

Myotonic dystrophy in the family

This leaflet gives information on the possible problems for a patient with a diagnosis of myotonic dystrophy.

Myotonic dystrophy

You have been given this leaflet because you, or a relative of yours, has been diagnosed as having myotonic dystrophy. This is an inherited (passed on from a parent) condition. If you have myotonic dystrophy, you may have relatives who would like to find out whether or not they have inherited the abnormal 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 one in two (50%) chance that you have the abnormal gene.

Myotonic dystrophy varies enormously in severity. Some people who have the gene have lots of problems, and some have no symptoms.

Should I still worry about it?

The reason is that sometimes there can be serious consequences even for people with no obvious symptoms, and that many of those consequences can be avoided given adequate knowledge.

The main problems with myotonic dystrophy

- Weak muscles (especially the fingers and/or hands)
- Muscle stiffness (especially the fingers and/or hands)
- Cataracts
- Heart rhythm problems (not angina or heart attack)
- Breathing problems (especially after anaesthesia)
- Excessive daytime sleepiness
- Premature balding (mainly in men)
- Reduced fertility
- Irritable bowel syndrome

Any of these can be the first sign of the condition. Three issues are very important to anyone who is unknowingly carrying the abnormal gene that causes the condition. They are listed here.

Anaesthetic problems

People with myotonic dystrophy have an increased risk of problems with anaesthetics. These can be prevented if the anaesthetist knows about the diagnosis and can allow for it when an anaesthetic is planned.

Heart problems

The electrical system of the heart, which is responsible for controlling the speed of the heartbeat, can be affected even when there are no other symptoms.

For some people this can cause dizzy spells and blackouts. Even if there are no symptoms, a problem with the heart's rhythm can still be there, and may need to be treated to stop it getting worse.

Affected children

This is perhaps the most important potential problem.

The condition tends to be more severe in the next generation. A person with few or no symptoms can have a child who is more severely affected.

This is particularly true when this condition is inherited from the mother. Even women who are not aware of any problems themselves can have a child who can be severely affected at birth (congenital myotonic dystrophy). These children might not survive, or might have major physical and/or educational difficulties later in childhood.

If people know that they carry this gene and are concerned, then there are tests that can be offered to see if a baby will be affected or not in a pregnancy. It is important that when you find out that you're pregnant, you contact your local genetics centre to inform them, or ask your GP or midwife for an urgent referral to your local genetics centre to have a discussion about the risks and options.

What should you do next?

If you decide that you would like further information about testing for myotonic dystrophy for you or your relatives, you could ask your GP to refer you to your local genetics department.

Useful sources of information

Myotonic Dystrophy Support Group

t: 0800 652 6352

w: www.muscular-dystrophy.org

Muscular Dystrophy Campaign

Helpline: 0115 987 5869

w: www.mdsguk.org

Contact us

If you live in the South East Thames Region:
Clinical Genetics Department, 7th Floor,
Borough Wing, Guy's Hospital, Great Maze
Pond, London, SE1 9RT

t: 020 7188 1364, **f:** 020 7188 1369

w:

www.guysandstthomas.nhs.uk/service/managednetworks/genetics

Pharmacy Medicines Helpline

If you have any questions or concerns about your medicines, please speak to the staff caring for you or call our helpline.

t: 020 7188 8748, Monday to Friday,
9am-5pm

Your comments and concerns

For advice, support or to raise a concern, contact our Patient Advice and Liaison Service (PALS). To make a complaint, contact the complaints department.

t: 020 7188 8801 (PALS)

e: pals@gstt.nhs.uk

t: 020 7188 3514 (complaints)

e: complaints2@gstt.nhs.uk

Language and accessible support services

If you need an interpreter or information about your care in a different language or format, please get in touch.

t: 020 7188 8815

e: languagesupport@gstt.nhs.uk

NHS 111

Offers medical help and advice from fully trained advisers supported by experienced nurses and paramedics. Available over the phone 24 hours a day. **t:** 111

NHS website

Online information and guidance on all aspects of health and healthcare, to help you take control of your health and wellbeing.

w: www.nhs.uk

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A list of sources is available on request