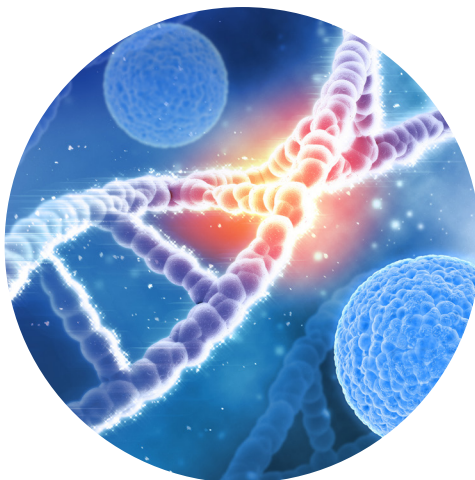




Reproductive Carrier Screening

What is genetic carrier screening?





Carrier screening is a genetic test to determine if a couple is at risk of having a child with a serious genetic condition that can significantly impact his or her quality of life or lifespan. It involves testing you and/or your partner to assess if either of you is a carrier for the tested genetic condition(s).

Most couples may not know if they carry a genetic condition until they have an affected child. This is because carriers are typically healthy and have no symptoms. Couples who are biological relatives (consanguineous) have a higher risk of being genetic carriers for the same condition. However, couples who are not related can still be at risk, with studies showing that 1-2% of these couples can have a child affected by a genetic condition.

For many couples, knowing their carrier status in advance helps them make informed reproductive decisions that can improve their chances of having a healthy child.



Who is suitable for carrier screening?

All couples planning to have children can consider carrier screening.

When should you get tested?

Carrier screening is ideally performed before conception, as this gives you the most time and reproductive options to make informed decisions. Testing can also be done in early pregnancy.

What kinds of carrier testing are there and how is it done?

Reproductive carrier screening is done with a simple blood test that can be performed on an individual basis or as a couple.

There are different types of carrier tests available:

+ Targeted screening

If you belong to certain ethnic groups that have higher risks for specific genetic conditions, we can test for those particular conditions that are most relevant to your background.

+ Expanded carrier screening

This screening tests for genetic changes in several hundred genes associated with early-onset genetic disorders. This test is useful for couples who have no known history of genetic conditions, regardless of ethnicity.

+ Comprehensive testing

These are more detailed tests using advanced technologies like Whole Genome Sequencing and Optical Genome Mapping. They analyse your entire DNA to identify a much broader range of conditions that you might be a carrier for.

If you or your family have a history of a genetic disorder, our genetic specialist will discuss and provide recommendations on the best genetic testing options for you.





How does a baby inherit a genetic condition from healthy parents?

A genetic condition may arise when there are changes to the genetic code of a gene that is associated with disease. Each person carries two copies of every gene in their cells. An individual who carries one non-working gene may be healthy if the other copy of the gene is functioning. However, if both parents are carriers for the same condition, there is a chance that their baby may inherit the non-working copy from each parent, which can lead to a genetic disorder.

There are different ways a genetic condition can be passed down or inherited in families. Carrier testing mainly tests for autosomal recessive and X-linked conditions.

Autosomal recessive (AR)

For AR conditions, an affected child inherits 2 copies of the non-working gene, one from each carrier parent. There is a 1 in 4 chance of the child being affected if both parents are carriers. An example of such a disorder is alpha or beta thalassaemia.

X-linked

X-linked conditions are caused by mutations in genes on the X chromosome.

- **Males** have one X and one Y chromosome. If a male carries a non-working gene on his X chromosome, he will usually be affected by the condition because he does not have a second X chromosome to compensate.
- **Females** have two X chromosomes. If a female carries a non-working gene on one X chromosome, she is usually healthy because her other X chromosome can compensate. However, she can still pass the condition to her children.

