Sonic Genetics
Quality is in our DNA

Reproductive carrier screening

When only one partner is a carrier for an autosomal recessive condition

Information for patients

The healthcare professional responsible for your care has given you this leaflet because Reproductive Carrier Screening shows that one of you is a carrier of a faulty gene which causes a genetic condition and that your partner is not. Whilst this result has little or no implications for your children, 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 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.

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

What is an autosomal recessive genetic condition and how is it inherited?

Every child inherits one copy of each autosome (and of each gene on the autosome) from each parent. If both parents are carriers of the same condition, the child could inherit the faulty gene from each parent and have no working copy of the gene. When the child does not have any working copy of the gene, he or she is likely to develop a genetic condition, due to the lack of genetic information required by the body. This type of condition is called 'autosomal recessive', because it is due to a gene on an autosome ('autosomal') and because neither gene copy is working ('recessive').

For a child to be born with an autosomal recessive condition, they must inherit a faulty gene copy from both mother and father. Autosomal recessive conditions can affect a child's physical and intellectual development and function. Symptoms may be present in babies or develop later in childhood.

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.

How to interpret a positive carrier result for one partner

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.

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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. As only one of you is a carrier and the other has had a normal screening test, this means that, in every pregnancy, there is:

- One chance in two (that is, a 50% chance) that you will have a child who inherits a normal copy of the gene from each of you. As your child will have two normal genes, the child will be not be affected by the condition due to this gene.
- One chance in two (that is, a 50% chance) that you will have a child who inherits the faulty gene copy from one of you and a working gene copy from the other. The child will be just like the parent who is the carrier: a genetic carrier who is unaffected by the condition.
- A very small chance (typically less than one in 10,000) that you, as a couple, will have a baby born with the condition due to this gene or who may develop it later in childhood.

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 one partner being a genetic carrier mean for us?

The test result for both you and your partner has shown that only one of you is a genetic carrier of a faulty autosomal gene. While there is no impact on your own health, the result means the genetic carrier parent can pass on the faulty gene to their child. But whilst that child would also be a carrier, he or she would only be affected with the condition if ***both*** parents were carriers of the same faulty gene and the child inherited an abnormal gene from each parent.

Hearing that one of you is a genetic carrier will usually be unexpected, and may lead to a range of physical and emotional reactions. It is important to discuss this result with your doctor so that you fully understand what it means for you and your partner. Genetic counsellors are also experienced in providing support and information and, if you want further genetic counselling regarding your test results, your doctor can refer you to genetic counsellors in your State or Territory. You can also access genetic counselling by phone. A list of private genetic counsellors is available at www.sonicgenetics.com.au/counsellingservices.

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.

Your test result is important for your relatives

You probably inherited the faulty gene from a parent. That means that other close genetic relatives, such as brothers, sisters and cousins, may be carriers 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