
# 隱性遺傳病帶病者基因檢測

隱性遺傳病帶病者基因檢測為客戶提供有關患者未來兒童患隱性遺傳疾病風險的信息。隱性遺傳病帶病者基因檢測利用新一代檢測技術 (Next Generation Sequencing) ，為有需要人仕作一項全面的「孕前篩查」。它遵循美國婦產科學院「ACOG」及美國醫學遺傳學院(ACMG)的建議，篩查患者能夠檢查到超過170多種隱性遺傳疾病項目。此項檢測可了解自己是否有更大風險將遺傳性疾病傳給子女或未來兒童的人仕，或某些族群、遺傳疾病家族史中的人仕也可受益於此測試。通過隱性遺傳病帶病者基因檢測，父母可以了解將隱性遺傳疾病傳播給其子女的風險。如果父母雙方均進行測試，便可以進行更全面的評估。

## 助您了解隱性遺傳病

- 每個人都有兩份的遺傳基因資料，父母雙方各遺傳一份給孩子。
- 許多遺傳性疾病是隱性的，這意味著該疾病的發生一定要由父母雙方遺傳了相同的突變基因給孩子而引起的。
- 如果父母在基因其中一方發生突變，則他或她便是遺傳疾病的帶病者。
- 帶病者本身不會有任何徵狀，但如果配偶同時也是同一個基因的帶病者的話，那麼孩子便有四分之一的機會遺傳了這兩種突變，從而成為隱性遺傳病的發病者。

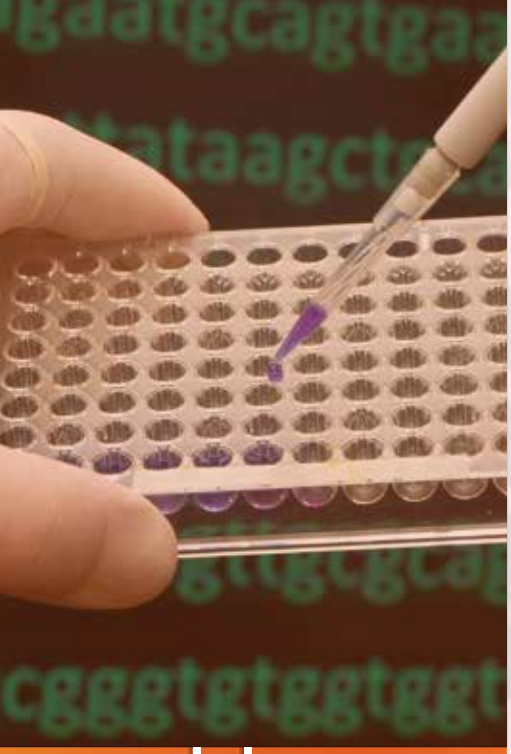
## 最常見隱性遺傳病一覽

### 脆性X綜合徵 (Fragile X Syndrome)

帶病率：女性1：151（一般人口）

脆性X綜合徵是導致一系列發育問題的遺傳病症，包括學習障礙和認知障礙。通常情況下，男性受到這種疾病的影響比女性更嚴重。如果引起疾病的突變基因位於X染色體上，這個情況便被形容為X連結的。由於男性只有一條X染色體，每個細胞中基因的X染色體(唯一拷貝)突變便會導致發病。

受影響的個體通常在2歲以前延遲發言和言語。大多數具有脆弱X綜合徵的男性具有輕度至中度的智力障礙，而大約三分之一的受影響的女性在智力上是有障礙的。脆弱X綜合症兒童也可能有焦慮和過度活躍，如躁動或衝動等。他們可能會出現注意力缺陷障礙（ADD），其中包括對注意力集中和特定任務困難的能力受損。約有三分之一的脆弱X綜合徵患者俱有自閉症譜系障礙的特徵，影響著溝通和社會互動。



