

Scottish Muscle Network

Scottish guideline for the management of Myotonic Dystrophy in adults

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Scottish guideline for the management of myotonic dystrophy in adults

Cardiac

- Patients, including asymptomatic carriers, should receive an annual automated ECG from diagnosis.
- Echocardiogram should also be performed at diagnosis. If normal, this should be repeated 3 to 5 yearly.
- If any baseline cardiac investigations are abnormal, the patient should be referred to a cardiologist. Current and previous ECGs should be provided with the referral.
- Patients with palpitations, presyncope or syncope, even if their ECG is normal, should be referred to a cardiologist. Patients/carers should also be educated that such symptoms require prompt attention.
- A low threshold should be kept to undertake extended ECG monitoring. It is anticipated that all patient under cardiology care will receive 24-hour Holter monitoring at regular intervals.
- For patients who meet criteria for referral to a cardiologist, cardiac care should be delivered by a consultant cardiologist with an expressed interest in myotonic dystrophy, who is therefore aware of disease-specific indications for device therapy (Appendix 1).
- Medications, including anti-myotonia agents and stimulants, may be associated with increased risk of cardiac complications. If there is doubt, prescribing of new drugs should be discussed with patient's cardiologist

Respiratory and Sleep

- Respiratory complications are the leading cause of death in patients with myotonic dystrophy and so spirometry and respiratory muscle assessment is recommended at baseline and at intervals.
- At annual review, patients should be asked if they have the following symptoms, which may suggest respiratory muscle weakness and/or sleep-disordered breathing: worsening dyspnoea, difficulty lying flat, not feeling refreshed in the morning, excessive daytime sleepiness, fatigue or morning headaches. Presence of any symptom should prompt Respiratory referral.
- The Epworth Sleepiness Scale is recommended as a screen for excessive sleepiness, though local practice may vary regarding use of other scales e.g. STOP-Bang or Respcheck.
- Referral to Respiratory should allow for assessment using spirometry, plethysmography, maximal inspiratory and expiratory pressure, sniff nasal/inspiratory force, peak expiratory cough flow, baseline capillary gases and nocturnal cardiorespiratory polygraphy (PGR). This may direct

intervention e.g. continuous positive airway pressure (CPAP) therapy or non-invasive ventilation.

- Recurrent chest infections may be due to ineffective cough. At each visit, patients should be asked: “Have you had respiratory infections requiring consecutive antibiotics in past year?” “Do you cough when you swallow food, more than once a week?” If yes to either, consider referral to Speech & Language Therapy (SLT).
- Seasonal influenza vaccine is advised annually and pneumococcal vaccine should be administered at least once in the patient’s lifetime. COVID-19 immunisation should also be offered in line with national guidance.
- Respiratory evaluation is recommended prior to any surgical procedure requiring anaesthetic or sedation.

Neuromuscular

- At each visit, patients should be examined and their muscle weakness classified using the Muscular Impairment Rating Scale (MIRS).
- SLT referral should be considered if dysarthria is impacting communication and social interaction. Some patients may benefit from a lightwriter.
- At each visit, patients should be asked, “Have you fallen since your last clinic visit?” If yes, consider referral to physiotherapy.
- Individuals with myotonic dystrophy should be encouraged to exercise regularly. Note the Scottish Muscle Network website (<https://www.smn.scot.nhs.uk/professionals/physiotherapy/>) contains information about appropriate stretches, exercise, and falls management including demonstration videos. Consider referral to physiotherapy for patients who wish further advice regarding appropriate exercise.
- Identify patients with foot drop who may benefit from orthotics assessment, for example by provision of ankle foot orthoses.
- Upper limb muscle weakness may impair daily function and compromise independence. Physical limitations may necessitate changes to the home environment for some patients. Consider referral to occupational therapy (OT) accordingly.
- Physical activity and social participation may be limited by central symptoms, such as fatigue or apathy. These symptoms can benefit from strategies such as cognitive behavioural therapy (via Clinical Psychology service) and/or graded exercise (via Physiotherapy). Discuss the possibility of referral if appropriate. NB fatigue symptoms may also be indicative of sleep-disordered breathing: see Respiratory section.
- Ask specifically about mood disturbance or cognitive changes. Consider referral to neuropsychology and/or psychiatry where relevant symptoms are disclosed.

MRI brain scan may be considered if there is a marked change in symptoms.

- Mexiletine may be considered to treat and manage myotonia. Mexiletine is a class IB anti-arrhythmic medication and there are no studies on long-term therapy. It is recommended that mexiletine should only be prescribed by a Neurologist and after cardiology review. An ECG is required prior to initiation and also after initiation and during dose incrementation. The cardiologist looking after the patient should be alerted to the fact that Mexiletine is being prescribed. Mexiletine should be taken with food to avoid the significant dyspepsia that can develop as a side effect.
- All individuals who drive are advised to inform the DVLA about their diagnosis of myotonic dystrophy.

