

The myotonic dystrophies

An overview of the condition, cause, and management

“MDUK provides us, who either have myotonic dystrophy or are otherwise affected by it, with hope. Importantly this hope is underpinned with practical help based on knowledge and determination to push boundaries, focusing on improvements to quality of life and ultimately to effect a cure.”

Jeanette Charlton, whose husband and daughter have myotonic dystrophy

What are the myotonic dystrophies?

Myotonic dystrophy is a genetic condition that causes progressive muscle weakness and wasting. It typically affects muscles of movement and commonly the electrical conduction system of the heart, breathing muscles, swallowing muscles, bowels, lens of the eye and brain. The age when symptoms start varies a lot and can be any time from birth to old age. In general, the later the condition starts, the milder it will be. There are two types of myotonic dystrophy. Type 1 tends to be more severe and more common in the UK than type 2. This factsheet will refer to only myotonic dystrophy type 1 apart from the section specific to myotonic dystrophy type 2 highlighted on page 4.

Myotonic dystrophy type 1 (DM1) - how will it affect me?

Muscles of movement

There are two problems that may affect muscles of movement or ‘skeletal muscle’.

The first is a gradual weakening of certain muscles, over time, caused by a ‘muscular dystrophy’. The muscles in the face, eyelids, jaw and neck are commonly affected. The muscles in the forearm that enable us to grip objects are often affected early on. The muscles around the ankles are also commonly affected, causing ‘foot drop’, which cause people to catch their toes and trip. The large, weight-bearing muscles of the legs and thighs are usually affected much later.

The rate of deterioration in muscle strength is typically slow over many years. Some people never have significant muscle weakness. Some people, who are more severely affected, may need a wheelchair, but many people with myotonic dystrophy do not.

The second problem affecting skeletal muscles is an electrical problem, called ‘myotonia’. Myotonia causes muscle stiffness (difficulty relaxing the muscle), which especially affects the hands and jaw. This can be helped with medication supplied by your specialist but usually does not need treatment.



Heart

The wiring or 'conduction system' of the heart is often affected and this can lead to attacks of dizziness or even blackouts. Sometimes a pacemaker or other device may be recommended. The mechanical system which pumps the heart may be affected but usually it does not cause a problem that needs treatment.

Lungs

The pumping of air in and out of the lungs, or 'ventilation', may be affected. This may cause daytime sleepiness and increase the risk of chest infections, especially after a general anaesthetic. Pneumonia is very common and requires prompt treatment. You should stay up-to-date with your immunisations, including the flu and pneumococcal vaccines.

Gastrointestinal (GI) tract

Because the swallowing muscles are affected, swallowing can often lead to coughing and spluttering when eating or drinking. This can be dangerous. If food or drink goes into the lungs rather than the stomach, this leads to 'aspiration pneumonia', which is a severe chest infection. The lower bowels are also typically affected by irritable bowel syndrome-like symptoms, with a mixture of constipation, crampy abdominal pain and diarrhoea.

Lens of the eye

It is very common for cataracts to form across the lens of the eye in people with myotonic dystrophy, but they usually cause few symptoms because they appear so slowly. However, early onset cataracts can be easily treated.

Brain

The brain can be affected in many ways and excessive daytime sleepiness is one of the most common consequences. Reduced motivation can also be a common problem.

Children with myotonic dystrophy may have learning difficulties, even if they have no muscle problems.

How is myotonic dystrophy inherited?

The condition follows a 'dominant' inheritance pattern, which means that children of a person with the condition have a 50 percent chance of inheriting it. Men and women are equally likely to be affected and to pass on the condition, but children of women with the condition are likely to be more severely affected. In general, though not always, the condition tends to be more severe with each generation OR in each younger generation. Please also see our factsheet on congenital myotonic dystrophy for more information on the most severe form of myotonic dystrophy.

