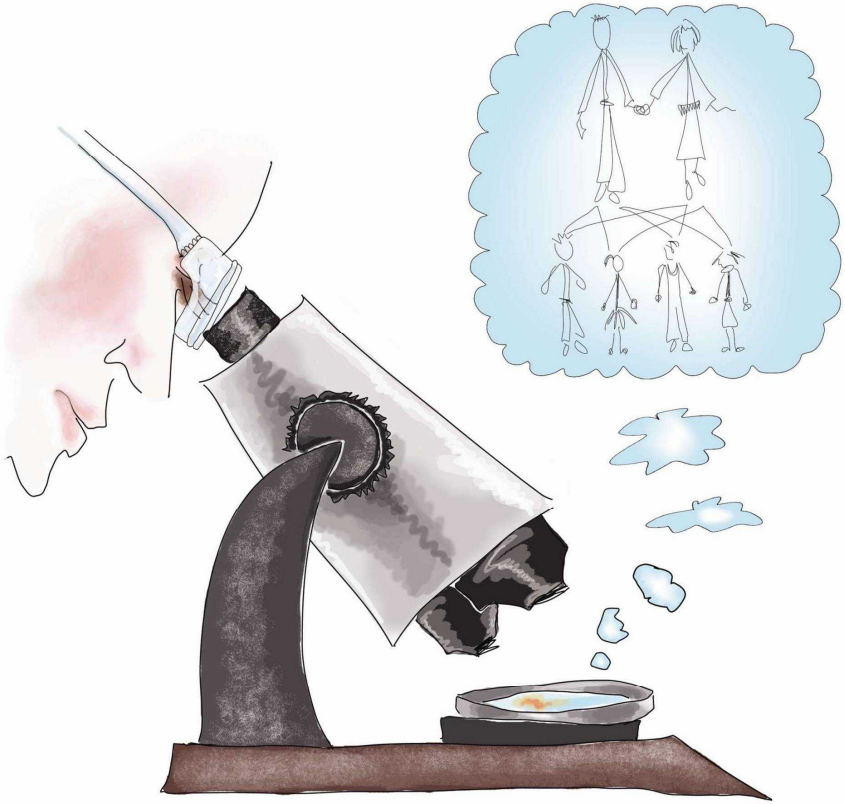
Carrier Testing



We are extremely grateful to all the people that allowed us to interview them during the making of this leaflet.

This information was developed by the Genetic Interest Group, a UK national alliance of patient organisations which supports children, families and individuals affected by genetic disorders.

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EuroGentest

Genetic Interest Group

Information for Patients and Families

Carrier Testing

This information is for people who are either considering taking a carrier test, or have already taken one. It provides you with information about what it means to be a 'carrier', how you find out if you are a carrier, and practical and emotional information about living with your test results. Much of this information has been gathered by speaking to people who have taken a carrier test. We hope you will find it helpful.

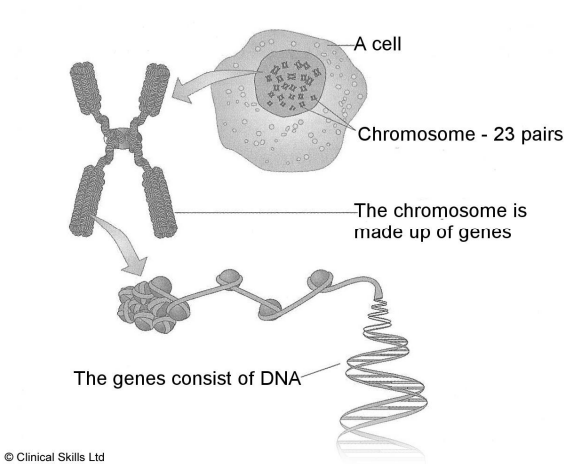
Section 1: What is a Carrier?

To understand what it means to be a carrier, it is helpful to understand what genes and chromosomes are.

Genes and chromosomes

Our bodies are made up of millions of cells. Most cells contain a complete set of genes. We have thousands of genes. Genes act like a set of instructions, controlling our growth and how our bodies work. They are responsible for many of our characteristics, such as our eye colour, blood type or height.

