Myotonic Dystrophy

What is Myotonic Dystrophy?

Myotonic dystrophy causes muscle weakness and wasting. Myotonic dystrophy is usually only slowly progressive. Myotonic dystrophy is an inherited genetic disorder. A genetic disorder is caused by one or more changed genes.

What are genes?

Our bodies are made up of millions of cells. Each cell contains a complete set of genes. We have thousands of genes. We each inherit two copies of most genes, one copy from our mother and one copy from our father. Genes act like a set of instructions, controlling our growth and how our bodies work. Any alteration in these instructions is called a mutation (or change). Mutations (or changes) can stop a gene from working properly. A mutation (change) in a gene can cause a genetic disorder. Genes are responsible for many of our characteristics, such as our eye colour, blood type or height.

What are the features of Myotonic Dystrophy?

The condition is very variable. These are the most common features.

- The muscles most frequently involved are in the face, jaw and neck. The legs and thighs are less commonly affected. Some people never have significant muscle disability.

- Muscle stiffness (or myotonia) is very characteristic, and especially affects the hands. People with myotonic dystrophy often have difficulty releasing a tight grip.

- Age of onset is variable and can be from birth to old age.

- Occasional associated problems may include tiredness, cataracts, constipation, frontal balding and infertility in males, disturbance of heart rhythm, diabetes, menstrual irregularities (irregular periods) in women and learning difficulties in children.

- Some people with myotonic dystrophy may experience problems following a general anaesthetic, so they are advised to carry a myotonic dystrophy alert card at all times.

Before an anaesthetic is given, an anaesthetist should be informed if a person has a family history of myotonic dystrophy.

What is wrong with the gene that causes Myotonic Dystrophy?

Genes contain information used by our bodies in the form of a code. The code is contained in the order of chemicals that make up the gene. In the normal myotonic dystrophy gene, a part of this code is repeated several times. In people with myotonic dystrophy, this repeated part of the gene is larger than normal (they have more repeats). This is called an expansion, because part of the gene has expanded, or grown bigger. If the size of this expansion is too great, it interferes with the normal genetic instructions and gives rise to myotonic dystrophy. People with myotonic dystrophy have a change (mutation) in a myotonic dystrophy gene.

This expansion (or repeated part) of the changed myotonic dystrophy gene appears to be unstable. It tends to expand when it is passed on to the next generation, particularly when it is passed on by the mother. This means that for many families, myotonic dystrophy becomes more severe in successive generations. Although myotonic dystrophy affects men and women equally, affected women are more likely to have a severely affected child. This severe form of myotonic dystrophy that affects children is called congenital Myotonic Dystrophy (this is not the same as congenital muscular dystrophy).

In some people, a diagnosis of myotonic dystrophy is not obvious, without a detailed medical assessment . It is therefore very important to offer counselling to all those members of a family who could carry the gene.

How is Congenital Myotonic Dystrophy different?

Congenital Myotonic Dystrophy is the early childhood form of myotonic dystrophy. Its features are present from birth (congenital means present from birth).

Congenital myotonic dystrophy is rare and is almost always inherited through a mother who has myotonic dystrophy (sometimes only very mildly) herself, rather than through an affected father.

Congenital myotonic dystrophy can vary in severity and is sometimes suspected before a baby's birth. In these cases, the condition is likely to be very severe and, sadly, some babies do not survive.

However, if medical staff are aware that an affected baby is expected, arrangements are made to give the best possible care in the early newborn period. A child who survives beyond his or her first year of life is likely to live to adulthood.

Features of Congenital Myotonic Dystrophy include:-

- Severe weakness, resulting in poor head control and, sometimes, a lack of facial expression.
- Breathing problems, which may require the baby to be on a ventilator at first.
- Difficulties with swallowing, and therefore feeding difficulties.
- Club foot or talipes may be present at birth.
- Developmental delay and learning difficulties.
- Problems with speech and pronunciation may arise, requiring the help of a speech therapist.
- Squint or impaired vision in some children.
- The development of bowel and bladder control may be delayed.

Like adults with myotonic dystrophy, babies and children with congenital myotonic dystrophy may have problems with general anaesthetics. It is advisable that they wear myotonic dystrophy alert bracelets or lockets.

How is Myotonic Dystrophy inherited?