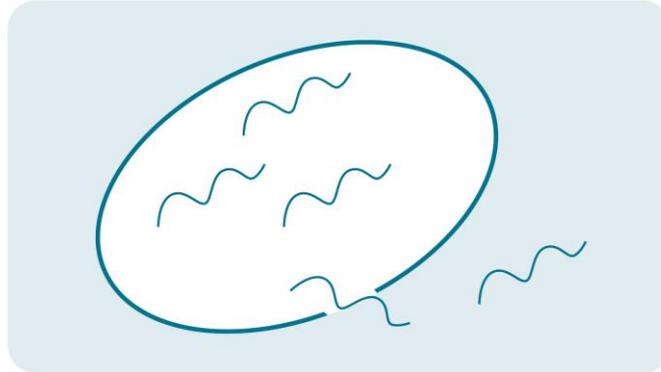
Very few cases of myotonic dystrophy occur 'out of the blue' with no evidence of family history. Almost always, one parent is affected, often very mildly. Some parents (or grandparents) may carry a very slight genetic change that will never cause any symptoms and may not be diagnosed. Careful study of the whole family often shows more members with the condition than would appear likely at first.



What happens inside cells affected by myotonic dystrophy?

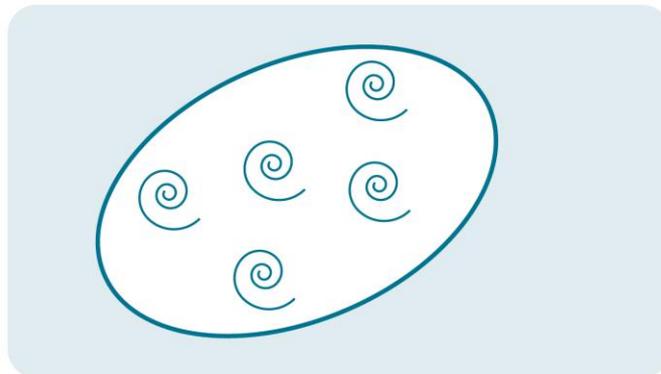
Healthy cell

DMPK RNA message leaves nucleus = normal cell functioning



Myotonic dystrophy cell

Trapped DMPK RNA forms clumps in nucleus = disrupted cell functioning





Myotonic dystrophy type 2 (DM2) or PROMM (proximal myotonic myopathy)

An important recent advance is the recognition of a second, milder condition with features resembling myotonic dystrophy type 1 (DM1). In this condition, weakness tends to occur in muscles close to joints (proximal muscles), such as those in the neck, fingers, elbows and hips. Facial weakness and myotonia (difficulty relaxing the muscle) is often mild or absent. Cataracts and heart problems can occur but are usually less severe than in DM1.

This condition was initially called proximal myotonic myopathy (PROMM) because affected individuals had clinical signs of myotonic dystrophy but did not have the increased number of CTG repeats in the DMPK gene. It was later discovered that these individuals instead had a similar genetic fault in a different gene. This led to the condition being renamed as myotonic dystrophy type 2 (DM2).

People with myotonic dystrophy type 2 have a genetic fault (mutation) in the CNBP gene (also called the ZNF9 gene) on chromosome 3. Although this gene is quite different from the DMPK gene that is mutated in myotonic dystrophy type 1, it contains a very similar, repeated section of DNA made up of lots of C's, T's and G's:

CCTGCCTGCCTGCCTGCCTGCCTGCCTGCCTGCCTG...

The number of CCTG repeats in the CNBP gene is increased in people with myotonic dystrophy type 2. This is thought to disrupt RNA production in the cell, in a similar way to myotonic dystrophy type 1 (see figure above). Clumps of CNBP RNA build up in the cell nucleus and interrupt the activities of important proteins and genes. Exactly how this leads to symptoms is not well understood, but researchers are looking into this.

In contrast with type 1, the number of CCTG repeats does not relate to the age of onset or severity of myotonic dystrophy type 2. In addition, type 2 does not tend to get worse with each generation.

However both types are dominantly inherited. This means that children of an affected person have a 50 percent chance of inheriting it.

Clinicians and scientists are still learning about myotonic dystrophy type 2, but it is probably uncommon and accounts for only a small proportion of people in the UK thought to have a myotonic dystrophy.

Treatment and management:

Although there is currently no cure for myotonic dystrophy, there are ways to help manage the condition. Since many doctors are unfamiliar with the condition (because it is so rare), it is essential that people who have myotonic dystrophy are aware of the problems and complications they may face and share this information with their health professionals. Some of these are mentioned on pages 1 and 2. They do not always all occur, and many people have few symptoms, but it is still important to be aware of them.



