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CASE REPORT

REGIONAL ANESTHETIC MANAGEMENT OF A PATIENT WITH CHARCOT MARIE TOOTH DISEASE WITH HIP FRACTURE

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Abstract

Introduction: Charcot-Marie-Tooth disease (CMT) is a hereditary peripheral neuropathy characterized by progressive peripheral muscular atrophy and muscle-sensitive disorders, especially in extremities. The choice of anesthesia in these patients is a great challenge, as the neurological symptoms may worsen.

Case presentation: Female S.G., 53 years old, with a previously diagnosed Charcot Marie Tooth disease, was admitted to the Clinic for Orthopedic Diseases in Skopje for the treatment of a basicervical fracture of the femur. Apart from the existing neurological disease, the patient had no other comorbidities. An indication for surgery was set, and regional, i.e. spinal anesthesia was the choice for the surgical management of the patient. In the postoperative period, the patient was treated with analgesic therapy. After 9 days of treatment at the Clinic for Orthopedic Diseases, the patient was discharged in good general condition, without worsening of the neurological symptoms.

Conclusion: Regional anesthesia has been shown to be a safe type of anesthesia in surgical treatment of the lower limb.

Keywords: *anesthetic treatment; Charcot-Marie-Tooth disease; postoperative complications; regional anesthesia; sedation.*

Introduction

Charcot-Marie-Tooth disease (CMT) is a hereditary peripheral neuropathy characterized by progressive peripheral muscle atrophy and muscle-sensory impairment, especially in the extremities (1). Pathophysiological, it is characterized by genetic changes that cause a defect in the myelin sheath (demyelination) of peripheral nerves (CMT1, CMT3 and CMT4), or damage to axons (CMT2) (1-3). As a result of this myelin or axonal degradation, the conduction of

nerve impulses in motor and sensory neurons is reduced, leading to progressive distal muscle weakness, wasting and atrophy, and loss of sensation, often accompanied by neuropathic pain. Clinical symptoms are similar in the different types of CMT, their severity, and progression (2-4).

In most cases, the disease begins with weakness in the lower extremities, followed by weakness in the hands and forearms. Due to muscle weakness and atrophy, deformities of the feet and lower legs, upper extremities, and in rare cases scoliosis may occur. In axonal degeneration, the phrenic nerve may be affected, accompanied by deterioration of respiratory function due to diaphragmatic weakness (6-8). Cardiac problems such as mitral valve prolapse, QT prolongation, AV block and dysrhythmias may also occur (4).

Diagnosis of CMT is made through history and physical exam, and can be confirmed with electromyography (EMG), nerve conduction studies (NCS), nerve biopsies and/or genetic testing (9,10).

However, in resource poor settings diagnosis and treatment are often delayed. This can lead to the need for surgical correction of soft-tissue contractures and bony deformities (9-11).

The choice of anesthesia in these patients is a major challenge, as each type of anesthesia has risks and can lead to a worsening of neurological symptoms and the patient's general condition (9-13).

Case Presentation

Female S.G., 53 years old, with previously diagnosed Charcot Marie Tooth disease, was admitted on August 12th, 2024, to the Clinic for Orthopedic Diseases in Skopje for treatment of a basicervical fracture of the femur. The patient was injured 10 days before admission to our clinic and was hospitalized in another hospital, where a contraindication for surgical treatment was set, due to high anesthetic risk and resource poor settings for the treatment of critical patients. An indication for surgical treatment was set at the Clinic for Orthopedic Diseases in Skopje. From the anamnestic data, it was determined that the patient's CMT was diagnosed in the early period of puberty. The patient reported weakness in the legs and difficulty walking, but the medical history was normal, with no respiratory or cardiovascular problems. She was not receiving any chronic therapy and was allergic to Penicillin.

