



Myotonic Dystrophy
SUPPORT GROUP

Congenital Myotonic Dystrophy

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Myotonic Dystrophy

What is Myotonic Dystrophy?

Myotonic Dystrophy is an inherited neuromuscular condition caused by an abnormality in the DPKM gene, which is located on chromosome 19. The condition is caused by an expansion (enlargement) in the gene. The size of this gene expansion determines the severity of the condition. At the mildest end of the spectrum, people who have a small expansion might complain only of diabetes and/or cataracts. More severely affected with larger expansions in the gene, complain of muscle stiffness and weakness, palpitations or dizzy spells, swallowing difficulties leading to choking, cataracts and glucose intolerance.

What is Congenital Myotonic Dystrophy?

Congenital Myotonic Dystrophy is the most severe form of the condition. Giving birth to

an affected baby is often unexpected and a great shock for parents. The first few weeks of the baby's life can be a difficult and frightening time for all the family.

The baby is affected with myotonic dystrophy because the mother has the condition. Frequently, she is completely unaware that there is anything wrong with her and she may have little or no symptoms. Usually, a grandparent is also affected and will have the mildest form of the condition with cataracts and/or diabetes.

There are a number of other conditions which can be present in newborn babies in a similar way to myotonic dystrophy. Many investigations, including a muscle biopsy are often necessary. A blood test to look for the abnormality in the DPKM gene, will confirm the diagnosis of myotonic dystrophy, but it can take up to two weeks for a result to be available, which can be a difficult and

frustrating time for parents. An electrical test, called an EMG, can be performed in the mother and will give an immediate clue to the diagnosis. However, this test is not available in all hospitals.

What are the symptoms?

The degree to which babies are affected is quite variable, some babies can be very severely affected, others less so. The newborn baby may not move very much before birth and the fluid surrounding the baby is frequently increased, this is called polyhydramnios. The baby is often born a few weeks prematurely.

The baby's head may seem very floppy and there may be joint deformities. Some babies are born with a club foot.

During the neonatal period (the first four weeks of life) the baby will have weak muscles and frequently will have breathing and feeding difficulties. Many babies will need a ventilator

to assist their breathing, it is important that the baby is sedated for this, morphine is the most common sedative used. For the first few days the baby may be fed by an intravenous drip or a tube is passed into the stomach to feed the baby, this is called a nasogastric tube. It is possible to feed the baby with breast milk via this tube and drugs may be given to speed bowel transit to prevent or reduce gastric reflux (domperidone, metoclopramide or erythromycin). Laxatives are not given to newborn babies, but if the baby becomes very constipated, a slither of glycerine can be given as a suppository.

After about 28 days, some improvement occurs in most babies, but not all. Sadly, a small number of babies are so severely affected that the condition is fatal. Babies who need to be ventilated for more than 90 days are the most severely affected. After one year of age, almost all children will survive to adulthood.

During the early childhood years the condition improves and is quite stable. However, during adulthood, the condition may deteriorate with time.

How is the condition treated?

There is no cure for congenital myotonic dystrophy, but much can be done to help the child and improve the outcome. As already mentioned, during the newborn period, support with feeding and breathing are most important, but these problems will improve with time.

During infancy (the first year of life) the child may be prone to chest infections because of poor swallowing, co-ordination and gastro-oesophageal reflux. Thickening the feeds can improve these symptoms and antibiotics should be given for chest infections. Ensuring the baby has all of the routine vaccinations is very important and some additional vaccinations may be offered to



protect the child from bronchiolitis, pneumonia and influenza.

The baby's developmental milestones (sitting, crawling, walking and speech) are frequently delayed. Physiotherapy is an important aspect of care and should be available to the child from a very young age. Almost all children will learn to walk independently, but some may need splints or special boots to achieve this.

All children should be assessed in a child development centre from two years of age. Multi-disciplinary care is required with input from a Paediatrician, Orthopaedic surgeon, Physiotherapist, Occupational therapist and Speech and language therapist. Assessment of the child's developmental level at this age enables planning for the child's future educational needs. Many children have learning difficulties and 'require a statement of educational needs'. The statement of educational needs determines the need

for extra support at school and is best undertaken BEFORE the child begins nursery school.

Weakness of the facial muscles causes the mouth to droop open, this may give the wrong impression that the child is unresponsive, teachers should be made aware of this.

Facial weakness may also lead to speech and feeding difficulties. A speech and language therapist can give helpful advice. Weight gain and growth should be monitored throughout childhood. Many children find it difficult to gain weight and some will need food supplements.

Some children may develop a squint, when this occurs the child should have a vision test to make sure that spectacles are not required. Some children may have reduced hearing due to Glue Ear which can be treated with grommet insertion. Visual and hearing assessments in early childhood are important.

