Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 1

Quick Reference Version

The studies & rigorous evidence needed to drive the creation of an evidence-based guideline for the clinical care of myotonic dystrophy type 1 (DM1) patients have not yet been executed for all affected body systems & manifestations. In order to improve & standardize care for this disorder now, more than 60 leading myotonic dystrophy (DM) clinicians in western Europe, the United Kingdom, Canada & the United States have created the *Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 1.*

Summary recommendations from the Consensus-based Care Recommendations are below. The full compendium of recommendations by body system & their disease manifestations is available here **http://www.myotonic.org/clinical-resources**.

LIFE THREATENING SYMPTOMS – CLINICAL CARE RECOMMENDATIONS

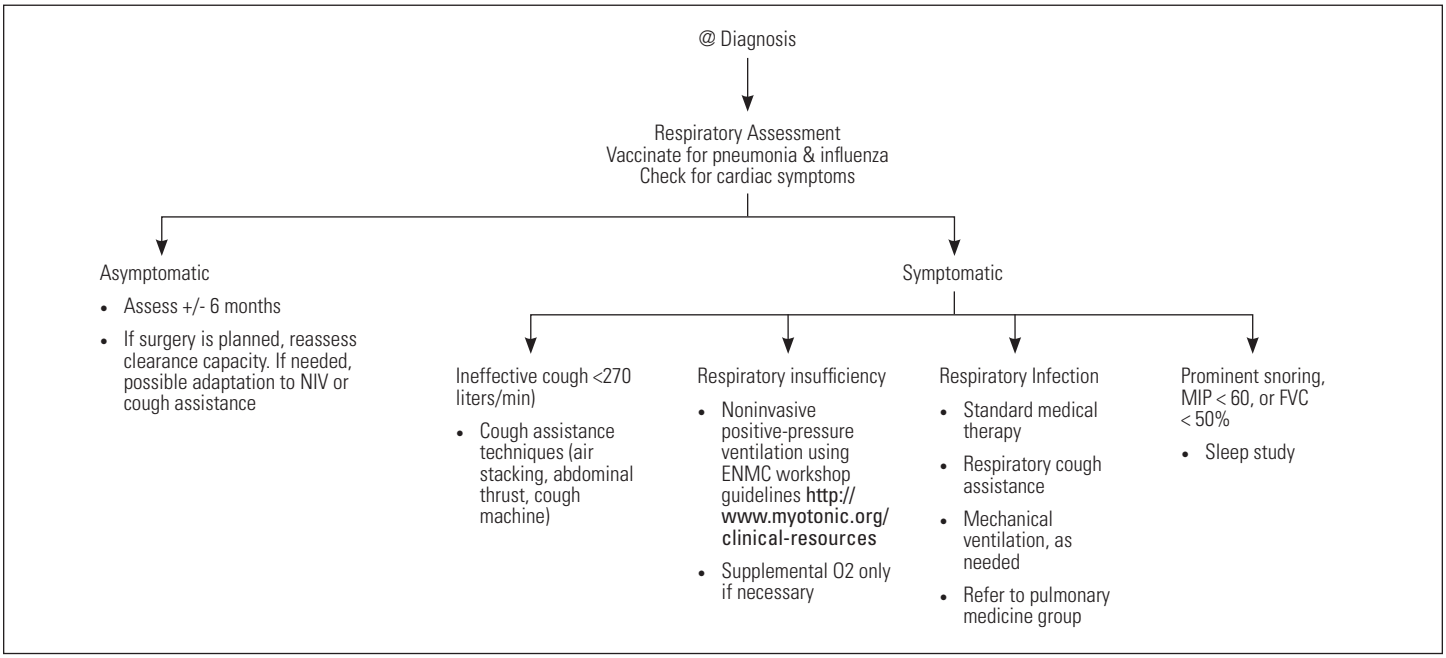
Surgery, anesthesia & pain

- See Myotonic Dystrophy Foundation's *Practical Suggestions for the Anesthetic Management of a Myotonic Dystrophy Patient* for anesthesia risks & recommendations before any surgeries or procedures requiring anesthesia **http://www.myotonic.org/clinical-resources**
- DM1 patients are far more likely to have adverse reactions to medications used for anesthesia & analgesia; interactions of the cardiac, respiratory, muscle & central nervous systems in each DM1 patient can lead to a variety of untoward responses before, during & after surgery
- Serious adverse events can occur throughout the course of DM1 & have been reported in patients whose DM1 symptoms were mild
- Behavioral & cognitive abnormalities need careful assessment & management preoperatively (if time permits & if it is possible) since these manifestations along with hypersomnia & preoperative sleep deprivation can complicate the patient's immediate postoperative care & long term recovery
- Most serious complications occur in the post-anesthesia period
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Respiratory symptoms