The initial assessment upon admission revealed stable respiratory status without recent coughing episodes, absence of wheezing or dyspnea, and no edema, hemodynamic or cardiac disorders were found. Chest X-ray was normal, SpO_2 - 97%, which is why spirometer analysis was not performed. The ECG was normal and without changes. A complete laboratory analysis and hemostasis were performed, which were also without significant changes. She denied previous surgical interventions. She was not smoker. She is classified in ASA II status. After 2 days, the patient was scheduled for implantation a subtotal prosthesis. The choice of anesthesia was regional (spinal) anesthesia. After explaining the risks associated with spinal

anesthesia and the growing evidence about the safety of this form of anesthesia in relation to the underlying condition, the patient accepted the technique and signed the informed consent. The patient had a normal constitution (height 163cm, weight 65kg) and we expected an easy performance of the intrathecal block. Of course, we considered that with regional anesthesia there was a lower risk of worsening of the neurological deficit compared to general anesthesia. Preoperatively, the patient was sedated with tbl diazepam 5mg. Before performing spinal anesthesia, basic monitoring such as ECG, non-invasive blood pressure, pulse oximetry and diuresis was performed. Initial parameters were: NIBP 100/60, HR 65b/min and SpO2 96%. Before the introduction to anesthesia, midazolam 1mg and ketamine 10mg were applied. Spinal anesthesia was performed under sterile conditions in the left lateral position, L3-L4 space, with a 25G needle. Clear cerebrospinal fluid was obtained and 1.5mg isobaric 0.5% bupivacaine was applied. The patient was managed on an oxygen mask with a flow rate of 4L/min, 1500ml saline 0.9% and one unit of erythrocytes at the end of the intervention. The surgical intervention lasted 2 hours and the patient was stable throughout the entire period with normal vital parameters. The block lasted 5 hours and after its discharge there were no changes from baseline in the motor and sensory status of the lower limb.
In the postoperative period the patient was treated with antibiotics, anticoagulant therapy and pain relief with paracetamol, NSAIDS and tramadol only on the first postoperative day. On the second postoperative day, physical therapy started. She was discharged from the surgical clinic after 9 days in good general condition.

Discussion

This type of disease is rare in anesthesiology practice, and as a result it is difficult to obtain relevant information on the type of anesthesia (6). Most of the published studies include a small number of patients and represent case reports. Regarding the anesthetic management of patients with CMT, there is controversial information regarding general versus regional techniques.
In the presented case, spinal anesthesia and preoperative sedation and analgesia with midazolam and ketamine were performed. The patient was stable perioperatively, without any disturbance of vital parameters (respiratory function and ECG changes) and without worsening of pre-existing preoperative neurological findings.
General anesthesia in patients with CMT carries risks when using certain anesthetics. Chronic denervation of peripheral nerves is a predisposing factor for hyperkalemia. However, in practice, significantly elevated potassium levels are rarely observed, probably because denervation occurs slowly. However, potassium levels should be checked preoperatively. For this reason, the use of succinylcholine for induction of anesthesia is not recommended (1-3). On the other hand, non-depolarizing muscle relaxants have a prolonged effect caused by muscle atrophy, which in turn leads to an increase in the number of acetylcholine receptors in the neuromuscular junction. Their careless use can worsen the weakness of the intercostal

