

隱性遺傳病基因篩查

擔心你下一代會有嚴重遺傳病？



全港
最全面

覆蓋高達 **700+**
疾病

針對最多700+種隱性遺傳病 | 準確率高達99.9%

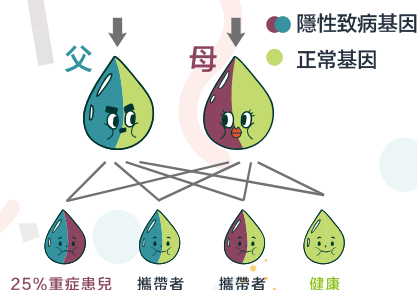
專業 | 全面 | 精確

每5人有2人
為隱性基因攜帶者

隱性基因疾病是如何遺傳的？

常染色體隱性疾病攜帶者父母個案

父母雙方是相同隱性疾病基因攜帶者



隱性遺傳病一般需要兩對隱性致病基因突變才會引致病症。如果只攜帶一個隱性致病基因，這種情況未必有症狀，而是“隱藏”於基因內。換句話說，父母雙方可能沒有症狀但如他們同時攜帶隱性致病基因，子女會有1/4的機率成為重症患兒。

NJ系列隱性遺傳病攜帶者篩查有什麼好處？

【做好家庭規劃】

1. 及早了解夫妻雙方隱性基因攜帶的狀況
2. 提示生育風險
3. 防止下一代患上可怕的遺傳病
4. 包含遺傳諮詢

NJ系列，**全港最全面**的隱性遺傳病攜帶者篩查，覆蓋高達**700+**遺傳病，您的醫生可以根據夫婦雙方的測試結果和家族遺傳病史，給予專業的建議。

適用對象

(建議夫妻雙方同時進行篩查)

- 準備結婚或計劃懷孕的夫婦
- 有家族性遺傳病史人士
- 有不明流產記錄的孕婦
- 近親通婚的夫婦
- 擔心自己攜帶隱性疾病基因
- 計劃進行人工受孕的夫婦
- 任何年齡人士希望了解相關疾病風險

- ✓ 針對隱性基因，預知潛在風險
- ✓ 最多700+種遺傳病

全面|優越組合

全港最全面	最精準	最可靠
<ul style="list-style-type: none"> 優秀組合：700+種 全面組合：400+種 提供專業報告解釋服務 	<ul style="list-style-type: none"> 使用市場獨家唯一“雙重驗證技術”以提升精準度 檢測準確率高達99.9% 	<ul style="list-style-type: none"> 香港首家專注基因檢測及擁有NGS設備的私營化驗所 100%香港本地化驗所，專業的教授和博士團隊

為您下一代檢測最多700+種遺傳病(包括以下常見類別)：

血液



- 常見疾病：
甲型及乙型地中海貧血
- 攜帶率：
1/12 (甲型) & 1/40 (乙型)

常見病徵

- 輕微貧血，容易勞累、虛弱
- 需要終生定期輸血
- 生長遲緩、骨頭畸形、肝脾腫大、黃疸、性腺功能低下

生殖



- 常見疾病：
先天性雙側輸精管發育不全
- 攜帶率：
1/4000

常見病徵

- 不育
- 輸精管發育異常
- 無精子症

心智發展



- 常見疾病：
脆性 X 綜合症
- 攜帶率：
1/1000 (男) 及 1/350 (女)

常見病徵

- 癲癇
- 結締組織發育不良
- 智力發展遲緩、自閉症

肌肉骨骼



- 常見疾病：
杜興氏肌肉營養不良症
- 攜帶率：
1/4900

常見病徵

- 肌肉萎縮
- 肺炎
- 心臟衰竭
- 青少年期已需倚助輪椅出入

代謝



- 常見疾病：
瓜氨酸血症
- 攜帶率：
1/42

常見病徵

- 痙攣
- 肌肉無力
- 胃口欠佳
- 呼吸急促



Carrier Risk Assessment

Are you worried that your children may inherit serious genetic diseases?



The **MOST** Comprehensive in Hong Kong

Covers up to **700+** diseases

Target up to 700+ Recessive Genetic Diseases
Accuracy up to 99.9%

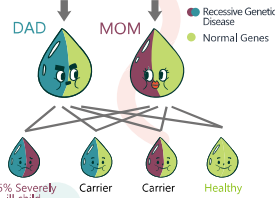
PROFESSIONAL | COMPREHENSIVE | ACCURATE

2 out of 5 people carry defective genes

How are recessive genetic diseases inherited?

Parents are autosomal recessive disease carriers

Both parents are carriers of the same defective gene



Recessive genetic diseases are caused by pathogenic gene mutations located on chromosomes; two copies of the same mutated gene from each parent may cause a disease. If only one defective gene is inherited in a recessive manner, the child may not have any symptoms.

In other words, If both patients are carriers of pathogenic gene mutations of the same recessive disease, they themselves may have no symptom at all, but the child has a 25% chance of being severely affected.

NJ Carrier Screening Test what are the benefits of this test?

Carrier Risk Assessment

1. Early understanding of you and your partner's carrier status
2. Assessment of fertility risks
3. Prevention of serious genetic diseases of your baby
4. Includes genetic counseling

NJ series, the most comprehensive carrier screening test in Hong Kong covers 400+ inherited diseases. Your doctor can give professional advice based on the test results of you and your partner, and your family genetic history.

Who is this test for?

(Couples are recommended to conduct the test at the same time)

- Couples who are planning to get pregnant
- Those with a personal or familial history of hereditary disease
- Couples with unexplained miscarriage history
- Consanguineous couples
- Those who want to know their carrier disease status
- Couples planning to have in vitro fertilisation (IVF) treatment
- People of any age want to know the risks of related diseases

Superior/
Prestige Panel

- ✓ Target recessive genes, predict potential risks
- ✓ Up to 700+ Genetic Diseases

The **MOST** Comprehensive in Hong Kong

The **MOST** Accurate

The **MOST** Reliable

Prestige Panel: 700+ diseases
Superior Panel: 400+ diseases

Provides professional genetic counselling

Use Market Unique "Dual Verification Technology" (DVT) to improve accuracy

Accuracy rate as high as 99.9%

The **FIRST** private laboratory in Hong Kong which focuses in genetic testing with own NGS facilities

100% Hong Kong local laboratory, with a professional team of Professors and Doctors

Detect up to 700+ inherited diseases, including the follow common categories:

Circulatory



- Common Disease : Alpha-thalassemia and Beta-thalassemia
- Carrier frequency : 1/12 (alpha) & 1/40 (beta)

Common symptoms

- Slight anemia, fatigue and weakness
- Requires blood transfusion throughout life
- Developmental delay, bone deformity, hepatosplenomegaly, jaundice, hypogonadism

Reproductive



- Common Disease : Congenital bilateral absence of vas deferens
- Carrier frequency : 1/4000

Common symptoms

- Infertility
- Van deferens not developed properly
- Azoospermia

Mental Development



- Common Disease : Fragile X Syndrome
- Carrier frequency : 1/1000 (Male) & 1/350 (Female)

Common symptoms

- Epilepsy
- Connective tissue dysplasia
- Intellectual disability, autism

Musculoskeletal



- Common Disease : Duchenne Muscular Dystrophy
- Carrier frequency : 1/4900

Common symptoms

- Muscles atrophy
- Pneumonia
- Heart failure
- Wheelchair-bound by adolescence

Metabolic



- Common Disease : Citrullinemia
- Carrier frequency : 1/42

Common symptoms

- Seizures
- Muscle weakness
- Poor appetite
- Rapid breathing