- Pulmonary complications are the leading cause of death in DM1 patients. Clinicians must monitor issues such as recurrent pneumonia at baseline & serially with pulmonary function tests, at least forced vital capacity (FVC)
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Fig. 1 Respiratory Care Recommendations Flowchart



Cardiovascular symptoms

- Cardiac complications are the second leading cause of death in DM1
- The most common cardiac issues are arrhythmias (sinus bradycardia, heart block, atrial fibrillation & flutter, & ventricular tachycardia)
- Palpitations, chest pain, dyspnea, orthopnea, lightheadedness, & syncope warrant cardiac investigation
- Significant cardiac involvement that subsequently leads to adverse cardiac events is often asymptomatic
- Impulse or conduction abnormalities on a standard 12-lead ECG including sinus rate < 50 BPM, PR interval > 200 ms, QRS duration > 100 ms, left anterior or posterior fascicular block, abnormal Q-waves, atrial tachycardia, fibrillation, or flutter, & ventricular arrhythmias are indicative of cardiac involvement
- Refer patients with cardiac symptoms, abnormal annual or biennial ECG indicative of cardiac involvement, & patients over the age of 40 years without previous cardiac evaluation to a center experienced in DM1 care
- Cardiology referral for all DM1 patients is reasonable if part of a multidisciplinary program or if the practitioners providing primary care are uncomfortable assessing cardiac history, exam, or ECG
- See full recommendations at **http://www.myotonic.org/clinical-resources**

SEVERE SYMPTOMS & CONDITIONS – CLINICAL CARE RECOMMENDATIONS

Skeletal muscle weakness & rehabilitation

- Evaluate annually for:
  - Swallowing & speech difficulties
  - Mobility, balance & falls
  - Activities of daily life – including self-care
  - Activities in home, school, work & community
- Refer to specialists, including PTs, OTs, speech pathologists, dietitians, social workers & others
- Encourage moderate intensity (aerobic & resistance training) exercise
- See *Role of Physical Therapy in the Assessment of Individuals with Myotonic Dystrophy* at www.myotonic.org.
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Skeletal muscle myotonia

- Myotonia can cause muscle stiffness, prolonged hand grip, speech & swallowing difficulties
- Mexiletine may be considered for myotonia treatment. Mexiletine is contraindicated for DM1 patients with cardiac involvement. See full recommendations regarding mexiletine at **http://www.myotonic.org/clinical-resources** for more information on cardiac implications.

Pregnancy & obstetric management

- Women with DM1:
  - Have increased risk of miscarriage, pre-term delivery, & respiratory insufficiency during pregnancy (especially in the 3rd trimester) & failed labor during delivery; extreme care should be taken with analgesics & sedating anesthetic drugs (see MDF Anesthesia Guidelines here **http://www.myotonic.org/clinical-resources)**
  - Should consult with a high-risk OBGYN prior to delivery & obtain ongoing antenatal care
  - Fatigue much more quickly during labor & are at risk of postpartum hemorrhage, particularly after prolonged first or second stage or if there has been polyhydramnios
  - Should be induced only at direction of obstetrician & after all necessary consultants assisting with the delivery are notified
- Sexually active patients with DM1:
  - Should be referred to genetic counseling & family planning services if of child-bearing age
  - Should receive parental counseling for prenatal genetic diagnosis or discussion of preimplantation genetic diagnosis
- Include pediatric or neonatal specialist at delivery; intensive neonatal care is recommended for neonates that may have DM1; anticipate need for feeding tube & ventilator support
  - Access to a pediatric or neonatal specialist is recommended even if fetus is known to be unaffected
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Excessive daytime sleepiness symptoms

- Assess for excessive daytime sleepiness (EDS) with Epworth Sleepiness Scale or similar standardized questionnaire instrument; prescribe sleep study if sleep disturbance is suspected
- Monitor periodic limb movements (muscle activity during sleep), as well as EEG, respiratory measures during sleep study to assess possible obstructive sleep apnea & central nervous system mediated sleep apnea
- Refer to pulmonologist &/or sleep specialist if EDS scores are positive on scales
- Question patients re: alcohol or caffeine consumption, medications & sleep habits for contribution to EDS
- Evaluate impact of possible respiratory muscle weakness (forced vital capacity value sitting & supine) on presence of EDS
- If nocturnal or daytime hypoventilation is suspected, consider non-invasive positive pressure ventilation, & refer to pulmonologist with experience in neuromuscular diseases re: possible need for NIV launching
- Consider modafinil for treatment if coexisting central nervous system alteration is suspected as the cause for EDS
- Consider cognitive behavioral therapy (CBT) or behavioral therapy for apathy; psychostimulant treatment can be considered if apathy is associated with an impairing level of fatigue or EDS
- See full recommendations at **http://www.myotonic.org/clinical-resources**

