



# Inheritance and genetics

Many muscle-wasting conditions have a genetic cause, which means that they can be passed down generations (inherited). However not all are inherited in the same way. It's important to have an understanding of what condition you or your family has. We'd recommend reading this factsheet together with our factsheet relating to your muscle-wasting condition. You can find all our condition factsheets on our [website](#).

This factsheet provides a brief summary of genetics and typical inheritance patterns. Understanding these patterns within a family tells us about the likelihood of another family member being affected or not. This information is particularly important in family planning.

If you have any concerns or questions about inheritance and genetic testing, do discuss these with your GP who can refer you to your local genetics counsellor.

## DNA, chromosomes and genes

Deoxyribonucleic acid (DNA) contains instructions for the functioning of living organisms. Inside our cells, DNA is organised into structures called chromosomes. These are found in the cell nucleus.

Imagine your DNA as a recipe book, and your genes as the recipes. Our cells use these [genetic] recipes to make proteins, which are essential for a wide range of biological processes in the body.

Almost all of our cells contain 23 pairs of chromosomes (46 in total). These include one pair of sex chromosomes (XX for females, XY for males) and 22 pairs of non-sex chromosomes (autosomes). For each pair of chromosomes, we inherit one from our mother and one from our father. The rest come from one parent or the other. For example, some genes involved in energy production are inherited from the mother only. These genes are known to be associated with mitochondrial diseases (you can find more information in our [Mitochondrial myopathies](#) factsheet).

## What is a genetic mutation?

A genetic mutation is a change in the DNA code. This can happen by complete chance, for example if the cell makes a 'mistake' during DNA replication. Sometimes this happens as a result of environmental factors, such as smoking, excessive exposure to sunlight or exposure to radiation. Our cells have natural mechanisms for recognising mutations and correcting them, but these are not always 100 percent effective.

Depending on where a mutation occurs, and the type of mutation it is, the effect could be harmless. Or it could 'disrupt' a gene and result in a genetic condition such as muscular dystrophy.

## How are muscle-wasting conditions inherited?

Muscle-wasting conditions have different inheritance patterns, depending on the type of condition and which gene is mutated. Please read the MDUK factsheet relating to your condition to find out its inheritance pattern before referring to the points below.

### ► **X-linked (or sex-linked) recessive inheritance**

Including, but not limited to:

- Duchenne muscular dystrophy
- Becker muscular dystrophy
- Emery-Dreifuss muscular dystrophy
- X-linked myotubular myopathy (X-MTM)

These conditions are caused by a mutation in a gene on the X chromosome, which is one of the sex chromosomes. Males have one X chromosome and one Y chromosome; females have two X chromosomes.

If a male's X chromosome has the mutated gene, then he will have the condition. He has only one copy of the gene and the mutation isn't on the Y chromosome.

