

DM2 Clinical Care Checklist

- Monitor for conduction block (yearly EKGs/ holters)
- Counsel about anesthesia
- Monitor for cancer
- Monitor for cataracts and respiratory decline
- Counseling about in vitro fertilization, and selective implantation
- Treat co-existing disorders (thyroid dysfunction, diabetes, elevated cholesterol)
- Sleep study (OSA)
- Discuss enrollment in clinical trials
- Ask about and treat specific symptoms (e.g.: pain, sleepiness, fatigue)
- Ask about and treat myotonia



Clinical Myotonia

- Prolonged time for relaxation after:
- 1) Voluntary muscle contraction (action myotonia)
- 2) External mechanical stimulation (e.g. percussion or puncture with a needle)



Patient Description

- “Stiffness”
- “I can’t release my grip after shaking someone’s hand”
- “Difficulty releasing a fist.”
- “Pain and Cramping”
- “Locking”



Smooth Muscle Myotonia

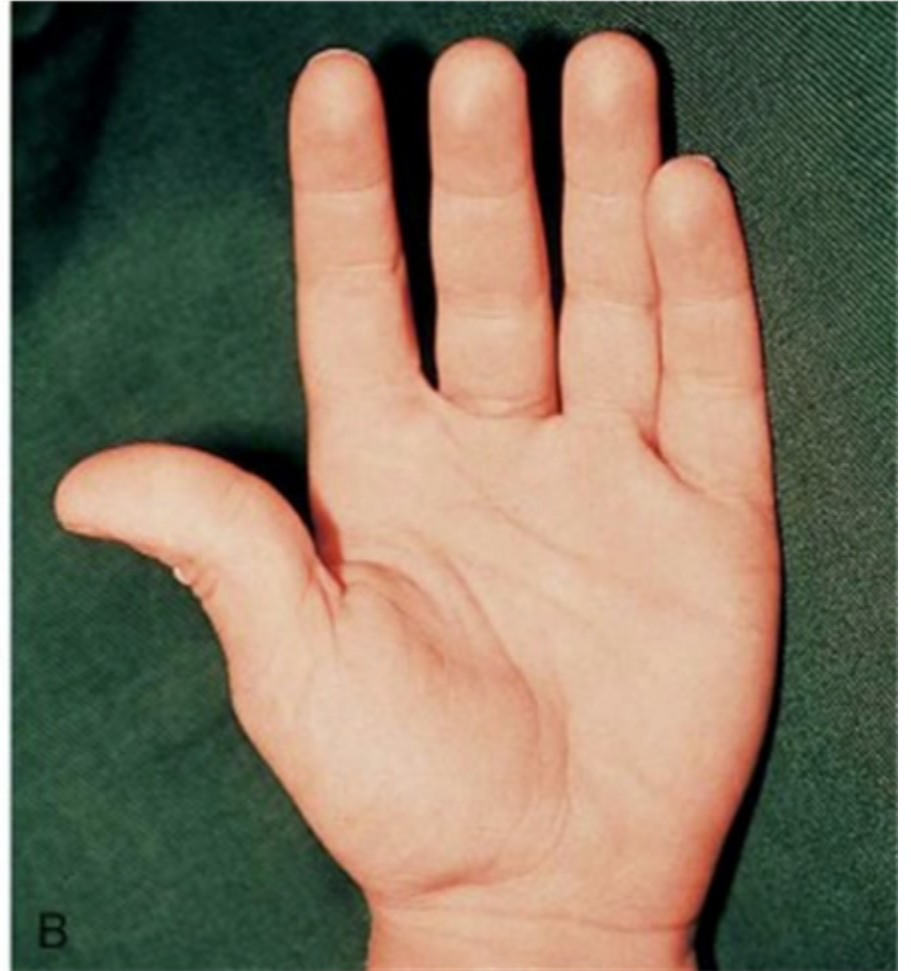
- Delayed pupillary response
- Gastrointestinal symptoms
- Abdominal pain
- Bloating
- Diarrhea
- Dysphagia



Cranial Muscle Myotonia

- Difficulty swallowing
- Difficulty chewing
- Impaired eyelid opening





<http://www.myotonia.com.hr/en/what-is-myotonia/>

Characteristic Feature of Clinical Myotonia

- Warm-up Effect
- In DM2 there can be a “ratchety” quality



What is Electrical Myotonia?

- Electrical hyperexcitability of the muscle
- Prolonged, spontaneous trains of muscle fiber action potentials that wax and wane in amplitude and frequency
- Recorded with EMG needle
- Usually appears as trains of positive waves
- In DM2 the most common location is the tibialis anterior muscle



What do people say it sounds like?

- WW2 Dive-bomber
- Revved motorcycle engine
- Chainsaw



Myotonic Treatment



Myotonia Trials in Myotonic Dystrophy Type-1 (off label)

- Carbamazepine (1983)
- Dilantin (1984)
- Nifedipine (1987)
- Imipramine (1989)
- Mexiletine (2010) (150 mg and 200 mg tid is safe, effective, and well-tolerated over 7 weeks)



Mexiletine

- First-line therapy
- Class 1B antiarrhythmic (enhances fast inactivation of sodium channels)
- Studied in DM1 and NDM
- 150mg TID
- Half life: 9 hours; bioavailability is 90%; peak concentration in 90 minutes.
- Tremor, diplopia, GI disturbances
- Take with food
- Isolated arrhythmogenesis has been reported (arrhythmia aggravation in patients with ventricular arrhythmias is lower with mexiletine)
- Still off label
- **Currently being investigated in a placebo controlled study**

