free fetal DNA. Using new generation DNA sequencing along with bioinformatics to calculate the risk of having a fetus with chromosomal abnormalities.**

Nifty / Nifty Pro+ is a non-invasive prenatal screening test (or cell-free DNA screening) for pregnant persons to assess the risk that the fetus will be born with certain chromosomal abnormalities. **Well-trusted in more than 137,000,000 pregnancies globally, Nifty / Nifty Pro+ can assess conditions that may affect your baby's health by blood collection from you.** It screens for common chromosomal abnormalities, including trisomies, sex chromosome aneuploidies, and microdeletion / duplication syndromes, and can provide information about the gender of your babies. **Nifty / Nifty Pro+** is available for singleton, twin, egg donor pregnancies.

According to Current ACOG Guidance, cell-free DNA screening should be offered to all pregnant patients regardless of maternal age or the risk of chromosomal abnormality. Cell-free DNA is the most sensitive and specific screening test for common fetal aneuploidies[1].



CONDITION		SENSITIVITY	SPECIFICITY	REFERENCE
T21		99.17%	99.95%	Ultrasound Obstet Gynecol. 2015 May;45(5):530-8.
T18		98.24%	99.95%	
T13		100%	99.96%	
Del/Dup	>10Mb	88.89%	99.32%	PLoS One.2016 Jul 14; 11(7):e0159233.
	<10Mb	72.73%	99.09%	

CONDITION	SENSITIVITY	SPECIFICITY	PPV	REFERENCE
XO	75%	99.9%	23.53%	BMC medical genomics vol. 5 57. 1 Dec. 2012
XXX	N/A	N/A	70%	
XXY	100%	100%	75%	Chinese medical journal vol. 133,13 (2020): 1617-1619.
XYY	100%	100%	80%	

[1] The data in the table are based on historical documents and internal data. They only reflect past testing conditions and do not represent the actual conditions or promised values of the samples being tested.

- **Risk Free:** Doesn't increase miscarriage risk.
- **Highly Accurate:** Accuracy > 99.9%, performed more than 13,700,000 clinical samples worldwide.
- **All Inclusive Screening:** Includes the screening of 6 autosomal chromosome abnormalities, 4 sex chromosome aneuploidies, 92 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 across 23 pairs of chromosomes.
- **Assurance:** Tests are all examined in Hong Kong and reports are issued by Hong Kong MLT board registered MLT.
- **Early Pregnancy Test:** Test could be done as early as 10 weeks.

NIFTY®

Conditions screen for :

- ✓ Trisomies (T21, T18, T13)
- ✓ Sex chromosomal aneuploidies
- ✓ Fetal sex

NIFTY pro™+

Conditions screen for :

- ✓ Trisomies (T21, T18, T13)
- ✓ Rare autosomal aneuplodies (T9, T16, T22)
- ✓ Sex chromosomal aneuploidies
- ✓ 92 types of microdeletions/ duplications syndromes
- ✓ Other aneuploidies
- ✓ Incidental findings
- ✓ Fetal sex

NIFTY® twins

Conditions screen for :

- ✓ Trisomies (T21, T18, T13)
- ✓ Rare autosomal aneuplodies (T9, T16, T22)
- ✓ 92 types of microdeletions/ duplications syndromes
- ✓ Other aneuploidies
- ✓ Incidental findings
- ✓ Chr-Y testing

Test Content

NIFTY® | NIFTY pro™+ TEST CONTENT

Chromosome Abnormality	Incidence Rate	Clinical Features	Accuracy Rate
Trisomies (NIFTY BASIC)			
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]
Rare Autosomal Aneuploidies (only for singleton pregnancy) (Nifty Pro / Nifty Pro+)			
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%
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 Identification	NA	The singleton gender identification sensitivity rate.	>99%

