FAQ

Q: **When should I undergo Expanded Carrier Screening (ECS)?**

A: ECS is recommended to be performed before pregnancy or in early pregnancy, allowing couples to make informed decisions regarding family planning.

Q: **If my test results are negative, does that mean my baby is free from all genetic disorders?**

A: ECS can only detect certain genetic mutations associated with known hereditary diseases. It does not detect all genetic disorders, nor does it encompass all known hereditary diseases.

This test cannot completely rule out the possibility of your baby being affected by genetic disorders; it can only reduce the risk of passing on specific genetic disorders to embryos. Therefore, it cannot guarantee that your baby will be completely healthy.

Q: **What are the options if my partner and I are carriers of a genetic disorder?**

A:
- Conceive naturally and perform a prenatal diagnostic test to determine if the fetus has the disorder.
- Consider using assisted reproductive technologies (e.g. IVF and PGT) to reduce the risk of having a baby with the disorder.
- Choose not to conceive or consider adoption.

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Expanded Carrier Screening (ECS)

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What is Expanded Carrier Screening (ECS)?

ECS Procedure:

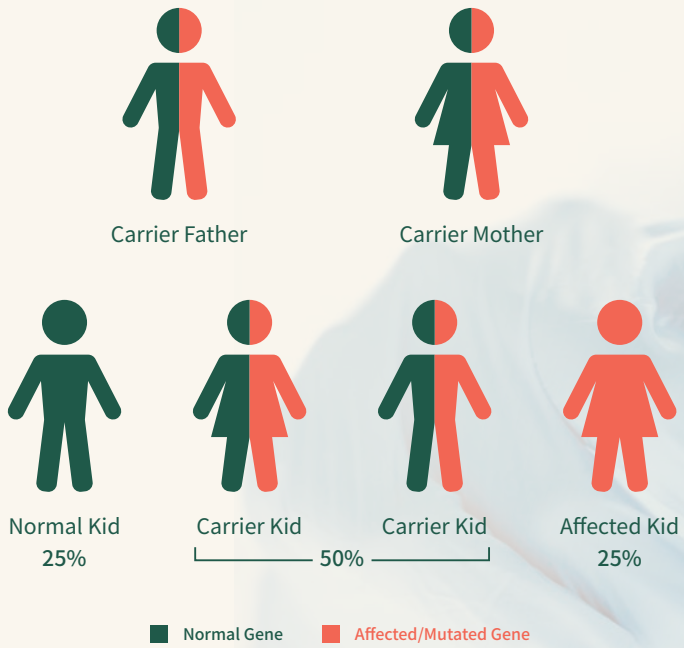
1. Pre-test Genetic Counselling
2. Blood Sampling
3. Samples sent to the lab for Expanded Carrier Screening
4. Report available (around 3-4 weeks)

Expanded Carrier Screening (ECS)

Carrier screening is a type of genetic test designed to identify whether you and/or your partner are carriers of autosomal or X-linked recessive genetic disorders that can be passed on to your next generation. A single blood sample can test for over 300 genetic disorders. ECS provides couples with additional genetic information and assesses the risk of passing on single-gene disorders to future generations.

What is a Carrier?

A carrier is an individual who possesses an affected or mutated gene without exhibiting any symptoms or who presents only mild symptoms. If both partners are carriers of the same disorder, there is a 25% chance that their baby will be affected by that disorder, except for X-linked recessive disorders.



Carriers are generally not aware of their carrier status, as they usually do not present with any symptoms or just present with mild symptoms.

Who Should Consider ECS?

1. Couples planning for a pregnancy or in early pregnancy who have never undergone genetic carrier screening
2. Couples considering conception through assisted reproductive technology
3. Egg and sperm donors for couples facing reproductive challenges
4. Patients with a history of recurrent miscarriages
5. Patients with a history of abnormal pregnancies
6. Couples who are biologically related
7. Patients with a family history of genetic disorders

Which Genetic Diseases Can Be Detected by the Screening?

The American College of Medical Genetics (ACMG) and the American College of Obstetricians and Gynecologists (ACOG) recommend that all women considering pregnancy or who are already pregnant undergo carrier screening for specific genetic disorders, such as spinal muscular atrophy (SMA), cystic fibrosis (CF), thalassemia, haemoglobinopathies, and fragile X premutation etc. Additional screening may be indicated based on family history or specific ethnic backgrounds. Please consult your doctor for more details.