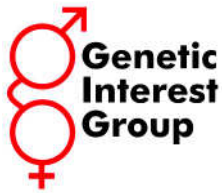


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**Genetic Interest Group (GIG)**

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**436 Essex Road, London, N1 3QP**

**Tel: 0207 7043141**

**[www.gig.org.uk](http://www.gig.org.uk)**

Registered Charity No 1114195

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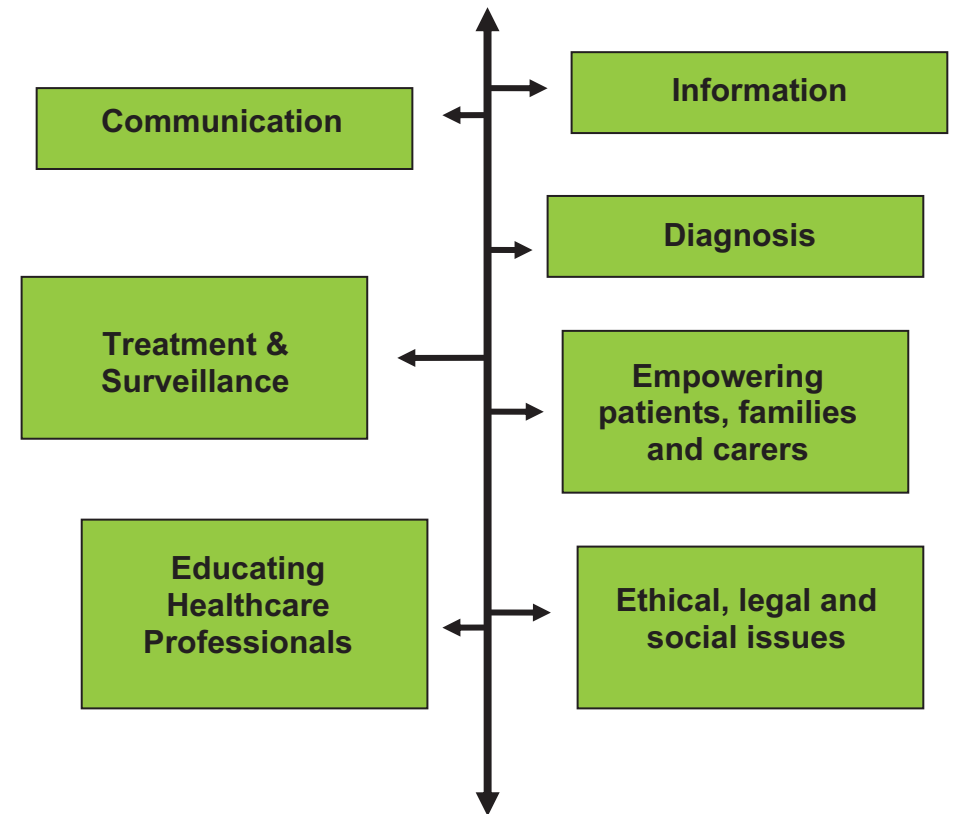
This leaflet has been prepared in good faith to provide patients with a guide to current services and information. Neither GIG nor MDSG can be held responsible for the accuracy of the information it contains. Links to other organisations are included for information purposes only and are not recommendations from GIG or MDSG.

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# Myotonic Dystrophy *Family Route Map*

This Family Route Map is a guide to current services and information for patients, relatives and professionals.



Charity Number 1073211

## INTRODUCTION

This leaflet is designed to provide signposts to sources of current information and appropriate services for patients, their families and carers, together with healthcare professionals. The issues and concerns raised during a series of patient focus groups in 2006 organised by the Genetic Interest Group (GIG) identified seven themes (see front cover) which have been used as the basis for developing this resource with the help of patients, families and carers, and clinical staff with experience and expertise of this condition.

## Myotonic Dystrophy

Myotonic Dystrophy is the most common and variable form of muscular dystrophy. It can affect either sex and can start at any age and both the range and severity of the symptoms vary widely. It is a neuromuscular genetic disease that affects around 1 in 8000 adults.

*Myotonia* is characteristic stiffness of the muscles that makes it hard to relax a muscle after it has contracted. For example it is difficult to let go after gripping an object. *Dystrophy* is weakening of the muscles.

The Myotonic Dystrophy Support Group (MDSG), a registered charity that aims to offer the hand of friendship and support to those affected by Myotonic Dystrophy, has medical professionals who are advisors to the group in the fields of; Genetics, Neurology, Anaesthetics and Research.

## MDSG contact details:

Mrs M A Bowler SRN, SCM — National Co-ordinator  
Myotonic Dystrophy Support Group  
35a Carlton Hill, Carlton, Nottingham, NG4 1BG  
Helpline Tel: 0115 987 0080 Office Tel: 0115 987 5869

Email: [mdsg@tesco.net](mailto:mdsg@tesco.net) Website: [www.mdsguk.org](http://www.mdsguk.org)

### TIP

A Medical Alert card is available from MDSG to carry on your person. This enables emergency medical staff to quickly read an important summary of information about your medical condition.

