

# Ulnar Nerve Decompression in a Patient with Myotonia Congenita under Regional Anaesthesia in a Daycase Setting: A Case Report

A Miglani<sup>a</sup>, L.M. Hughes<sup>b</sup>, C. Bossut<sup>a</sup>, C.C Nestor<sup>a</sup>

## Abstract

Myotonia congenita is a rare channelopathy characterized by impaired skeletal muscle relaxation after a depolarization causing myotonia. Several perioperative factors can trigger myotonia which can result in life-threatening muscle spasms with resultant ventilation difficulties, hence majority of these patients are not considered for ambulatory surgery. There is also a gap in the literature surrounding the application

of peripheral nerve blockade in this patient cohort. We report the case of a patient with Myotonia congenita who successfully underwent ulnar nerve decompression in a day-case setting under regional anaesthesia and sedation highlighting the importance of proper patient selection and preparation.

**Keywords:** Myotonia congenita, day case surgery, ambulatory anaesthesia, regional anaesthesia.

**Authors' Addresses:** <sup>a</sup>Tallaght University Hospital, Dublin, Ireland. <sup>b</sup>Alfred hospital, Melbourne, Australia.

**Corresponding Author:** Dr. Ankita Miglani, Department of Anaesthesiology, Tallaght university hospital, Dublin, Ireland.

Email: [ankitamiglani83@gmail.com](mailto:ankitamiglani83@gmail.com)

## Introduction

Myotonia congenita (MC) is a rare genetic disorder affecting skeletal muscle excitability, with a worldwide prevalence of about 1:100,000 (1). MC is caused by a mutation in the muscle chloride voltage-gated channel 1 gene (CLCN1) on chromosome 7q35. The clinical manifestations can vary widely, from subclinical symptoms to severe myotonic reactions and muscular weakness.

Neuromuscular disorders are rare but can have serious implications for these patients when undergoing surgery under anaesthesia (2). Association with musculoskeletal, respiratory, cardiovascular, gastrointestinal, metabolic, and central nervous system disorders warrants careful perioperative planning to achieve the best outcomes for this complex patient cohort. The anaesthetic management of a patient with MC should involve a careful emphasis on minimizing myotonic reactions, which can be triggered by several precipitants like, hypothermia induced shivering, pain (including pain on propofol injection), emotional stress, prolonged fasting and drugs such as depolarizing muscle relaxants (e.g., suxamethonium), adrenaline, beta-adrenergic agonists, and propranolol.

Most patients with MC are not considered for ambulatory surgery given the associated perioperative challenges. To our knowledge, there have been no published case reports of ambulatory anaesthesia as an option for managing patients with MC. In addition, there is a gap in the literature surrounding the efficacy of peripheral nerve blockade in this patient cohort. Our case report demonstrates that with proper patient selection and careful preparation successful ambulatory care under regional anaesthesia (RA) is possible for patients with MC. Written informed consent was obtained from the patient and the manuscript adheres to the applicable EQUATOR guidelines.

### Case description:

A 36-year-old female required decompression and transposition of the ulnar nerve. She was a known case of MC (Thomsen's type 1) since age 30, with a mutation in the CLCN1 gene with co-expression of sequence variant c.811T>C:P.Cys271Arg (reported as being consistent with but not diagnostic of autosomal dominant {AD}

inheritance). Electromyography and nerve conduction studies had been performed to confirm the diagnosis of MC, and a muscle biopsy had excluded muscular dystrophy.

She first noticed symptoms of stiffness aged 8, predominantly affecting her upper limbs, which was precipitated by hunger, stress, cold temperature, and infections. Worsening of myotonia was observed during her pregnancy 6 years ago when she had difficulty standing from seated. She reported stiffness in the neck which improved on movement, keeping with the warm-up phenomenon which is a well-known attribute of the disease. The patient was prescribed both phenytoin and mexiletine in the past, but therapy was discontinued due to the limited benefit.

Before undergoing this surgery, the patient was reviewed by the consultant anaesthetist assigned to the case. Following a multidisciplinary consultation with the surgical team regarding the duration of the procedure and anticipated postoperative complications, a plan was made to proceed under RA and propofol based sedation in a day case setting.

Preoperatively the patient was advised to minimize fasting time to 6 hours for solids and 2 hours for clear fluids as hunger had been noted as a precipitant of her myotonia. She was listed to be the first case on the list and malignant hyperthermia (MH) precautions were observed. Intraoperatively routine monitoring was used, warmed maintenance fluids were commenced, and a forced-air warming system was utilized to maintain temperature. The axillary block was performed with a 22G Stimuplex needle under ultrasound guidance with 20ml of 0.5% bupivacaine. After ensuring adequate anaesthesia of the limb, the tourniquet was inflated. Intravenous lignocaine 1% 3ml was given to minimize the pain of propofol infusion and a target-controlled infusion (TCI) of propofol using the Marsh Model to target a plasma concentration of 1-2 mcg.ml<sup>-1</sup> was commenced and titrated throughout the surgery aiming to achieve arousal to verbal stimulus. Surgery was uneventful with the tourniquet released at 59 minutes. The patient reported being comfortable in recovery and was safely discharged home the same day after routine day case discharge criteria were met. The patient was telephonically reviewed at days 1

and 2 postoperatively by the consultant anaesthetist. The block had fully resolved, pain was well controlled on oral analgesia and no post operative complications were noted.

