What types of dystonia are genetic?

More than 20 genes causing dystonia have been identified, most relating to early-onset dystonia. Examples include genes causing primary generalised dystonia (DYT1), dopa-responsive dystonia (DYT5a/DYT14), segmental dystonia with laryngeal involvement (DYT6), myoclonus dystonia (DYT11) and a number of genes causing paroxysmal dystonia. All of these genes are dominant. There are also some recessive genes that cause early-onset dystonia such as DYT5b which also causes dopa-responsive dystonia.

However, genes have not been identified for all cases of early-onset dystonia – for instance the number of cases of primary generalised dystonia that currently have no known cause exceeds the number of DYT1 cases. The same is true for adult-onset where most cases have no known cause.

This does not mean these cases are not genetic – simply that no gene has yet been identified. Neurologists believe that more genes will be discovered. One possible reason that it is difficult to identify such genes is that they might have a low penetrance or that there are a number of susceptibility genes that work together to make an individual more likely to develop dystonia.

If you would like a full list of dystonia-causing genes, please call our helpline.

What to do if you have concerns or need more information

If you have concerns about the inheritance of dystonia then you should talk to the consultant responsible for your care. There are genetic tests that can be done but, as the genes currently identified explain so few cases of dystonia, they are of limited benefit except for some very specific and unusual types of dystonia. The tests are not routinely available so it will be at your consultant’s discretion whether to put you forward for a test.

What the Society offers

The Dystonia Society provides information and support those affected by dystonia in the UK. Please call us if you would like to become a member.

Our website, www.dystonia.org.uk, has information on dystonia and a lively forum. You can also sign up on the website for our free e-newsletter.

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Supported by:
People with dystonia often want to know if their child will inherit dystonia. This is a difficult question to answer as there are many different types of dystonia. Some types of dystonia have been linked to a gene and so can be inherited while others have an identified non-genetic cause (such as a stroke) and so cannot be inherited. However, for most cases there is currently no known cause, genetic or otherwise - it is possible these may have a genetic contribution but the gene(s) have not yet been discovered.

It is important to remember that, even if the dystonia has a genetic cause, this does not mean that it will necessarily be passed on to children. The reasons why are explained below.

**How is genetic dystonia passed on?**

**Genes** hold the information that determines how humans develop. We all have around 24,000 pairs of genes and each parent passes half their genes to their children. These genes match up into pairs again which contain one gene from each parent. Each gene is the genetic code for a specific protein which has a unique role in the development and functioning of the human body.

In each gene pair, only one gene affects the development of the child. This is called the **dominant** gene. The other gene does not affect development and is called **recessive**. When the child has their own children, only one of the genes in the pair is passed on - with the dominant and recessive gene each having a 50% chance of being the gene from the pair passed on.

When genes are received by the child, very occasionally there is a copying error called a **mutation**. Sometimes these are beneficial but often they are harmful and can cause health problems. When someone with a mutation has their own child, there is a 50% chance that this unhealthy (mutated) gene will be passed on. This is how genetic health conditions arise.

In most types of genetic dystonia, the unhealthy gene is **dominant** which means it will be active in the development of the child and so may cause dystonia. However, as dystonia is rare, it is unlikely that a dystonia causing gene will be inherited from both parents so the recessive gene will in most cases be healthy. So usually there is a 50% chance of someone with these types of dystonia passing an unhealthy gene to their child.

In a few types of genetic dystonia, the unhealthy gene is **recessive**. These genes are only active if both genes in the pair are unhealthy. In these cases, the person with dystonia will certainly pass a dystonia-causing gene to their children but, unless their partner is also a carrier (which is possible but highly unlikely), the other gene the child receives will be a healthy dominant gene which will prevent dystonia from developing.

**If a child inherits genes that can cause dystonia, will dystonia develop?**

Even if the child inherits genes that cause dystonia, this does not guarantee that dystonia will develop. This only happens if there is also some other, as yet unknown, trigger in the environment that results in the dystonia appearing. The probability that this happens is called the gene’s **penetrance**.

For example, the estimated penetrance of the DYT1 gene, a dominant gene that is one cause of generalised dystonia, is 30%. This means that dystonia only appears in 30% of cases where the DYT1 gene is present. As the chance of passing on the gene is 50%, the chance that dystonia will develop in a child of someone who has the abnormal DYT1 gene is 15%.

Unfortunately, the penetrance of other types of genetic dystonia is not well known so the chance of passing on dystonia is more difficult to estimate.