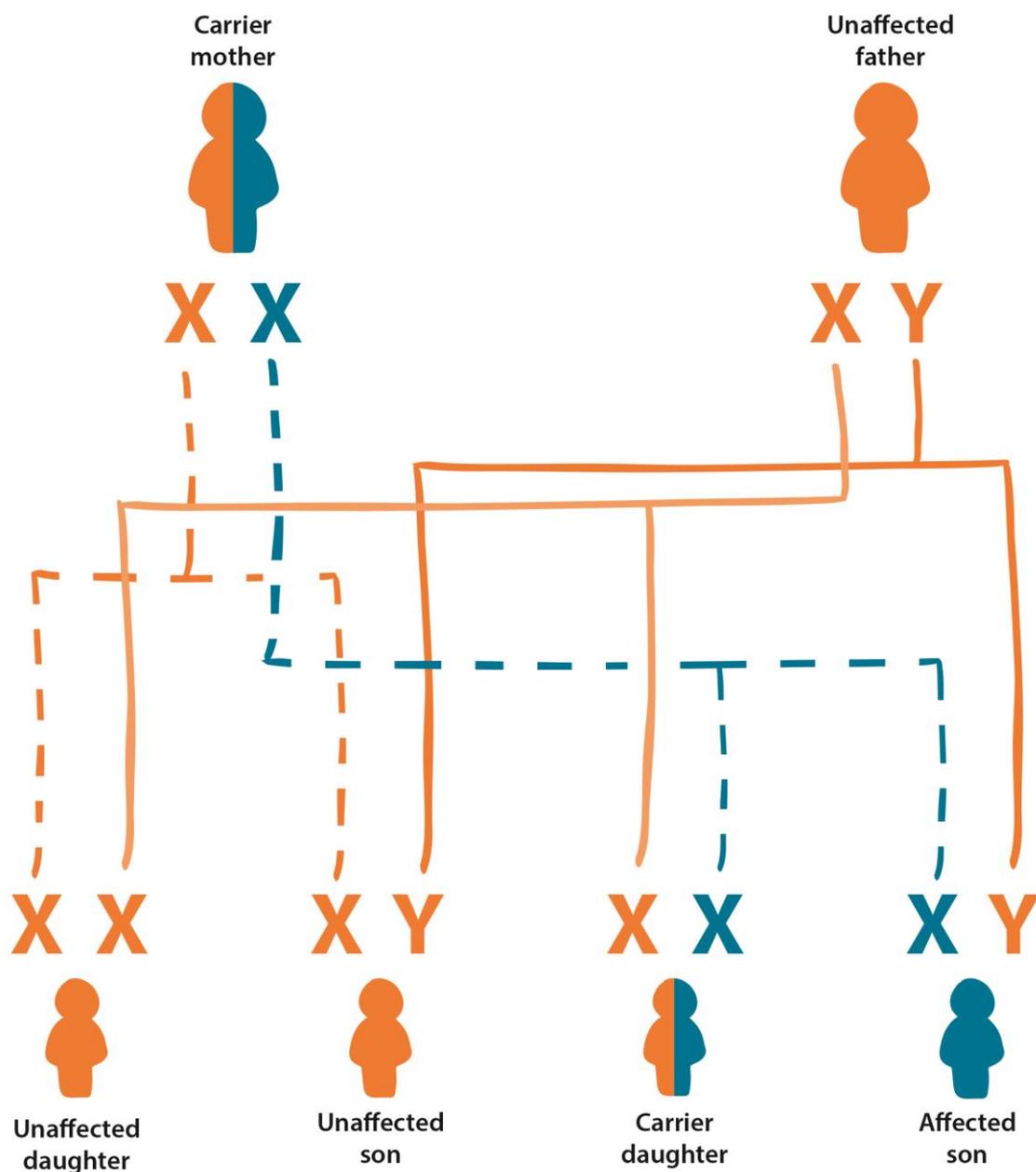
If a female has the mutated gene on one of her X chromosomes, she still has a healthy copy of the gene on her other X chromosome. This makes her a 'carrier' and she will not usually have symptoms of the condition.

If a carrier of an X-linked condition and a non-carrier have children, there is a 50 percent chance that each son will have the condition and a 50 percent chance that each daughter will be a carrier (see figure on p3). This probability is the same for each child born. So, for instance, if this couple have two sons, each son will have a 50 percent chance of inheriting the condition.

Unaffected males cannot pass on an X-linked condition. Affected males cannot pass an X-linked condition to their sons, but all of their daughters will be carriers (see figure on p4).



