Myotonic Dystrophy
SUPPORT GROUP

Basic Information for Midwives

by
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National Coordinator

Myotonic Dystrophy

I hope that this brief guide will be a positive step in providing help for mothers with myotonic dystrophy, and lead to a better understanding of how the condition can affect them and their baby.

Mrs M.A. Bowler. SRN SCM
National Co-ordinator

Myotonic Dystrophy
Written for Midwives

Women with myotonic dystrophy who are contemplating pregnancy should be given all the usual midwifery advice, but should also be advised to see a genetic counsellor.

Pregnant women who have myotonic dystrophy should be aware of the problems that may arise:-

1st Trimester

- Re-occuring miscarriages
- Extra tiredness due to myotonic dystrophy (more than the average person)
- The result of genetic testing from chorionic villus testing
- All other difficulties as with a non-affected mother must be noted i.e. Ectopic pregnancy

2nd Trimester

- Reduced fetal movements if baby is affected
- Extreme tiredness
- Results of genetic testing from amniocentesis

- Cardio-vascular problems
- Placenta previa may occur

3rd Trimester

- Hydramnios or Polyhydramnios; due to inadequate fetal swallowing
- Large for dates
- Reduced fetal movements
- Premature labour

Labour

- Premature labour
- Fetal distress
- Long and difficult labour due to abnormality of uterine muscular activity
- Out come is often a LSCS
- PPH

There should be an assessment of maternal respiration, before any type of anaesthesia or sedation. Epidural may be the most appropriate form of anaesthesia. This must be discussed with the anaesthetist **as anaesthetics and sedation are serious matters to be**

considered. NB Increased sensitivity to drugs.

People with myotonic dystrophy may find it helpful to carry an ALERT which notes:-

- Caution with anaesthetics and sedation
- May show extreme tiredness
- May have weakness and stiffness
- May have indistinct speech
- May have heart rhythm defects

Care in SCBU

There is very little recorded about the care of the babies with Congenital Myotonic Dystrophy. The baby has a 50% chance of having myotonic dystrophy in the severe form (CMD). The baby will be 'poorly' and the seriousness will need to be explained to the parents. Medical help will be needed to assess the fluid intake of the baby, as the baby may not be able to suck or tolerate milk.

Place of Delivery

Hospital under Consultant care, in a unit with neo natal intensive care.

Most babies born with CMD are from mothers who are undiagnosed. Following the delivery the baby may be diagnosed as having CMD, whilst still in the labour suite.

The mother should be seen by the Neurologist and an appropriate follow-up appointment made with the neurologist and/or geneticist, but she should have a detailed explanation of matters before discharge from hospital.

The diagnosis leaves the family in bewilderment.

General Information

The high spontaneous fetal loss must be borne in mind when early pregnancy and pre-implantation diagnosis are being considered for women with myotonic dystrophy.

Delivery of the Baby

The baby, if affected with Congenital Myotonic Dystrophy (CMD) will be a 'floppy baby'. There will be respiratory muscle involvement and pulmonary immaturity.

Bilateral talipes could be present in a baby that has CMD. The tent shaped mouth, and scoliosis may be apparent. The SCBU will monitor baby for several days, weeks or months. This depends on the severity of the condition (the genetic repeats).

As mother plans for the arrival of the new baby there is excitement from the extended family. When parents are told that the baby has a neuromuscular condition, and also that the gene has come from the mother (almost always in CMD) and grandparent, the family are distraught. Grandparents and mother feel guilty for passing on the condition.

Just a word about grandparents. They do seem to be left out of being given information at the time of diagnosis. The Myotonic Dystrophy Support Group receive a number of distressing phone calls from grandparents who feel very guilty for having passed on the faulty gene.

Extended family members are then advised to

seek genetic counselling. Often relatives do not want to know, but in later years find that they have the condition in their family.

Text Book

Myotonic Dystrophy (Third Edition)
by Prof. Peter S. Harper in the series
Major Problems in Neurology.
Published by W. B. Saunders.

This book will inform you in great detail of the progression of myotonic dystrophy.

Neonatal Deaths

by Dr David Hilton-Jones.

Sadly, some severley affected infants die at birth or within a few days. We are largely ignorant of the specific problems that lead to such early deaths although we know that the brain does not form normally. Our knowledge of the normal process of brain maturation has increased enormously in the last few years, but there have been no post mortem studies of infants dying from myotonic dystrophy in the newborn period in recent years. Seeking consent for post mortem study at what is clearly a very emotional time is difficult for doctors, midwives and family members. However, many mothers are aware of the need for further research and may be keen to see some good come out of such a tragic event. The possibility of post mortem examination should be discussed and The Myotonic Dystrophy Support Group and it's medical advisors would be pleased to help in any further discussions.

Dr David Hilton-Jones
Consultant Neurologist,
John Radcliffe Hospital, Oxford.

The following is a Facts leaflet which is available to all family members on request.

Myotonic Dystrophy is the most common 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 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, 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 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 leaflet 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. Flu and pneumonia jab advised.

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.

Information supplied by Prof. Peter Harper CBE FRCP DM Professor & Consultant in Medical Genetics, Cardiff (Retired)

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.myotonicdystrophysupportgroup.org

Other publications available from the Myotonic Dystrophy Support Group:

- Anaesthesia and Sedation for patients with Myotonic Dystrophy
- Bowel Problems in Myotonic Dystrophy
- Congenital Myotonic Dystrophy
- Excessive Daytime Sleepiness and Myotonic Dystrophy
- Facts for patients, family members and professionals
- 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?

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