Congenital myotonic dystrophy

An overview of the condition, cause and management

> "When my granddaughter was first diagnosed we, and everyone involved, did not have a clue what we were dealing with. Getting the right information to families and professionals through a factsheet like this is essential as so little is known about the condition."
>
> Linda Atkinson, whose granddaughter has congenital myotonic dystrophy

What is congenital myotonic dystrophy?

This is a form of myotonic dystrophy type 1, also known as Steinert's disease. *Congenital* means 'from birth' and the condition is usually identified at birth or soon after; *myotonic* means 'involving muscle tone' and *dystrophy* means 'wasting away'. The condition usually occurs when the mother already has myotonic dystrophy type 1 (although she may not be aware of it) and then it is passed on to her child in a more severe form. Very occasionally it can be passed on by an affected father, but this only occurs in one percent of cases. For more information about myotonic dystrophy type 1, please read our myotonic dystrophies factsheet.

The symptoms of congenital myotonic dystrophy appear from birth. In other kinds of myotonic dystrophy the symptoms usually appear in early adulthood.

(Please note: congenital myotonic dystrophy is not the same as congenital myopathy or congenital muscular dystrophy. For more information about these or other conditions please contact Muscular Dystrophy UK's care and support team.)

How common is it?

This condition is very rare. However there are families, groups and specialists known to Muscular Dystrophy UK who you can contact for support, advice and information.

How severe is it?

Up to 25 percent of affected children may not survive past their first birthday. Children who live beyond this are likely to live into adulthood.

What is the treatment/management of the condition?

Currently there are no treatments or cures for congenital myotonic dystrophy. However, physiotherapy and occupational therapy are very important in improving or maintaining a child's physical condition. It is important that the condition is managed by a neurologist, and referral to one can be made by your child's paediatrician.

Heart problems can develop with the condition, which are commonly treatable but can be serious if ignored. Speak to your child's neurologist or paediatrician about your child having an annual electrocardiogram (ECG). This can be performed by a cardiologist.

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Can my child have an anaesthetic?

Anaesthetics can be risky. It is very important that surgeons and anaesthetists are aware a child has congenital myotonic dystrophy before surgery is planned.

It is also helpful to complete a neuromuscular care plan, which contains information to alert emergency and other healthcare professionals to the specific issues that affect people living with congenital myotonic dystrophy. These are available for free from Muscular Dystrophy UK – call the freephone helpline on 0800 652 6352 or email info@musculardystrophyuk.org.

Can a child with congenital myotonic dystrophy have the usual immunisations?

Yes.

What are the symptoms?

Children born with congenital myotonic dystrophy often have problems breathing as soon as they are born and may need help to breathe, using a ventilator. Suction may also be necessary to remove any secretions in their lungs. Breathing problems may continue, and can be very severe and life-threatening, especially if the baby is premature. Once the neonatal period (28 days after birth) has passed, their breathing problems tend to improve.

Children with congenital myotonic dystrophy may have 'floppy baby syndrome', which means they have difficulty moving their arms, legs and head. For example, they may have little or no control of their neck muscles, meaning their head tends to flop forwards or backwards. This usually improves with age. It is important that they have physiotherapy from a very young age to help with breathing, and to encourage movement and strength.

The child may also have poor head control and, often, facial weakness. They may lack facial expression, simply because they may be unable to make the usual range of facial movements. This may improve with time. Parents, family, friends and care professionals, and later, teachers, should be made aware of this.

Older children with congenital myotonic dystrophy tend to have poor concentration and tire easily. Attending a therapeutic playgroup can often stimulate the child's learning and development.

The motor milestones (physical achievements, such as sitting unaided) and the intellectual milestones tend to be delayed in a child with congenital myotonic dystrophy. They may have difficulties with speech, particularly with clear pronunciation. Speech and language therapy can help.

Facial weakness may make it difficult for babies to suckle and feed but advice from a relevant health professional can help with this. Babies often have swallowing and therefore feeding difficulties. They may bring up their food, have bouts of colic (excessive crying) and need food supplements. A Haberman feeding teat can be helpful for some babies.

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Others may need a feeding tube (nasogastric tube) or – at times of illness – a drip to help with feeding.

Some children have a squint, and very occasionally children may have impaired vision.

Children commonly have club feet (talipes), or twisted feet. This may be mild or severe and will improve with physiotherapy. The more severe form, however, will require corrective surgery. Physiotherapy, with passive stretching, is important to help with foot problems.

In some cases, special shoes, walking aids and callipers may be needed. A few children with the condition may need to use a wheelchair.

Children sometimes take longer to control their bladder and bowels. Bladder control usually improves, however bowel problems – especially constipation – may persist, owing to problems with the muscle of the bowel wall.

Children with congenital myotonic dystrophy may have more trouble with other body functions than they do with their muscles. A symptom that may appear to be totally unrelated may in fact be connected to their condition. It is important that health professionals treating them are aware of the wide range of associated problems.

Will the condition improve?

The symptoms of the condition may improve during childhood, but are likely to deteriorate later in life.

Can the condition be diagnosed during pregnancy?

If a couple are concerned their baby may be affected, it is possible to have tests during 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.

What are your risks during pregnancy?

During her pregnancy, the mother of a child with congenital myotonic dystrophy may notice that the baby is not moving around in the womb as much as might be expected. She may have polyhydramnios (excessive amounts of amniotic fluid) and go into premature labour.

