

# Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 2

## Quick Reference Version

Due primarily to a significant lack of rigorous data, no evidence-based guidelines exist to inform the clinical care of people living with myotonic dystrophy type 2 (DM2). In order to improve and standardize care for this disease now, 15 leading myotonic dystrophy (DM) clinicians in Europe, Canada and the United States have created the Consensus-based Care Recommendations for Adults with Myotonic Dystrophy Type 2. A complete list of authors is available on page 4.

### Pain control

- Muscle pain can occur in the neck, back, shoulders, hip flexors, & upper legs. Statin-induced myopathy is often accompanied by muscle pain
- Treat with conventional pain medications to address the painful aspects of DM2. (Ibuprofen, etc.)
- Opioids can be used but should be avoided if possible. If implemented, low doses should be used with close monitoring for side-effects (See Anesthesia & surgery)
- Other remedies, such as massage, nerve blocks, heat/ice, or chiropractic may provide benefit in select patients. Anecdotally, some patients have reported that cannabis helps ease pain, however more research needs to be conducted (Jensen et al, 2008)

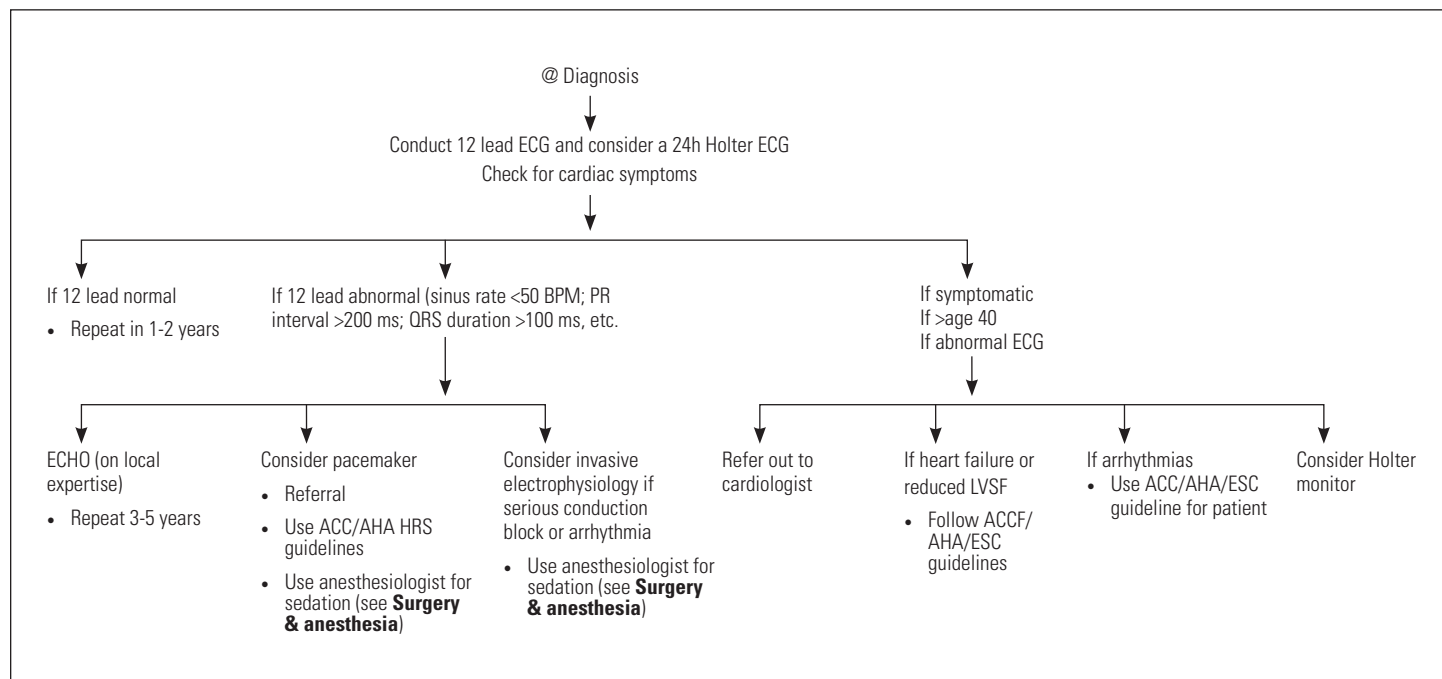
- Refer to physical therapy or occupational therapy if conventional treatment isn't successful
- See full recommendations at <https://www.myotonic.org/toolkits-publications>

### Cardiovascular management

DM2-related cardiac pathophysiology, although affecting all myocardial tissue, preferentially targets the cardiac conduction system. Conduction system defects are progressive & while initially asymptomatic, increase the risk for symptomatic arrhythmias.

