

Regional Contact



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MYOTONIC DYSTROPHY

FACT SHEET FOR PATIENTS, FAMILY MEMBERS AND PROFESSIONALS

Myotonic dystrophy is the commonest muscular dystrophy of adult life and also the most variable. Some patients have disabling neuromuscular symptoms, especially weakness and to a lesser extent stiffness (myotonia); others have minimal muscle symptoms. Onset varies from birth to old age.

Important features involving other body systems may be more troublesome than muscle symptoms (see overleaf) and can seriously affect health. Specialists in these other systems are often unfamiliar with myotonic dystrophy and may not diagnose it.

Not all patients need to be followed by a specialist clinic, but all should have a thorough initial assessment, should be given full information about the condition and its consequences, including genetic aspects, and should have a clear plan for when medical help is needed. Specialist advice should always be sought in relation to surgery and anaesthesia, pregnancy, or serious illness. A wide range of disability aids exists. It is important that these are chosen with expert assistance.

At present, there is no cure for myotonic dystrophy, but this may change as a result of rapid progress in our understanding of how the disorder is caused – meanwhile all patients can help themselves by knowing about their condition, recognising and avoiding hazards, and by informing doctors and other professionals. We hope that this fact sheet will help in this process.

Myotonic Dystrophy - Important Clinical Aspects

Muscle weakness

- May range from mild to severe
- Particularly involves face, jaw, neck, distal limb muscles
- Can affect speech and give lack of facial expression

Heart

- May be involved in adults, even without symptoms
- Rhythm disturbance
- Regular ECG advised for early detection of conduction defect

Chest Problems

- Chest infections may result from diaphragm weakness or from food entering the lungs
- Impaired ventilation, especially at night, can occur

Anaesthetics and surgery

- Essential for anaesthetist and surgeon to be aware of the disorder and its risks
- Only undertake surgery in centre with full facilities
- High risk of post-operative respiratory problems - avoid depressant drugs

Gastro-intestinal problems

- Widespread and common - involvement of bowel smooth muscle
- Swallowing problems a common cause of food entering the lungs
- Large bowel pain, constipation, sometimes dilation of the bowel

Other problems

- Cataract - may be the only feature
- Hormonal problems - male hypogonadism, infertility, insulin resistance
- Excessive sleepiness

Affected children

- Muscle involvement can be severe, especially when onset from birth
- Important educational and behavioural aspects

Inheritance

- Can affect and be passed on by both sexes
- Other family members frequently affected
- Accurate genetics tests possible, including in early pregnancy
- Specialist genetic counselling advised if genetic testing is being considered

Support and further information

- Available from Myotonic Dystrophy Support Group (UK) (www.comcom.org/mdsg) and from Muscular Dystrophy Campaign (www.muscular-dystrophy.org)