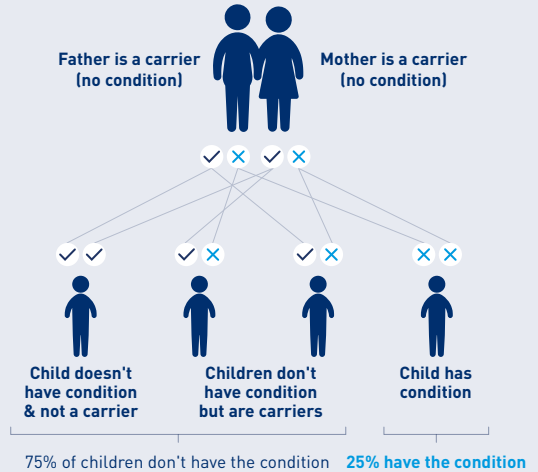
When a carrier mother has children with an unaffected father, with each pregnancy, there's a:

- 1 in 2 chance of having an affected son
- 1 in 2 chance of having a daughter who is a carrier

An example of this condition is Duchenne Muscular Dystrophy.

Autosomal Recessive Inheritance Pattern

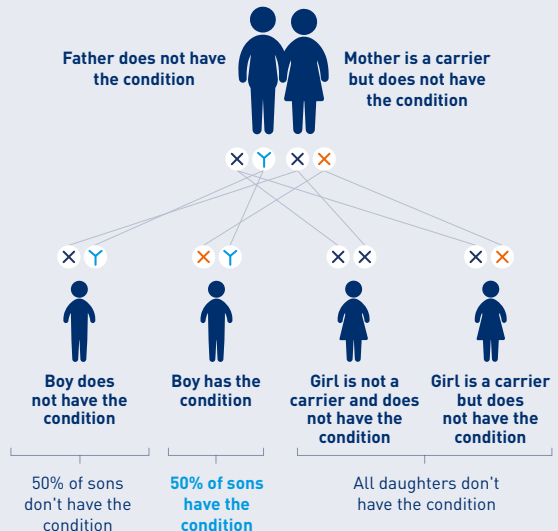
- ✓ Working gene
- ✗ Non-working gene



X-Linked Recessive Inheritance Pattern

Mother is a carrier of the condition

- ✗ X chromosome with working gene
- ✗ X chromosome with non-working gene



Do I still need to do aneuploidy screening once pregnant?

(e.g. Down's syndrome screening)

Yes. Carrier screening does not check for extra or missing chromosomes that may occur in the child.

What are the possible outcomes of carrier testing?

There are two outcomes:

- + You are a genetic carrier for a condition. Your reproductive risks may depend on whether your partner is also a genetic carrier for the same condition.
- + You are not a genetic carrier for the conditions tested. This does not mean that the chance of having a child affected with a genetic condition at birth is zero, as not all genetic conditions are inherited.

Rarely, it could be possible for you to be identified with personal risk for a genetic disorder. Your genetic counsellor or genetic specialist will discuss this risk with you before proceeding with the test.

What happens if I am a carrier for a genetic condition?

Learning that you're a carrier can feel overwhelming, but it's important to remember that being a carrier simply means you have more information to help you make informed decisions about your family planning.

If your carrier screening results indicate an increased risk for a genetic condition in your future children, your genetic counsellor will explain the specific risks based on the inheritance pattern involved.

We strongly recommend genetic counselling prior to pregnancy to explore your available options such as:

- Conceiving naturally and doing prenatal genetic testing for the condition
- In Vitro Fertilisation (IVF) and preimplantation genetic testing (PGT) to select embryos unaffected by the genetic condition that both partners carry
- IVF with donor sperm or eggs
- Conceiving naturally and evaluating the child for the condition after birth
- Exploring other family-building options such as adoption



What is the cost of carrier testing?

The costs of carrier screening vary depending on the test you choose. For example:

- + A karyotype that checks the number and structures of chromosomes costs approximately \$800 SGD.
- + Carrier screening tests can range from \$800 SGD to \$5000 SGD depending on the test you choose. Your doctor will go through the options in detail with you.

How do I arrange for carrier screening if I am interested?

+ **Speak to your doctor**

If you decide to proceed with carrier screening, a blood test can be done on the same day. It will take approximately 4 weeks for the results to be ready.

