Sonic Genetics
Quality is in our DNA

Reproductive carrier screening

When neither partner is a carrier for an autosomal or 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 was negative. This result means, as a couple, the chance of having a child with an inherited condition due to these genes is very low. This fact sheet contains more information about the result mentioned in your report. We recommend that you 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 on autosomes 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.

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.

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

How does a person inherit a genetic condition?

If a child is affected by an autosomal or X-linked condition, then either both parents have the same faulty autosomal gene that they have passed to their child, or a mother has passed her X-linked faulty gene to her sons (or to her daughters who may be mildly affected).

There are thousands of genetic conditions that are due to a child inheriting the same faulty autosomal gene from each parent, and so 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. There are also many hundreds of X-linked genetic conditions that are also rare.

How to interpret a negative carrier result

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.

You and your partner have had a carrier screening test to see if both of you are genetic carriers of the same faulty autosomal gene. If both partners were shown to be carriers of the same autosomal condition, there is a one in four chance in every pregnancy of their having a child with the genetic condition due to that gene.

The female partner will also have had screening to see if she is a carrier of an X-linked faulty gene. If a woman were a carrier of an X-linked condition, there is a one in two chance of her son being affected. There would also be a one in two chance of her daughter being a carrier, and possibly mildly affected.

If the screening test for a couple does not identify any faulty genes, the chance of that couple having a child who is affected is very much lower than the chance for the general population. But the chance is not zero. 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 being negative genetic carriers mean for us?

The test result for both you and your partner was negative, that is, the test did not identify that either of you is a carrier of faulty autosomal genes, and it did not find that the woman is a carrier of a faulty X-linked gene. This means that the chance of your having a child who is affected by an autosomal or X-linked condition is at least 20 times lower than it was before you had the test, and very much less than the chance in the general population.

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, your overall chance of having a child with any inherited genetic condition is very much lower than the chance for the general untested population.

It is important to discuss this result with your doctor so that you understand fully what it means for you and your partner. 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.

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