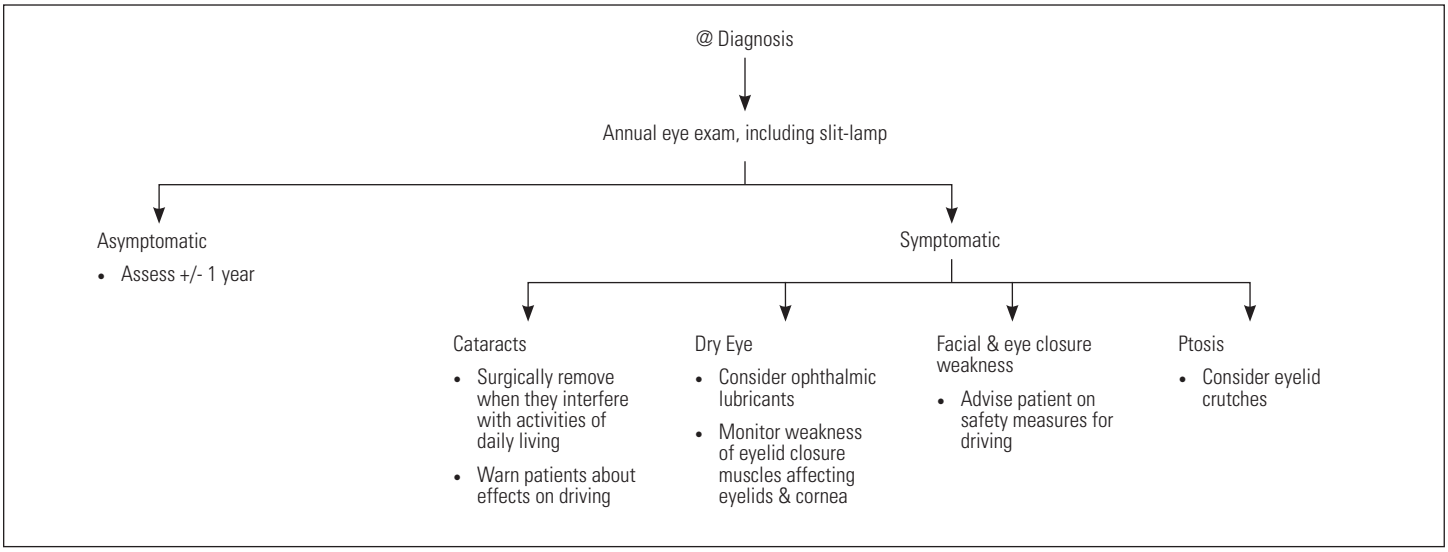
Gastrointestinal symptoms

- Ask about problems with chewing, swallowing, drooling, reflux, bloating, abdominal pain, bowel movement frequency & characteristics, diarrhea & incontinence
- Physical exam should include abdominal palpation, including around gall bladder, & rectal exam for anal sphincter spasm & dyssynergic defecation for symptomatic patients
- DM1 patients are at risk for pseudo-obstruction, & experience other problems that may cause actual obstruction of small or large intestine, including endometriosis, acute gallbladder inflammation, ruptured ovarian cysts, sigmoid volvulus. Monitor potential obstructions to determine whether they are pseudo or actual & treat accordingly
- Non-medical interventions:
  - High-fiber diet for diarrhea or constipation; increase water intake
  - Nutritional supplement for weight loss, weight gain or dysphagia
  - Dysphagia therapy referral for oral pharyngeal dysphagia
- Medical interventions:
  - Loperamide (gentle use) for diarrhea control
  - Laxatives for constipation:
  - First line therapy: Miralax, Senna, Ducosate or Linaclotide
  - Second line therapy: Bisacodyl, Lubiprostone, Linaclotide
  - Avoid oils – if above fails, refer out for anal manometry
  - Metoclopramide for gastroparesis, pseudo-obstruction, reflux
  - Antibiotics for bacterial overgrowth-induced diarrhea (based on breath testing)
  - Enteral feeding only for recurring pneumonia or severe dysphagia causing weight loss or causing inability to swallow safely without recurrent aspiration
  - Mexiletine can be considered to treat diarrhea or constipation. Mexiletine is contraindicated for DM1 patients with cardiac involvement. See full recommendations regarding mexiletine & cardiac involvement.
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Ocular symptoms

