

# MYOTONIC DYSTROPHY

## INFORMATION FOR FAMILY MEMBERS

You have been given this information because you or a relative of yours has been diagnosed as having myotonic dystrophy type 1. This is an inherited condition. If you have myotonic dystrophy, you may have relatives who would like to find out whether or not they have inherited the alteration in the gene that causes it.

If you have an affected relative who is a parent, brother/sister, or a child of yours, then there is a 50% (1 in 2) chance that you might have inherited the altered gene.

Myotonic dystrophy is a very variable condition. Some people who inherit the gene have no symptoms – so why worry about it? The reason is that sometimes there can be serious consequences, even for people with no obvious symptoms, and many of those consequences can be avoided given adequate knowledge.

### THE MAIN PROBLEMS WITH MYOTONIC DYSTROPHY

- Muscle stiffness (especially the fingers/hands)
- Weak muscles used in movement (especially the fingers/hands)
- Involvement of muscles not under our control, including those of the gastrointestinal tract causing irritable bowel type symptoms and the muscles involved in breathing which can affect the quality and amount of sleep
- Heart rhythm problems (not angina or heart attack)
- Breathing problems (especially after anaesthesia)
- Excessive daytime sleepiness
- Problems with certain anaesthetics, sedation and use of strong painkillers following surgery

- Cataracts at a younger age than the general population
- Premature male pattern balding
- Reduced fertility
- Having a child who is much more severely affected and associated learning disability

Three issues of particular importance to those who unknowingly carry the abnormal gene that causes the condition:

### 1. Problems before, during and after an operation

People with myotonic dystrophy have an increased risk of problems with certain **anaesthetics**. These can be prevented if the diagnosis is known and taken into account when an anaesthetic is planned. Strong painkillers or **opiates** are often used during or after an operation and can affect the breathing muscles which may be affected in myotonic dystrophy. Smaller operations often involve **sedation** rather than a general anaesthetic. Closer monitoring of the heart and breathing muscles is required.

### 2. Heart problems

The electrical system of the heart, which is responsible for controlling the heartbeat, even when there are no other symptoms. The heart rate may run too slow, fast or irregularly causing dizzy spells or blackouts. However, problems with the heart rhythm can be there even without these symptoms and may need to be treated to stop it getting worse.

### 3. Affected children

The condition tends to get more severe in successive generations. So, a person with few or no symptoms can have a child who is more severely affected. This is particularly true for women. Even women who are not aware of any problems themselves can have a child who can be severely affected at birth (a condition called **congenital myotonic dystrophy**). These children are often born prematurely and critically unwell. These babies may not survive. If they do they may have substantial mobility problems and will have a degree of learning

disability. If people know that they have this risk and are concerned, then there are tests that can be offered before and during pregnancy to see if a baby would be affected or not. Ideally, people at risk or known to have myotonic dystrophy should seek advice early when planning to have children in order to get the most up to date advice.

If you decide that you would like further information about testing for myotonic dystrophy for yourself or your relatives, you could do one of the following:

- If you live in Scotland, you can contact your local genetics department for local referral processes (see the table at the end of this leaflet)
- Alternatively, you could take this leaflet to your GP and ask to be referred to your local regional genetics centre. Though the genetic alteration can be picked up by a simple blood test it is important to be seen in the genetic clinic by someone who knows about the condition.

#### MORE INFORMATION ABOUT MYOTONIC DYSTROPHY

##### **Myotonic Dystrophy Support Group**

19-21 Main Road

Gedling

Nottingham NG4 3HQ

Helpline: 0115 987 0080

National Office: 0115 987 5869

Email address: [contact@mdsguk.org](mailto:contact@mdsguk.org)

Website: [www.myotonicdystrophysupportgroup.org](http://www.myotonicdystrophysupportgroup.org)

*Other information leaflets are also available from the myotonic dystrophy support group and also the Scottish Muscle Network ([www.smn.scot.nhs.uk](http://www.smn.scot.nhs.uk))*

##### **Muscular Dystrophy UK**

61A Great Suffolk Street

London SE1 0BU

Helpline: 0800 652 6352 (Mon to Fri, 8.30am-6.00pm)

Website: [www.musculardystrophyuk.org](http://www.musculardystrophyuk.org)

Email address: [info@musculardystrophyuk.org](mailto:info@musculardystrophyuk.org)

### Your local genetics service:

South East of Scotland Clinical Genetic Service:	MMC, Western General Hospital Crewe Road South, Edinburgh EH4 2XU Telephone: 0131 537 1116
North of Scotland Genetics Service:	Department of Clinical Genetics Ashgrove House, Foresterhill Aberdeen AB25 2AZ Telephone: 01224 552120
East of Scotland Genetics Service:	Human Genetics Unit Level 6, Ninewells Hospital Dundee DD1 9SY Telephone: 01382 632035
West of Scotland Genetics Service:	Level 2A Laboratory Medicine The Queen Elizabeth University Hospital 1345 Govan Road, Glasgow G51 4TF Telephone: 0141 354 9201