Sonic Genetics
Quality is in our DNA

Reproductive carrier screening

When the woman is a carrier for an X-linked recessive condition

Information for patients

The healthcare professional responsible for your care has given you this leaflet because Reproductive Carrier Screening of specific genes for both you and your partner has shown that one of you, the female, is a carrier of a faulty gene that causes a genetic condition. This result has implications for your children and this fact sheet contains more information about the result mentioned in your report. We recommend that you read the leaflet carefully and also discuss your result with an experienced doctor or genetic counsellor.

Every cell in our bodies has genes, which tell the body how to function, grow and develop. In each cell, there are 20,000 or so genes, which are packaged in 22 bundles called chromosomes. Everyone has two copies of each of these chromosomes.
We all inherit one copy of each chromosome from each of our parents. These paired chromosomes are also referred to as *autosomes*. Each cell also has two *sex chromosomes*: women have two copies of the X chromosome (XX) and men have one X and one Y chromosome (XY).

In every person, the great majority of the genes on the chromosomes will be working as they should, sending the correct information to the body. However, everyone will have some genes in which one of the gene copies is faulty. These gene changes are called *mutations*.

The chance of there being a mutation in a specific gene is usually low, however, with 20,000 genes in every cell, every person will have a number of gene pairs in which one copy is faulty. But as there is still a second working copy of the gene, a person can carry the faulty gene, but will not be affected by that fault, because their second copy of the gene is working normally and sends enough correct information to the body to avoid any medical consequences. This person, carrying the faulty gene, is described as being a 'genetic carrier' of that gene.

We are all carriers for a number of conditions, but we usually do not know this unless we have had carrier testing.

There are thousands of genetic conditions that are due to a child inheriting the same faulty autosomal gene from each parent, and thus having no working copy of the gene. These genetic conditions are often very rare and there is usually no family history of the disorder to suggest that the parents were carriers and were likely to have an affected child.

What is an X-linked genetic condition and how is it inherited?

Genetic conditions that are carried on chromosomes 1–22 are called autosomal, but those that are carried on the sex chromosomes are called sex-linked, and those that are specifically related to the X chromosome are called X-linked. A woman may be described as 'a carrier' if she carries a faulty gene on one X chromosome and a normal gene on the other X chromosome. Because she has two X chromosomes, her second normal copy of the gene will usually send enough correct information to her body for it to function correctly. Thus, women who are carriers of an X-linked faulty gene are usually not affected by the condition, or may have only mild features of it.

On the other hand, a male has only one X chromosome. He does have a Y chromosome, but this does not provide a spare copy of the genes on the X chromosome, so if he inherits a faulty gene on his X chromosome, there is nothing to protect his cells from this fault. He will, therefore, usually display symptoms of the condition caused by the fault. These conditions can affect a boy's physical and intellectual development and function. Symptoms may be present in babies or develop later in childhood. This type of disorder is called 'X-linked recessive', because it is due to a gene on the X chromosome ('X-linked') and because a woman is protected by the presence of a normal X chromosome ('recessive').

As with autosomal traits, there are many hundreds of X-linked genetic conditions, most of which are very rare. You and your doctor may never have heard about them. Sometimes, but not always, there is a family history that suggests that a woman may be a carrier or may have an affected son.

info@sonicgenetics.com.au • www.sonicgenetics.com.au

How to interpret a carrier result for an X-linked condition

Reproductive carrier screening involves testing many different genes in reproductive partners. For each gene tested, the result could be (in decreasing order of likelihood):

- Normal screen in both partners
- Abnormal screen in one partner, normal in the other
- Abnormal X-linked screen in the female partner
- Abnormal screen in both partners.

As the female partner, you have had screening to see if you are a carrier of an X-linked faulty gene and, since you are, this means that you can pass on the faulty gene to your children, even though you yourself might not experience any of the symptoms of the condition.

If a woman is a carrier of an X-linked condition, the chance of having an affected child differs, depending upon whether that child is a son or a daughter. In every pregnancy, there is:

- One chance in four (that is, a 25% chance) that she will have a son who inherits the X chromosome containing the faulty gene from his mother and the Y chromosome from his father. As no working gene information will be produced, this son will be affected with the X-linked condition at birth or may develop it later in childhood.
- One chance in four (that is, a 25% chance) that she will have a son who inherits the X chromosome containing the working gene from his mother and the Y chromosome from his father. As working gene information will be produced, this son will not be born with the condition due to the faulty X-linked gene, nor will he develop it later in childhood.
- One chance in four (that is, a 25% chance) that she will have a daughter who inherits the X chromosome containing the faulty gene from her mother and the other X chromosome containing a working gene from her father. This daughter will be just like her mother: a genetic carrier of the faulty X-linked gene, who is usually unaffected by the condition.
- One chance in four (that is, a 25% chance) that she will have a daughter who inherits the X chromosome containing the working gene from her mother and the other X chromosome containing a working gene from her father. As she will have full working gene information produced, this daughter will be not be affected by the condition due to this X-linked gene and cannot pass a faulty gene onto her child.

This test identifies more than 97% of the faults in each gene tested. However, no screening test is perfect. There is always a small possibility that a person is a carrier, despite the screening test being normal.

What does it mean for us if the mother is a carrier for an X-linked genetic condition?

The test result has identified that you, as the female partner, are a carrier of a faulty X-linked gene and this means that you can pass this faulty gene on to your children.

Hearing about the carrier result will usually be unexpected and it may lead to a range of physical and emotional reactions. In the midst of this emotional confusion, you will probably need to make some decisions and it's vital that you have as much information and support as possible when doing so. It is also important to discuss this result with your doctor so that you fully understand what it means for you and your partner.

If you are not pregnant, you have time to think about the test result when discussing your family planning options. These options include testing a baby in pregnancy for this gene (prenatal testing). Or you could consider using IVF and then testing the embryo for this gene before implantation. You should also be aware that having no testing at all for this gene is also an option.

If you are already pregnant, your options include prenatal testing or, again, not testing the baby at all for this gene.

The Beacon expanded carrier screen has been developed to concentrate on relatively common conditions that cause serious childhood disorders. There are other conditions that are not included in this screen because they are exceedingly rare or are not childhood conditions. This test result does not alter your chance of having a child with one of those other disorders. Nonetheless, since you have now had the test and understand your options, your overall chance of having a child with any inherited genetic condition is very much lower than the chance for the general untested population.

Genetic counsellors are also experienced in providing support and information. If you want further genetic counselling regarding your test results, your doctor can refer you to genetic counsellors in your State or Territory, or you can access genetic counselling by phone. A list of private genetic counsellors is available at www.sonicgenetics.com.au/counsellingservices. Sonic Genetics offers free genetic counselling for couples where the woman is identified as a carrier for an X-linked recessive disorder. If you are eligible for this service, your doctor may choose to refer you for genetic counselling. Conditions apply and can also be discussed with your doctor.

Your test result is important for your relatives

You probably inherited the faulty gene from your mother. That means that close female relatives on your mother's side, such as sisters and female cousins, may also be a carrier of this faulty gene. Telling them about the availability of the carrier test may help them make informed family planning decisions as well, if they wish to do so.

What if I have another partner in the future?

The information summarised in this leaflet, and the results in your report, apply only to a specific couple. If either of you were to have a different reproductive partner in the future, your chances of having a child with an inherited genetic condition would need to be re-assessed for that new partnership.

Further information and contacts

The Centre for Genetics Education www.genetics.edu.au