Club foot and joint deformities are managed



by orthopaedic surgeons in specialised clinics. Many children have a tendency to turn their feet inwards when they walk, however, this tends to improve with time.

Muscle stiffness (myotonia) is not present in new born babies or infants. It develops in childhood and is worse in cold weather. It predominantly effects the hands. There are some drug treatments which can help with the stiffness, but they tend to have unpleasant side effects and they are not recommended for children. Most people are able to manage their stiffness without using medication.

Many children with congenital myotonic dystrophy have bowel problems, either diarrhoea or constipation. Diarrhoea can be treated effectively with a drug which slows down the gut motility, called loperamide (imodium), constipation is more common and can be more problematic. Severe untreated constipation can lead to incontinence caused by overflow of liquid stools, when this happens

the child can be mistaken to have diarrhoea. The correct treatment, however, is a laxative. Paediatric Movicol is a good choice because it softens the motion and stimulates the gut to move. If the constipation is allowed to go untreated, eventually the child will become incontinent and very strong laxatives in high doses will be required to reverse the problem. Early diagnosis and treatment of constipation is advisable.

Behaviour in some children with myotonic dystrophy can be difficult. Young children can be prone to temper tantrums. Some children can have a stubborn personality and at the severe end of the spectrum, there may be features of Autism. Most Paediatric Units will have access to a special needs behaviour team, who can provide support and advice on how to manage difficult behaviour. Don't be afraid to ask for help with behaviour management.

Children with myotonic dystrophy can tire



much more easily than other children of the same age and they usually require longer periods of sleep and rest, this is a feature of the condition. For mothers of new born babies, tiredness can be a big problem and they should not be shy about asking for help. A stimulant drug called Modafanil can be used, it is not recommended for children, however it might be useful for mothers and some older teenagers who are troubled by sleepiness.

The heart is not usually affected in children with myotonic dystrophy.

Maternal Health

The mother of a child with congenital myotonic dystrophy can easily forget her own health. Discovering that she also has the condition soon after the birth of a child can be very shocking. Even though the mother may feel perfectly well, it is very important that she receives an annual check up. In particular she should have an ECG to check for cardiac rhythm abnormalities, which can be effectively

treated. A check for diabetes and cataracts should also be undertaken.

Anaesthetic Issues

It is very important to be aware that anaesthetics can be very risky in anyone who has myotonic dystrophy, whether they are mildly or severely affected. This is because some anaesthetics, especially muscle relaxants, can cause a severe reaction. It is very important to let any surgeon or anaesthetist know about the diagnosis of myotonic dystrophy. It is advisable to carry a myotonic dystrophy care card or wear a medic alert bracelet in case of any emergency.

How is the condition inherited

Myotonic dystrophy affects both men and women. It is inherited as a dominant gene which means that an affected adult (male and female) has a 50% chance of passing the condition on to each of their offspring. The condition usually tends to get worse with



each successive generation. This is called 'anticipation' and it occurs because the gene expansion grows with each generation. Very rarely, the opposite may happen and the gene expansion can decrease in the next generation leading to a milder form of the condition.

The severe congenital form only occurs when the mother is affected. This is because only the mother's eggs can carry gene expansions large enough to cause the congenital form. Some affected pregnancies can be so severe that they will end in miscarriage.

When a mother is known to have myotonic dystrophy, pre-natal genetic testing can be performed in the first 12 weeks of pregnancy. The procedure is called Chorionic villous sampling.

Myotonic Dystrophy support group

Myotonic Dystrophy is a relatively rare disorder, babies who have the severe congenital form are very rare. It is not

uncommon for parents to feel alone and isolated when a diagnosis of myotonic dystrophy is made. The National Myotonic Dystrophy Support Group can provide information and support. They can be contacted via the website or office.

Further information about Myotonic Dystrophy can be obtained from:

- **Myotonic Dystrophy Support Group**

35a Carlton Hill, Carlton,
Nottingham. NG4 1BG

Tel/Fax: 0115 987 5869

Email: contact@mdsguk.org

www.myotonicdystrophysupportgroup.org

**Other publications available from the
Myotonic Dystrophy Support Group:**

Anaesthesia and Sedation for patients with
Myotonic Dystrophy

Basic Information for Midwives

Bowel Problems in Congenital Myotonic
Dystrophy

Excessive Daytime Sleepiness and Myotonic
Dystrophy

Facts for patients, family members and
professionals

Myotonic Dystrophy Support Group

Relatives Information

The Heart and Myotonic Dystrophy

Why do we get new families with Myotonic
Dystrophy?



Myotonic Dystrophy
SUPPORT GROUP

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