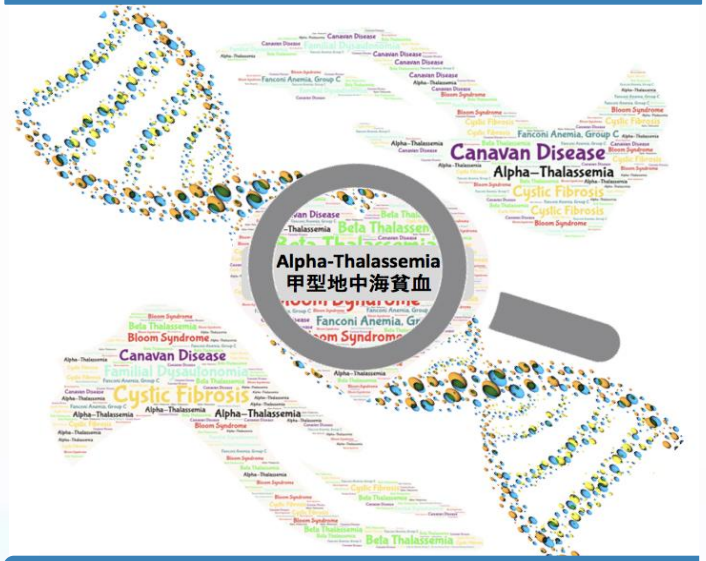
香港中文大學　婦產科學系

Department of Obstetrics and Gynaecology
The Chinese University of Hong Kong

擴展性攜帶者篩查
Expanded Carrier Screening (ECS)



什麼是擴展性攜帶者篩查？

攜帶者篩查是針對夫婦雙方是否攜帶某種遺傳病致病突變而進行的檢測。**擴展性攜帶者篩查**是一種基於二代測序技術，不限於種族和地域，可一次性檢測289種常見嚴重遺傳病的新服務（另可自選其他13種臨床表現多樣的遺傳病）。

Enquiries 查詢電郵及電話: obsgyn@cuhk.edu.hk

(852) 3505 1557 (general enquiries 一般查詢)

(852) 3505 4416 (appointments 預約診症)

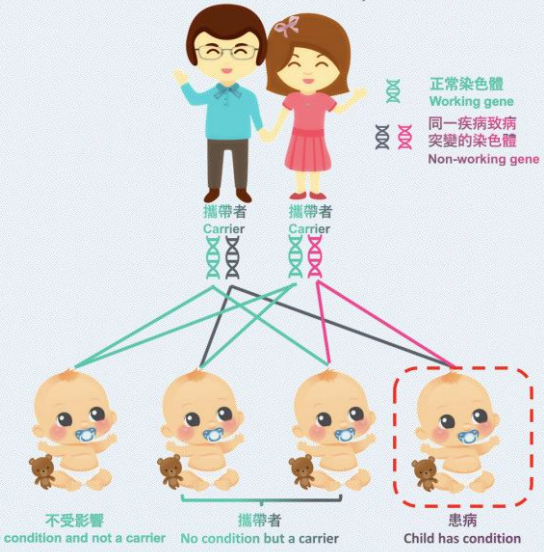
For more detailed information, please refer to your clinician or visit our website　如需獲取更多資料，可向您的專科醫生查詢，或請瀏覽我們的網頁:
http://www.obg.cuhk.edu.hk/_services/laboratory_service/ecs/

Printing material sponsored by (印刷材料贊助者):
Liauw's Family Reproductive Genomics Programme

什麼是隱性遺傳病攜帶者？

隱性遺傳病攜帶者是指某些人沒有或僅有輕微症狀，但是其一條染色體上具有致病突變，而另一條染色體正常。大多數隱性遺傳病攜帶者在篩查前並不知道自己是致病突變的攜帶者。

如果夫婦雙方均攜帶同一疾病的致病突變，那麼其後代將有25%的概率患該種疾病（X連鎖的隱性遺傳病除外）。



檢測流程

檢測前遺傳諮詢

↓

採集DNA樣本
（例如口水、外周血等）

↓

擴展性攜帶者篩查

↓

報告（10-21天）

選擇此項檢測的好處

- 若ECS的結果顯示夫婦雙方或其中一方未攜帶這些疾病的致病突變，則其寶寶罹患這些疾病的風險將大大降低；
- 若正在準備懷孕的夫婦均檢測出攜帶同一疾病的致病突變，夫婦可選擇在人工受孕相關技術的輔助下，降低寶寶的患病風險；
- 若在早孕期檢測出夫婦均攜帶同一疾病的致病突變，則可及早進行絨毛取樣或羊水穿刺，進一步明確胎兒是否受累，根據醫生建議在懷孕期間或寶寶出生後預早作出針對處理。

哪些人適用該項檢測？

ECS將為以下人群提供重要的指導建議：

- 選擇婚前或懷孕前檢查的（準）夫婦或早期孕婦；
- 欲通過人工輔助生殖技術生育健康寶寶的夫婦；
- 人工輔助生殖技術中的供卵者、供精者；
- 有不明原因不良生育史的夫婦；
- 血緣關係相近的夫婦。

