UNDERSTANDING REPRODUCTIVE CARRIER SCREENING

Mercy Health
Care first

What is reproductive carrier screening?

Genetic screening helps people understand their chance of having a child with a serious genetic condition. Although each genetic condition is uncommon, genetic conditions as a group affect about 1 in 400 people. Most couples with an affected child have no family history of the condition, and healthy parents can unknowingly pass on serious genetic conditions to their children.

People who have a genetic change that can affect their children's health are called carriers. Testing to find out if you or your partner are carriers for a condition is called **Reproductive Carrier Screening (RCS).**

How are genetic conditions passed from parent to child?

There are two main inheritance types that can cause healthy, unaffected biological parents to have a child with a serious genetic condition:

1. **Autosomal Recessive conditions:** These types of conditions require both parents to be carriers of the same faulty gene. These couples have a 1 in 4 chance of having an affected child. An example of an autosomal recessive condition is cystic fibrosis.
2. **X-Linked conditions:** These conditions result from a faulty gene on the X chromosome. Males are usually more severely affected than females because males only have one X chromosome. An example is fragile X syndrome, where female carriers have a chance of having an affected child.

What should you do if you have a family history of genetic conditions?

If there's a family history of a genetic condition like thalassemia, cystic fibrosis, or fragile X syndrome, discussing it with your GP is very important. Your future child may have an increased chance of a genetic condition if a relative has them. Your GP will refer you for genetic counselling if you have a family history of a genetic condition.

When is the best time to do Reproductive Carrier Screening?

This test is best performed on the woman before she gets pregnant, or in early pregnancy (before 14 weeks).

If she is a carrier for a condition, then the biological father of the baby should be tested for that condition. Their combined results will tell them the chance of passing an autosomal recessive or X-linked genetic condition to their children.

Carrier screening can be done at any time but is preferably **done before pregnancy** for better planning. It only needs to be performed once in a couple's lifetime.

DATE FOR REVIEW : 18/04/2027
Prompt doc number: MER0223089

Community Reviewed Making information easier to read.
This document provides general information only and is not intended to replace advice about your health from a qualified practitioner.
If you are concerned about your health, you should seek advice from a qualified practitioner.

Is reproductive carrier screening the same as NIPT (non-invasive prenatal testing)?

No, reproductive carrier screening is different to NIPT. They test for different types of genetic conditions. NIPT is only performed **during** pregnancy. It is a blood test on the pregnant person to detect Down Syndrome or other chromosome conditions **in the baby**. These conditions are usually **not** inherited from the parent.

Carrier screening can be performed **before or during pregnancy**. It is a blood test to detect genetic changes **in the biological parents** that can be passed to their child. The baby may not be affected by the genetic condition even if the parents are carriers of the same condition.

How do you arrange carrier screening?

You should see your GP to discuss carrier screening. The general antenatal clinics at Mercy Health are **not** able to order carrier screening for you.

How much does it cost?

Medicare now covers screening for three common genetic conditions (cystic fibrosis, fragile X syndrome, and spinal muscular atrophy). This means that you will not have to pay for this test if you have a Medicare number. There are some carrier screening tests that detect more genetic conditions, but these will cost extra. Mercy Health suggests discussing your options with your GP, including their costs, when you're planning to get pregnant or as soon as you know you're pregnant.

Your result says you are a carrier: What next?

We recommend genetic counselling for people who have an increased chance of a child with a genetic condition. This includes couples where they each carry a variant gene for the same autosomal recessive condition, or if the woman is a carrier of an X-linked condition.

If you discover that you have an increased chance of a child with a genetic condition, you can choose how to plan your family. You could choose natural conception with newborn testing, natural pregnancy with diagnostic testing during pregnancy, in vitro fertilization (IVF) with preimplantation genetic testing (PGT), or adoption. Talk to your doctor or genetic counsellor for more information.

Do you have to do RCS?

No, carrier screening is an optional test. Deciding to do RCS is personal, and it’s helpful to think about how the information might be useful to you. This can be different for each person or couple. If you do have a family history of a genetic condition, it’s important to talk to a genetic counsellor about what it could mean for your pregnancy.

DATE FOR REVIEW : 18/04/2024
Prompt doc number: MER0223089

This document provides general information only and is not intended to replace advice about your health from a qualified practitioner. If you are concerned about your health, you should seek advice from a qualified practitioner.

Where can you find more information and resources?

For additional information please visit:

- Online information at carrierscreening.org.au
- Royal Australian and New Zealand College of Obstetricians and Gynaecologists https://ranzcog.edu.au/wp-content/uploads/2022/06/Reproductive-carrier-screening.pdf
- NSW Centre for Genetics Education, NSW Health https://www.genetics.edu.au/PDF/Reproductive_carrier_screening-fact_sheet-CGE.pdf

For more information

For a plain language version of this information, please ask one of our friendly staff for a copy of the Mercy Health patient information sheet:

"Genetic testing for people planning to have children"

If you have any questions regarding this information, please contact your doctor or midwife.

In the event of an emergency, please dial **000** for an ambulance or go to your nearest Mercy Health or other hospital emergency department.

DATE FOR REVIEW : 18/04/2024
Prompt doc number: MER0223089

This document provides general information only and is not intended to replace advice about your health from a qualified practitioner. If you are concerned about your health, you should seek advice from a qualified practitioner.