- The onset of symptoms in DM2 is later, typically around the fourth decade
- The muscle weakness is proximal (hips and shoulders) rather than distal (hands and ankles) and rarely involves the face and throat muscles
- Pain is much more prominent and often a presenting feature – some patients have initially been thought to have "fibromyalgia"
- Pain is usually a more disabling problem than weakness
- Brain involvement (e.g. learning difficulties) is very uncommon
- Anticipation (where the disease obviously gets more severe in the next generation) is much less striking
- Myotonia is much less evident
- Cataracts are less common
- Diabetes is very infrequent
- Life expectancy is normal

Information supplied by Dr. David Hilton-Jones MD FRCP FRCPE. Department of Neurology, John Radcliffe Hospital, Oxford - First Edition April 2010.

Further information about Myotonic Dystrophy can be obtained from:

Myotonic Dystrophy Support Group
19/21 Main Road, Gedling, Nottingham.
NG4 3HQ

Tel/Fax: 0115 987 5869
Email: contact@mdsguk.org
www.myotonic**dystrophy**support**group**.org

Other publications available from the Myotonic Dystrophy Support Group:

- Anaesthesia and Sedation for patients with Myotonic Dystrophy
- Basic Information for Midwives
- Bowel Problems in Myotonic Dystrophy
- Congenital Myotonic Dystrophy
- Excessive Daytime Sleepiness and Myotonic Dystrophy
- Myotonic Dystrophy and the Eye
- Myotonic Dystrophy Support Group
- Relatives Information
- The Heart and Myotonic Dystrophy
- Why do we get new families with Myotonic Dystrophy?

Myotonic Dystrophy
SUPPORT GROUP

National Co-ordinator
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Open Tues/Wed/Thurs 9am to 1pm

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Myotonic Dystrophy
SUPPORT GROUP

Facts

for patients, family members and professionals.

Myotonic Dystrophy

Myotonic Dystrophy is the most common and most variable muscular dystrophy of adult life. 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 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 (1) have a thorough initial assessment, (2) be given full information about the condition and its consequences, including genetic aspects, and (3) have a clear plan for when medical help is needed.

Specialist advice should always be sought in relation to surgery and anaesthesia, sedation, 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 progress is being made in our understanding of how the disorder is caused. All patients can help themselves by knowing about their condition, recognising and avoiding hazards, and by informing doctors and other professionals.

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.
- Swallowing

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.
- Flu and pneumonia jab advised.

Anaesthetics and Surgery

- Caution with Anaesthetics and Sedation.
- Essential for anaesthetist and surgeon to be aware of the disorder and its risks.
- **Sedation** - It is very important that an anaesthetist is available for procedures.
- 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.
- Soiling due to weak anal sphincter.

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 and autistic tendencies.

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.

Information supplied by Sir Prof. Peter Harper CBE FRCP DM Professor & Consultant in Medical Genetics, Cardiff (Retired) - Second Edition August 2009.

MYOTONIC DYSTROPHY TYPE 2 (DM2)

Sometimes also called proximal myotonic myopathy (PROMM) this condition shows similarities to, but also important differences from, classical myotonic dystrophy (which is now referred to as type 1, or DM1). It involves a different gene to DM1, but the underlying, and very complicated, molecular disturbance is similar.

DM1 and DM2 never occur in the same family. DM2 seems to be more common in some countries (e.g. Germany) than others, and so far seems to be rare in the UK. The major differences include:

- The severe congenital form is not seen in DM2