



Carrier testing and reproduction: your options

Muscular Dystrophy UK supports people living with around 60 different muscle-wasting conditions. If you have one of these conditions, or one of your relatives has, and you are planning to start a family, you may want to ask yourself: am I a carrier, and if so, will I pass the condition on to my children?

The answers to these questions will vary according to the type of condition, its inheritance pattern, and how much is known about its genetic causes. To find out if carrier testing is available for the condition in which you are interested, you can contact your local genetic counselling centre. A geneticist can discuss with you your particular circumstances, and how they might affect the possibility and accuracy of testing.

It's important to note that genetic tests may take several weeks or months to complete. With this in mind, it is advised that you do these before planning a family and a pregnancy.

Once you know the results, you will have time to think through the various options open to you, and then discuss these with specialists at a genetic counselling centre.

This factsheet is designed as a reference guide for some of the general techniques and genetic tests that can be used to find out if you may be a carrier.

What is a carrier?

A carrier is a person who usually shows no symptoms of a condition, but carries the genetic mutation (sometimes called a 'faulty' or 'altered' gene) that causes it. A carrier may have very mild symptoms of a condition, and may not have been diagnosed as having it, but still carries the genetic mutation that causes it.

There is a risk that a 'carrier' will pass the mutation on to his or her children, and that they themselves might be affected by, or be carriers of, the condition. The level of risk depends on the inheritance pattern of the condition. (You can find this information in the Muscular Dystrophy UK factsheet relating to the condition in which you are interested, as well as our *Inheritance and the muscular dystrophies* factsheet, or you can discuss this with your geneticist.)

What is 'carrier testing'?

When someone is affected by a particular condition, this is usually evident on clinical examination. Carrier tests are designed to discover whether or not a person who has no symptoms of a condition carries the genetic mutation that causes it.



Direct genetic tests

These are carried out on the part of a person's genetic code (DNA) which, when changed or altered (mutated), causes a particular condition. Once scientists have identified the genetic cause of a condition, they know where to look for the genetic mutation(s) among the thousands of genes that make up our genetic code (DNA).

Genetic mutations may take the form of a repeated piece of genetic code (called a 'duplication' or an 'expansion'), a missing chunk (called a 'deletion'), or a tiny alteration in the genetic code (called a 'point mutation'), depending on the condition.

Different types of mutations are found in different conditions, or sometimes in different families with the same condition. It is usually necessary to find the specific genetic mutation responsible for the condition in a particular family before it can be used to test other members of that family.

DNA is obtained by taking a blood sample. If your relative who has the condition is alive, he or she may be asked to give a blood sample, which can be compared directly with yours.

Linked genetic markers or 'family studies'

Sometimes, if there is no obvious genetic mutation (a duplication, deletion or point mutation) in an affected person, direct genetic tests are not possible/informative. In these cases, 'markers' are used to find out whether someone has inherited the crucial part of the genetic code that carries the genetic mutation within it. These are pieces of DNA, situated close to the mutated gene, which can be used to distinguish it; they are said to be 'linked' to the gene. If you are found to have inherited the same 'linked' markers as the affected person, you are also likely to have inherited the mutated gene.

The accuracy of this depends on how close the marker is to the mutated gene. Sometimes, a reliable marker is not available, so the test does not give any useful information. In order to use linked markers, DNA samples – from an affected person and from as many other family members as possible – need to be available. This will help to establish which 'markers' are linked to the mutated gene in that particular family.

Where can I get advice about carrier testing?

Contact your nearest genetics centre to find out if testing is available for the condition in which you are interested. If you want to be tested, your family doctor can refer you to the genetics centre. The genetics centre and your family doctor can then arrange for other possible carriers and family members to visit the appropriate centre, and/or to give blood samples for testing.

Once you know you are a carrier, you have the option to undergo Preimplantation genetic diagnosis (PGD), which involves using In vitro fertilisation (IVF):



In vitro fertilisation (IVF)

In vitro fertilisation (IVF) is a reproductive medicine technique used to assist couples who cannot, or have difficulty, conceiving naturally. Embryos are created in the laboratory from the eggs and sperm of a couple, then transferred into the womb in the hope that a pregnancy will occur. First, the ovaries of the female partner are stimulated using fertility drugs to produce several eggs. Once the eggs are mature, they are collected from the ovaries. A semen sample is produced by the male partner and the sperm is then used to fertilise the eggs in the laboratory. Eggs which are successfully fertilised begin to grow and divide; they are now called embryos, and can be transferred into the womb in the hope a pregnancy will occur.

Pre-implantation genetic diagnosis (PGD)

Pre-implantation genetic diagnosis (PGD) is a technique that has been developed to help couples who are at risk of having a child with a serious genetic condition to have an unaffected child. PGD involves using in vitro fertilisation (see above) to create embryos in the laboratory from the eggs and sperm of that couple. The embryos are then tested for the particular genetic disorder.

PGD is currently offered in only a few centres in the UK. It is a relatively complicated and lengthy procedure with some associated risks. The chances of a successful cycle of PGD are relatively low compared with the chances of conceiving naturally. National guidelines have been developed, which lay out certain criteria that must be met for a couple to be eligible for PGD. If you think this is an option you might wish to pursue or find out more about, you need first to be seen by your nearest Regional Genetic Service.

Other related publications:

- ▶ Muscle biopsies
- ▶ Inheritance and the muscular dystrophies
- ▶ Pregnancy & fertility
- ▶ Prenatal diagnosis and testing

We're here for you at the point of diagnosis and at every stage thereafter, and can:

- ▶ give you accurate and up-to-date information about your or your child's muscle-wasting condition, and let you know of progress in research
- ▶ give you tips and advice about day-to-day life, written by people who know exactly what it's like to live with a muscle-wasting condition
- ▶ put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences
- ▶ tell you about – and help you get – the services, equipment and support you're entitled to.

Disclaimer

While every reasonable effort is made to ensure that the information in this document is complete, correct and up-to-date, this cannot be guaranteed and Muscular Dystrophy UK shall not be liable whatsoever for any damages incurred as a result of its use. Muscular Dystrophy UK does not necessarily endorse the services provided by the organisations listed in our factsheets.

Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on **0800 652 6352** or **info@muscular dystrophyuk.org** from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.muscular dystrophyuk.org