Scottish guideline for the management of Myotonic Dystrophy type 2 (DM2) in adults

NOTE
This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined based on 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.

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Cardiac

- Patients, including asymptomatic carriers, should receive an annual automated ECG from diagnosis.
- An 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, pre-syncope or syncope, even if their ECG is normal, should be referred to a cardiologist. Patients and 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 patients under cardiology care will receive 24-hour Holter monitoring at regular intervals.
- For patients who meet the criteria for referral to a cardiologist, cardiac care should be delivered by a consultant cardiologist with an expressed interest in myotonic dystrophy.
- Medications, including anti-myotonia agents and stimulants, may be associated with an increased risk of cardiac complications. Prescribing of new drugs should be discussed with the patient's cardiologist.

Respiratory and Sleep

- Respiratory complications are much less common than in Myotonic Dystrophy type 1 but 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. The presence of any symptom should prompt a Respiratory referral.
- A range of sleep disturbances including daytime sleepiness, insomnia, REM behaviour disorders, and restless leg syndrome have been observed in case reports and case series of individuals with DM2. Daytime sleepiness can be associated with restless leg syndrome.
- The Epworth Sleepiness Scale is recommended as a screen for excessive sleepiness, though local practice may vary regarding the use of other scales including STOP-Bang or Respicheck.
- 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 the 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 before any surgical procedure requiring anaesthetic or sedation although adverse reactions to anaesthesia are much less frequent than in Myotonic Dystrophy type 1 (DM1).

Additional features

- Hearing impairment. Cochlear sensorineural hearing impairment is reported in different series as affecting 20 - 60% of individuals with DM2, suggesting an early presbycusis.

- Cancer risk. Retrospective studies have shown that individuals with DM2 appear to be at a higher risk of developing cancer. A cross-sectional analysis of a large DM study showed that tumour risk is higher in DM1 than in DM2. Cancer in individuals with DM2 may involve the colon, brain, thyroid, pancreas, ovary, prostate, and endometrium. Adherence to routine population screening is strongly advised.

- Autoimmune disorders have been reported in 21% of cases in one series (only 31 cases) - a feature distinguishing DM2 from DM1 although both disorders are associated with hypogammaglobulinaemia. No particular autoimmune disorder was more prevalent.

Neuromuscular

- Distal weakness is less frequent than in patients with DM1. Proximal upper and lower limb muscle weakness is commoner in DM2 and 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.

- Muscle pain which does not appear to be related to myotonia may be a prominent feature and difficult to treat. Axial weakness may contribute to low back pain which is reported to affect about 50% of patients in one series.

- Individuals with DM2 should be encouraged to exercise regularly. Note the Scottish Muscle Network website. www.nn.nhs.scot/smn/podcast-and-videos 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. The QR code below will take you to the Scottish Muscle Network:

- Physical activity and social participation may be limited by central symptoms, such as fatigue or apathy. These symptoms are usually much less severe than in DM1 but may benefit from strategies such as cognitive behavioural therapy (via Clinical Psychology service) and/or graded exercise (via Physiotherapy). Fatigue symptoms may also be indicative of sleep-disordered breathing: see Respiratory section.

- 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 a cardiology review. An ECG is required before initiation and also after initiation and during dose incrimination. 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 they must inform the DVLA about their diagnosis of myotonic dystrophy.

Ocular system

- All patients should see 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

- Non-insulin-dependent diabetes is more common than in Myotonic Dystrophy type 1 and all patients should have HbA1c measured annually. Abnormal results should be followed up by their 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

- Hypogonadism in males has been reported and all affected men should be advised of this and referred if symptomatic.

- Patients should be made aware of the Scottish Government's guidance of Vitamin D supplementation. Some may meet the criteria for year-round supplementation.

- Enquire about bone health and correlate with the 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 a MIRS scale is 4 or 5. Consider local protocols when referring patients for DEXA scans. If the DEXA scan is abnormal and/or if the 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

- Dysphagia and constipation may occur and biliary tract disease may be more common than in the general population, if these symptoms develop consultation with Gastroenterology teams is advised
- 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/m^2), and 20% are obese (BMI > 30 kg/m^2)
- if age 50 or over, confirm the patient is participating in the general population bowel screening programme

Genetics

- A severe congenital form of DM2 has not been reported to date. However, a paediatric onset with muscle pain or arthrogryposis-like features has been recently reported so it is important patients and families are counselled at each visit by an individual with expertise in unstable trinucleotide repeat disorders.
- 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.

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 the availability of PGD and testing in pregnancy
    - confirm the importance of making contact as soon as pregnancy is confirmed
- 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 before 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. Care should include assessments of maternal respiratory and cardiovascular function and anaesthetic risk and allow for delivery with available maternal 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 2025 by a working group of the Scottish Muscle Network Managed Clinical Network when the Scottish Intercollegiate Guidelines Network grading system was used.

Additional information is available at

Schoser B. Myotonic Dystrophy Type 2. 2006 Sep 21 [Updated 2020 Mar 19]. In: Adam MP, Feldman J, Mirzaa GM, et al., editors. GeneReviews. Seattle (WA): University of Washington, Seattle; 1993-2024.

JAMA. 2024 Apr 9;331(14):1227-1228.

Specialist clinics

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

South East of Scotland Clinical Genetic Service Covering Lothian, Borders, and Fife (except the East Neuk)	MMC, Western General Hospital, Crewe Road South, Edinburgh EH4 2XlJ Telephone: 0131 537 1116 https://services.nhslothian.scot/geneticservice/clinical-genetic-service/
North of Scotland Genetics Service Covering NHS Grampian, NHS Highland, NHS Orkney, NHS Shetland and the northern half of the Western Isles	Ashgrove House, Foresterhill, Aberdeen, AB25 2AZ Telephone: 01224 552120 https://www.nhsgrampian.org/service-hub/north-of-scotland-medical-genetics/
East of Scotland Genetics Service Covering Tayside and the East Neuk of Fife	Level 6, Ninewells Hospital, Dundee, DDI 9SY Telephone: 01382 632035 https://www.nhstayside.scot.nhs.uk/OurServicesA-Z/Genetics/index.htm
West of Scotland Genetics Service Covering Greater Glasgow and Clyde, Forth Valley, Ayrshire and Arran, Lanarkshire, Dumfries and Galloway and the southern half of the Western Isles	Level 2A Laboratory Medicine, Queen Elizabeth University Hospital, 1345 Govan Road, Glasgow G51 4TF Telephone: 0141 354 9201

	https://www.nhsggc.scot/staff-recruitment/staff-resources/laboratory-medicine/clinical-genetics/

Additional sources of support and information:

Scottish Muscle Network	Email: nss.smn@nhs.scot www.nn.nhs.scot/smn/
Myotonic Dystrophy Support Group (19-21 Main Road, Gedling, NG4 3HQ)	Helpline: 08081691960 Email: contact@mdsguk.org www.myotonicdystrophysupportgroup.org/
Muscular Dystrophy UK (32 Ufford Street, London, SE1 8QD)	Helpline: 0800 652 6352 Email: info@musculardystrophyuk.org www.musculardystrophyuk.org/

Appendix 1

Cardiologists with an interest in Myotonic Dystrophy

- Ayrshire
  - Dr Joanne Simpson, University Hospital Crosshouse
- Borders
  - Dr Paul Neary, Borders General Hospital
- Dumfries & Galloway
  - Dr Jenna McMinn, Dumfries & Galloway Royal Infirmary, Dumfries
- 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 Will Jenkins, 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