Sonic Genetics
Quality is in our DNA

Reproductive carrier screening

When both partners are carriers for the same autosomal recessive condition

Information for patients

The healthcare professional responsible for your care has given you this leaflet because Reproductive Carrier Screening has identified both you and your partner as having a faulty gene which causes a genetic condition. This result has potential 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), however, we do not need to consider the sex chromosomes further in this leaflet.

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.

How does a person inherit an autosomal recessive genetic condition?

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

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. The test result for both you and your partner was positive, that is, the test identified that both of you are carriers. This means that, as you and your partner are carriers of the same autosomal recessive condition, there are three possible outcomes in every pregnancy:

- One chance in four (that is, a 25% chance) that you will have a child who inherits both copies of the normal gene from each of you. As your child will have two working genes, they will be not be affected by the condition associated with this gene.
- One chance in two (that is, a 50% chance) that you will have a child who inherits the faulty gene from one of you and a working gene from the other. The child will be just like you: a genetic carrier who is unaffected by the condition.
- One chance in four (that is, a 25% chance) that you will have a child who inherits the faulty gene from each of you. As the child has no working copy of the gene, they will be born with the condition or will be likely to develop it 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 being genetic carriers mean for us?

The test result has identified that you are both genetic carriers of the same faulty autosomal 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 and your doctor may choose to refer you for genetic counselling. Sonic Genetics offers genetic counselling free-of-charge^ for eligible patients. 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.

Your test result is important for your relatives

You and your partner probably inherited the faulty gene from a parent. That means that the close genetic relatives of both of you, 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

^Terms and conditions apply. Please refer to www.sonicgenetics.com.au/rcs/gc