## X-linked recessive inheritance



X-chromosome with mutated gene



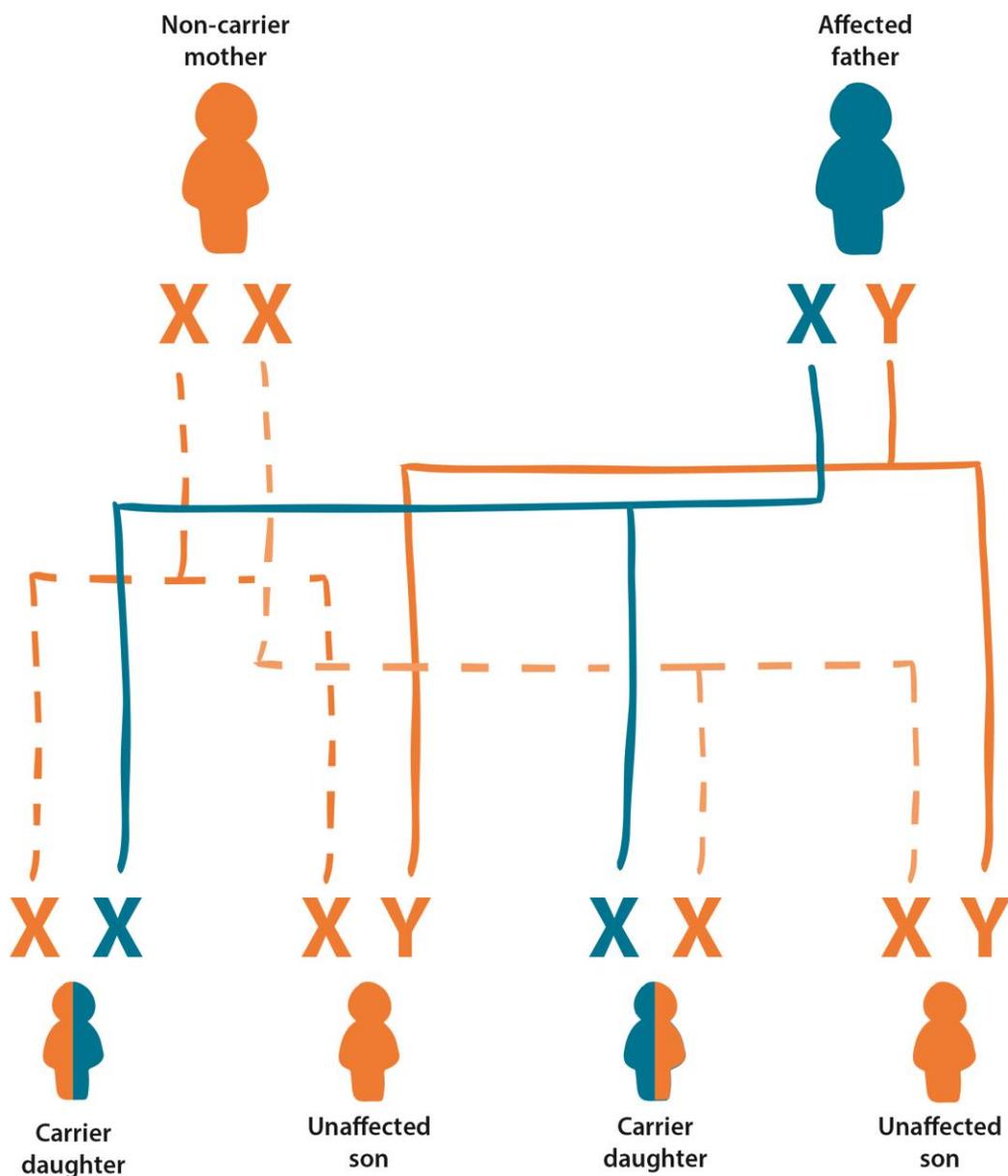
X-chromosome with healthy gene



Y-chromosome that does not contain the gene



## A father with X-linked muscular dystrophy



**X** X-chromosome with mutated gene    **X** X-chromosome with healthy gene    **Y** Y-chromosome that does not contain the gene

► **Autosomal recessive inheritance**

Including, but not limited to:      Type 2 limb girdle muscular dystrophies  
   Spinal muscular atrophy (SMA)  
   Some congenital muscular dystrophies

‘Autosomal’ means the mutation is in a gene on the non-sex chromosomes, so children of either sex can inherit the condition. ‘Recessive’ means both copies of that gene have to be mutated for someone to have the condition. If only one copy of the gene is mutated, that person is a carrier and will have no symptoms.

