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# Anaesthetic Management of Two Patients of Steinert Syndrome (Myotonic Dystrophy-Type 1) for Emergency Surgery

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## Case Study

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**ABSTRACT**

Anaesthesia for Myotonic dystrophy type 1 (DM1), also known as Steinert's disease, is a genetic disorder that mainly affects muscle function and is characterized by progressive muscle weakness and loss of muscle mass. The involved muscles often goes into sustained contraction and fail to relax (myotonic crisis) under physical, chemical, thermal or electrical stimulations. Patients with DM1 are hypersensitive to too many anesthetic drugs, especially muscle relaxants and opioids. Anesthesia for DM1 patients often results in cardiovascular, skeletal and pulmonary complications. Postoperative respiratory complications, such as hypoxia, atelectasis, aspiration and respiratory failure are very high in patients with DM-1. Thorough patient evaluation and proper anaesthetic planning and active postoperative care are very important in managing such patients. We encountered two patients with Steinert myotonic dystrophy syndrome for emergency surgeries, one for exploratory laparotomy for perforated appendix and another of acute rectal prolapse for manual reduction and rectopexy. Both cases were successfully managed without using any opioid and muscle relaxant.

## INTRODUCTION

Myotonic dystrophy is an autosomal dominant genetic disorder caused by abnormal repetition of genetic codes (trinucleotide). There are repeats of two or three nucleotides in certain areas of DNA. It's a disorder mainly affecting the muscular system characterized by gradual wasting of muscle, weakness and periodic muscle spasm called as myotonia (incomplete muscle relaxation). Muscular dystrophy are of two main types; type 1 (DM-1), also known as Curshmann-Batten-Steinert-disease and type 2 (DM-2), Proximal myotonic myopathy (PROMM) which is generally milder than type 1.

Primary manifestations and clinical progress depend on the amount of genetic codes repetition. Clinically DM 1 is characterized by myotonia (incomplete muscle relaxation), marked wasting of the muscles of mastication, neck, pharynx and distal limbs. Extra muscular features may include cataracts, frontal baldness, conduction abnormalities in heart and cardiomyopathies, restrictive lung disease, central and obstructive sleep apnea syndrome, dysphagia, delayed gastric emptying and endocrine abnormalities such as hypothyroidism, primary hypogonadism infertility and diabetes mellitus [1].

Myotonic crisis can be trigger by physical, chemical, thermal or electrical stimulation. Patients with DM-1 are hypersensitive to sedative, anaesthetic and neuromuscular blocking agents which may contribute to delayed recovery from anaesthesia and increased intraoperative and early postoperative respiratory and cardiovascular complications. Myotonic crisis (sustained muscle contraction) can be induced by any triggering medications, electrolyte imbalance, hypothermia, shivering or any mechanical or electrical stimulus [1-4]. Due to progressive muscle atrophy and degeneration involving the respiratory muscles, it is not uncommon for patients of DM-1 to have a history of hypoxia, dyspnea on exertion, sleep apnea requiring CPAP or marked ventilatory muscle weakness necessitating BIPAP. Gastrointestinal (GI) involvement in DM-1 is frequent, may affect at any level and may be a presenting symptom in some undiagnosed DM-1. GI involvement in DM remains a complex pathophysiology which bears no correlation with disease duration and severity. It may be a direct muscular and or neural involvement. Patients with muscle dystrophy may have oedema, atrophy and disappearance of smooth muscle cells and fibrosis. Common GI symptoms are dysphagia flatulence, heartburn, regurgitation and dyspepsia, abdominal pain, bloating and changes in bowel habits [5-7].

Patients with DM-1 are often abnormally sensitivity to opioid, inhalation anesthetics and muscle relaxants (both depolarizing

and non-depolarizing). Therefore, selecting safe drugs with minimal risk of complication is paramount in perioperative management of such patient. The objective of this case report is to highlight the perioperative issues in patients with DM-1 and to discuss the anesthetic implications in managing such patients.