Everybody has two copies of the myotonic dystrophy gene, but a person only has to have one changed copy of the gene to develop the disorder. The normal copy cannot compensate for the effects of the copy with the change (mutation). When people who have a myotonic dystrophy gene with a mutation (change) have children, they can pass on either their normal copy of the gene or the copy with the mutation (change). This means there is a 1 in 2, or 50% chance of their child inheriting the gene with the change (mutation). There is also a 1 in 2 or 50% chance of their child inheriting the normal copy of the gene.

Is a genetic test available for Myotonic Dystrophy?

There is a direct test to look at the length of the myotonic dystrophy gene of any person who may have a chance of developing myotonic dystrophy. It is important, however, that this test is offered only after careful medical assessment and genetic counselling, together with patient consent.

Prenatal diagnosis

Prenatal diagnosis involves testing a baby for a genetic disorder during the mother's pregnancy. Prenatal diagnosis is sometimes requested to check whether a baby has the myotonic dystrophy gene with the change (mutation), especially in women who have had babies with the severe form of the disorder.

Where can I get more information about Myotonic Dystrophy?

This is a brief guide to myotonic dystrophy and only the most common features of the disorder have been covered here. More information can be obtained from your local regional genetics centre or from these addresses:

The Myotonic Dystrophy Support Group
35A Carlton Hill
Carlton
Nottingham, NG4 1BG
Tel: 0115 987 0080 (Helpline)
Email: *mdsg@tesco.net*
Web: *www.mdsguk.org*

The Genetic Interest Group
Unit 4D,
Leroy House,
436 Essex Rd.,
London, N1 3QP
Telephone: 020 7704 3141
mail: *mail@gig.org.uk*
Web: *www.gig.org.uk*

Contact a Family
209-211 City Rd.,
London,
EC1V 1JN
Telephone: 020 7608 8700 FAX: 020 7608 8701
Helpline 0808 808 3555 or Textphone 0808 808 3556
(Freephone for parents and families, 10am-4pm, Mon-Fri)
e-mail: *info@cafamily.org.uk*
Web: *www.cafamily.org.uk*

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Glossary (difficult words and their meanings): Myotonic Dystrophy

This glossary is intended only to explain terms used in the information: Myotonic Dystrophy. Words shown in **bold** are defined elsewhere in the glossary.

cell. The human body is made up of millions of cells, which are like building blocks. There are many specialised types of cells. These include skin cells, brain cells, and blood cells. Cells in different parts of the body look different and do different things. Every cell (except for eggs in women and sperm in men) contains all the body's **genes.**

club foot. An abnormality in position and shape of the foot, which will need treatment to correct it.

congenital. Present at birth.

dystrophy. Developmental changes in the muscles leading to weakness and wasting.

expansion. Increased number of **repeats** in the myotonic dystrophy **gene.**

gene. Information needed for the body to work, stored in a chemical form. Changes or **mutations** in genes alter the information and this can change how the body works. Most genes are in pairs: one from the mother, one from the father. (As an analogy: a gene is like a story in a book, a change or **mutation** in a gene is like a missing or extra letter in a word in the story).

general anaesthetic. Treatment given to remove sensation while a procedure (such as an operation) takes place. The patient is not awake under general anaesthetic.

mutation. A change in a **gene**. Some mutations are not harmful. Sometimes when a **gene** is changed, its information is altered so it does not work properly. (As an analogy: a change or mutation in a **gene** is like a missing or extra letter in a word in a story). In myotonic dystrophy, the **mutation** (change) is an increase in the number of **repeats** of part of a **gene**. This increased number is called an **expansion**.

myotonia. Difficulty in relaxing muscles.

prenatal diagnosis. Test during a pregnancy for the presence or absence of a **genetic** disorder in the baby.

repeats. In myotonic dystrophy, the change (**mutation**) that causes the disease is in a part of the myotonic dystrophy **gene** that is repeated. A test can show the size of the repeated part of the myotonic dystrophy **gene**. People who have myotonic dystrophy have too many repeats. (As an analogy, repeats are like repeated letters or syllables in a word; if more repeated letters or syllables are added, the sense of the word may be lost).

speech therapist. Specialist who helps children with learning to talk, for children who start to talk late, or who make slow progress with speaking.

squint. A condition of the eyes. The eyes do not move together as expected, one eye turns in a different direction to the other.

ventilator. Equipment to help maintain breathing.

This glossary is intended only for use by patients and families, with the **genetic** information to which it refers.

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