Figure 1: Genes, Chromosomes and DNA



Haemophilia Society
Tel: 020 7831 1020
Email: info@haemophilia.org.uk
Web: www.haemophilia.org.uk

The Jennifer Trust (For Spinal Muscular Atrophy)
Tel: 0800 975 3100 (9am-5pm)
Email: jennifer@jtsma.org.uk
Web: www.jtsma.org.uk

The Sickle Cell Society
Tel: 020 8961 7795
Tel: 0800 0015660 (24-hour
Email: info@sicklecellsociety.
Web: www.sicklecellsociety.org

These contact details may no longer be correct.
Please check them online.
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UK Thalassaemia Society
Tel: 020 8882 0011
Email: office@ukts.org.uk
Web: www.ukts.org

Unique - The Rare Chromosome Disorder Support Group
Tel: 01883 330766
Email:info@rarechromo.org
Web: www.rarechromo.org

EuroGentest
Free-access website providing information about genetic testing and links to support groups across Europe.
Web: www.eurogentest.org

Orphanet
Free-access website providing information on rare diseases and orphan drugs, and links to support groups across Europe.
Web: www.orpha.net

Further information
For further information, contact your local regional genetics service. Their contact details can be found at:
www.gig.org.uk/services.htm

Additional information can also be found at:

Climb
The National Information Centre for Metabolic Diseases
Have information about Tay-Sachs disease.
Tel: 0800 652 3181
Email: info@svcs@climb.org.uk
Web: www.climb.org.uk

Contact a Family
Provides advice, information and support to the parents of all disabled children.
Helpline 0808 808 3555
(Mon-Fri, 10am-4pm and Mon, 5.30pm-7.30pm)
Email:ihelpline@cafamily.org.uk
Web: www.cafamily.org.uk

Cystic Fibrosis Trust
Tel: 0845 859 1000
Email: enquiries@cftrust.org.uk
Web: www.cftrust.org.uk

Fragile X Society
Tel: 01371 875100
Email: info@fragilex.org.uk Web: www.fragilex.org.uk

Genetic Interest Group
Provides information about specific genetic conditions and contact details of support organisations.
Tel: 020 7704 3141
E mail: mail@gig.org.uk
Web: www.gig.org.uk

Genes are carried on thread-like structures called chromosomes. Usually, we have 46 chromosomes in most cells. We inherit our chromosomes from our parents, 23 from our mother and 23 from our father, so we have two sets of 23 chromosomes, or 23 'pairs'. Because the chromosomes are made up of genes, we therefore inherit two copies of most genes - one copy from each parent. This is the reason why we often have similar characteristics to our parents. The chromosomes, and therefore the genes, are made up of a chemical substance called DNA.

Changes in genes or chromosomes are called **mutations**, and we all carry a number of them. However, because we have two copies of most genes, the normal copy compensates for the copy with the mutation. Being a carrier therefore means that you do not have the condition, but carry a changed copy of the gene on one of a pair of chromosomes. Being a carrier usually does not affect your health in any way. The only time when being a carrier can cause problems is if it can lead to your children having a genetic condition.

When can being a carrier lead to our children being affected by a genetic condition?

There are three situations where being a carrier could result in a child being affected:

1. Autosomal recessive conditions
For these conditions, there will only be a risk of children having the genetic condition if both parents carry the same mutation in the same gene or a mutation for the same condition. If this is the case then there is a 25% chance (1 in 4) that each child will inherit one copy of the mutation from each parent and be affected. Common conditions inherited in this way include cystic fibrosis, sickle cell, beta-thalassaemia and Tay-Sachs disease. More information about this can be found in the ***Recessive Inheritance*** leaflet**.**

2. X linked conditions

For these conditions, if a woman is a carrier there is a 50% chance (1 in 2) that her sons can be affected by the genetic condition. There is also a 50% chance (1 in 2) that her daughters will inherit the mutation and be carriers, like herself. In rare cases a daughter can be affected by the condition. If a man who has an X linked condition has a daughter, his daughters will **always** inherit the mutation and be carriers, but his sons will never inherit the mutation. More information about this can be found in the ***X linked Inheritance*** leaflet. Common conditions inherited in this way include fragile X, Duchenne muscular dystrophy and haemophilia.