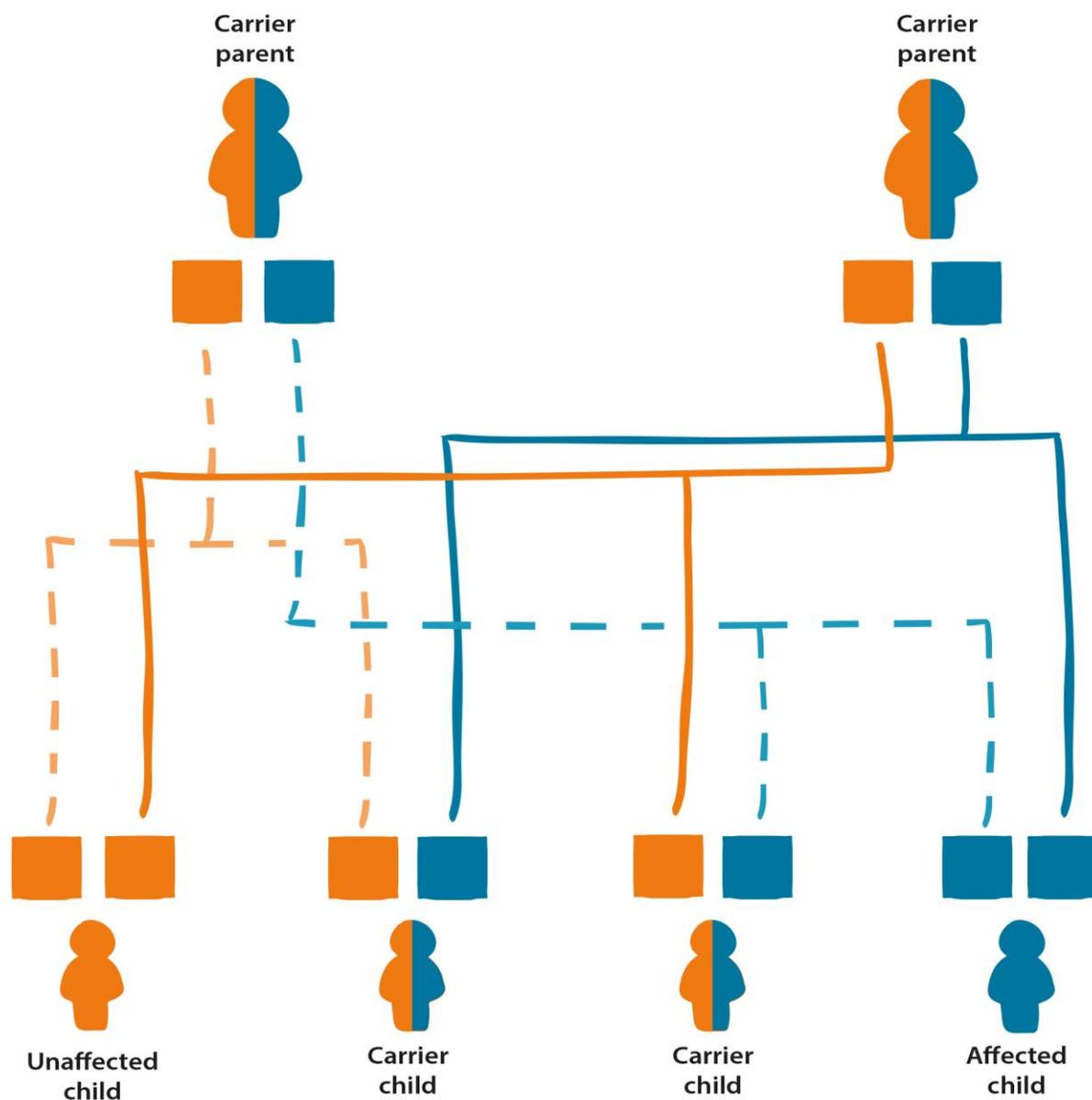
If mother and father are both carriers, (see figure on p6), there is a:

- 25 percent chance that each child will be unaffected
- 50 percent chance that each child will be a carrier
- 25 percent chance that each child will have the condition.

Parents with autosomal recessive conditions will pass on one copy of the mutated gene to their children (see figure on p7). It is very unlikely their children will be affected (unless the other parent is a blood relative).



