The Leeds Teaching Hospitals NHS Trust

Yorkshire Regional Genetics Service

Myotonic Dystrophy (DM1)

Information for patients

Leeds Clinical Genomics Service

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 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. Myotonic dystrophy can be abbreviated to DM1.

What causes DM1?

DM1 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 DM1. The muscles may feel weak or stiff, especially in cold weather. This 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 difficulty in relaxing a muscle after it has been contracted. People with DM1 sometimes find it difficult to release their grip on things such as shopping bags, steering wheels or cups.

Heart problems

DM1 can affect the heart. Sometimes there are no symptoms at all, or it may cause the heartbeat to be abnormally slow or irregular (this is called an arrythmia). Regular ECG tests (heart tracings) may be offered to find out if the heart is affected.

Breathing problems and sleepiness

People who are severely affected may get chest infections. Inadequate breathing during the night can lead to disturbed sleep, snoring, difficulty waking, and morning headaches. Daytime sleepiness is a common problem.

Digestive problems

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

- Swallowing problems
  (which can cause food to enter the lungs).
- Pains in the bowels with constipation or diarrhoea.
- Gallstones, which can cause painful spasms after eating fatty foods.

Eye problems

Cataracts can cause blurring and dimming of vision. In some people this is the only problem that myotonic dystrophy causes. Droopy eyelids can cause a problem with reading and watching television. You should have regular eye checks at the opticians and see a medical eye specialist if there is any concern. Cataracts can be effectively treated with surgery. Surgery is usually completed as a day-case under local anaesthetic.

Other problems

- Male fertility problems.
- Diabetes is more common in people who have DM1.
- Pregnancy problems (the muscles in the womb can be involved, leading to a long labour). It would be important for a pregnant woman's obstetric and midwifery team to be aware of her diagnosis of DM1.
- When DM1 is present at birth, muscle involvement can be very severe. Babies may live only a short time.
- Learning problems can occur, especially when DM1 begins in childhood.

Anaesthetics and surgery

People with DM1 may have problems with surgery when certain anaesthetic drugs are used.

Make sure the surgeon and anaesthetist know about your DM1 before an operation that involves a general anaesthetic.

Alert Cards

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 UK Support Group (see back page for contact details).

Inheritance

- DM1 is caused by a genetic expansion that can run in families.
- A person who carries a DM1 gene expansion has a 50% (1 in 2) chance of passing it on. This is called 'autosomal dominant inheritance'.
- 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 (called "congenital DM1").
- Broadly speaking, a larger DM1 gene expansion can be associated with more severe clinical features.
- The DM1 expansion can occasionally get bigger from one generation to the next.
- Accurate genetic tests are possible for healthy young adults who are at risk of developing DM1 because they have an affected relative. This is called 'predictive' testing.
- A healthy person who has genetic testing and is found to have a familial DM1 expansion can be offered clinical check ups such as heart, respiratory and diabetes checks.
- Tests are also available in pregnancy.
- Having the expansion may affect people very differently, even within the same family.
- Before having a genetic test for DM1, it is important 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.

Research and DM1 registry

Research into DM1 therapies is ongoing. You may be interested to know that a registry exists for patients with DM1. This was set up to help advance the research and development of treatment, therapies and care for people affected with DM1. You can self-register for this by following the link on the back of this leaflet.

For more information:

If you need more information please contact your local Genetics Department. If you live in the Yorkshire region please contact:

Leeds Clinical Genomics Service
www.leedsth.nhs.uk/services/pathology/clinical-genetics-2/

Muscular Dystrophy UK
www.musculardystrophyuk.org/conditions/a-z/myotonic-dystrophy-type-1-dm1/

Myotonic Dystrophy Support Group
www.myotonicdystrophysupportgroup.org
National Office: 0115 987 0080
Helpline: 0808 169 1960
Email: contact@mdsguk.org

The UK Myotonic Dystrophy Patient Registry
www.dm-registry.org.uk
Email queries:
myotonicdystrophyregistry@newcastle.ac.uk

Leeds Clinical
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What did you think of your care?

Visit: www.leedsth.nhs.uk/patients/support/feedback-complaints/friends-family-test/

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