

What is dystonia?

Dystonia is a neurological movement disorder. It causes involuntary muscle contractions, which lead the affected parts of the body to develop abnormal postures, with or without tremor. Dystonia may affect just one part of the body or several different parts. However, when it develops in adults it usually confines itself to one part of the body.

Dopa-responsive dystonia

Terms used to describe dopa-responsive dystonia include: DRD, Segawa's dystonia, Segawa's disease, DYT5 dystonia.

Dopa-responsive dystonia (DRD) is a broad term used to describe genetic inherited forms of dystonia that respond to a medication called **levodopa**. In many patients, onset is characterized by an abnormal, 'stiff-legged' manner of walking, with upward bending of the sole of the foot or turning of the foot outward at the ankle and a tendency to walk on the toes.

DRD may also extend to involve muscles of the arms and trunk. In addition, DRD is typically characterized by signs of Parkinsonism that may be relatively subtle. Such signs may include slowness of movement, stiffness and resistance to movement (rigidity), balance difficulties, and postural instability.

Many patients experience improvement with sleep, are relatively free of symptoms in the morning, and develop increasingly severe symptoms as the day progresses. Accordingly, this disorder has sometimes been referred to as '**progressive hereditary dystonia**'. It is a common characteristic of the condition that the symptoms will fluctuate and worsen as the day progresses.

DRD usually presents itself in childhood between the ages of 6 and 16 and is 2–4 times more likely to affect girls than boys. As with generalised dystonia, DRD has reduced penetrance. This means that only about 30–40% of people who carry the gene will actually go on to develop DRD.

"Much research is ongoing and is progressing towards a greater understanding of the condition..."

What causes dystonia?

Dystonia is believed to be due to a problem in an area of the brain called the *basal ganglia*, which are structures deep within the brain that control voluntary movements and postures.

The most common form of DRD, is often referred to as DYT5 dystonia. DYT5 is a dominantly inherited condition caused by mutations in a specific gene. A dominantly inherited disorder means that only one parent needs to have the DYT5 genetic mutation, in order for the child to inherit the disorder. Individuals with DYT5 DRD often have a parent who also has the mutation, with or without symptoms. The DYT5 mutation has variable penetrance, meaning that not everyone who inherits the mutated gene will develop symptoms. Children of a parent with the DYT5 mutation have a 50% chance of inheriting the mutation. At this time, there is no way to predict whether a person with the mutation will go on to develop dystonia.

The gene abnormality in DRD causes a problem with the brain's production of the chemical dopamine, and this is what causes the dystonia and other movement disorders.

How may DRD be treated?

Symptoms of DRD can usually be treated effectively with **levodopa** (Sinemet). In many cases, full physical functionality is restored. Even those whose symptoms have been untreated for up to 50 years respond to Levodopa and a stable response after years of continuous treatment is typical. Side effects of the medication may include light-headedness, nausea, constipation, and, in some cases, fatigue.

- The goal of drug therapy is complete remission of symptoms, including dystonia and Parkinsonism when present.
- Dyskinesia may very rarely occur with initiation of treatment or in older individuals who are treated with relatively higher doses of levodopa. Dyskinesia (involuntary movements) may be reduced or eliminated by reducing the dose of levodopa.
- Physical therapy may be helpful if problems with walking have been identified or if shortening of the muscles occurred whilst DRD was uncontrolled. However, the role for physical therapy is limited, because most patients have complete response to drug therapy.

How do I live with Dopa-responsive dystonia?

As this form of dystonia may be genetic, some people find it helpful to speak to a genetic counsellor, regarding the genetic issues, and what this may mean for you and your family.

What the Society may offer

The Dystonia Society is dedicated to providing information and support to everyone affected by dystonia in the UK and to raising awareness of the condition and the needs of everyone affected. The Society is also committed to ensuring that everyone with dystonia has access to the treatment they need.

Call the Dystonia Helpline on:

0845 458 6322

Important note *The contents of these pages are provided only as information and in no way intended to replace the advice of a qualified medical practitioner. The Society strongly advises anyone viewing this material to seek qualified medical advice on all matters relating to the treatment and management of any form of medical condition mentioned. Furthermore, rapid advances in medicine may cause information contained here to become outdated after some months.*



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