Manchester University
NHS Foundation Trust

Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Myotonic Dystrophy

Myotonic dystrophy has a wide range of effects

This leaflet is designed to explain how myotonic dystrophy affects people who have it. It lists a large number of possible effects.

It is important to remember that it is extremely unlikely that any one person will have all of these problems.

Myotonic dystrophy may be very mild with virtually no symptoms. Some people, however, are quite severely affected.

What causes myotonic dystrophy?

Myotonic dystrophy is caused by a change in a gene. People who have this gene change (sometimes called a mutation or expansion) may develop a variety of symptoms. These symptoms can affect various parts of the body.

Not everyone with the gene change will develop all the symptoms and some people with the gene change may have no symptoms at all.

Muscle weakness

Muscle problems are often the first symptom of myotonic dystrophy. The muscles may feel weak, although this weakness is very variable and can range from mild to severe. It particularly involves the face and eyelids, jaw, neck, forearms and hands, lower legs and feet. If the muscles of the mouth and tongue are affected, it can make it difficult to speak clearly. Weakness in the muscles of the face may make someone's face appear 'droopy' or 'expressionless'.

Myotonia

Myotonia is a word that is used to describe a difficulty in relaxing a muscle after it has been contracted. People with myotonic dystrophy sometimes say that they find it difficult to release their grip on things such as shopping bags or steering wheels. This is a common symptom that usually does not cause much of a problem.

Heart problems

Myotonic dystrophy can affect the heart. Sometimes there are no symptoms at all or it may cause the heartbeat to be abnormally slow or irregular.

Regular ECG tests (heart tracings) are the best way to find out if the heart is affected. We recommend that everyone with myotonic dystrophy should have an ECG every year or so.

Chest and breathing problems

Chest infections may result from weakness of the breathing muscles, including the diaphragm, or from food entering the lungs as a result of choking. Inadequate breathing during the night can lead to disturbed sleep, snoring, difficulty waking, morning headaches and daytime sleepiness.

Digestive problems

These are common as the muscles of the digestive system may be affected. This may lead to:

- Swallowing problems (which can be a cause of food entering the lungs).
- Pains in the bowels with constipation and diarrhoea.
- Gallstones, which can cause painful spasms after eating fatty food, can be a problem in myotonic dystrophy.

Eye problems

Cataracts can cause blurring and dimming of vision. This may be the only problem that myotonic dystrophy causes.

Droopy eyelids can cause a problem with reading and watching television. You should have regular checks at the optician and see a medical eye specialist if there is any concern.

Other problems

- Male fertility problems.
- Diabetes (we recommend a urine test once a year).
- Pregnancy problems (the muscles in the womb can be involved, leading to an increased miscarriage risk or to a long labour.
- When myotonic dystrophy is present at birth, muscle involvement can be very severe. Babies may live only a short time.
- Speech, educational and behavioural problems can occur, especially when myotonic dystrophy begins in childhood.

Anaesthetics and surgery

People with myotonic dystrophy may have problems with surgery when certain anaesthetic drugs are used.

Make sure the surgeon and anaesthetist know about your myotonic dystrophy before an operation.

They may wish to contact a specialist centre for advice. Carry an Alert Card in your wallet or purse or wear a Medic Alert bracelet or necklace at all times in case of an accident or emergency. You can obtain a free card from the Myotonic Dystrophy Support Group (see contact details).

Inheritance

Myotonic dystrophy is caused by a genetic change that can run in families. Other family members are frequently affected.

It can affect and be passed on by both sexes, but women with the condition are more at risk of having a seriously affected child.

Accurate genetic tests are possible for healthy young adults who are at risk of developing myotonic dystrophy because they have an affected relative.

Tests are also available in pregnancy.

Before having a genetic test for myotonic dystrophy, it is important to see a genetic counsellor to find out all the facts.

Work, employment and mobility

If your ability to work or get around is affected by myotonic dystrophy, you may be entitled to special benefits. You can find out more about this from a local Citizen's Advice Bureau, a Family Care Officer of the Muscular Dystrophy Campaign or the Benefits Agency (see contact information).

For more information

If you need more advice about any aspect of Myotonic Dystrophy, you are welcome to contact:

Manchester Centre for Genomic Medicine
Sixth Floor
Saint Mary's Hospital
Oxford Road Manchester M13 9WL
Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

Department staffed Monday - Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

Muscular Dystrophy Campaign
61, Southwark Street, London SE1 0HL Telephone: 0207 803 4800
E-mail address: info@muscular-dystrophy.org
Website: www.muscular-dystrophy.org

Myotonic Dystrophy Support Group
35a Carlton Hill, Carlton, Nottingham NG4 1BG Office:
Telephone: 0115 987 5869
24 hour helpline: 0115 987 0080
E-mail address: contact@mdsguk.org
Website: www.myotonicdystrophysupportgroup.org

Benefits Agency Helpline: 0800 88 22 00

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

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Seen in clinic by (doctor): _____

And (Genetic Counsellor): _____

Telephone number: _____

Family reference number: _____