

Congenital myotonic dystrophy

With thanks to Professor Peter Harper for his help in producing this Fact Sheet for the Muscular Dystrophy Campaign

1 What is congenital myotonic dystrophy?

Congenital myotonic dystrophy is the early childhood form of myotonic dystrophy (also known as Steinert's disease). Usually in myotonic dystrophy the symptoms begin to show in childhood or later in life, but symptoms of congenital myotonic dystrophy are evident from birth. It occurs only when the mother already has myotonic dystrophy (although she may not be aware of this) and she passes it on to the child in a more severe form. *Congenital* means "from birth" because the condition is usually identified at birth or soon after; *myotonic* means "involving muscle stiffness"; and *dystrophy*, "muscle wasting and weakness".

(Congenital myotonic dystrophy is not the same as congenital myopathy or congenital muscular dystrophy. For more information about these or other conditions please contact the Muscular Dystrophy Campaign's Information Officers.)

2 How common is it?

This condition is rare, but a family affected by congenital myotonic dystrophy is not alone: there are many other families, groups and specialists who can be contacted and who offer support, advice and information.

3 What are the symptoms?

Often babies with congenital myotonic dystrophy have problems with breathing after delivery and may need to be helped, using a ventilator. Suction, to remove any secretions in the lungs may also be necessary. Respiratory problems may continue after the birth, and can be very severe and life threatening, especially if the baby is premature. Once the neonatal period (28 days after birth) has passed, the respiratory problems tend to improve.

The baby is often floppy which means that she or he has poor muscle tone. This usually improves with age. It is important that physiotherapy should be practised on the baby from a very young age to help with breathing and lung function, and to encourage movement and strength.

The baby may have poor head control.

There is commonly facial weakness and a lack of facial expression. This does not mean that the child is unresponsive, it is simply that he or she may be unable to make the usual range of facial movements although these may improve with time. Parents, family, friends and care professionals, and later, teachers, should be aware of this.

Older children tend to have poor motivation and concentration and are easily tired. If a child can attend therapeutic playgroup, this can have an important effect on stimulating learning and development, which will stand the child in good stead later in life.

The motor milestones (physical achievements such as sitting unaided) and the intellectual milestones that a healthy child reaches by a certain age, tend to be delayed in a child with congenital myotonic dystrophy.

There may be speech difficulties, particularly with clear pronunciation. Speech therapy can be of help.

A baby often has swallowing and therefore feeding difficulties. She or he may regurgitate food, have bouts of colic and need food supplements. A haberman-feeding teat can be helpful for some babies. Some babies may need a feeding tube (nasogastric tube) or even at times of illness a drip (intravenous infusion) to help with feeding.

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

Children commonly have club feet (talipes). This may be mild or severe; both improve with physiotherapy but the severe form will require corrective surgery. Physiotherapy is important, with passive stretching to help with the foot problems.

The development of control over the bladder and bowel are sometimes delayed. Bladder control usually improves to become normal but bowel problems especially constipation may be persistent due to the muscle of the bowel wall being involved.

As you can see, youngsters with congenital myotonic dystrophy may have more trouble with other body systems than they do with their muscles. A symptom that may appear to be totally unrelated may in fact be connected. It is important that whoever treats them is aware of the wide range of associated problems.

4 how early is diagnosis made?

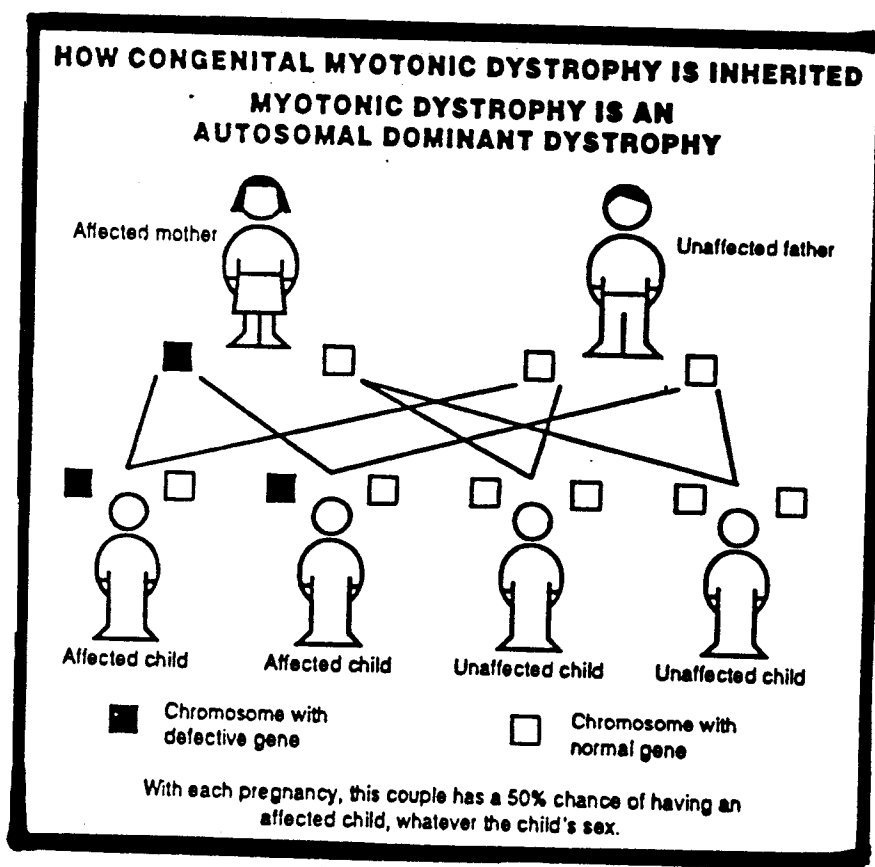
During her pregnancy, a mother of a child with congenital myotonic dystrophy may have noticed that the baby was not moving around in the womb as much as normal, and had reduced fetal movements. She may have had hydramnios (excessive amounts of amniotic fluid) and premature labour. The mother may not be aware of having myotonic dystrophy herself until after the birth of her affected baby. At the time of delivery, (if the baby is known to have congenital myotonic dystrophy antenatally), staff should be aware that the baby may need immediate intensive care and the parents should be made aware of the procedures.