Ocular System

- All patients should attend an optician every year. Patients should be advised to seek early advice from optometry if they experience any visual problems before their annual review.
- Patients with ptosis interfering with vision should be referred to an ophthalmologist for discussion of surgery.

Endocrine System

- All patients should have HbA1c measured annually. Abnormal results should be followed up by GP according to local protocols.
- TFTs should also be measured at baseline and annually. More frequent testing is suggested if thyroid dysfunction is suspected. Consider ultrasound screening of the thyroid gland if abnormal on clinical examination.
- Bone profile should also be measured at baseline and annually. If calcium is elevated, check PTH. If low, consider vitamin D deficiency.
- Patients should be made aware of Scottish Government guidance of Vitamin D supplementation. Some may meet criteria for year-round supplementation.
- Enquire about bone health and correlate with level of mobility. Ask about fractures in the last 12 months and document where the fracture was. Consider a DEXA scan where there is a history of fracture, or MIRS scale is 4 or 5. Consider local protocols when referring patients for DEXA scans. If the DEXA scan is abnormal and/or if patient's mobility status is poor, refer to the relevant Mineral Metabolism or Rheumatology specialist for guidance on preventative/proactive therapy. NB Patients may be more susceptible to side-effects of bisphosphonates (dysphagia).

Gastrointestinal System

- At each visit, ask:
 - Do you choke on or cough after swallowing food or drink more than once per week? If yes, consider SLT referral.
 - Do you soil underwear more than once a month? With this or new bowel symptoms consider referral to a Gastroenterologist.
 - Do you have a yearly check-up appointment with your dentist? If not, encourage them to attend.
- Also enquire about dyspepsia after meals, abdominal pain and bloating, constipation and diarrhoea. Persisting, troublesome symptoms or weight loss may justify Gastroenterology referral.
- Liver function tests (LFTs) should be measured annually. If any abnormalities are detected, follow local protocols for further investigation.
- Record BMI yearly. Evidence suggests approximately one third of all patients with DM1 are overweight (BMI > 25 kg/m²), and 20% are obese (BMI > 30 kg/m²).
- If age 50 or over, confirm the patient is participating in the general population bowel screening programme.

Genetics

- At each visit, review full pedigree to identify new at-risk or uncounselled individuals.
- Individuals who have not received formal genetic counselling should be referred to their local Genetics Centre. Reproductive implications should be discussed and re-referral made to Genetics if patients are considering a pregnancy or if testing of at-risk relatives is requested.
- Unusual clinical presentations, atypical results from diagnostic genetic testing, or a negative genetic test in an individual felt to have clinical signs of myotonic dystrophy should be discussed with a clinical geneticist, to consider the possibility of variant repeat sequences.

Fertility and reproductive health

- For individuals of reproductive age, at every visit if appropriate:
 - Offer pre-conception counselling
 - Reaffirm potential issues around subfertility
 - Ask about recurrent pregnancy loss or difficulties conceiving
 - Confirm awareness of availability of PGD and testing in pregnancy

- Confirm the importance of making contact as soon as pregnancy confirmed
- Confirm awareness of risks associated with pregnancy for both mother and foetus
- Men may benefit from treatment for erectile dysfunction. Be mindful of possible adverse cardiovascular effects associated with such treatments.
- Women may require referral to gynaecology for menstrual irregularity problems and management of subfertility.

Pregnancy

- All patients should be offered pre-conception genetic counselling with information about antenatal testing and PGD.
- Obstetric and Genetic teams should be made aware of the patient as early in the pregnancy as possible.
- Repeat ECG and echocardiogram in the first trimester, and refer to cardiology if there is any abnormality. Plan to repeat these prior to delivery, in the third trimester. Ensure the Obstetric team responsible for the patient is also aware of this guidance.
- Keep a low threshold to request or update baseline respiratory assessments in pregnancy.
- Pregnant patients should be managed in a specialist centre with a fetal medicine unit. Care should include assessments of maternal respiratory and cardiovascular function and anaesthetic risk, and allow for delivery with available maternal and neonatal Intensive Care facilities.