- See full recommendations at <https://www.myotonic.org/toolkits-publications>

Fig. 1 DM2 Cardiac Care Recommendations Flowchart



## Neuropsychiatric management

- Advise patients that DM2 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, & patients with cognitive complaints to mental health care professionals for testing & follow up; patients may have limited insight into these issues – consider input from partners & family members as appropriate
- Consider the use of psychostimulants if apathy is associated with an impairing level of fatigue or excessive daytime sleepiness (see excessive daytime sleepiness) or antidepressive medication (cardiac examination before starting treatment, including a 12 lead ECG)
- See full recommendations at <https://www.myotonic.org/toolkits-publications>

## Respiratory management

Some DM2 patients have significant breathing problems that can result from muscle weakness of the diaphragm, abdominal & intercostal muscles & myotonia of these muscles, leading to poor ventilatory force & resulting in low blood oxygen & elevated blood carbon dioxide levels.

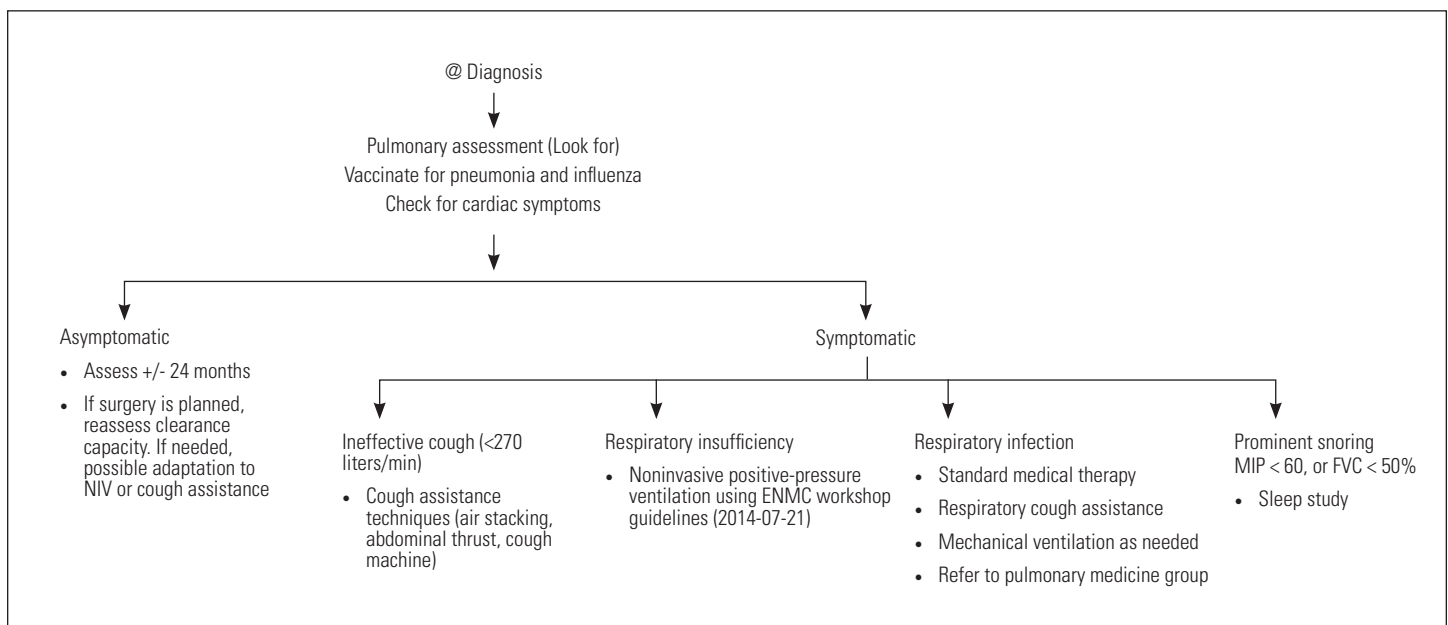
- See full recommendations at <https://www.myotonic.org/toolkits-publications>

## Excessive daytime sleepiness

Excessive daytime sleepiness (EDS) & fatigue may be life altering & disabling in DM2.

