

HG Non-Invasive Prenatal DNA Testing (103 Types)

Special Price HK\$4,280

~~(Original Price HK\$7,000)~~



Only requires a blood sample



No Risk of Miscarriage



High accuracy > 99%



Test Contents:

1.	Trisomy Syndromes	Down Syndrome Trisomy 21 Edwards Syndrome Trisomy 18 Patau's Syndrome Trisomy 13
2.	Rare Autosomal Aneuploidies (RAAs)	Trisomy 9 Trisomy 16 Trisomy 22
3.	Sex Chromosome Aneuploidies (SCAs), 4 types (only for singleton pregnancy).	
4.	Microdeletions & Micoduplications, 92 types .	
5.	Incidental findings	
6.	Gender Identification (only for singleton pregnancy)	

Recommended for:

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| <ul style="list-style-type: none"> ✓ Pregnant women who are 10 weeks or more pregnancy. ✓ Pregnant women carrying Twins. ✓ Pregnant women who refuse invasive testing and are at risk of miscarriage. | <ul style="list-style-type: none"> ✓ Women undergoing in vitro fertilization or experiencing recurrent miscarriages. ✓ Pregnant women aged 35 and above. ✓ Pregnant women at high risk for Down Syndrome screening. |
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Not suitable for:

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| <ul style="list-style-type: none"> X Pregnancy <10 weeks pregnancy X Pregnant women carrying triplets or higher, regardless of subsequent reduction. X Pregnant women with chromosomal abnormalities themselves or in their partners. X Pregnant women during pregnancy or with a history with malignant tumors. X Patients who have received a blood transfusion within 1 year before the testing date. X Patients who have had stem cell therapy. X Pregnant women with a Body Mass Index (BMI) >40. X Pregnant women carrying a fetus with Robertsonian translocation. | <ul style="list-style-type: none"> X Maternal, fetal and/or placental mosaicism (mixtures of chromosomally normal and abnormal cells in pregnancy). X Patients who have had transplant surgery. X Vanishing twin syndrome: <ul style="list-style-type: none"> • 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 women should rest for less than 8 weeks. |
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What is Non-Invasive Prenatal DNA testing?

- **Non-invasive, simple, and advanced testing technique**
- **HG-NIPT is a non-invasive prenatal DNA testing technique** that detects chromosomal abnormalities in the fetus. The testing **only requires 10ml of maternal peripheral blood**. It analysis of cell-free fetal DNA by using next-generation DNA sequencing technology combined with bioinformatics to determine the risk of the fetus having a chromosomal abnormalities such as Down syndrome.

What is Non-Invasive Prenatal DNA testing?

- ✓ **Risk-free:** It only requires a blood sample from the pregnant woman, eliminating the risk of miscarriage.
- ✓ **High accuracy:** The testing **accuracy is > 99%**.
- ✓ **Large dataset:** **Over 1 million people** have undergone HG-NIPT testing.
- ✓ **Early detection:** Testing can be performed **as early as 10 weeks of pregnancy**, providing early peace of mind.



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Chromosome Abnormality	Incidence Rate	Clinical Features	Accuracy Rate
Trisomies			
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%
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%
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			
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, dysmorphism in the heart system, nervous system 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 .	Rare Sample Sensitivity rate not yet validated
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 .	Rare Sample Sensitivity rate not yet validated
Sex Chromosome Aneuploidies (only for singleton pregnancy)			
Gender Identification	NA	The singleton gender identification.	>99%
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 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%
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 as 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%



Chromosome Abnormality	Incidence Rate	Clinical Features	Accuracy Rate
XXX 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%

92 types condition list for microdeletions & microduplications

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