The Royal Australian and New Zealand College of Obstetricians and Gynaecologists
Excellence in Women's Health

Reproductive Carrier Screening

Genetic screening options for healthy couples who are planning a pregnancy, or who are in the early stages of pregnancy, are becoming more available.

Inherited genetic conditions

There are hundreds of inherited genetic conditions that can affect human health, and most are very rare. However, when all of these inherited conditions are considered together, they affect up to 1 in 400 people. Most couples who have an affected child have no family history of the condition and were not aware they had an increased chance of having a child with the condition. This occurs because a healthy couple can pass on genetic changes to their child without knowing they are carriers of that condition. Therefore, carrier screening is relevant to everyone regardless of whether or not they have a family history of a genetic condition.

What screening is currently available for genetic conditions?

The newborn screening programs in Australia and New Zealand offer screening of all newborns for a range of genetic conditions using the "heelprick test". This is a voluntary, government-funded test that does not require any payment. The majority of parents choose to have this screening for their baby.

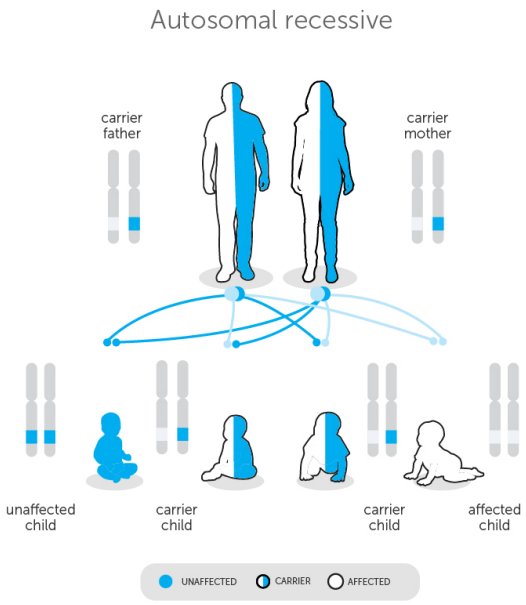
Screening can also be performed on adults to see if they are at increased chance of having a child with a genetic condition. When a healthy couple or individual have screening to see if there is a chance of passing a genetic condition to their children, this is called "reproductive carrier screening". This is usually not government funded unless there is a family history of the condition.

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

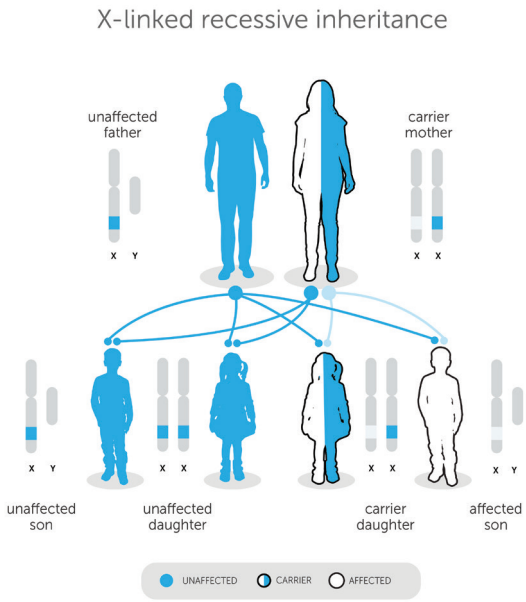
There are two major types of inheritance that can lead to a healthy couple having a child with a serious genetic condition. These are referred to as autosomal recessive and X-linked recessive inheritance.

Autosomal recessive conditions

For autosomal recessive conditions, a person only develops the disease if they inherit the same faulty gene from each parent. In this case, each parent has one faulty gene and one healthy or functioning gene, they do not have the condition; but are healthy "carriers" of the condition. If both members of a couple are carriers of the same faulty gene there is a 1 in 4 chance of having a child affected by that condition. The most common autosomal recessive conditions in our community are thalassemia and cystic fibrosis.