Some specialist centres now recommend an active monitoring process of people with myotonic dystrophy type 1 so that the effects of the condition can be found and managed before they become a severe problem.

What are the risks with anaesthetics?

Operations and anaesthetics can be risky, even for people mildly affected. It is very important that surgeons and anaesthetists know that a person has myotonic dystrophy before any surgery is planned. Problems only usually occur when doctors are unaware of the condition.

It is also helpful to carry a myotonic dystrophy alert card, and a care plan, both of which contain information to alert emergency and other healthcare professionals to the specific issues that affect people living with myotonic dystrophy. These are available for free from Muscular Dystrophy UK – call the freephone helpline on 0800 652 6352 or email info@muscular dystrophyuk.org to order one.

Related health issues:

Many people can develop heart problems, which are commonly treatable but can be serious if ignored. Speak to your neurologist or GP about having an annual electrocardiogram (ECG). This can be performed by a cardiologist.

Some people who have myotonic dystrophy may have more trouble with other body systems than they do with their muscles. For example, excessive daytime sleepiness, swallowing difficulties and a range of bowel symptoms may appear unrelated, but they may all be connected to myotonic dystrophy. They may be helped by specific drugs. It is important that people with myotonic dystrophy make sure that whoever treats them is aware they have the condition and knows the wide range of associated problems.

Things to think about during a pregnancy:

Women with the condition need careful management if planning a pregnancy: not only is there a risk of a baby being severely affected, but sometimes serious problems during pregnancy and delivery may also affect the mother. If a couple are concerned their baby may be affected it is possible to test a pregnancy. Your GP can refer you to your local clinical genetics department for more information and testing. Please see our [genetic counselling and family planning](#) factsheets for more information.

Will I need specialist equipment?

Equipment for mobility and adaptations in the house can be very useful. Weak neck muscles may require a head-rest when driving.

The Joseph Patrick Trust (JPT) is a welfare fund within Muscular Dystrophy UK that provides grants towards the costs of specialist equipment such as powered wheelchairs, adapted computers and electric beds, for children and adults with muscular dystrophy or a related neuromuscular condition.

For information about Joseph Patrick Trust grants, email: JPTgrants@muscular dystrophyuk.org

Our advocacy team is here to help anyone with a muscle-wasting condition to get the care and support, services, benefits and equipment they are entitled to. Contact them on 0800 652 6352 or info@muscular dystrophyuk.org

Research into myotonic dystrophy

Advances in research over the past 10 years have increased our understanding of the underlying cause of the condition. This knowledge is important in order to develop effective treatments. Some potential treatments have shown promising results in myotonic dystrophy mouse models. However, it is important to remember that mice are very different to humans, so these treatments now have to go through several rigorous clinical trials to show that they are safe and effective in people with myotonic dystrophy.

Find out the latest news on myotonic dystrophy research and clinical trials by visiting our website at: www.muscular dystrophyuk.org/news/news/research/

To find out more about the research that Muscular Dystrophy UK is funding, please visit: www.muscular dystrophyuk.org/progress-in-research/research-projects/current-grants/

If you have any questions, please contact the research team at research@muscular dystrophyuk.org

Other related publications and resources:

- ▶ Congenital myotonic dystrophy
- ▶ [Inheritance and muscular dystrophies](#)
- ▶ [An introductory guide for families with a child newly diagnosed with a neuromuscular condition](#)
- ▶ [Inclusive education guide](#)
- ▶ [Muscle biopsies](#)
- ▶ [Heart check](#)
- ▶ [Pregnancy and fertility](#)
- ▶ [Prenatal testing and diagnosis](#)
- ▶ [Carrier testing and reproduction: your options](#)
- ▶ [Exercise advice for adults with muscle wasting conditions](#)
- ▶ www.muscular dystrophyuk.org/talkmd/
- ▶ Joseph Patrick Trust: www.muscular dystrophyuk.org/jpt



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 you 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.

If you would like your GP or other healthcare professional to have more information about myotonic dystrophy, we have some relevant materials. We've developed an online training module for GPs, as well as one for physiotherapists working with adults with muscle-wasting conditions. Contact our helpline or email us to find out more.

If you have feedback about this factsheet or would like to request any references used to produce it, please email info@musculardystrophyuk.org.

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@musculardystrophyuk.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.musculardystrophyuk.org