

Anesthesia for Patients with Myotonia Congenita



Quick Reference Guide

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Myotonia Congenita (MC) is an autosomal dominant or autosomal recessive genetic mutation of **CLCN1**.^{1,2,3,4,5,6,7} MC may be inherited as a mild form as an autosomal dominant trait (Thomsen's disease) or a more severe form with a recessive genetic mutation (Becker's disease).^{1,2,3,4,5,7,8} Patients with MC often have an abnormally enlarged muscular physique with percussion myotonia on physical exam.^{3,4,5,6,7} Thomsen's = greater effect on upper body (i.e.. Dysphagia, Mastication, & Airway); Becker's = Impacts the lower extremity muscles.^{6,7} Triggers precipitate a delayed muscle reaction causing transient muscle stiffness and impaired mobility.^{2,3,4} The primary symptom is delayed muscle relaxation following voluntary contraction, with some Becker patients also having brief weakness (a few seconds) at the onset of movement.^{3,4,5} Patients note that the stiffness is most prominent with the first movement after a period of rest but repeated muscle contractions may alleviate the stiffness (warm up).^{3,4,5,8} With occurrence of less than 1:100,000 in the general population, most treatment comes from anecdotal reports and not all patients respond alike.^{3,6,9}

Daily Prevention/Treatment: Avoid pharmacological and environmental triggers.^{2,4,5,7} Sodium channel blockers (mexiletine or lamotrigine) or a carbonic anhydrase inhibitor (acetazolamide) help reduce the frequency and severity of episodes.^{2,3,4,5,7,8,9}

Malignant Hyperthermia (MH): Though myotonic episodes may resemble an MH event, MH susceptibility is believed to be no different than that for the general population, **UNLESS** the patient has an RYR1 mutation, personal history, or close family history of MH.^{10,11,12}

Pregnancy: No contraindications for normal childbirth, but pregnancy may worsen the symptoms.^{2,3,5,7,11} Need to form a comprehensive birth plan with a multidisciplinary team.^{4,5,7} Labor epidurals for pain/anxiety and spinals for cesarean sections work well.^{6,7} Movement during labor may be beneficial.² Keep patient normothermic and hydrated.^{2,7} Guideline below for general anesthesia.

Anesthetic Plan

Standard of Care:	Maintain normothermia, prevent shivering, appropriate hydration, anxiety control, and avoid attack triggers. ^{2,5,6,7,8,12}
Common Attack Triggers:	Pregnancy, cold temperatures, hunger, emotional stress, pain, fatigue, exercise, rest after exercise, succinylcholine, colchicine (especially with renal insufficiency), and propranolol. ^{2,3,4,5,6,7} Caution with solutions containing epinephrine and high doses of beta agonist medications. ^{4,5,7,8}
Regional / Local Anesthesia:	Preferred anesthetic choice! ^{6,7,8,12} Normal dosing of spinals, epidurals, regional nerve blocks, and local infiltration (without epinephrine) with appropriate sedation and monitoring. ^{8,11,12}
Pre-op Testing / Consultations:	Physical exam, electrolytes, blood gas analysis, airway exam, chest X-ray, and EKG. ⁷ Multidisciplinary consultations needed. ^{4,5,6}
Preparations:	Fluid warmer, forced air warmer, fluid with minimal potassium, and nerve stimulator. ^{2,7,8,12}
Intra-op Monitoring:	Core temperature, electrolyte levels (avoid hyperkalemia) and nerve stimulation. ^{2,3,6,7,8,12}

Anesthesia Process for Myotonia Congenita Patient

Preoperative	Keep patient normothermic in preoperative area and during transport. ² IV access for fluids (Normal Saline and Lactated Ringers each used in a case study). ^{6,8} Sedation with benzodiazepine (midazolam used in case study) ⁸ .
MH Susceptible	Follow MH guidelines. Propofol for induction. ^{2,6,12} Case reports with successful use of propofol infusion and remifentanyl infusion for maintenance. ^{2,6,8,12}
<u>NOT</u> MH Susceptible	Propofol for induction. ^{2,6,12} Case reports show successful use with individual or combinations of sevoflurane, isoflurane, nitrous, propofol infusion and remifentanyl infusion for maintenance. ^{2,6,8,12}
Paralysis	No Succinylcholine – may cause masseter spasm, exaggerated contracture, and laryngospasm. ^{2,4,5,7,8,12,13,14} Low dose nondepolarizing muscle relaxant may be used. ^{2,8,12} Avoid use of anticholinesterase agents. ^{2,8,12,14} Sugammadex reversal (successful use case study with Beckers MC) with diligent neuromuscular monitoring. ^{8,15}
Maintenance	Maintain normothermia and adequate hydration. ^{2,8,12}
Pain Management	Regional and local anesthesia are preferred! ^{6,7,8,12} A successful outcome was reported using low dose fentanyl IV injection combined with a peripheral nerve block. ⁸ Remifentanyl infusions with propofol show positive results. ^{2,6,8,12}
Antiemetics	No known case reports or studies. No known contraindications.
Emergence	Continue strict temperature control and shivering to avoid a myotonia trigger. ^{6,7,8,12} Patient may be weak from anesthesia/disease. ⁶ Use great care with extubation. ⁶
Recovery	Have ICU availability. ⁶ Concern is prolonged post-operative weakness and extubation may be prolonged. ⁶ Prevent shivering. ^{6,8,12} Need good pain control. ^{6,7} Monitor electrolytes and blood gases as needed. ^{6,11} Movement as soon as possible is beneficial but be careful due to high fall risk. ^{2,5}

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This document gives anesthesia providers basic guidelines designed to increase the safe anesthesia care for this patient population. Being a rare disease, most of the information is based on the lower level of the evidence hierarchy and does not consider comorbid conditions. Feedback and comments may be forwarded to PeriodicParalysisResearch@gmail.com

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