### TIP

Drivers have a duty to inform DVLA and car insurance companies about their medical diagnosis or risk non-payment in the event of a claim.

## Links

### Other useful organisations:

British Heart Foundation [www.bhf.org.uk](http://www.bhf.org.uk) or phone Heart Information Line (charged at local call rate) 0845 0708070 for a booklet about 'Pacemakers'

British Society for Human Genetics (BSHG): [www.bshg.org.uk](http://www.bshg.org.uk)

Carers UK [www.carersuk.org](http://www.carersuk.org) Tel: 0808 808 7777

Citizens Advice Bureau (CAB) can help with advice on benefits for individuals and carers. Your local branch is listed in your telephone directory. Their website address is [www.citizensadvice.org.uk](http://www.citizensadvice.org.uk)

Contact-A-Family: [www.cafamily.org.uk](http://www.cafamily.org.uk) Tel: 020 7608 8700

Department of Health have produced a set of 'Questions to ask' to take with you to your appointment with a specialist. Available in several different languages; use the Search option on their website. [www.dh.gov.uk](http://www.dh.gov.uk)

Directgov [www.direct.gov.uk](http://www.direct.gov.uk) provide a website with help on benefits and entitlements

Disability Benefits Office Tel: 08457 123456

Disablement Income Group Scotland [www.digscotland.org.uk](http://www.digscotland.org.uk)  
Tel: 0131 555 2811

GIG (Genetic Interest Group) have a range of patient leaflets on their website [www.gig.org.uk/eurogentest\\_patientleaflets.htm](http://www.gig.org.uk/eurogentest_patientleaflets.htm) Tel: 020 7704 3141

Muscular Dystrophy Campaign [www.muscular-dystrophy.org](http://www.muscular-dystrophy.org) Tel: Freephone 0800 625 6352 for information about The Joseph Patrick Trust that provides grants to individuals who need adaptations and support.

The Family Fund [www.familyfund.org.uk](http://www.familyfund.org.uk) Tel: 0845 1304542

The UKGTN (UK Genetic Testing Network) has produced a patient leaflet [www.ukgt.nhs.uk/gtn/UKGTN-information/Patient-leaflet.html](http://www.ukgt.nhs.uk/gtn/UKGTN-information/Patient-leaflet.html)

Wellchild [www.wellchild.org.uk](http://www.wellchild.org.uk) Tel: 0808 801 0330

## OTHER INFORMATION

### Education

Children with myotonic dystrophy may have learning problems at a time when there are no muscle complaints. Again, be sure that myotonic dystrophy is borne in mind if this disorder is in the family. 'Educational Guidelines' for children with muscular dystrophies are available from the website of The Muscular Dystrophy Campaign

[www.muscular-dystrophy.org](http://www.muscular-dystrophy.org)

### Parents and Carers

Parents and carers have needs too and sometimes it may be difficult to identify yourself as a carer. Your health and wellbeing are important, especially if you are a carer to more than one family member. The Myotonic Dystrophy Support Group have a leaflet available on 'Information for Relatives'.

### Psychological help

Psychological counselling is not commonly offered on diagnosis or even later on. If you feel that you need extra help in coming to terms with your condition or any other aspect of your life, don't be afraid to ask for this via your GP.

### Insurance

It is common to have difficulties finding adequate and affordable insurance policies once diagnosed with a genetic condition. Companies may offer savings plans as an alternative to Life Cover. It is important to shop around. The following companies offer travel insurance for people with pre-existing conditions. There may be others available and this list should *not* be taken as a recommendation:

- The insurance group Banner - see [www.bannergroup.com](http://www.bannergroup.com) have a simple policy that anyone declared fit for travel by Interhealth, see [www.interhealth.org.uk](http://www.interhealth.org.uk) is covered as normal by them. They don't have extra rates for particular conditions.
- Free Spirit (AXA Insurance UK plc) [www.free-spirit.com](http://www.free-spirit.com) for people with medical conditions and/or disabilities  
Tel: 0845 2305000, email: [sales@pjhayman.com](mailto:sales@pjhayman.com)
- AllClearPlus (Groupama Insurance Co Ltd) [www.allcleartravel.co.uk](http://www.allcleartravel.co.uk) provides a special policy.  
Tel: 08712 088500, email: [info@bishopscurtas.co.uk](mailto:info@bishopscurtas.co.uk)
- Freedom Insurance [www.freedominsure.co.uk](http://www.freedominsure.co.uk) offer travel insurance. Tel: 01223 454290

## INFORMATION

Often this is a condition that affects many family members to different degrees and regular reviews and assessments of your condition by your consultant are very important. Helpful resources and information for your different life-stages and decision-making are available from MDSG. A range of leaflets, including facts about the condition and information for relatives can be ordered using a form on the MDSG website or by phoning the Helpline or their office number. A DVD of the Myotonic Dystrophy Annual Conference covers the main events of the programme and includes talks by medical professionals.