3. Chromosome rearrangements

If an individual is a carrier of a chromosome rearrangement (such as a balanced chromosome translocation), there is an increased chance that a pregnancy will end in miscarriage, or that the child will be born with physical disabilities and learning difficulties. More information about this can be found in the leaflets ***Chromosome Changes*** and ***Chromosome Translocations***.

Could I be a carrier?

There are a number of reasons why you might have an increased chance of being a carrier of a particular genetic condition or chromosome rearrangement.

- Someone in your family has a recessive condition, X linked condition, or chromosome rearrangement.
- Someone in your family has found out that they are a carrier of a particular recessive condition, X linked condition, or chromosome rearrangement.
- You have a child who has a recessive condition, X linked condition, or chromosome rearrangement.
- You have a particular ethnic background which means you are more likely to be a carrier of a particular genetic condition. Examples of this include sickle cell in people of

Other sources of support

Genetic specialists and other health professionals (such as counsellors and psychologists) are experienced in helping people talk through the emotions that relate to receiving carrier test results. They can be a good source of information and support.

"Seeing Emma [the genetic counsellor] was very reassuring because before then I hadn't really understood that there was this thing called a CVS or an amniocentesis. So it was just good to have a professional explain all the different options. It was very helpful." (Tay-Sachs carrier)

Some people also find it helpful to contact a patient support group. Patient support groups can provide information about the practical and emotional aspects of being a carrier of a condition. Many have a website and helpline giving information and advice. They can often put people and families in touch with others who are in a similar situation. They may have a chat forum on which members can email other members.

"We've got quite a lot of information from the patient support group and so it's been helpful. Also, knowing that some nights they work late, if you want to ring them up and have a chat to them, they will chat to you over the phone, send things in the post, work with the schools and so on." (fragile X carrier)

"I think my parents want to get tested because they have brothers and sisters who have children, and so would like to know if there's a chance they might have passed it on too." (spinal muscular atrophy carrier)

For some people, sharing information with other family members is a positive experience. It can bring families closer together, and family members can provide a good source of support. For others it can be difficult to share carrier test results with other family members, and the experience can be difficult or distressing.

"You kind of feel like you're bringing a blight into someone else's family and that really did affect me because, you know, as a daughter-in-law you don't want to do that really…" (haemophilia carrier)

For grandparents, it can be particularly difficult. They may not want to accept that the gene mutation is something that could have been passed down from them. It is also not uncommon for grandparents to feel guilty because they feel it is their fault for having passed the gene on. These are reactions you should be aware of.

"My mum spoke to my nan and she said 'it's not from me full stop'. So she blocked it off, she said 'I didn't bring anything into the family.'" (fragile X carrier)

"She was really cut up about it. She felt guilty. And I said look, it's just one of those things, its not your fault." (cystic fibrosis carrier with an affected child)

It can be helpful to have information provided by your genetic specialist to show to other family members to help explain what it means to be a carrier, and that being a carrier is something that occurs by chance.

Afro-Caribbean descent, beta-thalassaemia in people of Mediterranean descent, and Tay-Sachs in people of Ashkenazi Jewish descent. These conditions are more prevalent in these particular ethnic groups, but may occur in others.

Even if none of these reasons exist, you may decide to have a carrier test because your partner knows he or she is a carrier of a particular recessive condition. In this case the results of your test will help you to find out if your future children are at an increased risk of having the condition.

How can I find out if I am a carrier?

If you think there is a chance that you could be a carrier of a particular genetic condition you should speak to your family doctor. Your doctor can then refer you to a genetic specialist. The genetic specialist will ask you questions about your family history and any genetic conditions that have occurred in your family. They will discuss what it means to be a carrier and the implications of knowing your carrier status. It is important to remember that carrier testing is a personal choice and is entirely voluntary – you should not feel pressured into taking the test if it would not be useful to you.

If you want to go ahead with carrier testing, and the specialist agrees that there is a reason for testing, you will be offered a genetic test. A genetic test can identify if there is a mutation in a particular gene or chromosome that would mean you are a carrier. The test is usually performed on a blood sample, although sometimes a saliva sample can be used. A sample will be taken from you and sent off to the laboratory to be analysed.

