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Autosomal recessive inheritance and carrier testing

This information sheet from Great Ormond Street Hospital (GOSH) explains autosomal recessive inheritance of genetic conditions and when testing may be helpful and when it might not.

Background information

Genes consist of chemical 'instructions', which determine how our bodies develop and function. We have about 20,000 pairs of genes in total. We have two copies of every gene - one inherited from our mother and one from our father.

We all have several genes where one copy is altered or changed (the medical word for this is a gene mutation). This usually has no effect as the other copy of the gene is normal so cancels out the change.

Genes are complicated messages and a change can happen anywhere in the message. Some genes tend to have changes in the same place, but most genes can have changes anywhere, making testing of the gene to look for any changes more difficult.

What is autosomal recessive inheritance?

The term autosomal recessive inheritance is used to describe one way that some genetic conditions are inherited. An example of a condition inherited in an autosomal recessive manner is cystic fibrosis.

In autosomal recessive inheritance, both parents have a change in one copy of the same pair of genes so a child inherits an altered copy of the gene from both parents. As both genes are altered, there is no normal gene to cancel out the change so the child can develop the genetic condition.

The way that autosomal recessive inheritance happens is shown in the diagram on page two.

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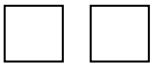
What is a carrier?

A 'carrier' is a person with one normal and one altered copy of a gene. Being a carrier of an autosomal recessive condition does not cause medical problems. In the diagram below, both parents and two of the children are carriers.

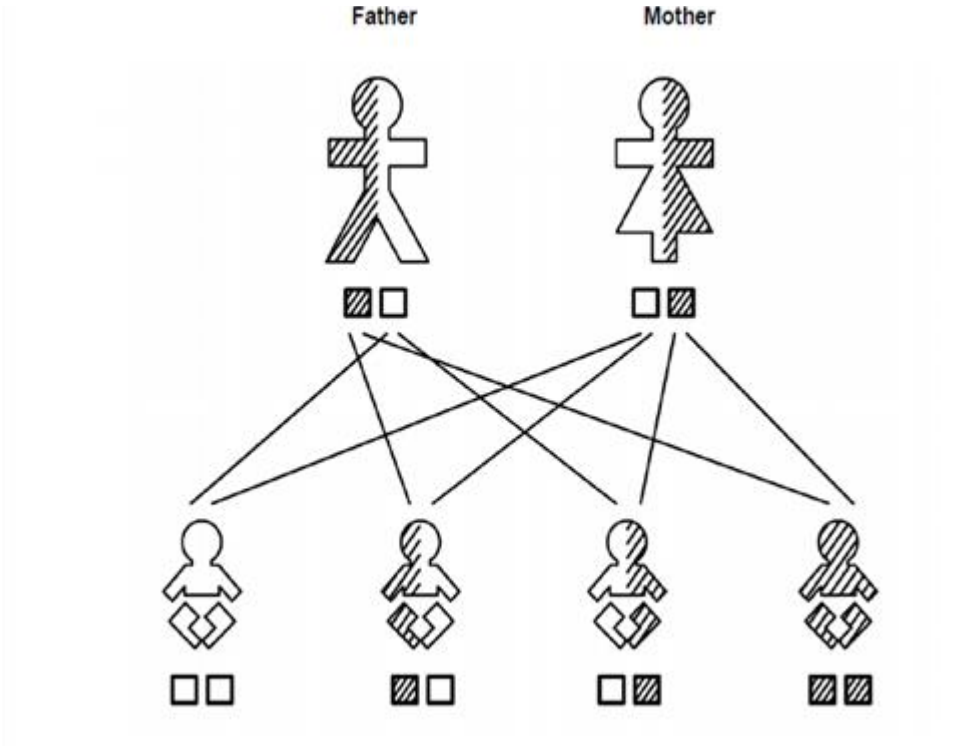


These shapes represent a pair of genes

This shape represents a normal copy of the gene

This shape represents an altered (mutated) copy of the gene

Both parents are carriers of the same genetic condition. They both have an alteration (mutation) in one copy of the gene as well as one normal copy of the gene so they do not have the medical condition. There are four possible ways for the parents to pass on their genes.



Unaffected Two normal copies of the gene	**Unaffected** Carrier	**Unaffected** Carrier	**Affected** two altered copies of the gene

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If I am a carrier, what is the chance of my child having a genetic condition?

This depends on whether your partner is a carrier or not.

If your partner **is not** a carrier your children will be unaffected by the condition, although each child will have a 50% (1 in 2) chance of also being a carrier.

If your partner **is** a carrier there is a 25% (1 in 4) chance that a child will inherit two altered copies of the gene and will be affected by the genetic condition. If a child inherits only one altered copy of the gene they will be a healthy carrier like their parents. A child may also inherit normal copies of the gene from each parent. This is shown in the diagram above.

The chances of inheriting the altered gene are the same for each pregnancy. If one child in the family already has the condition, there is still a 25% chance that the next child will have it too. In the same way, the child will have a 50% chance of being an unaffected carrier and a 25% chance they will inherit two normal genes.

Should my partner and I have genetic testing for the recessive condition in my family?

If one of your relatives is affected with or is a carrier for a recessive genetic condition and the genetic alteration that has caused this has been identified, we may be able to offer you and your partner carrier testing for the condition.

Most of the time, there is a low chance of having a child affected with the condition even when one of the partners is a carrier. This is because their child is only at risk of having the condition if both parents are carriers.

There are some situations where the chance may be higher and we recommend testing. These are:

- When the condition is common within your ethnic group, more than 1 in 70 people are carriers for it and an accurate genetic test is possible
- If you and your partner share common relatives such as grandparents and an accurate genetic test is possible
- There are also a few conditions with a very accurate carrier test, for example genes that always have alterations in the same place.

If any of these situations apply to you, testing may be recommended.

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Appointments in the Genetics Clinic

We offer appointments when carrier testing is indicated based on the information above except for a small number of conditions where there are clinics at other hospitals able to offer testing.

If you have any questions, we are happy for you to contact us. The contact details are:

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