- Relevant eye manifestations of DM1 include cataracts, strabismus & other ocular motility problems, myopia, & astigmatism in congenital & juvenile-onset patients
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Fig. 3 Ocular Recommendations Flowchart



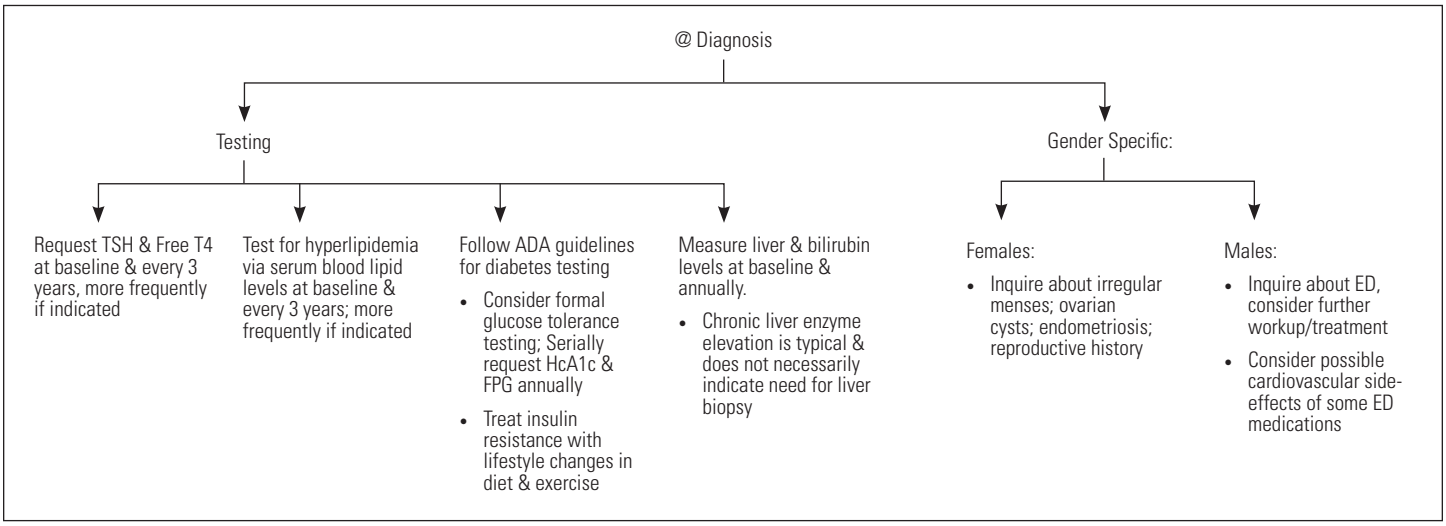
Tumors

- Look for pilomatrixomas (skin tumors); refer to surgeons for safe removal
- Train patients to detect pilomatrixomas (small, hard lump under skin on head, neck, arms, torso, legs)
- Follow general population cancer screening guidelines, particularly for breast, testicular, cervical & colon cancer
- Evaluate suspicious new CNS, abdominopelvic & thyroid symptoms for possible cancer; consider cancers of the brain, uterus & ovary
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Endocrine & metabolic symptoms

- See full recommendations at **http://www.myotonic.org/clinical-resources**

Fig. 4 Endocrine & Metabolic Care Recommendations Flowchart



Neuropsychiatric symptoms

- Advise patients that DM1 is also a 'brain disorder'
- Include psychiatric & behavioral examination at baseline, & during regularly-scheduled follow up appointments or when symptoms appear
- Refer patients with psychiatric or behavioral disorders, those with late-onset phenotype & patients with cognitive complaints to mental health care professional for testing & follow up; patients may have limited insight into these issues – consider input from partners & family members as appropriate
- DM patients with late-onset phenotype can exhibit fast decline in certain cognitive functions
- See full recommendations at **http://www.myotonic.org/clinical-resources**

Psychosocial symptoms

- Assess patient's social circumstances in household; consider & be aware of possible child neglect, acute financial need, unsafe driving, unsafe or unsanitary home; refer to social services, support programs & organizations

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A full list of authors & an overview of the methodology used to develop consensus for these recommendations can be found here: **http://www.myotonic.org/clinical-resources**