

Reproductive Carrier Screening

Every couple hopes to have a healthy baby however about 1 in 30 (3%) babies are born with a medical condition. Although some medical conditions are genetic and will be inherited from parents, over 80% of children with genetic conditions are born to parents with no known family history of the condition.

Although there is no test that can be done before or during pregnancy that can guarantee a healthy baby, new testing called 'Reproductive Carrier Screening' may provide useful information which can help couples make informed reproductive choices. The Royal Australian and New Zealand College of Obstetricians and Gynecologists (RANZCOG) recently recommended that everyone planning or early in pregnancy be told that this testing is available.

What is Reproductive Carrier Screening?

Reproductive carrier screening is genetic testing performed on a blood or saliva sample to tell if a couple have a high chance of having a child with certain genetic conditions. The conditions screened for are conditions that cause serious medical issues which usually begin in childhood. This testing is different to screening tests offered during pregnancy for chromosome conditions such as Down syndrome.

This test can tell with high accuracy if a person is a carrier of the genetic conditions screened for. Being a carrier of a genetic condition means that you have an abnormal gene that generally will not cause you any health problems, but may increase your chance of having a child with a genetic condition.

Most of the conditions screened for by carrier screening are caused by a child inheriting a copy of the same abnormal gene from **both** healthy parents who are carriers of the condition. These are called recessive conditions. Examples of these conditions include cystic fibrosis and spinal muscular atrophy. If a couple are both carriers of the **same** recessive genetic condition they have a 1 in 4 (25%) chance of having a child affected by that condition.

Some of the conditions screened for in women only affect male children and are caused by the child inheriting an abnormal gene from a healthy carrier mother. These are called X-linked conditions. Some examples of these conditions are fragile X syndrome and Duchene muscular dystrophy. If a woman is a carrier of an X-linked condition, with each pregnancy she has a 1 in 4 (25%) chance of having a son affected by that condition.

Who should consider Reproductive Carrier Screening?

This screening should be considered by everyone planning a family even if they do not have a personal or family history of genetic conditions, as everyone is likely to be a carrier of several genetic conditions. (although not all of these will be detected by current carrier screening). The screening is relevant to people of all ethnic backgrounds.

Before considering reproductive carrier screening it is important that you make your doctor aware of situations which may increase your chance of having a child with a genetic condition as you may require more specialized advice and testing. These include:

- if you have a personal or family history of a genetic condition
- if you have a personal or family history of being a carrier of a genetic condition
- if you and your partner are related

There is no routine testing that is offered in pregnancy to show a baby is affected by any of the conditions screened for, unless reproductive carrier screening has identified that a couple have a high chance of this.

When should this testing be done?

The ideal time for reproductive carrier screening is before a couple plan a pregnancy so that they can be informed about all their reproductive options. Screening can also be done during pregnancy, ideally before 12 weeks gestation. Screening later in a pregnancy may be possible but you should speak to a genetic counselor about the potential implications before going ahead.

How can I access Reproductive Carrier Screening?

You can discuss reproductive carrier screening further with your GP or Private Obstetrician. In South Australia, reproductive carrier screening for couples or individuals is currently offered by a number of Private services, including (in alphabetical order): Abbott Pathology, Australian Clinical labs, Clinpath/Sonic, Eugene, Invitae and Repromed.

The cost of reproductive carrier testing is not currently covered by Medicare or private health insurance. A range of different screening options exist, and costs vary between different options and service providers, ranging from about \$400 to \$900 per person. A reduced cost may apply if a partner also has the test.

Detailed information is available on the service provider websites regarding the testing they offer, so you can compare the available tests to see which option best suits your needs. The test must be ordered by your doctor and is done on a blood or saliva sample, dependent on the laboratory used. Results take about 4 weeks to be available. The testing can be performed either individually or as a couple.

Some aspects to consider when selecting a test and service provider include:

- the number and type of genetic conditions the test screens for
- the cost of the testing per person or as a couple
- the availability of pre and post test genetic counseling through the service provider
- the time taken to get results

What can I expect from the results?

A detailed report of your results will be released to the doctor ordering the test. Once the results have been discussed with you, you will receive a copy of the detailed report.

About 2.5% (1 in 40) couples tested will be found to have a high chance of having a child with one of the genetic conditions tested for because they are both carriers of the same recessive condition or because the woman is a carrier of an X linked condition. About 70% (7 out of 10) of people will be found to be a carrier of at least one of the genetic conditions tested for. Because genetic conditions are inherited, other family members may also carry the same genetic condition as you and may then also wish to have carrier screening.

There is a small chance that the test may show you are a carrier for a genetic condition which may effect your future health or that you have a genetic condition which requires medical follow up.

What happens if I am found to have a high chance of having a baby with one of the conditions screened for?

Genetic counseling is available through the public health system for couples found to have a high chance of having a baby with a genetic condition. Your doctor can arrange a referral. A referral may also be made to an appropriate specialist who can discuss how the condition would affect a baby and the current treatments available if a baby is affected.

A genetic counselor will discuss available reproductive options related to the condition which may include:

- accepting the chance of having a baby affected by the condition and preparing for this possibility in conjunction with a team of specialists to support you and ensure the best possible outcome for the baby.
- Choosing not to have biological children or possibly considering alternative options for parenting including fostering and adoption.

Some couples may wish to be informed about options to reduce the chance of having a baby affected by the condition. These options may include:

- testing in pregnancy for the condition (called prenatal diagnosis)
- choosing to have IVF with embryos being screened for the condition (called preimplantation genetic diagnosis)
- using donor sperm/eggs or embryos to reduce the risk of having a baby affected by the condition.

If you want more information about reproductive carrier screening please refer to the online fact sheet available at:

<https://www.genetics.edu.au/publications-and-resources/factsheets/FS65REPRODUCTIVECARRIERSCREENING.pdf>
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