For an explanation of and information about the genetics of Myotonic Dystrophy please see the MDSG leaflet :

***Why do we get new families with Myotonic Dystrophy?***

Written by Prof. Darren G Monckton PhD

### Genetic Testing:

This condition follows an autosomal dominant inheritance pattern, which means that each pregnancy has a 50% chance of the child having the myotonic dystrophy gene if either of the parents has myotonic dystrophy. The Genetic Interest Group (GIG) website has a leaflet 'Dominant Inheritance' and a glossary of genetic terms plus other information on genetics. Please see the website [www.gig.org.uk/eurogentest\\_patientleaflets.htm](http://www.gig.org.uk/eurogentest_patientleaflets.htm) or phone their office for a copy Tel: 020 7704 3141

Genetic testing and counselling is available at 23 regional NHS genetic centres throughout the UK; please see the website of [www.bshg.org.uk](http://www.bshg.org.uk) for a list. In their section 'For Patients' there is an explanation of what Genetic Counselling is and is not. A referral to a genetic centre is usually made through your GP or specialist. If you already have a family member who has been seen in the Genetics Unit it may be possible to contact the Unit directly as a self-referral. All patients should be seen and counselled by the clinical genetics service as early on as possible. Tests in pregnancy can be undertaken in addition to the testing of individuals, however, there are a number of issues surrounding genetic testing particularly in relation to children. Results of genetic tests should be received face-to-face wherever possible.

## DIAGNOSIS, TREATMENT AND SURVEILLANCE

Although no 'cure' for myotonic dystrophy exists at present, there is a lot that can be done to help those affected. If troublesome, muscle stiffness due to myotonia can be helped with certain drugs.

Some patients can develop heart problems, which are commonly treatable but can be serious if ignored. A regular electrocardiogram (ECG) is strongly recommended.

Some people who have myotonic dystrophy may have more trouble with other body systems than they do with their muscles. A symptom that appears quite unrelated may be connected.

Excessive daytime sleepiness, swallowing difficulties and a range of bowel symptoms are examples. It is important that people with myotonic dystrophy should make sure that whoever treats them is aware that they have the condition and knows the wide range of associated problems.

Contact the **Myotonic Dystrophy Support Group** (MDSG) for further information on centres of expertise or clinicians, ideally Neuromuscular Consultants, who have experience of diagnosing and managing the condition:

Helpline Tel: 0115 987 0080 Office Tel: 0115 987 5869

Email: [mdsg@tesco.net](mailto:mdsg@tesco.net) Website: [www.mdsguk.org](http://www.mdsguk.org)

### TIP

Genetic Counsellors (part of the NHS Clinical Genetics Services) can help with concerns around the impact on you and your family of being diagnosed with a genetic condition and may be able to give you information about where to go for support.

In the UK, patients have the right to ask for a second opinion via their first specialist or through their GP. They are not, however, entitled to receive that second opinion if the clinicians do not think it necessary. A patient may appeal against any decision via their Primary Care Trust (PCT). For more information on the Referral Process please see: [www.gig.org.uk/docs/referrals.pdf](http://www.gig.org.uk/docs/referrals.pdf)

## EDUCATING HEALTH PROFESSIONALS

**Operations and anaesthetics** can be risky, even for mildly affected people. It is most important that any surgeon or anaesthetist should know a person has myotonic dystrophy before surgery is planned. Anaesthetic Guidelines are available from [www.gla.ac.uk/muscle/dmanaesthesia.htm](http://www.gla.ac.uk/muscle/dmanaesthesia.htm) and the leaflet 'Anaesthesia and Myotonic Dystrophy' is available from MDSG.

Affected women need careful management if undertaking a pregnancy. Not only is there a risk of a baby being severely affected, but problems in pregnancy and delivery may affect the mother. MDSG provides a leaflet 'Information for Midwives'.

The Scottish Muscle Network [www.gla.ac.uk/muscle/dm.htm](http://www.gla.ac.uk/muscle/dm.htm) have developed a Care Card offering advice for those living with Myotonic Dystrophy, their carers and healthcare professionals. MDSG have also produced a leaflet 'Facts for patients, family members and professionals'.

Prof. Peter S. Harper, University Research Professor in Human Genetics at Cardiff University, has published three books; Myotonic Dystrophy (3rd Ed.)-Major Problems in Neurology. Myotonic Dystrophy-The Facts. Oxford University Press (OUP). The above text book is suitable for lay people. Myotonic Dystrophy-Present management, future therapy. OUP

### Other clinical information can be found at:

- TREAT NMD Project is a European initiative [www.treat-nmd.eu](http://www.treat-nmd.eu)
- National Organization for Rare Diseases [www.rarediseases.org](http://www.rarediseases.org)
- Orphanet (European database) [www.orpha.net](http://www.orpha.net) Free-access website providing information on rare diseases
- 107th ENMC International Workshop: the management of cardiac involvement in muscular dystrophy and myotonic dystrophy. 7th-9th June 2002, Naarden, the Netherlands. K. Bushby, F. Muntoni, J.P. Bourke. *Neuromuscular Disorders* 13 (2003) 166-172

### TIP

Say how you feel: if you don't want your picture taken; or if you don't want medical students participating in your appointments; or clinical staff discussing your child in front of them, do say so.