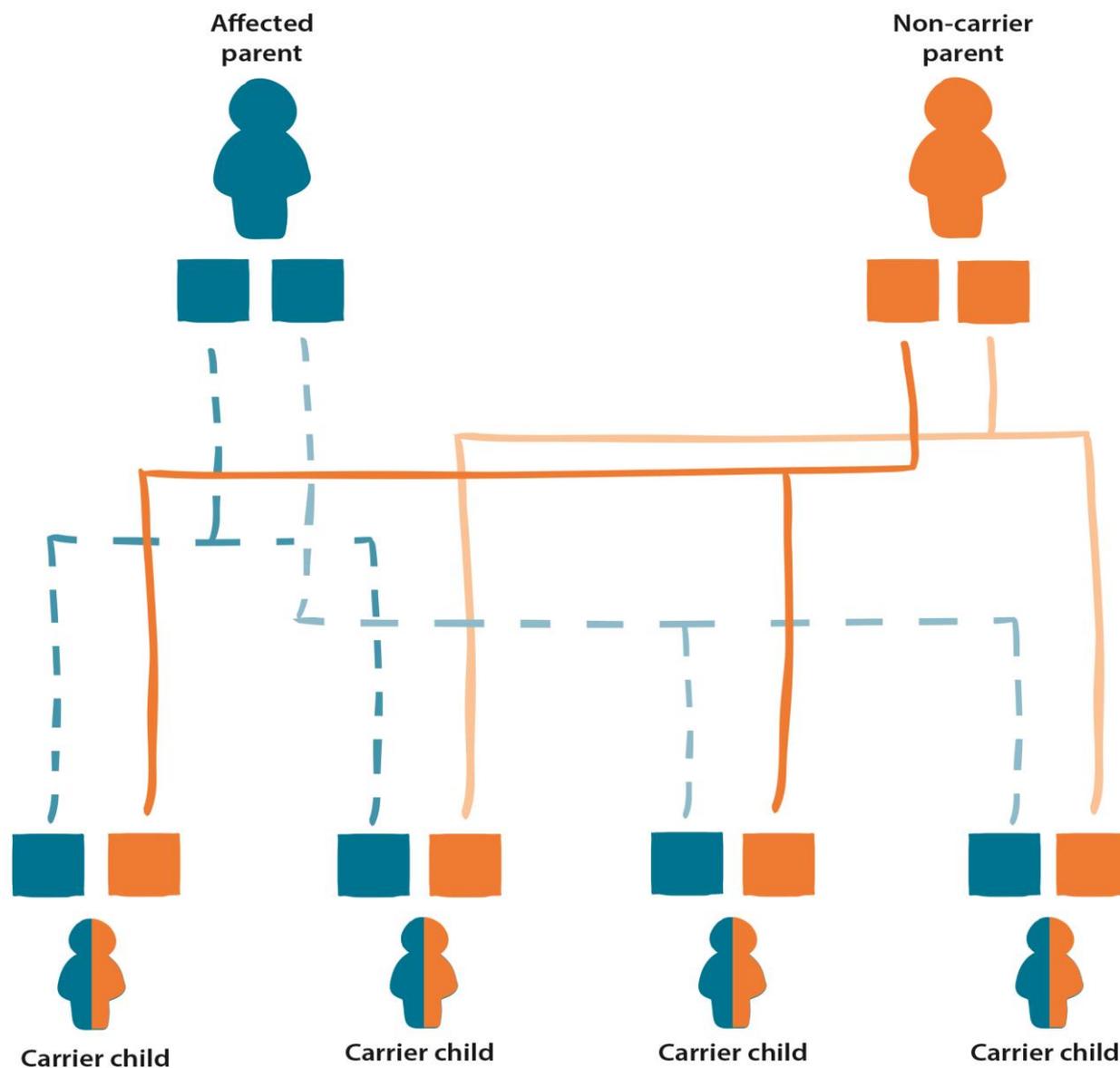
## Autosomal recessive inheritance



 Chromosome with mutated gene  
 Chromosome with healthy gene



## A parent with a recessive condition



-  Chromosome with mutated gene
-  Chromosome with healthy gene

▶ **Autosomal dominant inheritance**

Including, but not limited to:

- Facioscapulohumeral muscular dystrophy (FSHD)
- Myotonic dystrophy
- Most oculopharyngeal muscular dystrophies
- Type 1 limb girdle muscular dystrophies