Anaesthesia / Care card

- Patients should be advised to carry an alert card or jewellery.
- Ask the patient to produce their alert card at each clinic visit.
- Patients should be advised of general anaesthetic and sedation risks (see Anaesthetic Guideline: www.smn.scot.nhs.uk)
- Alerts should also be added to the patient's electronic record according to local protocols (e.g. KIS, ACP)

Guideline Development

These guidelines were developed in 2009 by a working group of the Scottish Muscle Network Managed Clinical Network, when the Scottish Intercollegiate Guidelines Network grading system was used. The guidelines were updated by the Scottish Muscle Network Myotonic Dystrophy Subgroup in January 2014, January 2017 and in May to December 2021.

The subgroup are most grateful for advice from colleagues in the preparation of this guidance, including Drs Jan Kerr, Mark Wright and Peter Cackett, (Consultant Ophthalmologists, Edinburgh), Dr Alison Kelly (Consultant Biochemist, Glasgow) AND Dr Caroline Coats (Consultant Cardiologist, Glasgow).

Specialist Clinics

Specialist management clinics are located throughout Scotland. Details of your nearest clinic are available from:

- West of Scotland Regional Genetics Service, Laboratory Medicine building, Queen Elizabeth University Hospital, 1345 Govan Road, **Glasgow**, G51 4TF
0141 354 9200
- South East Scotland Regional Genetics Service, Department of Clinical Genetics, Western General Hospital, Crewe Rd, **Edinburgh**, EH4 2XU
0131 537 1116
- Department of Clinical Genetics, Ninewells Hospital, **Dundee**, DD1 9SY
01382 632035
- Department of Medical Genetics, Ashgrove House, Aberdeen Royal Infirmary, Foresterhill, **Aberdeen**, AB25 2ZN
01224 552120

Other contacts

- **Myotonic Dystrophy Support Group**,
19-21 Main Road, Gedling, Nottingham NG4 3HQ
0115 987 0080
www.myotonicdystrophysupportgroup.org
- **Muscular Dystrophy UK**,
61A Great Suffolk Street, London, SE1 0BU
020 7803 4800
www.musculardystrophyuk.org

Appendix 1

Cardiologists with an interest in DM1

- **Ayrshire-** Dr Victor Chong, Crosshouse Hospital, Kilmarnock
- **Borders-** Dr Paul Neary, Borders General Hospital
- **Dumfries & Galloway-**
- **Fife-** Dr Mark Francis, Victoria Hospital, Kirkcaldy
- **Forth Valley-** Dr Catherine Labinjoh, Forth Valley Royal Hospital, Larbert
- **Grampian-** Dr Paul Broadhurst, Aberdeen Royal Infirmary
- **Greater Glasgow & Clyde**
 - Dr Caroline Coats, Queen Elizabeth University Hospital, Glasgow
 - Dr David Murdoch, Queen Elizabeth University Hospital, Glasgow
- **Highland-** Dr Stephen Cross, Raigmore Hospital, Inverness
- **Lanarkshire-**
 - Dr Andrew Docherty, Wishaw General Hospital
 - Dr Brian O'Rourke Hairmyres Hospital, East Kilbride
 - Dr Graeme Tait, Wishaw General Hospital
- **Lothian-**
 - East** - Dr Martin Denvir, Royal Infirmary of Edinburgh
 - West** - Dr Alan Japp, Royal Infirmary of Edinburgh / St. John's Hospital, Livingston
- **Tayside-** Dr Anna Maria Choy, Ninewells Hospital, Dundee

Appendix 2

Respiratory/Ventilation Physicians with an interest in DM1

- **Ayrshire** - Dr David Sword, University Hospital Ayr
- **Borders** - Dr Roberto Rabinovich and Dr Melanie Cross – Royal Infirmary of Edinburgh
- **Dumfries & Galloway** - Dr Stuart Little, Dumfries and Galloway Royal Infirmary
- **Fife** - Dr Roberto Rabinovich and Dr Melanie Cross - Royal Infirmary of Edinburgh
- **Grampian** - Dr Patrick Fitch, Aberdeen Royal Infirmary
- **Greater Glasgow & Clyde** -
Dr Scott Davidson, Queen Elizabeth University Hospital, Glasgow
Dr Eric Livingstone, Glasgow Royal Infirmary
Dr Caroline O’Dowd, New Victoria Hospital, Glasgow
Dr Chris Carlin, Gartnavel Hospital, Glasgow
Dr Douglas Grieve, Royal Alexandra Hospital, Paisley
- **Highland** - Dr Lorna Murray, Raigmore Hospital, Inverness
- **Lanarkshire** - Dr Ken Dagg, Wishaw General Hospital
- **Lothian** - Dr Roberto Rabinovich and Dr Melanie Cross - Royal Infirmary of Edinburgh
- **Tayside** - Dr Morven Wilkie, Ninewells Hospital, Dundee

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NOTE

This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined on the basis of all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient’s case notes at the time the relevant decision is taken.