## 脊髓性肌萎縮 (SMA)

帶病率 1:54- 72

脊髓性肌萎縮（SMA）是導致漸進性肌肉無力和癱瘓的遺傳性疾病。病發率為1/10,000，男女皆受影響。

SMA有三種類型。最嚴重的類型通常在出生的頭幾個月內診斷。受影響的兒童患有嚴重的肌肉無力，通常在2歲以前無法生存。

另外兩種不同的SMA類型肌肉無力程度較小。大多數患者需要使用輪椅或走路輔助器。不太嚴重的人群的預期壽命從青少年到成年。

## Pendred綜合徵

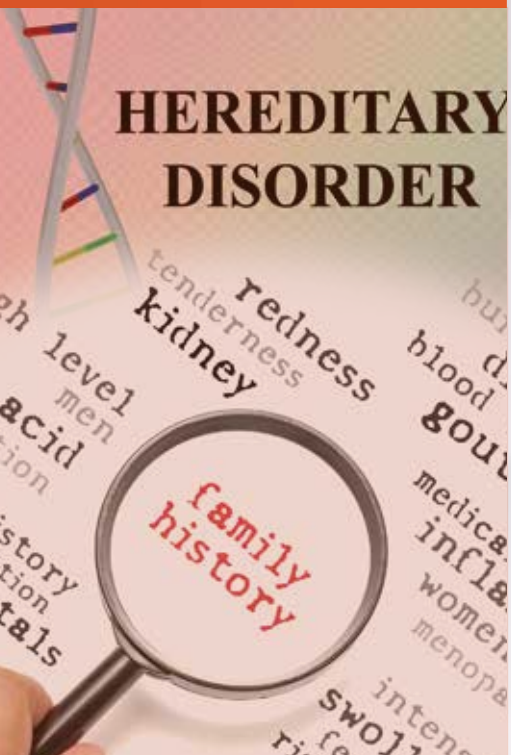
帶病率：1:50（中國人）

這是一種通常與聽力損失相關的疾病和稱為甲狀腺腫的甲狀腺疾病。如果一名甲狀腺腫發展成為Pendred綜合徵患者，則通常會在兒童期和成年早期之間形成。在大多數Pendred綜合徵患者中，在出生時便有明顯目內耳變化引起的嚴重聽力損失（感覺神經性聽力損失）

## 苯丙酮尿症（俗稱PKU）

帶病率：1:53（中國人）

苯丙酮尿症是一種遺傳性疾病，可以增加血液中稱為苯丙氨酸的物質的含量。苯丙酮尿症的症狀不同可從輕度到嚴重。這種疾病的最嚴重形式被稱為典型的PKU。具有典型PKU的嬰兒在幾個月大時不易察覺。若果沒有治療的話，這些孩子會發展成為永久智障。癲癇發作，發育延遲，行為問題和精神疾病也很常見。




## 史密斯 - 萊姆 - 奧皮茲綜合徵 (Smith-Lemli-Opitz syndrome)

帶病率：1：68（一般人口）

史密斯 - 萊姆 - 奧皮茲綜合徵是影響身體許多部位的發育障礙。這種情況的特點是面部特徵異常，頭部變小（小頭症），智力障礙或學習問題以及行為問題。許多受影響的兒童具有自閉症的特徵，影響孩子溝通和社會互動的能力。

Smith-Lemli-Opitz綜合徵的體徵和症狀差別很大。輕度受影響的個人可能只有輕微的身體異常與學習和行為問題。嚴重的病例可能危及生命，涉及深度的智障和重大身體異常。

## 威爾遜氏病 (Wilson's disease)

帶病率：1：90（亞洲）

威爾遜氏病是一種遺傳性疾病，由於會影響排銅異常，過多的銅會積聚在體內，特別是在肝，腦和眼的位置。威爾遜氏病的症狀通常與腦和肝有關。肝臟相關症狀包括嘔吐，虛弱，腹部液體積累，腿部腫脹，皮膚黃色和瘙癢。腦相關症狀包括震顫，肌肉僵硬，說話麻煩，人格變化，焦慮，以及看到或聽到其他人沒有的東西。大多數人會在5歲至35歲期間發病或被診斷，但也有機會影響年輕人和老年人。

## β-貧血症 (Beta -Thalassemia)

帶病率：1：100（中國人）

地中海貧血症包括各種遺傳性血液疾病，其中一些是相對溫和的，另一些可能導致嚴重的貧血症和其他嚴重問題。地中海貧血症的體徵和症狀主要出現在2歲前。兒童會發展至危及生命的貧血。孩子體重會減慢或甚至不會增加，或不能以預期的速度增長（未能發育），並可能會導致眼睛皮膚變黃（黃疸）。