+ **Meet with a genetic counsellor or genetic specialist**

You will receive an appointment with a genetic counsellor or genetic specialist who will walk you through your genetic report and explain what the results mean. Your Obstetrics and Gynaecology (O&G) doctor will address your specific reproductive risks and help you understand your options going forward.

Alternatively, you may arrange an appointment with the O&G clinic by calling +65 6908 2222.

About the National University Centre for Women and Children

The National University Centre for Women and Children (NUWoC) empowers women, children, and families to lead healthier lives. It brings together the [Department of Obstetrics & Gynaecology \(O&G\)](#) and [Khoo Teck Puat – National University Children’s Medical Institute \(KTP-NUCMI\)](#) at National University Hospital, working alongside [National University Polyclinics \(NUP\)](#) to strengthen continuity of care across the community.

Supported by the National University Health System (NUHS), NUWoC delivers comprehensive medical and surgical services across the life course – from pre-conception through maternal, child, adolescent, and women’s health.

Through a generous gift from the Estate of Khoo Teck Puat, KTP-NUCMI has established an integrated outpatient facility offering medical, diagnostic, and rehabilitation services for children. NUWoC is Singapore’s only public specialist centre providing paediatric kidney and liver transplant programmes.

For more information about us, visit www.nuh.com.sg/NUWoC

Emergency (24-hr)

Location NUH Main Building, Zone F, Level 1

Women’s Clinic – Emerald/Ruby

Location NUH Kent Ridge Wing, Zone D, Level 3, D03-06
Operating Hours 8.30am – 6pm (Mon to Thu), 8.30am – 5.30pm (Fri), 8.30am – 12.30pm (Sat)
Email appointment@nuhs.edu.sg

Women’s Clinic – Sapphire

Location NUH Kent Ridge Wing, Zone D, Level 3, D03-03
Operating Hours 8.30am – 6pm (Mon to Thu), 8.30am – 5.30pm (Fri), 8.30am – 12.30pm (Sat)
Email appointment@nuhs.edu.sg

Women’s Clinic – Jade [Former Clinic G]

Location NUH Kent Ridge Wing, Zone C, Level 3, C03-02
Operating Hours 8.30am – 6pm (Mon to Thu), 8.30am – 5.30pm (Fri)
Email appointment@nuhs.edu.sg

Fetal Care Centre

Location NUH Kent Ridge Wing, Zone D, Level 3, D03-04
Operating Hours 8am – 5.30pm (Mon to Thu), 8am – 5pm (Fri)
Email appointment@nuhs.edu.sg

Clinic for Human Reproduction

Location NUH Kent Ridge Wing, Zone D, Level 4, D04-02
Operating Hours 8am – 5pm (Mon to Fri), 8.30am – 12.30pm (Sat)

NUWoC Women’s Clinic @ JMC

Location Jurong Medical Centre, Level 2
Operating Hours 8.50am – 11.30pm (Tue & Thu), 2pm – 5pm (Mon & Fri)

Jurong Clinic for Women

Location 130 Jurong Gateway, #01-231
Operating Hours 9am – 12pm, 2pm – 5pm (Mon to Sat), 6pm (Mon to Thu)
General Enquiry +65 6665 4277
Appointment Line +65 6908 2222
Email appointment@nuhs.edu.sg

NUWoC Women’s Clinic @ NTFGH

Location Ng Teng Fong General Hospital, Tower A – Specialist Outpatient Clinics, Level 7
Operating Hours 8.30am – 5.30pm (Mon to Fri), 8.30am – 12.30pm (Sat)
Appointment Line +65 6908 2222
Email appointment@nuhs.edu.sg

Our Patient Care Institutions

National University Hospital

Ng Teng Fong General Hospital

Alexandra Hospital

Tengah General and Community Hospital (upcoming)

Jurong Community Hospital

National University Polyclinics

Jurong Medical Centre

National University Cancer Institute, Singapore

National University Heart Centre, Singapore

National University Centre for Oral Health, Singapore

NUHS Diagnostics

NUHS Pharmacy



Scan the QR code for more information on our patient care institutions.

OneNUHS Hotline: (65) 6908 2222

OneNUHS General Enquiries: contactus@nuhs.edu.sg

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