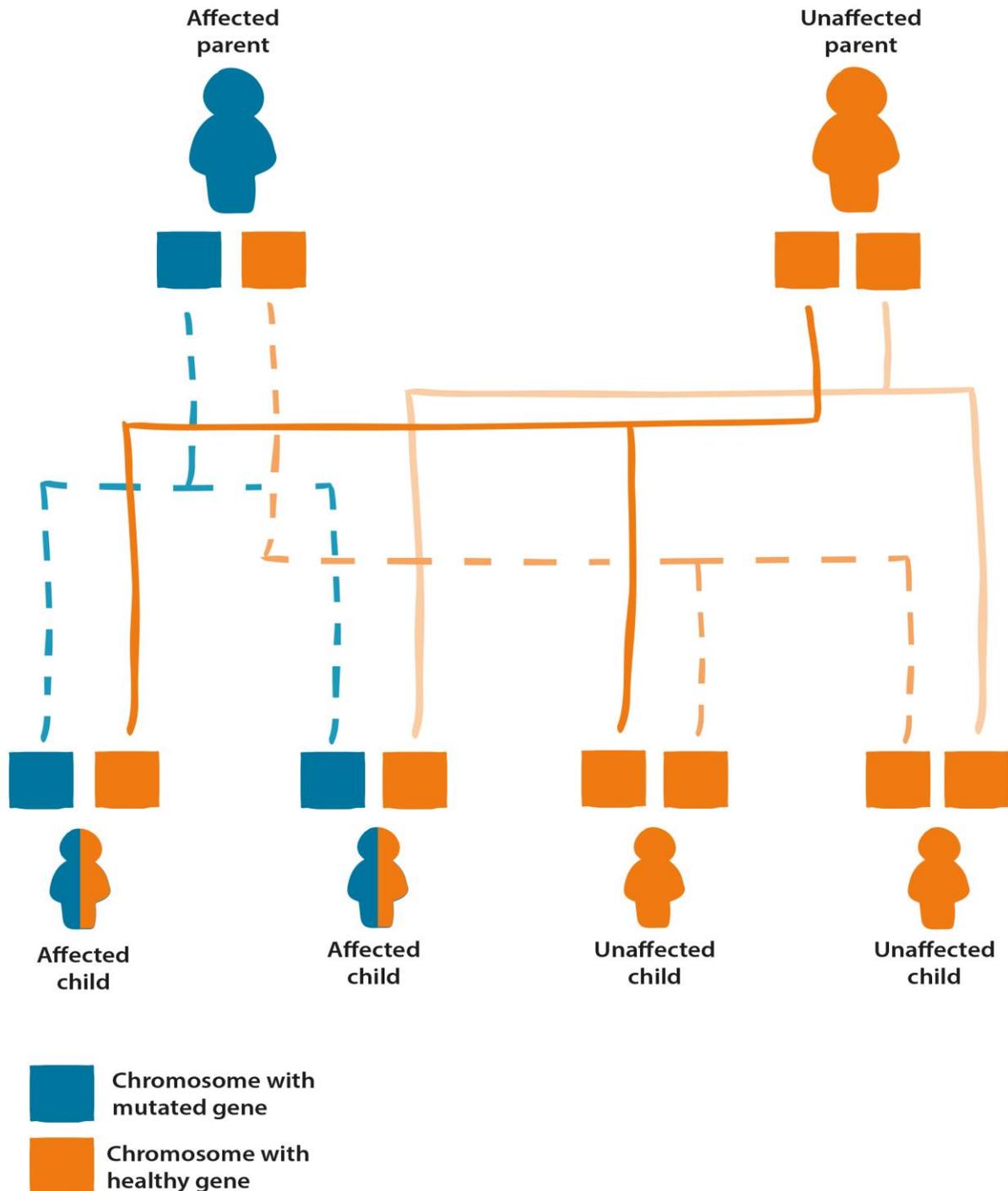
‘Autosomal’ means the mutation is in a gene on the non-sex chromosomes, so either sex can inherit the condition. ‘Dominant’ means the mutation has to occur in only one copy of the gene for someone to inherit the condition.

If one parent has the condition and the other doesn’t (see figure on p9), there is a:

- 50 percent chance that each child will have the condition
- 50 percent chance that each child will be unaffected.



# Autosomal dominant inheritance





## Are there ways of not passing the condition on to my children?

There are options available for some couples affected by muscle-wasting conditions to have an unaffected child. You can read about these in our [Genetic counselling and family planning](#) factsheet. We recommend speaking to your local clinical geneticist or genetic counsellor about what options might be available to you.

## Where can I get advice?

There are a number of specialist genetic centres throughout the country that do genetic tests and offer advice. Your GP can arrange a referral to a clinical geneticist or genetic counsellor at your local genetics centre.

### **Disclaimer**

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## 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 8.30 am to 6 pm 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](http://www.muscular dystrophyuk.org)**