More information about genetic testing can be found in the leaflets ***What is a Genetic Test?*** and ***What Happens in a Genetics Laboratory?***

It is important to remember that, for conditions which are recessive, both partners have to be carriers of the same genetic condition for there to be a chance that children will inherit the condition. In the case of X linked conditions, only the mother needs to be a carrier for there to be a chance that her sons will be affected, although daughters of carrier men will always be carriers. In some rare cases women can be affected by X linked conditions.

Uncertainty of test results

Sometimes the results of a genetic test can be uncertain.

For some conditions, such as cystic fibrosis, there will still be a very small risk of being a carrier, even though no mutation is found through carrier testing. This is known as **residual risk**. This is because there are hundreds of known mutations that can cause the condition. However, genetic tests usually only test for the most common mutations.

In other cases a mutation may be found through genetic testing, however it is not clear what effect the mutation will have. In these cases no definite conclusions can be made.

Section 2: Living with your Carrier Status

The following information discusses the various experiences of people who have taken a carrier test. It will be of use to people who have recently found out their carrier status, but it will also be useful to people who are thinking about taking a carrier test. We have tried to highlight a range of issues and emotions that knowledge of one's carrier status can cause, although they may not all be relevant to you.

just listen and talk about it, just really being there and supporting. There is no solution so you just have to go through it, and that takes a lot of time." (partner of haemophilia carrier)

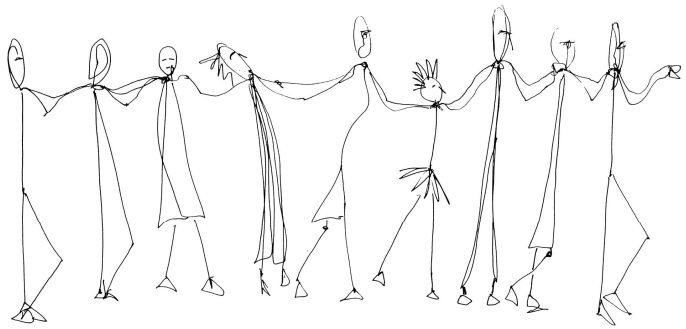
However, knowing your carrier status can also cause tension and strain within the relationship. Sometimes couples will have to discuss issues that are very difficult and upsetting.

"I feel like I can cope with a lot, but to have to pull in someone you really care about, that felt horrible. That felt worse than dealing with it myself." (haemophilia carrier)

Other family members

If you find out that you are a carrier, you may wish to discuss this with other family members. This gives other family members the opportunity to have a blood test to see if they are also carriers, if they wish. This information may also be useful in helping diagnose other family members. It might also be particularly important to family members who are likely to have children in the future. Discussing your carrier test results is your choice and your test results will never be shared with other family members without your permission.

"They now know that it's in the family and they can be tested if they want to, they have that choice. And it's useful for them when they want to have children." (cystic fibrosis carrier)



What if you already have children?

Some parents who already have a child with a genetic condition, say that one of the emotions that they experience when they find out they are a carrier is feeling 'guilty' for having 'passed on' the child's condition. It is entirely natural to feel this way. Mothers of boys who have X linked conditions sometimes say that they feel 'blamed' by their male partners for having passed on the faulty gene, or blame themselves. If you are having these kinds of feelings you should talk them through with your genetic specialist. It is important to remember that genes are distributed by chance and having a gene mutation is not your fault. Over time it has been shown that these feelings usually lessen in intensity.

"I do feel sort of that I've let my family down or let my husband down, and obviously let my son down because obviously I gave him something that he didn't really need to deal with for the rest of his life." (fragile X carrier)

If you find out that you are a carrier and you have children, even if they are not affected by the condition, there is still a chance that they could be carriers. It is important that you discuss this risk with your children at an age which is suitable. For some parents this might be when they think the child is old enough to understand. For others it might be when their children begin to have serious relationships. The age at which a young person can have a carrier test varies, but the person having the test has to make their own decision about it.

Relationship with partner

Knowing about your carrier status can have an effect on your relationship with your partner. For some people it can bring them closer together and they can help support one another.