The mother may not be aware of having myotonic dystrophy herself until after the birth of her baby. If she does know she has myotonic dystrophy, staff should be made aware during the pregnancy and at the time of delivery, that the baby may need immediate intensive care. This may mean that the baby will need to be resuscitated or taken away for extra help with their breathing. The parents should also be made aware of these procedures. Please see our *Genetic counselling and family planning* factsheets for more information.

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How is congenital myotonic dystrophy inherited?

In 99 percent of families, congenital myotonic dystrophy is inherited from an affected mother. In one percent of cases, the father is the affected parent. Myotonic dystrophy follows a 'dominant' inheritance pattern. This means that people with the condition (including the congenital form) have a 50:50 chance of passing it on to their children.

Once a mother has had one child affected with congenital myotonic dystrophy, any of her other children are also very likely to have the same condition. Children who do not inherit the genetic change that causes it will not be affected by myotonic dystrophy and cannot pass it on to their children either. Congenital myotonic dystrophy can affect both sexes, with the mother almost always being the affected parent.

What is the cause?

Congenital myotonic dystrophy is caused by a specific genetic change (mutation) within the DMPK gene on chromosome 19. Our DNA is made up of lots of individual building blocks represented by the letters A,C,T and G. The DMPK gene contains lots of Cs,Ts and Gs repeated over and over like this:

CTGCTGCTGCTGCTGCTGCTGCTGCTGCTGCTG…

The CTG ‘triplet’ in the DMPK gene is usually repeated between five and 34 times in a healthy person. However, in someone with myotonic dystrophy type 1 (including the congenital form), the CTG triplet is unstable and is repeated more than this.

The number of CTG repeats in the DMPK gene generally correlates with the severity of the condition and the age of onset. For example, individuals with 50 to 100 repeats are usually mildly affected and symptoms may not arise until adulthood. In contrast, individuals with thousands of repeats are likely to have the severe, congenital form described in this factsheet.

The number of CTG repeats, and hence the severity of the condition, usually increases from generation to generation. This is why it is possible for a mildly-affected parent to have a child with congenital myotonic dystrophy.

Recent scientific advances have shown that too many CTG repeats leads to abnormal RNA production in the cells of people with myotonic dystrophy type 1. When a gene is expressed (or active), RNA copies of the gene are produced in the cell nucleus (see figure below). This RNA acts as a messenger that travels out of the nucleus and then instructs the cell’s machinery to make proteins.

In myotonic dystrophy type 1, this RNA production process is disrupted. Too many CTG repeats in the DMPK gene cause the resulting RNA to have an unusual shape (see figure below). It becomes trapped in clumps in the nucleus and affects the function of important proteins that regulate other genes. It is the disruption of these other genes that is thought to cause the body-wide symptoms of myotonic dystrophy type 1.

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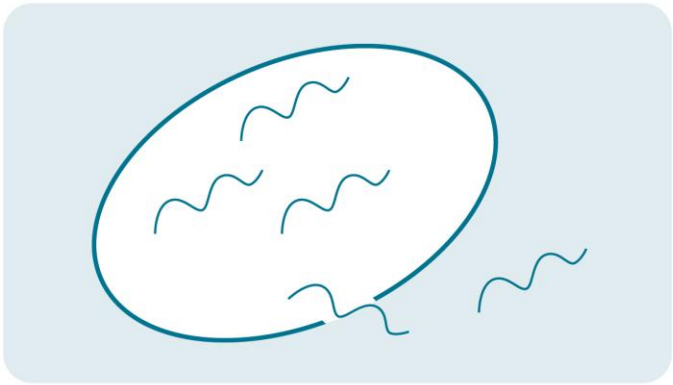
Muscular Dystrophy UK
Fighting muscle-wasting conditions

What happens inside cells affected by myotonic dystrophy?

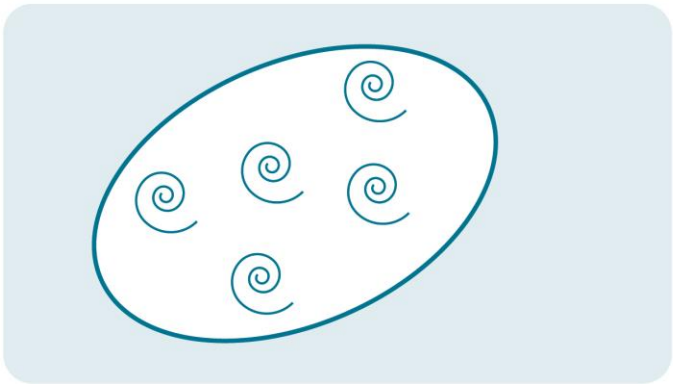
Healthy cell

DMPK RNA message leaves nucleus = normal cell functioning



Myotonic dystrophy cell

Trapped DMPK RNA forms clumps in nucleus = disrupted cell functioning



Find out the latest news on congenital myotonic dystrophy research by visiting our website at: www.musculardystrophyuk.org/news/news/research/

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

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

Other related publications and resources:

- The myotonic dystrophies
- Inheritance and muscular dystrophies
- An introductory guide for families with a child newly diagnosed with a neuromuscular condition
- Inclusive education guide
- Muscle biopsies

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Health & care information you can trust
The Information Standard Certified Member

- Heart check
- Pregnancy and fertility
- Prenatal testing and diagnosis
- Carrier testing and reproduction: your options
- Exercise advice for adults with muscle wasting conditions
- www.musculardystrophyuk.org/talkmd/
- Joseph Patrick Trust: www.musculardystrophyuk.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 your 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 health professional to have more information about congenital 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.

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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

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