選擇此項檢測的注意事項

- ECS只能檢測部分已知遺傳病相關的基因突變；
- ECS無法檢測每一種疾病相關的所有基因突變，也不能用於全部已知的遺傳疾病；
- 該項檢查只能降低寶寶患某些疾病的風險，但不能完全排除患病可能性，陰性結果並不能保證寶寶一定健康。

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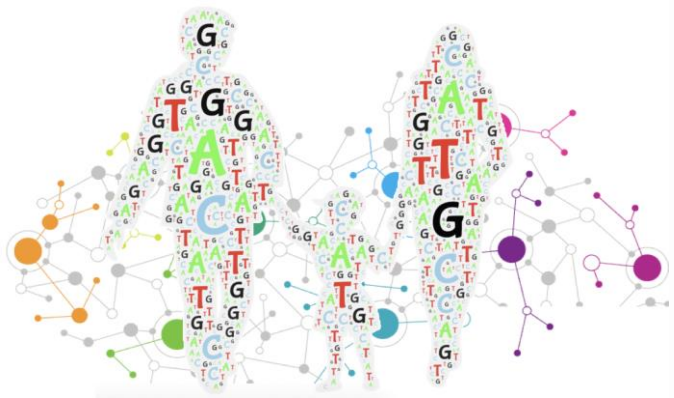

What is expanded carrier screening (ECS)?

Carrier screening is a type of genetic test which identifies whether you and/or your partner are carriers of a genetic disorder which can be passed on to your baby**. ECS** is based on next-generation sequencing technology and offers carrier screening for 289 genetic disorders (plus 13 add-on genetic disorders with variable presentation) using a single panel regardless of ancestry or geographic origin.

What is a carrier?

A **carrier** is a person who does not have symptoms or only has mild symptoms, but carries one copy of non-working gene for a genetic disorder. Most individuals do not know their carrier status prior to screening.

If you and your partner both are carriers of the same disorder, there is a **25% chance** that your baby will be affected with the disorder except for X-linked recessive disorders.

Genetic conditions recommended for screening by ACMG and ACOG

美國醫學遺傳學與基因組學協會、美國婦產科協會推薦篩查的疾病

Condition 疾病	Gene 檢測基因	ACMG[1]	ACOG[2]
Alpha-Thalassemia 甲型地中海貧血	*HBA1/HBA2*		Y
Beta Thalassemia 乙型地中海貧血	*HBB*		Y
Bloom Syndrome 布魯姆綜合徵	*BLM*	Y	
Canavan Disease 海綿狀腦白質營養不良	*ASPA*	Y	Y
Cystic Fibrosis 囊腫型纖維化症	*CFTR*	Y	Y
Familial Dysautonomia 家族性自主神經失調症	*IKBKAP*	Y	Y
Fanconi Anemia, Group C 范克尼貧血C型	*FANCC*	Y	
Gaucher Disease 高雪氏症	*GBA*	Y	
Mucolipidosis, Type IV 黏脂質症IV型	*MCOLN1*	Y	
Niemann-Pick Disease, Types A/B 尼曼匹克症 A/B型	*SMPD1*	Y	
Sickle Cell Disease 鐮狀細胞貧血	*HBB*		Y
Spinal Muscular Atrophy 脊髓性肌肉萎縮病	*SMN1*	Y	
Tay-Sachs Disease 黑蒙性家族癡呆症	*HEXA*	Y	Y

1 American College of Medical Genetics & Genomics. *Genet Med*. 2013 Jun;15(6):482-3.
2 American College of Obstetricians & Gynaecologists. www.acog.org/About-ACOG/ACOG-Departments/Genetics/Carrier-Screening

How is the test carried out?

Pre-test Genetic Counseling

↓

DNA sampling
(e.g. saliva or peripheral blood)

↓

Expanded Carrier Screening

↓

Report (within 10-21 calendar days)

Advantages of ECS

- For couples who are not carriers, ECS provides reassurance that their child will be at a significantly reduced risk of developing any of the genetic disorders covered by this test;
- If the test indicates both you and your partners are carriers of the same disorder(s) before pregnancy, you can choose assisted reproductive technologies to have a baby with a reduced risk of disorder(s).
- If the test indicates both you and your partner are carriers of the same disorder(s) during the early pregnant, you can pursue prenatal testing to determine whether your pregnancy is affected, and to ensure the appropriate management for your baby.

Who will benefit from this test?

People who (are):

- choose premarital screening, planning a pregnancy or in early pregnancy ;
- planning to bear a healthy baby by assisted reproductive technology;
- planning to donate eggs, sperm, or embryos;
- have a history of abnormal pregnancy for unknown reason;
- consanguineous marriage.

Important notes for this test

- ECS detects known disease-causing mutations on ECS panel;
- It is unable to detect all disease-causing mutations for each disease, nor for all known genetic disorders;
- It does not eliminate 100% of the risk of having an affected pregnancy. A normal test result does not guarantee a normal baby.