## CASE DESCRIPTION 1

A nineteen years old female with DM-1 (facial muscle) was admitted with fever, vomiting and pain abdomen for two days. She had frequent loose watery stools for one day. She was leading a relatively normal life without any limb or muscle weakness and at that point she was not on any active medication for DM-1. General examination was unremarkable, except for temperature 37.8°C, tachycardic – HR-115/min, mild tachypnea - respiratory rate- 22/min systemic examination- revealed tender and distended abdomen. Ultrasound showed dilated bowel loops with interloop free fluid. Her total counts were elevated. ECG showed sinus tachycardia. She was started on broad spectrum antibiotics (piperacillin and tazobactam), but patient continued to have fever, pain abdomen and vomiting. The next day, a computed tomography (CT) scan was performed which showed pelvic abscess, possibly appendicular in origin and bilateral minimal pleural effusion. She was taken for emergency laparotomy with appendectomy. In view of her myotonic condition, spinal anaesthesia was planned. She was given a lumbar subarachnoid block with 2 ml 0.5% bupivacaine heavy along with fentanyl 25 mcg. Through a lower midline incision, 200 ml of purulent fluid in the pelvis was evacuated and appendectomy was done. Postoperative course was complicated by pneumonia and sepsis with septic shock and coagulopathy. Antibiotic therapy was revised and other supportive care. Patient's condition gradually improved over five days after revision of antibiotic therapy (meropenem and Clindamycin) along with other supportive care for sepsis in the form of hemodynamic support with aggressive fluid and vasopressor therapy, high flow oxygen, active chest physiotherapy and nutritional support. She was shifted out of ICU on 6$^{th}$ postoperative day and started on oral diet. After few meals on an oral diet, she developed vomiting, regurgitation and constipation. Examination revealed mild abdominal distension and absent bowel sounds. Abdominal x-ray showed gas filled grossly dilated stomach reaching up to the pelvis. Placement of nasogastric tube done for continuous aspiration to relief the gastric distension and patient was kept in propped up position. Two liters of fluid was drained out. An oral dye study was performed to confirm the diagnosis and nasojejunal tube was inserted with the tip at duodenojejunal flexure under fluoroscopic guidance. She was kept nil by mouth, continuous nasogastric aspiration and was initiated on nasojejunal feeds which she tolerated well. There was significant improvement in her saturation and her chest became clear. With these measures her gastroparesis improved and her nasogastric tube was removed. Her repeat oral dye study before discharge showed that stomach emptied after some hold up with positional changes. Nasojejunal tube was removed and she was started on liquid and later a semisolid diet.

## CASE DESCRIPTION 2

A 46 year old male patient presented with acute pain and bleeding per rectum, diagnosed as prolapse of rectum with hemorrhoids. Patient was posted for emergency manual reduction of prolapse. Patient was a known case of Myotonic dystrophy type 1. He had family history of Myotonic Dystrophy. Patient had a history of ear surgery under General Anesthesia in his home country 3 years back done without muscle relaxants and history of inguinal hernia repair done under spinal anesthesia. Both surgeries were uneventful.

On evaluation, patient was conscious, oriented, but anxious and apprehensive, profound atrophy of limb and trunk muscles. There was slurring of speech on talking, he could walk with support. Vitals parameters were within the normal limit with room air saturation of 98%.

ECG showed normal sinus rhythm without any significant ST-T changes and Chest X ray was normal and routine blood investigations (hematology and biochemistry) were within normal limits.

Patient was taken up for emergency manual reduction of prolapse under sedation-analgesia with Dexmedetomidine infusion (1 mcg/kg.h) and target control infusion (TCI) of Propofol. Total procedure time was approximately 10 min. Plasma Propofol level was maintained within 3-4 ng/ml. Patient was stable during the procedure. Total Dexmedetomidine and Propofol consumption were 25 mcg and 110 mg respectively. Post procedure, he was sent to the high dependency unit (HDU) for further monitoring. He was scheduled for Elective Haemorrhoidectomy and Rectopexy after 2 days. Cardiology and Medical evaluation was done. Echocardiography showed normal Ejection Fraction of 60% with no valvular involvement or regional wall abnormalities. He was trained with deep breathing exercise and incentive spirometry. Arterial Blood Gas (ABG) showed $pO_2$ of 78 on room air.

He was taken up for Elective Haemorrhoidectomy with Rectopexy under spinal anaesthesia and was given Spinal in L3 L4 Interspace with 12.5 mg of 0.5% Bupivacaine heavy in sitting position. Level of block achieved was T12 for sensory. Patient was sedated with 2 mg of Midazolam and oxygen was administered with nasal canula with 2 L/min oxygen. Patient remained stable throughout the procedure which lasted for one hour. Post-operative analgesia was achieved with multimodal analgesia that includes peri-rectal local anaesthetic infiltration with 0.25% bupivacaine 20 ml by surgeon along with regular Paracetamol, diclofenac and PRN intravenous tramadol. Postoperative course was uneventful. Patient was discharged from hospital on the third day postoperatively without any complications.

## DISCUSSION

The muscular dystrophy is one of the predisposing factors for the development of acute gastric dilatation secondary to motility disorder and muscular degeneration [6]. The causes of acute gastric dilatation in our 1st case are multifactorial, in addition to DM-1, anxiety, medication, dyselectrolytemia (hypophosphatemia), sepsis, postoperative bowel dysfunction precipitating acute gastroparesis.