muscles and diaphragm, and thus respiratory function. Therefore, they should be dosed carefully (3). Regarding the use of inhalation anesthetics, there are data on triggering malignant hyperthermia (MH) (3), although there is no report of a case of MH so far. Considering that CMT is a peripheral neuropathy and not a myopathy, it is considered that the occurrence of malignant hyperthermia triggered by inhalation anesthetics is unfounded (4). In a case report from 2021, a 24-years-old male with a previous diagnosis of CMT at the age of 14, was presented for the removal of a renal calculus. General anesthesia was performed, with induction with midazolam, fentanyl and propofol, and maintenance of anesthesia with dexmedetomidine infusion 0.5μg/kg/h. Atracurium was used as a muscle relaxant. Capnography was used as an indicator for repeating the dose of the muscle relaxant. At the end of the intervention reversal of the neuromuscular block was performed with neostigmine. The patient demonstrated all clinical signs of adequate reversal and satisfactory recovery (4). Vinci P., Lapi G. in a study from 2011, presented a case of a 73-years-old woman with two surgical interventions at an interval of 22 months (umbilical hernioplasty; transvaginal hysteroannexiectomy (5). The patient has previously been diagnosed with CMT2 (axonal degeneration), and reported severe sensory impairment in the lower extremities, particularly worsened after an epidural anesthesia performed several years ago for osteosynthesis of a hip fracture. Total intravenous anesthesia was performed for both interventions. Fentanyl and propofol were used for induction and maintenance of anesthesia, and atracurium was used as a muscle relaxant. The reversal of the block was performed with atropine and prostigmin. The patient was successfully extubated, but during both interventions in the immediate postoperative period she developed respiratory failure, which is why she was re-intubated and mechanically ventilated for 2½ hours after the first and 10 hours after the second surgical intervention (5). This study confirms the findings that patients with CMT2 have a higher risk of developing intercostal and diaphragmatic muscle weakness and are at higher anesthetic risk.

For minor surgical interventions in patients with CMT disease, light sedation can be safely used, as referenced in a study in 2016, in which midazolam and propofol were used for tooth extraction and implant placement in a 51-years-old man. After sedation, the patient was administered the local anesthetic lidocaine 2% (6).

Of course, there is also concern about the application of regional anesthesia, which is associated with peripheral nerve denervation. It has been thought that neuraxial blocks can worsen pre-existing nerve deficits. In the study by Rodrigez et al., a single subarachnoid (epidural) block was successfully performed in a 63-years-old male with diabetes mellitus and pulmonary comorbidity for fixation of a hip fracture. The choice for regional anesthesia was made in order to avoid respiratory deterioration with the use of general anesthesia and intubation (7). Roriz et al. also reported the safe performing of spinal anesthesia in a patient undergoing lower extremity orthopedic surgery (8), while Brock et al. demonstrated the successful use of combined spinal-epidural anesthesia for labor and cesarean section in patients with CMT (10).

Several studies have demonstrated safe and successfully performed peripheral nerve blocks (11-13). Localization of the nerve using ultrasound has been shown to be simpler and more reliable, demonstrated in a small group of 3 patients (11), compared to the use of a neurostimulator (12,13). Regardless of the technique used to perform the block, successful anesthesia and postoperative analgesia were achieved in patients with CMT in all three studies. Schmitt et al. used a catheter to achieve a continuous distal sciatic nerve block for foot surgery in a group of 27 patients with CMT. Localization of the sciatic nerve was guided by a nerve stimulator (13). In this study, it was determined that patients with the highest pain threshold required the least analgesia during the block. The authors believe that this phenomenon is due to demyelination of sensory and peripheral nerves. Patients with CMT, due to demyelination, have different pain sensitivity and therefore an individual approach to each patient is required. Local anesthetic was successfully applied through the placed catheter in the patients during three postoperative days, without complications and worsening of the previously determined neurological deficit (13).

Ritter and colleagues used a combined neuraxial block and ultrasound-guided peripheral nerve block for surgery for a foot deformity, in a patient with pronounced muscle atrophy, limb deformities and facial indicators for difficult intubation. With this combination of two regional anesthetic techniques, the authors report successful management of anesthesia in the patient, with minimal perioperative sedation and avoidance of the risk of difficult or impossible intubation.

The choice of the type of anesthesia in patients with CMT remains a dilemma. An individual approach to each patient is required, depending on the severity of symptoms and the type of trauma or disease to be treated surgically.

Conclusion

In this presented case, spinal anesthesia was performed without worsening neurological or sensory symptoms in the immediate and later postoperative period and proved safe type of anesthesia for lower extremity surgery.

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