## 隱性遺傳病帶病者全基因檢測項目 (>173 疾病)
## 4C Recessive Disease Carrier Status Full gene screening (>171 diseases)

SCAD Deficiency
17-beta-hydroxysteroid dehydrogenase X deficiency
2-methylbutyryl-CoA Dehydrogenase Deficiency
3-hydroxyacyl-CoA dehydrogenase deficiency
3-Methylcrotonyl-CoA carboxylase 1 deficiency (MCC1D)
3-Methylcrotonyl-CoA carboxylase 2 deficiency (MCC2D)
3-methylglutaconic aciduria type I (MCGA1)
3-methylglutaconic aciduria, type III
3-methylglutaconic aciduria, type V
Adrenoleukodystrophy Hb EE
Alpha-methylacetoacetic aciduria (3-ketothialase deficiency)
Argininemia (Arginase Deficiency)
Arginosuccinic Aciduria
Ault-onset citrullinemia Type II
Autosomal dominant deafness Type 3A
Autosomal dominant deafness Type IIB
Autosomal dominant deafness Type IIIB Autosomal dominant persistent hypermethioninemia due to methionine adenosyltransferase I/III deficiency
Autosomal recessive deafness
Autosomal recessive deafness Type 1A
Autosomal recessive deafness Type IB
Autosomal recessive deafness type IV
Autosomal recessive Methionine adenosyltransferase deficiency
Barth Syndrome
Bart-Pumphrey Syndrome
Beta thalassemia major
BH4-deficient Hyperphenylalaninemia A
BH4-deficient Hyperphenylalaninemia B BH4-deficient Hyperphenylalaninemia C
BH4-deficient Hyperphenylalaninemia D
Biotinidase deficiency
Bloom Syndrome
Canavan disease
Carnitine-acylcarnitine translocase (CACT) deficiency
cbl E complementation type homocystinuria-megaloblastic anemia
cbl G complementation type homocystinuria-megaloblastic anemia
cblB complement type Vitamin B-12 responsive methylmalonic aciduria (due to defect in synthesis of adenosylcobalamin)
cblD complement type homocystinuria (Variant 1)
cblD complement type homocystinuria (Variant 2)
cblD complement type Methylmalonic aciduria and homocystinuria
cblJ Type Methylmalonic aciduria and homocystinuria
Citrullinemia
Clouston type ectodermal dysplasia Type II Cogenital bilateral absence of the vas deferens (CVAD)
Combined malonic and methylmalonic aciduria
Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency
Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia
Congenital nongoitrous hypothyroidism 1
Congenital nongoitrous hypothyroidism 4
Congenital nongoitrous hypothyroidism 6
Hb Barts
Hb C disease (Hb CC)
Hb C/ Beta° thalassemia
Hb C/Beta$^{+}$ thalassemia
Hb D disease (Hb DD)
Hb D/ Beta° thalassemia
Hb D/Beta$^{+}$ thalassemia
Hb E/ Beta° thalassemia
Hb E/Beta$^{+}$ thalassemia
Hb H (3 gene deletion)
Hb H/Constant Spring disease
Hb S/ Beta° thalassemia
Hb S/Beta$^{+}$ thalassemia
Hb Variant/Beta° thalassemia
Hb Variant/Beta$^{+}$ thalassemia
Hb variants/Alpha thalassemia
Hemolytic anemia due to G6PD deficiency
Hepatic carnitine palmitoyl transferase deficiency Type I
Hepatic carnitine palmitoyl transferase deficiency Type II
Hereditary persistence of fetal hemoglobin
Hex A pseudodeficiency
HMG-CoA Lyase Deficiency
Holocarboxylase synthetase deficiency
Homocystinuria due to MTHFR deficiency
Homocystinuria, B6-responsive and nonresponsive types
Hyperhomocysteinemic thrombosis
Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase
Hystrix-like ichthyosis with deafness
Isobutyryl-CoA dehydrogenase deficiency
Isovaleric acidemia
Keratitis ichthyosis deafness syndrome
Krabbe disease
LCHAD deficiency
Lethal neonatal CPT2 deficiency
Malonyl-CoA decarboxylase deficiency
Maple syrup urine disease type II
Maple syrup urine disease, type Ia
Maple syrup urine disease, type Ib
MCAD Deficiency
Mental retardation X-linked syndromic 10 (MRXS10)
Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic aciduria due to Methylmalonyl-CoA Mutase deficiency
Methylmalonic aciduria due to transcobalamin receptor defect
Methylmalonyl-CoA epimerase deficiency
Mucolipidosis IV
Mucopolysaccharidosis Ih
Mucopolysaccharidosis Ih/s
Mucopolysaccharidosis Is
Neonatal hypertrypsinemia