X-linked conditions



X-linked conditions occur when the faulty gene is on the X chromosome. Males have an X and a Y chromosome while females have two X chromosomes. Since males have only one X chromosome, if there is a faulty gene on their X chromosome they are more severely affected by the condition since they do not have a second normal X chromosome to compensate.

If a woman is a carrier for an X-linked condition, there is a 1 in 2 chance of having an affected son and 1 in 2 chance of the daughter being a carrier.

The most common X-linked condition is fragile X syndrome. For fragile X, female carriers have up to a 50% chance of having a child with fragile X syndrome. Both males and females can have fragile X syndrome.

What should we do if we have a family member with a genetic condition?

If you or your partner have a relative with a genetic condition, you may have an increased chance of having a child with that genetic condition. Examples of inherited genetic conditions include thalassemia, cystic fibrosis, fragile X syndrome, spinal muscular atrophy, and haemophilia. Some genetic conditions occur more frequently in certain ethnic groups. If you have a relative with a genetic condition, you should discuss this with your family doctor (general practitioner (GP)). Your GP can refer you to a genetic counsellor or medical geneticist for further advice and testing if needed.

We don't have a genetic family condition. Is there a risk?

Carrier screening is relevant to all people planning a pregnancy or in early pregnancy. Most people who are a carriers of a genetic condition/s do not have a family history of a genetic condition/s. This is because carriers are generally healthy and because usually both members of the couple need to carry the same condition in order to have an increased chance of having a child with that condition. This means these conditions can be passed down through families for many generations before a person is affected by the condition.

How often do these genetic conditions occur?

The chance of a child being born with a genetic condition varies depending on the ethnicity of the population. The numbers of carriers and affected individuals for the more common conditions in a Caucasian population are listed below. As technology improves and people are having screening for a large number of conditions, it is becoming clear that most people are carriers for one or more inherited conditions.

	Number of people who are carriers	Number of people with the conditions
Cystic fibrosis	1 in 25	1 in 2,500
Fragile X syndrome	1 in 250	1 in 4,000
Spinal muscular atrophy	1 in 40	1 in 6,000 - 1 in 10,000

When should I have screening?

Carrier screening can be performed at any time, but it is preferable to screen before pregnancy so that prospective parents have time to consider their reproductive options.

What are the costs?

The cost of testing for three of the most common genetic conditions - cystic fibrosis, spinal muscular atrophy, and fragile X syndrome - is currently out of pocket. At the moment there is no rebate for these tests.

How do I access screening?

A range of carrier screening options are available. These generally fall into two groups:

- Screening for a small number of common inherited conditions (such as cystic fibrosis, fragile X syndrome and spinal muscular atrophy)
- Screening the common inherited conditions as well as a large number of rare conditions

If you are considering carrier screening, speak to your GP, obstetrician or midwife. They can discuss your options with you and may refer you to a genetic counsellor. Some genetic testing laboratories and clinical genetics services offer genetic counselling for people considering carrier screening.

What can we do if we have an increased chance of having a child with a genetic condition?

If you and your partner are carriers of the same genetic condition or the female partner is a carrier of an X-linked condition, then you should seek genetic counselling prior to getting pregnant. This will give you time to consider all the options available to you, including:

- Getting pregnant naturally and having the baby tested after birth
- Getting pregnant naturally and having diagnostic testing during pregnancy, with the option of considering an abortion if the baby will be affected
- Having in vitro fertilization (IVF) and preimplantation genetic testing (PGT) in order to selected unaffected embryos to get pregnant
- Using IVF and sperm, eggs or embryos from donors who are not carriers of the condition
- Adoption
- Not to have children at all

If you are already pregnant, it is recommended that you speak to a genetic counsellor. They can discuss options for testing in early pregnancy to determine whether the developing baby is likely to be affected.