 瑪麗醫院 QUEEN MARY HOSPITAL	Department of Obstetrics and Gynaecology	Document No.	OGRM244d
		Issue Date	June 2025
	Subject Information on Expanded Carrier Screening-English	Next review date	April 2026
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Information on Expanded Carrier Screening

Indication: Couples who are planning for or in early pregnancy and have not done any genetic carrier screening before

Nature

Expanded carrier screening (ECS) is a genetic screening test designed to identify asymptomatic individuals who carry variants associated with various single-gene conditions, including autosomal recessive conditions (e.g. Thalassemia, Non-syndromic Hearing Loss, Congenital Adrenal Hyperplasia, Spinal Muscular Atrophy) and X-linked recessive conditions (e.g. Fragile X Syndrome, Haemophilia, Duchenne Muscular Dystrophy) (Figure 1).

ECS aims to provide supplementary genetic information to assess the risk of an affected child for couples and assist in making informed reproductive decisions.

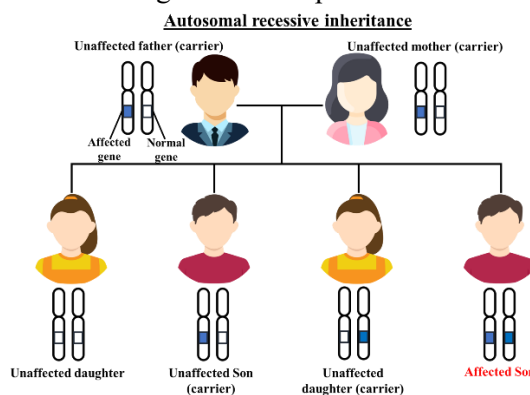


Figure 1a. Inheritance pattern of autosomal recessive conditions

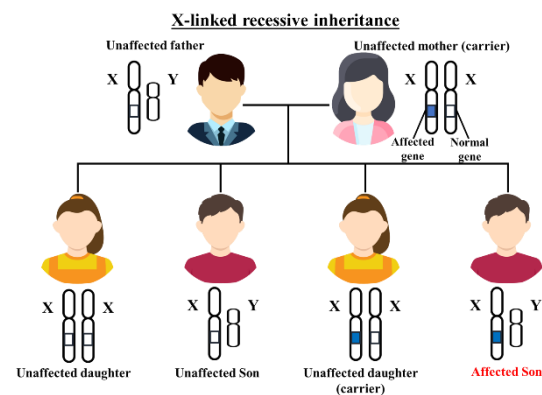


Figure 1b. Inheritance pattern of X-linked recessive conditions

Procedure

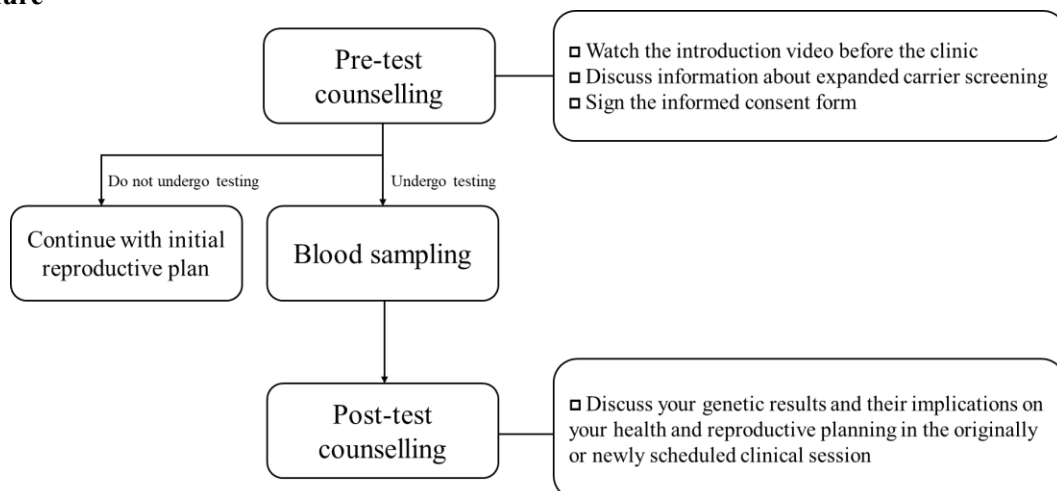



Figure 2: Flowchart of Procedures in Expanded Carrier Screening



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We recommend that the couple undergoes ECS together. Ideally, the test should be performed at least two months prior to any fertility treatment or natural conception to allow time for post-test counselling.

- a) Pre-test counselling:
 - You will watch an introduction video of ECS before the clinic.
 - The content, benefits, limitations, and potential outcomes of the test will be discussed at the clinic.
 - Informed consent forms will be signed.
- b) Sampling
 - You will follow the instructions and provide a blood sample at the clinic.
- c) Post-test counselling
 - The genetic report will normally be returned to the clinic within 3-4 weeks after the sampling.
 - The genetic results and their implications on your health and reproductive planning will be discussed in the originally or newly scheduled clinic session.

Genetic test options

We offer expanded carrier screening services from the HKUOG-Translational Research and Diagnostic Laboratory (HKUOG-TDL) (uniogtdl@hku.hk).

Test name	Uni-ECS	Uni-ECS Plus
Number of genes covered	453	708
Gene list		

Charging


1. Uni-ECS (single/couple) \$3,500 / \$6,500
2. Uni-ECS Plus (single/couple) \$4,500 / \$8,000
3. Processing fee for failure samples (single/couple) \$500 / \$1,000

Genetic tests done before

Have you or your family members done genetic testing before?

- ☐ Yes, and the result is _____.
- ☐ No.

Please ✓ in the appropriate box.

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I acknowledge that the above information concerning my test has been explained to and discussed with me by the medical staff and I fully understand them. I have been given the opportunities to ask questions pertinent to my condition and management, and satisfactory answers have been provided by the medical staff.

(Affix label)

Patient's name, ID number,
and date of birth

Patient's signature : _____

Date : _____