5 how severe or mild is it?

Congenital myotonic dystrophy can vary considerably in severity from child to child. If a child is diagnosed with the condition soon after birth, symptoms are likely to be severe. In these cases, special shoes, walking aids and calipers may be needed. A few affected children need to use a wheelchair. Sadly, congenital myotonic dystrophy can be fatal, especially in the early weeks of life, but a child who lives beyond his or her first birthday is likely to live to become an adult.

6 How is congenital myotonic dystrophy inherited?

The condition follows a 'dominant' inheritance pattern which means that on average, half of the children of a woman with myotonic dystrophy will be affected themselves. It affects both sexes, but the mother is usually the affected parent.



7 Is there a treatment or cure?

Not at present, however,

- **physiotherapy** and **occupational therapy** are very important ways of improving or maintaining a child's physical condition,
- **diagnosis** is increasingly accurate,
- **pre-natal testing** is available at an early stage of pregnancy,
- and **expert care** is on hand at Muscle Centres, funded by the Muscular Dystrophy Campaign, and may also be available at other hospitals inspecialist units.

8 Is there any research being conducted into the condition?

The good news is that research into this condition is very active and productive. The Muscular Dystrophy Campaign is funding research projects and scientists collaborate world-wide to share their knowledge and accelerate their understanding of the condition.

In 1992 Muscular Dystrophy Campaign then Group researchers pin-pointed the genetic defect which causes myotonic dystrophy. This was a crucial milestone because it allows researchers to study the gene and the protein(s) it codes for and it is only by understanding the way that the gene works that they can begin to think of ways of developing a treatment. We now know that the type of genetic defect responsible for myotonic dystrophy is virtually identical in all those so far tested which indicates that the condition may have sprung from a single original change in the gene that happened many thousands of years ago. The gene responsible for myotonic dystrophy seems to govern an important protein, which has an effect on many of the body's functions. This is why myotonic dystrophy has so many varied effects.

It has been shown that the larger the disruption of the gene, the more severe the symptoms are likely to be. Although there is some overlap, three main categories can usually be predicted from the genetic studies from very mild, barely noticeable symptoms to average and very severe symptoms. Researchers based in Cardiff and London have also discovered that in very rare cases (2-3%) the genetic mutation actually decreases through the generations.

The severity of myotonic dystrophy tends to increase through the generations (except in rare cases mentioned above). This means that a grandfather might only have cataracts but his grandchild could be very severely affected with the congenital form of myotonic dystrophy. It is very important therefore, to trace

relatives of someone affected by myotonic dystrophy in order to give them the option of genetic counselling.

9 Are anaesthetics a risk?

Operations and anaesthetics can be risky. It is very important that any surgeon and anaesthetist should know a child has congenital myotonic dystrophy before surgery is planned. Problems usually occur when doctors are unaware of the disorder; if care is taken, surgery is usually safe. Patients may wish to wear a bracelet or locket stating their condition. A specific warning card is available that can be carried in a purse or wallet. This can be obtained from the Myotonic Dystrophy Support Group (address below).

A factsheet on anaesthetics is available from the Muscular Dystrophy Campaign

10 Can a child with congenital myotonic dystrophy have the usual inoculations?

Yes

11 Will the condition improve?

It often improves during childhood but may deteriorate again later in life.

FINDING THE BEST HELP

Muscle Centres

A GP or consultant may be able to refer a child with congenital myotonic dystrophy to a Muscular Dystrophy Campaign Muscle Centre if there is one in the area. A Muscle Centre, always based in a hospital is staffed by specialists in neuromuscular conditions. They have the facilities to provide the most up-to-date diagnostic information and clinics are held there which the child may be able to attend. If there is no Muscle Centre nearby, there may nevertheless be a specialist in the area who will be able to provide the care the child needs.

Family Care Officers

The Muscular Dystrophy Campaign has a nation-wide team of Family Care Officers. They provide practical support and information. They also help families liaise between the various professionals e.g. physiotherapists, occupational therapists, social workers, teachers. The MDC's National Occupational Therapy Advisor may also be able to help.

For more details about the Muscular Dystrophy Campaign's support services, please write to or telephone our Information Officers (details on front page).

Support Group

Myotonic dystrophy and congenital myotonic dystrophy are fairly rare conditions and ones about which people often have not heard. Parents of a child with congenital myotonic dystrophy can feel rather isolated. However, they will find that there are many people in similar situations who have already dealt with problems they are encountering. A national support group is run by and for families affected by myotonic dystrophy.

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