Information on Expanded Carrier Screening

Indication: Couples who are planning for 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).

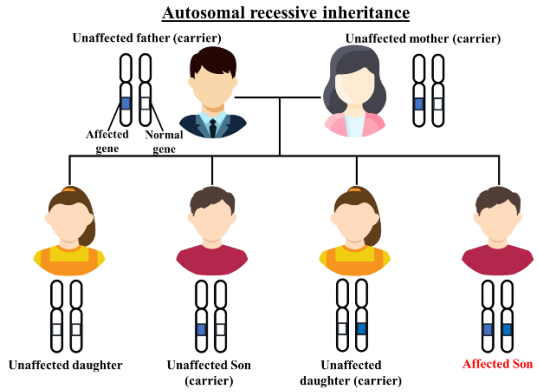

Figure 1a. Inheritance pattern of autosomal recessive conditions

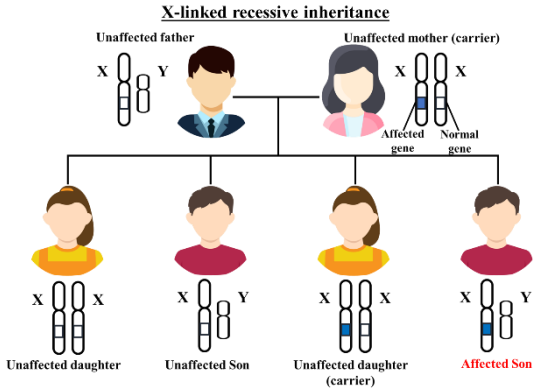

Figure 1b. Inheritance pattern of X-linked recessive conditions

ECS aims to provide supplementary genetic information to assess the risk of an affected child for couples and assist in making informed reproductive decisions For this purpose, the Invitae Comprehensive Carrier Screen (Test code: 60100, up to 569 genes, https://www.invitae.com/en/providers/test-catalog/test-60100) has been selected.

Procedure

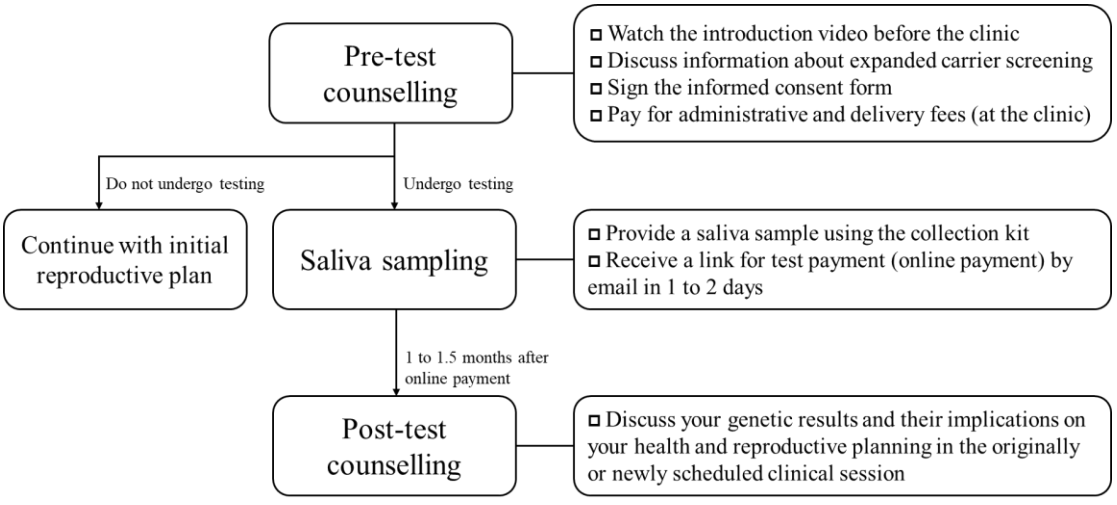

Figure 2: Flowchart of Procedures in Expanded Carrier Screening

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:
- The couple 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.
- Administrative and delivery fees will be paid at the clinic.

b) Sampling
- You will follow the instructions and provide a saliva sample using the collection kit (OrageneTM self-collection kit OGD510).
- *Please do not eat, drink, smoke or chew gum for 30 minutes before collecting the saliva sample.
- A link will be forwarded to your email address for test payment (online payment) in 1 to 2 days.

c) Post-test counselling
- The genetic report will be returned to the Centre within 4 to 6 weeks after the online payment.
- The genetic results and its implications on your health and reproductive planning will be discussed in the originally or newly scheduled clinical session.
  - Positive: the test reveals that you carry pathogenic/likely pathogenic genetic variant(s) which your child could inherit or increase the risk of a genetic condition for you.
  - Negative: the test does not identify a significant genetic variant which reduces, but not eliminate, your risk of being a carrier of the tested conditions.
- In some situations, the test results may alter your original reproductive plan.

Risk

The sampling procedure is non-invasive. Nevertheless, the awareness of genetic information can give rise to psychosocial consequences, including but not limited to psychological distress, family dynamics, insurance policies, stigmatisation, and alteration of initial treatment plans. In rare situations, certain variations with direct ramifications for your personal health may be detected.

Test accuracy and limitations
- The test achieves >99% analytical sensitivity and specificity for the DNA regions covered, including single nucleotide variants, insertions and deletions <15bp in length, and exon-level deletions and duplications in coding exons and +/-10 to 20 base pairs of adjacent intronic sequence of coding exons.
- Certain types of variants, such as structural rearrangements or variants embedded in sequence with complex architecture, may not be detected.
- Unless explicitly guaranteed, variants in the promoter, deep intronic regions, and other non-coding regions are not covered.
- Based on current literature and databases, the test result only reports pathogenic or likely pathogenic variants identified within the tested genes.

Alternative

You can choose to undergo ECS in other institutions, preferably utilising an accredited laboratory for conducting the test. Alternatively, you can choose not to undergo ECS and continue with your initial reproductive plan without knowing further genetic information.

Charging

1. Administration fee per person (at the clinic) HKD 500
2. Sample delivery fee per person (at the clinic) HKD 500
3. Sample delivery fee per couple (at the clinic) HKD 600
4. Invitae Comprehensive Carrier Screen per person (Online payment) USD 299
5. Invitae Comprehensive Carrier Screen per couple (Online payment) USD 524

Genetic test done before

Have you or your family members done genetic testing before?

❑ Yes, and the result is _____.

❑ No.

Please ✓ in the appropriate box.

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

(Affix label) Wife's name & ID number	(Affix label) Husband's name & ID number

Signature of Wife:_____ Signature of Husband: _____

Date:_____