"I just had to try and be there. She was very upset. You have to

How might I react if the result is positive?

People react in all sorts of ways when they receive their test results. Many people say that they feel angry or worried when they first find out that they are a carrier. Some people report that they feel sad, surprised or shocked. All these reactions are normal. For most people these feelings subside after a few months.

"To find out I was a carrier and my partner was also a carrier, I felt very gutted and also when you look at the statistics… to be one then your partner to be one is so rare I just felt really cross about it, it was just such awful bad luck." (Tay-Sachs carrier)

"I was actually quite surprised. I don't know why because my sibling is a carrier so I guess I should have expected it in a way, but I was quite shocked that I was a carrier." (spinal muscular atrophy carrier)

It is quite normal to feel somehow different about yourself when you find out you are a carrier. Some people say that it is strange to find out something new about yourself when you think you know everything. It can take time to adapt to this new information.

"It just kind of felt like some of the genetic burden I've been given, along with very short sight and a tendency to get fat and things like that." (Tay-Sachs carrier)

"At the time it seemed of enormous importance that this diagnosis had been found. But then as time went on it seemed like just a quirk." (carrier of a chromosome translocation)

Some people say that finding out they are a carrier makes them feel 'less healthy'. Other carriers say that they are concerned that, in the future, they will be more likely to develop health problems. These reactions are very normal but it is important to

remember that your carrier status has no effect on your health. We are all carriers of a number of gene mutations.

Studies have shown that carriers cope better with their results if they recognise that being a carrier is something that you cannot change, and that the information can be used positively.

"I've just accepted it now. I've moved on from that. Life throws things at you and you just get on with it really. It's just another one of life's twists." (cystic fibrosis carrier)

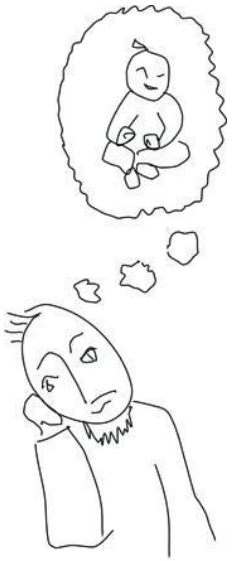
"My mother said to me, 'well how do you feel about it?', and I said 'fine, at least I know'." (cystic fibrosis carrier)

How might I react if the result is negative?

For most people, finding out that they do not carry the changed gene brings joy and relief. However, some find it difficult to communicate the 'good news' to their siblings and other relatives who may themselves be carriers or have an affected child. They wonder why they 'escaped' when other family members did not. Sometimes it is difficult to accept that you have been fortunate when others have not.

Future children

For some people, finding out they are a carrier is a source of worry because it affects their plans to have children. For other people, it is a relief to know so that they can plan ahead. Knowing that there is an increased risk that your future children might have a genetic condition means that you can be prepared and take the time to make important decisions. There are a number of options you may wish to consider.



If you and your partner are both carriers for the same recessive condition, or if you are female and a carrier of an X linked condition, there are a number of options available to you. For some genetic conditions, it is possible to perform a test during pregnancy to see if the baby has inherited the changed gene (prenatal testing). For more information you should look at the leaflets about ***Amniocentesis*** and ***CVS***. If you think this might be an option for you, speak to your doctor about whether these tests are available for the condition you are concerned about. If possible do this before the pregnancy as the laboratory may have to make preparations that can take several months. If you are considering prenatal testing you should think about what you would do if the foetus was found to have a genetic condition, and how you might feel about a termination of pregnancy.

It may be possible to perform a technique called Preimplantation Genetic Diagnosis (PGD) as an alternative to testing the foetus during pregnancy. This involves the couple undergoing medically assisted reproduction, after which the fertilised eggs are tested to see if they have the changed gene. Only those eggs without the changed gene are implanted into the woman's womb. This is a demanding process and is not suitable for everyone. For more information about PGD, and whether it is available to you, you should speak to your doctor.

Other options you might want to consider include adoption, conceiving using donor eggs or sperm, or the possibility of not having children.

"The good thing is when I went for the genetic counselling I found out there were choices. If you want to have more children, there are ways you can go about it." (cystic fibrosis carrier with an affected child)