## Discussion

MC is a rare channelopathy caused by a mutation in CLCN1 causing interference with the appropriate relaxation of skeletal muscle fibers after depolarization, resulting in prolonged muscle contraction or myotonia. The disease was first described by Thomas Thomsen in Schleswig, Denmark in 1876 who himself was affected by this disorder. Two main forms of MC are described: Thomsen's myotonia (AD) and Becker's myotonia (autosomal recessive) (2). Thomsen's myotonia presents with less severe symptoms of muscle stiffness predominantly affecting the upper limb muscles beginning from infancy to 2-3 years of age. Patients generally do not develop muscle weakness later in life. In contrast, Becker's myotonia is twice as common and presents later with more severe symptoms predominantly affecting the lower limbs. Symptoms are apparent between 4-12 years of age and can lead to muscle weakness later in life. Myotonia is often reported by patients as muscle stiffness, most pronounced in the extremities which ameliorates with continuous activity also known as the "warm-up phenomenon" and worsens after rest (3). The binding of acetylcholine to postsynaptic acetylcholine receptors causes the opening of sodium channels and sodium influx, leading to depolarization. The action potentials are propagated along the sarcolemma and t-tubule system. The arriving action potential, in turn, activates voltage-dependent calcium channels in the adjacent sarcoplasmic reticulum, causing a rise in cytosolic calcium levels and thus muscle contraction. Normally chloride influx stabilizes the action potential but in MC, this chloride conductance is impaired leading to prolonged contraction (4). Myotonic reactions in patients with MC may range from subclinical findings and periodic symptoms to disabling muscle weakness. Muscle stiffness may respond to sodium channel blockers such as mexiletine (currently the medication with the best documented effect), lamotrigine carbamazepine or phenytoin. Beneficial effects have also been reported with quinine, dantrolene, and acetazolamide (3).

In general, available literature outlining the anaesthetic considerations for the myotonic patient is relatively scarce. The few published case reports outlining anaesthetic considerations for these patients highlight potential issues of masseter spasm at induction leading to difficulty intubating, aspiration events and precipitation of myotonic contractions (4-7). Given these risks, MC is a condition that traditionally would have been considered a relative contraindication for day-case anaesthesia. Our case describes how a RA technique in combination with sedation using short acting drugs such as propofol, carefully titrated using a TCI can be used without any respiratory side effects and with careful planning and patient selection, the patient can be safely discharged home the same day of surgery.

Neither RA nor muscle relaxation can control myotonic contractions once they begin, emphasizing the importance of prevention. The anaesthetic plan for patients with MC must involve a consideration for minimizing triggers of myotonic reactions, some of which include hypothermia-induced shivering, pain (including pain on propofol injection), and certain drugs (4). Regional nerve block used in our report had the advantage of providing postoperative analgesia which is a key consideration for preventing myotonias in the postoperative period. Whilst a regional technique was used successfully here, the perioperative plan must include a plan for conversion to GA in the instance of a failed regional block. An awareness of the effects of the different neuromuscular blocking drugs on this condition is essential for the anaesthetist. Patients with MC are at high risk of developing

a severe myotonic response with generalized muscle spasms when depolarizing muscle relaxants (suxamethonium) are used. This could include masseter spasm, opisthotonos and decerebrate posturing making intubation and even ventilation difficult (7,8). In general, the response to non-depolarizing muscle relaxants (NDMRs) is reported to be normal, but dose reduction is advised in cases of established muscle weakness and wasting (late stages of Becker's disease). Anticholinesterase drugs have been shown to precipitate myotonia so short-acting NDMRs should be used when possible. Sugammadex for the reversal of rocuronium-induced neuromuscular block has been successfully employed but strong evidence suggesting its use is lacking. In vitro studies have shown that the administration of propofol had an anti-myotonic effect on skeletal muscle bundles but the clinical importance of this is unclear (9). Traditionally, concerns about susceptibility to MH have been described in patients with MC. Whilst this predisposition to MH is not clearly established, if sustained muscle contraction occurs during the induction or the course of anaesthesia, MH should always be suspected. Familiarity with MH protocol and availability of dantrolene should be ensured for all patients with MC presenting for anaesthesia.

Perioperative management of a patient with MC can be particularly challenging. Whilst no definitive recommendations exist recommending regional or GA for these patients, the combination of a regional anaesthetic technique and TCI of a short acting sedative agent should be considered.

## Acknowledgements

Published with the written consent of the patient.

## References

1. Emery AE. Population frequencies of inherited neuromuscular diseases – a world survey. *Neuromuscular Disorders* 1991;1:19–29.
2. Bissonnette B, Luginbuehl I, Marcinick B. Syndromes : **Rapid Recognition and Perioperative Implications, Second Edition**. McGraw Hill, 2019.
3. Dunø M, Vissing J. Myotonia Congenita, Updated 2021. Seattle: GeneReviews [Internet], 2005.
4. Bandschapp O, laizzo PA. Pathophysiologic and anesthetic considerations for patients with myotonia congenita or periodic paralyses. *Pediatric Anesthesia* 2013;23:824–33.
5. Hasan SS. Anesthetic management of a patient with myotonia congenita. *Anaesthesia, Pain & Intensive Care* 2021;25:816–8.
6. Abut YC, Şimşek S, Köse S, et al. Anesthesia Experience in a Patient with Myotonia Congenita. *Bağcilar Medical Bulletin* 2019;4:53–5.
7. Farbu E, Sjøfteland E, Bindoff L. Anaesthetic complications associated with myotonia congenita: case study and comparison with other myotonic disorders. *Acta Anaesthesiologica Scandinavica* 2003;47:630–4.
8. Looi I, Bakar A, Lim C, Khoo T, Samuel P. Anaesthetists' nightmare: masseter spasm after induction in an undiagnosed case of myotonia congenita. *Medical Journal of Malaysia* 2008;63:423–5.
9. Bandschapp O, Ginz H, Soule C, et al. In vitro effects of propofol and volatile agents on pharmacologically induced chloride channel myotonia. *Anesthesiology* 2009;111:584–90.