- Assess for excessive daytime sleepiness (EDS) with Epworth Sleepiness Scale or similar standardized questionnaire instrument; prescribe sleep study if a 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 as needed 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 the presence of EDS
- If nocturnal or daytime hypoventilation is suspected, consider non-invasive positive pressure ventilation, & refer to a pulmonologist with experience in neuromuscular diseases re: possible need for NIV treatment
- Consider modafinil for treatment if coexisting central nervous system alteration is suspected as the cause for EDS

**Fig. 2 DM2 Pulmonary Care Recommendations Flowchart**



- 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 <https://www.myotonic.org/toolkits-publications>

### Skeletal muscle weakness & rehabilitation

Skeletal muscle weakness & myalgia are major features of DM2 if bilateral & progress at a relatively slow rate.

- Initial weakness is typically in the proximal hip girdle & neck (flexors > extensors) muscles. Axial muscle weakness is frequent in DM2 & may result in lower back pain
- Mild ptosis might be occasionally present. Calf hypertrophy may occur
- Myalgic pains can be the most prominent clinical feature in the early stages & may severely affect occupational performance
- Neck flexor weakness can occur causing difficulty raising the head from a surface
- Treat with moderate- or low-intensity aerobic & resistance exercise, orthoses or braces. It is advisable to obtain a cardiac evaluation prior to starting a new exercise routine
- See full recommendations at <https://www.myotonic.org/toolkits-publications>

### Skeletal muscle myotonia

- Myotonia – sustained muscle contraction & difficulty relaxing muscles may be present
- Myotonia can contribute to muscle stiffness, muscle locking, pain, prolonged hand grip, speech & swallowing difficulties, & GI issues & may be associated with hand tremor
- Mexiletine may be considered for myotonia treatment. Mexiletine is contraindicated for DM2 patients with cardiac involvement
- See full recommendations at <https://www.myotonic.org/toolkits-publications>

### Gastrointestinal management

Ask about problems with chewing, swallowing, drooling, reflux, bloating, abdominal pain, bowel movement frequency & characteristics, diarrhea & incontinence

- DM2 patients are at risk for pseudo-obstruction, & may experience other problems that 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, Docusate or Linaclotide
    - Second line therapy: Bisacodyl, Lubiprostone, Linaclotide
  - Metoclopramide for gastroparesis, pseudo-obstruction, reflux
  - Antibiotics for bacterial overgrowth-induced diarrhea (based on breath testing)
- See full recommendations at <https://www.myotonic.org/toolkits-publications>

## Ocular management

Major & clinically relevant eye manifestations in DM2 can include the following: cataracts, eyelid ptosis & incomplete eyelid closure, retinal changes & changes in intraocular pressure.