## 隱性遺傳病帶病者全基因檢測項目 (>171疾病) (續...)
## Appendix 4C Recessive Disease Carrier Status Full gene screening (>171 diseases) (con't)

cblJ Type Methylmalonic aciduria and homocystinuria
Citrullinemia
Clouston type ectodermal dysplasia Type II Cogenital bilateral absence of the vas deferens (CVAD)
Combined malonic and methylmalonic aciduria
Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency
Congenital hypothyroidism due to thyroid dysgenesis or hypoplasia
Congenital nongoitrous hypothyroidism 1
Congenital nongoitrous hypothyroidism 4
Congenital nongoitrous hypothyroidism 6

Mental retardation X-linked syndromic 10 (MRXS10)
Methylmalonic aciduria and homocystinuria, cblC type
Methylmalonic aciduria due to Methylmalonyl-CoA Mutase deficiency
Methylmalonic aciduria due to transcobalamin receptor defect
Methylmalonyl-CoA epimerase deficiency
Mucolipidosis IV
Mucopolysaccharidosis Ih
Mucopolysaccharidosis Ih/s
Mucopolysaccharidosis Is
Neonatal hypertrypsinemia

CPT2 deficiency associated myopathy
Cystic Fibrosis
Digenic deafness GJB2/GJB3
Digenic GJB2/GJB6 deafness Dihydrolipoamide dehydrogenase deficiency DOPA-responsive dystonia (with or without hyperphenylalaninemia)
Erythrokeratodermia variabilis et progressiva
Fabry disease
Familial dilated cardiomyopathy Familial dysautonomia

Neonatal onset citrullinemia Type II
Niemann-Pick disease, type A
Niemann-Pick disease, type B
Niemann-Pick disease, type C1
Niemann-pick disease, type C2
Niemann-Pick disease, type D
Nonautoimmune hyperthyroidism
Non-classic hyperandrogenism due to 21-hydroxylase deficiency
Non-PKU hyperphenylalanemia
Optic atrophy 3 with cataract

Familial gestational hyperthyroidism
Familial hyperinsulinemic hypoglycemia type 4
Favism
Galactokinase deficiency with cataracts
Galactose epimerase deficiency Galactosemia
Gaucher disease Type I
Gaucher disease Type II
Gaucher disease Type III
Gaucher disease Type IIIC

Ornithine transcarbamylase deficiency
Palmoplantar keratoderma with deafness
Partial adenosine deaminase deficiency
Pendred syndrome
Perinatal lethal Gaucher disease
Phenylketonuria
Propionicacidemia
Severe combined immunodeficiency (SCID) due to adenosine deaminase deficiency (ADAD)
Sickle cell anemia (S/S)
Sickle cell disease variants

Glutaric acidemia IIA
Glutaric acidemia IIB
Glutaric acidemia IIC
Glutaric aciduria Type I
Glycine N-methyltransferase deficiency
Glycogen storage disease Ia Glycogen storage disease II
GM2-gangliosidosis
Hawkinsinuria

Sickle hemoglobin C disease
Sickle hemoglobin D disease
Sickle hemoglobin E disease
Susceptibility to acute-infection induced encephalopathy
Susceptibility to autoimmune thyroid disease Type III
Systemic primary carnitine deficiency
Tay-Sachs disease
Thryoid dyshormonogenesis 6
Thyroid dyshormonogenesis 1

Fragile X Syndrome
Spinal Muscular Atrophy
Vitamin B-12 responsive methylmalonic aciduria
VLCAD deficiency
Vohwinkel syndrome
X-linked mental retardation with methylmalonic acidemia and homocysteinemia
X-linked severe combined immunodeficiency (SCID)

Thyroid dyshormonogenesis 2A
Thyroid dyshormonogenesis 3
Thyroid hormone resistance
Transcobalamin II deficiency
Trifunctional protein deficiency
Tyrosinemia, type I
Tyrosinemia, type II
Tyrosinemia, type III

本檢測在經CLIA國際認証的美國基因實檢室進行，
檢測標準附合國際水平或以上。

Clinical Laboratory Improvement Amendments

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