6. 檢測可能得出的結果
Possible test results

表一 / Table 1

FMR1 CGG 重覆次數 Maternal repeat size	結果 Result	表現型 Phenotype	後代繼承全突變擴增的機會 Risk of full mutation expansion in next generation
< 45	正常 Normal	正常 Normal	0 %
45 - 54	中間 Intermediate	正常 Normal	0 %
55 - 200	前突變 Premutation	FXPOI 風險（女性） FXTAS 風險（女性及男性） Risk of FXPOI (females) Risk of FXTAS (females and males)	4 - 98 % (取決於重複次數) (depending on the repeat size)
> 200	全突變 Full mutation	FXS（男性） FXS（約50% 女性攜帶者） FXS (males) FXS (~50% female carriers)	–

7. 選擇是項檢測還需考慮的重點
Considerations in choosing this test

◊ 遺傳學檢驗可能會揭露您的敏感健康資訊。檢驗結果或顯示您的家人有較大可能攜帶致病性突變基因。以上情況可能造成心理負擔。

◊ Genetic testing may reveal sensitive information about your own health. Your results may indicate increased likelihood that other family members also carry disease mutations. The above may impose psychological distress.

8. 費用
Charge

Fragile X testing 脆性 X 檢驗	HK$1,100

The above service is provided by Mrs Wu Chung Prenatal Diagnostic Laboratory of Tsan Yuk Hospital.
以上服務由贊育醫院胡忠夫人產前診斷化驗室提供

Please ask your doctor if you have any questions about the information in this brochure.
如果您對本小冊子的內容有任何疑問，請詢問您的醫生。

Please visit
http://www.obsgyn.hku.hk/prenatal_diagnosis
for other information

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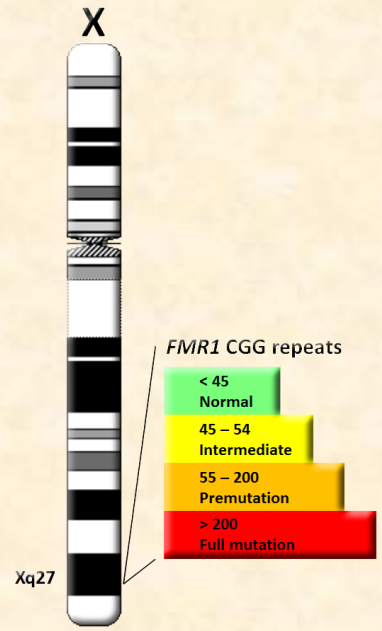
脆性 X 綜合症 (FXS) 與 脆性 X 相關疾病

Fragile X syndrome and Fragile X associated disorders



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1. 甚麼是脆性X綜合症(FXS)和脆性X相關疾病？
What are FXS and Fragile X associated disorders?

◊ 脆性X綜合症(FXS)和脆性X相關疾病是導致遺傳性智力障礙最常見的原因。它是由X染色體上*FMR1*基因的CGG重複次數增加而導致。男性發病率為1:4000，女性發病率則為1:5000-8000。

◊ 所有男性全突變基因攜帶者均會患上FXS，出現學習困難(智商低於70)、生長遲緩和行為問題，亦會有某些表徵，例如高額頭、大睾丸、大下巴和長耳朵。

◊ 約50%的女性全突變基因攜帶者會患上FXS，出現輕至中等程度的智力障礙，嚴重程度一般較男性患者低。約1/5女性前突變基因攜帶者會在40歲前停經(脆性X相關的原發性卵巢功能不全症，FXPOI)。

◊ Fragile X syndrome (FXS) and Fragile X associated disorders are the most common cause of inherited intellectual impairment. It is transmitted as an X-linked disorder caused by an increase in number of CGG repeats of the *FMR1* gene. The estimated incidence of FXS is 1 in 4000 males and 1 in 5000-8000 females.

◊ All male carriers with full mutation suffer from FXS. They present with learning difficulties (IQ less than 70), delayed milestones and behavioural problems, along with some physical features such as high forehead, large testicles, large jaw and long ears.

◊ About 50% female carriers with full mutation suffer from FXS, with mild to moderate mental disabilities, generally less severe than males. Around 1 in 5 female premutation carriers will have no periods before 40 years of age (Fragile X associated primary ovarian insufficiency, FXPOI).

◊ 至於脆性X相關的運動失調症候群(FXTAS)的患者，則會出現晚發型神經性紊亂，症狀包括顫抖、認知力下降或神經受損、柏金遜症和大小便失禁。

◊ 前突變基因攜帶者的後代亦有患上FXS的風險。由於風險高低取決於母親前突變的重覆次數(表一)，或需接受產前診斷。

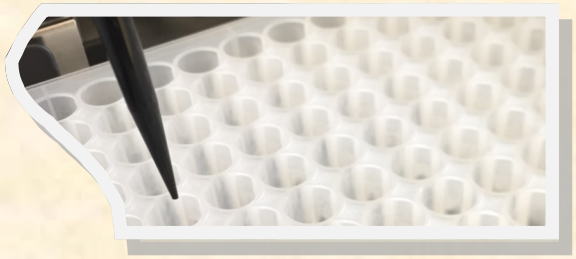
◊ In fragile X associated tremor / ataxia syndrome (FXTAS), there will be late onset neurodegenerative disorder including tremor, cognitive decline or nerve impairment, Parkinsonism, and urinary and bowel incontinence.

◊ Premutation carriers are also at risk of having FXS offspring. This risk of transmission is dependent on the size of the maternal premutation (Table 1). Thus, prenatal diagnosis may be required.

2. 我應該考慮接受檢驗嗎？
Should I consider having this test?

◊ 如家族成員曾患上 FXS 或脆性 X 相關疾病、原因不明的智力障礙、發育遲緩、自閉症或 40 歲前開始更年期，或經諮詢後擔心患上 FXS，您可考慮接受檢驗。

◊ If there is a family history of FXS or Fragile X associated disorders, unexplained mental retardation, developmental delay, autism or menopause before the age of 40; or if you are worried about FXS after counselling, you may consider testing for carrier status.



3. 需要檢驗甚麼樣本和所需時間
Types of sample and Turnaround time

◊ 3毫升血液樣本。 檢測結果約需2週。

◊ 3mL of blood sample. Test results will be available in 2 weeks.

4. 怎樣獲知結果？
How do I get to know the results?

◊ 檢驗結果將送交您的醫生，由他/她向您解釋。

◊ 如發現前突變或全突變，會轉介給臨床遺傳學科作進一步諮詢。

◊ The test result will be reported to your doctor, who will explain the results to you.

◊ Referral to clinical geneticist for further counselling shall be arranged if premutation or full mutation is found.

5. 檢驗有甚麼局限？
What are the limitations of the test?

◊ 檢測不能排除*FMR1*基因或基因組其它可能導致FXS的區域出現的點突變、缺失或嵌合。

◊ The test does not exclude point mutation, deletion, or mosaicism in the *FMR1* gene or other regions of the genome that cause fragile X syndrome.