[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

	Condition	Region		Condition	Region
1	Chromosome 1p36 deletion syndrome	1p36	47	Chromosome 10q26 deletion syndrome	10q26
2	Chromosome 1q41–q42 deletion syndrome	1q41–q42	48	Chromosome 10p12–p11 deletion syndrome	10p11.21–p12.31
3	Chromosome 1p32–p31 deletion syndrome	1p32–p31	49	Chromosome 10p duplication	10p
4	Chromosome 2p16.1–p15 deletion syndrome	2p16.1–p15	50	Chromosome 11p13 deletion syndrome	11p13
5	Chromosome 2q33.1 deletion syndrome	2q33.1	51	Chromosome 11p11.2 deletion syndrome	11p11.2
6	Chromosome 2q31.1 duplication syndrome	2q31.1	52	Jacobsen syndrome	11q23–25
7	Chromosome 2q37 deletion syndrome	2q37	53	Chromosome 11q23 deletion syndrome	11q23
8	Chromosome 2q31.1 microdeletion syndrome	2q31.1	54	Chromosome 12q14 microdeletion syndrome	12q14
9	Chromosome 2q duplication	2q	55	Chromosome 12p12.1 microdeletion syndrome	12p12.1
10	Chromosome 3pter–p25 deletion syndrome	3pter–p25	56	Chromosome 12p duplication	12p
11	Dandy–Walker syndrome	3q22–q24	57	Chromosome 13q14 deletion syndrome	13q14
12	Chromosome 3q13.31 deletion syndrome	3q13.31	58	Distal chromosome 13q deletion	13q32–qter
13	Distal chromosome 3p duplication	3pter–p25	59	Chromosome 14q11–q22 deletion syndrome	14q11–q22
14	Chromosome 3q duplication	3q	60	Chromosome 14q22 deletion syndrome	14q22.1–q22.3
15	Chromosome 4p16.3 deletion syndrome	4p16.3	61	Proximal chromosome 14q deletion	cen–14q22
16	Chromosome 4q21 deletion syndrome	4q21	62	Chromosome 14q duplication	14q
17	Chromosome 4p duplication	4p	63	Angelman syndrome	15q11–q13
18	Distal chromosome 4q duplication	4q21–q35	64	Prader–Willi syndrome	15q11–q13
19	Distal chromosome 4q deletion	4q31–qter	65	Chromosome 15q26–qter deletion syndrome	15q26–qter
20	Cri–du–Chat syndrome	5p15	66	Levy–Shanske syndrome	15q26–qter
21	Chromosome 5q14.3 deletion syndrome	5q14.3	67	Chromosome 15q14 deletion syndrome	15q14
22	Chromosome 5q12 deletion syndrome	5q12	68	Chromosome 15q24 microdeletion syndrome	15q24
23	Chromosome 5p13 duplication syndrome	5p13	69	Chromosome 15q26 overgrowth syndrome	15q26
24	Chromosome 5p duplication	5p	70	Distal chromosome 15q deletion	15q22–q26
25	Chromosome 6pter–p24 deletion syndrome	6pter–p24	71	Chromosome 16p12.2–p11.2 deletion syndrome	16p12.2–p11.2
26	Chromosome 6q24–q25 deletion syndrome	6q24–q25	72	Chromosome 16p12.2–p11.2 duplication syndrome	16p12.2–p11.2
27	Chromosome 6q11–q14 deletion syndrome	6q11–q14	73	Chromosome 16p13.3 deletion syndrome	16p13.3
28	Chromosome 6p deletion	6p	74	Chromosome 16p13.3 duplication syndrome	16p13.3
29	Chromosome 6q15–q23 deletion syndrome	6q15–q23	75	Proximal chromosome 16q duplication	16q11–q13
30	Chromosome 6q25–qter deletion syndrome	6q25–qter	76	Smith–Magenis syndrome	17p11.2
31	Chromosome 6q26–q27 deletion syndrome	6q26–q27	77	Chromosome 17p13.3 deletion syndrome	17p13.3
32	Chromosome 7q deletion	7q	78	Potocki–Lupski syndrome	17p11.2
33	Chromosome 7q11.23 deletion syndrome	7q11.23	79	Chromosome 17p13.3 duplication syndrome	17p13.3
34	Chromosome 7q21–q32 deletion	7q21–q32	80	Yuan–Harel–Lupski syndrome	17p12–p11.2
35	Chromosome 7q31–q32 deletion	7q31–q32	81	Chromosome 17p duplication	17p
36	Chromosome 8p23.1 deletion syndrome	8p23.1	82	Chromosome 18p deletion syndrome	18p
37	Chromosome 8p23.1 duplication syndrome	8p23.1	83	Distal chromosome 18q deletion syndrome	18q22.3–q23
38	Langer–Giedion syndrome	8q23.3–q24.11	84	Alagille syndrome 1	20p12
39	Chromosome 8q22.1 deletion syndrome	8q22.1	85	Chromosome 20p duplication	20p
40	Chromosome 8q22.1 duplication syndrome	8q22.1	86	Chromosome 21q22 deletion	21q22
41	Chromosome 8p duplication	8p	87	Chromosome 22q11.2 deletion syndrome	22q11.2
42	Chromosome 8q duplication	8q	88	Chromosome Xp11.23–p11.22 duplication syndrome	Xp11.23–p11.22
43	Chromosome 9p deletion syndrome	9p	89	Chromosome Xp21 deletion syndrome	Xp21
44	Chromosome 9p duplication	9p	90	Chromosome Xq27.3–q28 duplication syndrome	Xq27.3–q28
45	DiGeorge syndrome 2	10p14–p13	91	Chromosome Xq21 deletion syndrome	Xq21
46	Chromosome 10q22.3–q23.2 deletion syndrome	10q22.3–q23.2	92	Chromosome Xq22.3 deletion syndrome	Xq22.3

Comparison of Prenatal Screening Tests:

Test	Accuracy	Gestation	Miscarriage Risk	False Positive Rate	TAT (Working Days)
Maternal Serum Screening -1st trimester	80-90%	$11\text{-}13^{+6}$	0%	5%	1-2
Maternal Serum Screening -2nd trimester	60-90%	$16\text{-}19^{+6}$	0%	5%	1-2
Fetal Nuchal Translucency	60-80%	$11\text{-}13^{+6}$	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® / NIFTY pro™+	**>99%**	**≥10**	**0%**	**<1% (Trisomy 21)**	**5**

Nifty / 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.

Nifty / 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 receival 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 qh+/-, ps+/-, 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 / Nifty Pro+, please contact your doctor.

*If the test result is "**HIGH RISK**", it is recommended to perform prenatal diagnosis.

Niftyhk

BGI www.hkbgi.com

For further inquiries about Nifty Pro / Nifty Pro+, please contact us at:

Web: www.nifty.com.hk Tel: 3610 3525 Email: p_hkhealth@bgi.com