

It is also important to know that just as some people or children who have the gene mutation may not have any symptoms, people or children may have the above conditions but do not carry the known gene(s). This may be because the gene that is responsible for their dystonia has not yet been found.

Why do we need to know if the dystonia has a genetic basis?

Understanding the genetics of dystonia allows specialist neurologists and geneticists to make better diagnoses. They are then able to suggest the best available treatment. As more is learnt and understood about the genetic types of dystonia, treatment options will improve.

What do I do to find out if my children will inherit my dystonia?

First of all speak to your specialist neurologist who has a better understanding of your dystonia. Your specialist can refer you and/or your family to see a geneticist if there is a likelihood your dystonia has a genetic basis. Where there is a possible genetic cause, the specialist will discuss the possibility of genetic testing and counselling. This will help you understand the up to date information on your dystonia so that you can make decisions on your and your family's future.

Help us help people with dystonia by becoming a member, assisting with fundraising or joining/starting up a support group.

Go to our website or contact the association directly for information.

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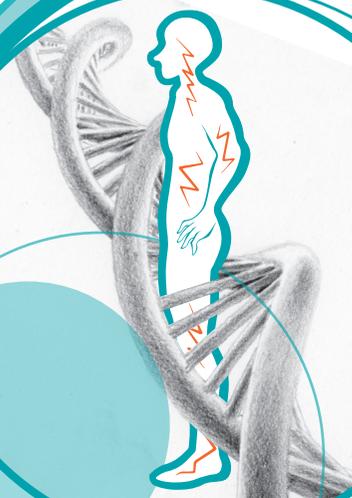
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Disclaimer

The information contained in this leaflet is of a general nature only. Please consult your Movement Disorder specialist or neurologist if you have specific questions regarding your condition.



Genetics in Dystonia



What causes Dystonia?

Many people with a diagnosis of dystonia do not have a reason for their symptoms. When no cause is found, it is generally accepted that their dystonia is related to a problem with the functioning and connections of various areas of the brain, including the basal ganglia and the cerebellum, that are involved in movement. For some people dystonia may be the result of certain medications, trauma or other neurological conditions such as stroke or infection. However, a genetic abnormality is sometimes found in people with certain types of dystonia.

How is Dystonia inherited?

People are born with 46 chromosomes. 44 of these are called autosomes or non-sex chromosomes (apply to either sex) and 2 are sex chromosomes (2 “X” chromosomes for females and an “X” and a “Y” for males). The chromosomes are present in pairs, with one of each pair inherited from each parent; such that half of the genetic material is from the mother and half is from the father.

Chromosomes are thread-like strands, arranged in pairs that are made up mainly of DNA (deoxyribonucleic acid) and contain the person’s genetic information. They are found in the nucleus (central part) of almost every cell in the body. There are many thousands of genes, and together they are responsible for each person’s genetic make-up, such as hair and eye colour, facial features etc. Just as each chromosome is present as a pair, each gene is also present as a pair; with one version of each gene inherited from the mother and the other version inherited from the father.

Each gene is responsible for a particular protein. Proteins have an important function in our bodies so if a gene is changed (such as occurs in a mutation), the protein for that gene may not work normally. This can result in a disease such as dystonia.

The mutations (changes) in the genes discovered so far in dystonia are mostly inherited in a manner we call autosomal dominant. This means that only one parent (mother OR father) needs to have the mutated gene for children to inherit it and each child has a 50% chance of inheriting that gene. However, in dystonia not all people with the gene mutation will have symptoms of dystonia or they may have very mild symptoms – this is called reduced penetrance. For instance the child or adolescent who carries the gene mutation causing DYT1 dystonia has only a 30-40% chance of showing many of the symptoms of generalised dystonia.

If the gene for dystonia is inherited in a manner we call autosomal recessive, then both versions of the gene (inherited from both mother and father) need to have a mutation present for dystonia to occur. In this case the parents are known as carriers, but will not have dystonia themselves. For each child that is born where both parents are carriers, there is a 25% chance that the child will inherit both copies of the mutated dystonia gene.

What types of Dystonia have a genetic basis?

At present, mutations in around 25 genes have been found in various forms of dystonia. However, it is important to understand that this represents a very small proportion of people with dystonia. Included in these is early-onset generalised dystonia in children and adolescents (DYT1) who carry a mutated TOR1A gene. This was the first gene isolated for dystonia. Among other genes found in dystonia are those causing dopa-responsive dystonia (DYT 5a / 5b / 14) who carry a mutated GCH1 or TH gene, myoclonus dystonia (DYT 11) who carry a mutated SGCE gene, and segmental dystonia with spasmodic dysphonia (laryngeal dystonia - DYT 6), who carry a mutated THAP1 gene.