- See full recommendations at <https://www.myotonic.org/toolkits-publications>

## Pregnancy & obstetrics management

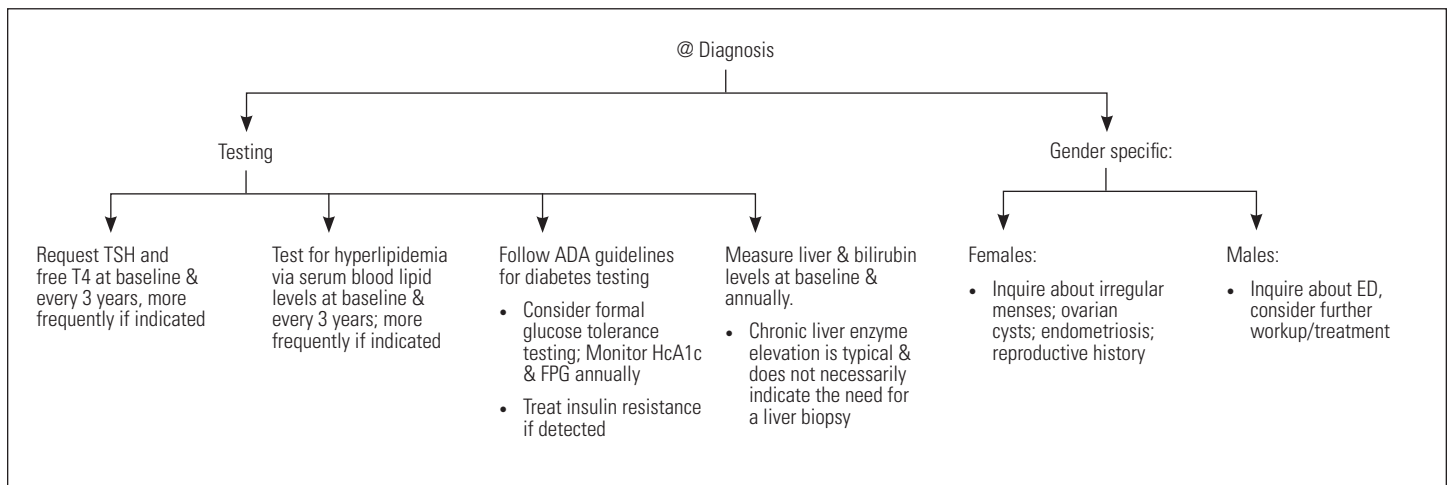
The effects of DM2 on both smooth & striated muscle can complicate pregnancy, labor & delivery.

Prenatal & preimplantation genetic diagnosis can allow for termination of the pregnancy or selective implantation of unaffected embryos if desired.

- See full recommendations at <https://www.myotonic.org/toolkits-publications>

## Endocrine & metabolic

**Fig. 3 Endocrine and Metabolic Care Recommendations Flowchart**



## Acknowledgments

This project would not have been possible without the tireless and longterm commitment made by the 15 international professionals involved in its development. Each is experienced in the care and treatment of people living with myotonic dystrophy type 2. They included:

Guillaume Bassez, M.D., Ph.D.  
Institut de Myologie

Barbara Fossati, M.D.  
U.O. Neurologia, IRCCS Policlinico San Donato

Josep Gamez, M.D., Ph.D.  
Vall d'Hebron University Hospital

Chad Heatwole, M.D., MS-CI  
University of Rochester

James Hilbert, M.S.  
University of Rochester

Cornelia Kornblum, M.D.  
University Hospital of Bonn

Anne Kostera-Pruszczyk, M.D.  
Medical University of Warsaw

Ralf Krahe, Ph.D.  
University of Texas MD Anderson Cancer Center

Anna Lusakowska, M.D., Ph.D.  
Medical University of Warsaw

Giovanni Meola, M.D.  
Department of Biomedical Sciences for Health  
University of Milan

Federica Montagnese, M.D.  
Friedrich-Baur-Institute, Ludwig-Maximilians-University Munich

Richard Moxley III, M.D.  
University of Rochester

Benedikt Schoser, M.D.  
Friedrich-Baur-Institute, Ludwig-Maximilians-University Munich

Charles Thornton, M.D.  
University of Rochester

Bjarne Udd, M.D., Ph.D.  
Tampere University

An overview of the methodology used to develop consensus for these recommendations can be found here:

<https://www.myotonic.org/toolkits-publications>

## Surgery & anesthesia

Although a higher incidence of adverse reactions to medications used for anesthesia & analgesia has been reported for DM1 (about 8%), it is yet not clear whether similar risks occur also in DM2 patients. However, given this uncertainty & the potentially serious complications reported in some DM2 patients, our advice is to adopt similar anesthesia guidelines as suggested for DM1.

For procedures requiring anesthesia, see *Myotonic Dystrophy Foundation's Practical Suggestions for the Anesthetic Management of a Myotonic Dystrophy Patient and Anesthesia Quick Reference Guide* at <https://www.myotonic.org/toolkits-publications>