Providing anaesthesia to patients suffering from DM-1 pose series of challenges due to involvement of multiple systems (respiratory, cardiac, gastrointestinal and central nervous systems) as the disease progress. Increased sensitivity of central nervous system to sedatives, anxiolytics, and analgesics, further reduces the ventilatory drive and airway protection.

Rapid sequence intubation is recommended for General Anaesthesia as there is higher risk of regurgitation and aspiration due to delayed gastric emptying time [4]. Succinylcholine should be omitted at all as succinylcholine effects are unpredictable and it can itself trigger myotonic crisis in DM-1 [8,9]. Opioids may also induce muscle rigidity. Therefore, omitting opioids is also recommended. Myotonic crisis characterized by sustained muscle contraction with incomplete relaxation can lead to an increase in oxygen consumption and cardiac output, which may cause cardiorespiratory insufficiency, is one of the first things to be avoided in patients with DM-1 [10]. Several things can trigger Myotonic crisis in perioperative period such as, anxiety, fear, prolonged fasting, hypoxemia, hypercarbia, increased mechanical pressure, pain, adrenergic discharge, electric scalpel, hypothermia, peripheral nerve stimulator, shiveriing, medications, electrolytes imbalance and voluntary effort [5-7]. Involvement of cardiovascular conduction system may lead to cardiac arrhythmias and sudden cardiac deaths in these patients [8-11].

Preoperative electrocardiogram shows conduction disturbances in 50% of the patients, while electrophysiological studies are abnormal in 90% of the cases [6].

Surgery under balanced general anesthesia is associated with serious perioperative complications mainly related to the pulmonary system. Patients who were symptomatic of muscular dystrophy at the time of surgery and who underwent upper abdominal surgery were especially at risk. Therefore, “careful monitoring during the early postoperative period, protection of the upper airways, chest physiotherapy, and incentive spirometry are mandatory”.

Neuraxial anaesthesia supplemented with intravenous dexmedetomidine as an alternative to general anaesthesia have been described in the literature as successful anesthetics technique for cholecystectomy in a DM-1 patient [4]. However, neuraxial is also not without any risk; there are few case reports that describe shivering sufficient to stimulate myotonic contractures with neuraxial anesthesia, as well as incomplete blocks in patients with DM [12-14].

If a muscle relaxant is necessary for the surgery, depolarising agent should be avoided at all. Non-depolarizing agent with a short recovery index should be chosen (e.g. Rocuronium, Cis-atracurium) [15]. Laryngoscopy and jaw manipulation should be done with care as there is risk of temporo-mandibular joint dislocation during forceful laryngoscopy [16,17]. The use of muscle relaxant without reversal was shown to be an independent risk factor for an adverse perioperative event as risks of residual muscle relaxation in this patient group are significant [18]. Therefore, reversal should be considered when non-depolarizing muscle relaxants are used; Sugammadex has been used uneventfully to reverse muscular blockade in MD-1 patients [16,18]. Neuromuscular reversal agent’s neostigmine can induce Myotonia and therefore should be avoided if possible.

Total intravenous anaesthesia with propofol and ultrashort opioid (remifentanil) has been described in the medical literature [16,18]. Electrolyte imbalance (hypocalcemia and hyperkalemia) also can precipitate myotonic crisis. DM-1 patients have reduced $Na^+$-$K^+$ pump capacity and may be prone to the development of hyperkalemia [17]. Crystalloid fluids as well as colloid without added potassium can be safely used.

Opioid both systemic and neuraxial should be avoided for postoperative analgesia. Multimodal analgesia with combination of acetaminophen, non-steroid anti-inflammatory drugs (NSAIDs) and regional techniques using local anesthetics should be used for postoperative analgesia. If opioids are employed (systemic or neuraxial), then ICU care and continuous pulse oximetry must be considered given the high risk for respiratory depression and aspiration.

The anaesthetic management in both cases were slightly different, but in both cases were managed without using opioid and muscle relaxant. In the first case, we choose spinal anaesthesia to avoid the consequences of general anaesthesia. In the 2nd case, we could have given spinal anaesthesia, but as the manual reduction of rectal prolapse was of short duration with minimal invasion, we opted for procedural sedation; the 2nd case was again managed with spinal anaesthesia when he was scheduled for elective haemorrhoidopexy.

## CONCLUSION

Patients with Muscular Dystrophy (DM-1) present great challenges for anaesthesiologist as the anaesthesia for such patients are associated with issues to multiple of system mainly, musculoskeletal, respiratory, cardiovascular, nervous, gastrointestinal system. Greater use of regional anaesthesia or anaesthesia technique where there is minimal interference with self-respiratory mechanics should be used for patients with muscular dystrophy.

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