



CASE REPORT

Several challenges associated with the anesthetic management of Emery-Dreifuss muscular dystrophy patients: case report

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Abstract Emery-Dreifuss Muscular Dystrophy is a very rare type of muscular dystrophy, associated with contractures, atrophy, and muscle weakness, besides cardiomyopathy with severe arrhythmias. Published studies focusing on this disorder are scarce. We describe the anesthetic management of a male patient with Emery-Dreifuss Muscular Dystrophy, to be submitted to umbilical and inguinal hernioplasty and hydrocele repair under epidural anesthesia. The anesthesia approach enabled us to circumvent the patient's susceptibility to malignant hyperthermia and his potentially difficult airway, in addition to maintaining hemodynamic stability. The day after surgery the patient resumed walking, and two days later he was discharged from the hospital.

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Introduction

Emery-Dreifuss Muscular Dystrophy (EDMD) is a very rare condition, with an estimated incidence ranging from 1:100,000 to 1:300,000.¹ Its clinical presentation is characterized by a triad of symptoms consisting of early

contractures of muscles, particularly in the regions of the elbow, Achilles tendon and posterior cervical muscles, weakness of muscles with humeroperoneal distribution, and cardiomyopathy, more frequently atrial conduction disorders with the development of severe arrhythmias.^{2,3} Cardiac disorder progresses as age increases, without correlation with the degree of muscle involvement.

The mutated gene is directly related to the production of nuclear membrane proteins present especially in skeletal and cardiac muscle. The way EDMD is inherited depends on the gene involved. Thus, when the mutation affects the

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emerin gene (EMD, Xq28), EDMD shows X-linked inheritance. Conversely, the disorder has both recessive and dominant autosomal inheritance, when the mutated gene encodes the production of Lamin A/C (LMNA, 1q21.2). Approximately 45% of patients do not carry these mutations, which suggests the presence of additional mutated genes yet to be detected.²

Diagnosis relies on the identification of the clinical trial; nonetheless typical symptoms may be mild or even nonexistent. Investigation by imaging, such as MRI, and electromyographic tests are crucial to document the atrophy of major muscle groups. Heart function can be assessed by echocardiography, and by exercise stress test and cardiac angiography/catheterization in the case of previous ischemic events. Diagnostic confirmation is obtained by genetic tests identifying mutations in the EMD gene (forms linked to the X chromosome) or in the LMNA gene (autosomal forms).¹ Molecular tests are based on the immunodetection of emerin in various tissues (muscles, lymphoblasts, skin), which is reduced or absent in EDMD.

Anesthetizing patients with EDMD is challenging as they are susceptible to Malignant Hyperthermia (MH) and may have cardiac abnormalities. Moreover, Airway (AW) management and the neuraxial technique may be difficult due to contractures of cervical and lumbar muscles.

Our literature review revealed a scarce number of cases reported.^{1,3-5}

Case report

A male, 59-year-old patient, ASA (American Society of Anesthesiologists) physical status III, with EDMD, pulmonary sarcoidosis, arterial hypertension, dyslipidemia, and renal lithiasis was scheduled for umbilical and right inguinal hernioplasties and hydrocele repair surgery with total surgical time expected to exceed 2 hours.

With no known family history, EDMD was diagnosed at the age of 49 years as the patient presented unsteady gait with permanent extension of the feet related to a pronounced contracture of the Achilles tendon, associated with a motricity disorder of lower and upper limbs. The patient had no cardiovascular or other signs or symptoms. Over 10 years, the patient was submitted to several surgeries to repair musculoskeletal defects on lower limbs. The first surgery was conducted under General Anesthesia (GA) and the subsequent surgeries under Spinal Anesthesia (SA), which were uneventful, except for some difficulty in performing the neuraxial technique.

The patient was admitted the day before surgery. He presented BMI of 24.2 kg.m^{-2} (weight 76 kg and height 178 cm), moderate muscle atrophy of upper and lower limbs and cervical and lumbar contractures, leading to lumbar hyper lordosis. He was hemodynamically stable, with normal cardiopulmonary auscultation. Airway assessment revealed Malampati, a few missing teeth, and Thyromental Distance (TMD) $>6 \text{ cm}$. Cervical mobility was reduced, with significant limitation of neck extension and flexion, but bilateral movement of the head preserved.

Chest radiography and laboratory tests were within normal parameters, except for Lactate Dehydrogenase (LDH) of 248 UI.L^{-1} (reference values between 120 and 240 UI.L^{-1}).

Cardiac evaluation comprised a 12-lead ECG, showing sinus rhythm with a heart rate of 86 bpm, and a transthoracic echocardiogram revealing preserved systolic Left Ventricular (LV) function, LV ejection fraction of 61% and grade II diastolic dysfunction of the LV. After consulting the cardiologist, we judged that placement of a perioperative pacemaker was not required. No preanesthetic medication was prescribed.

The anesthesia technique chosen was an epidural anesthesia that was promptly consented by the patient. In the operating room standard ASA monitoring was installed. Lateral decubitus positioning was challenging and painful for the patient. We identified the epidural space and inserted the epidural catheter in L2-L3 via a Touhy needle and median approach, on first attempt and without the need for administering anxiolytic medication. Following the test dose with 3 mL of 2% lidocaine, we injected 16 mL of 7.5 mg.mL^{-1} ropivacaine and 2 mL of 5 mcg.mL^{-1} sufentanil, in divided doses totaling 18 mL.

The sensory block level was T6, enough for umbilical hernia repair. The two-hour long surgery was uneventful. During the procedure, the patient remained hemodynamically stable with no vasopressor requirement. A total of 700 mL of 0.9% sodium chloride was administered. At the completion of surgery, 5 mL of 0.2% ropivacaine was injected via epidural catheter.

The patient stayed in the Postanesthetic Recovery Room (PACU) for 4 hours, without requiring additional epidural analgesia.

The morning following surgery, the patient resumed walking and was discharged from the hospital on the second postoperative day and was very satisfied with the anesthetic technique and postoperative analgesia.

Discussion

EDMD is a very rare disease characterized by a triad of symptoms consisting of contractures of the Achilles tendons, elbow, and cervical region. The disorder usually starts at childhood and progressively worsens to severe limitation of joint movement. Muscle atrophy and weakness slowly establish, initially at the humeroperoneal site, and as the disease progresses, present a diffuse distribution. Cardiac abnormalities comprise conduction and/or rhythm disorders (atrioventricular node) and dilated cardiomyopathy, which can lead to sudden death (occasionally the first symptom of the disorder), and ischemic accidents due to embolic phenomena.¹ Cardiac abnormalities, however, are rare before the second decade of life.

No specific treatment is available. Existing treatment options are based on orthopedic orthotics and physiotherapy to mitigate contractures and prevent severe muscle atrophy. As the disorder progresses, surgery may be necessary to correct contractures that significantly impair gait.

This case describes a patient with EDMD undergoing surgery under regional anesthesia to avoid several potential anesthetic problems.

Severe arrhythmias associated with conduction disturbances are the most common cardiac issue for patients with EDMD. The patient we reported did not present symptoms or test results compatible with cardiomyopathy or heart fail-

ure, thus we decided not to implant a cardiac pacemaker. However, due to the high risk of severe bradyarrhythmia's, a pacemaker (temporary or permanent) should be available perioperatively.¹ In patients with significant cardiomyopathy, myocardial depressant agents should be avoided, and the risk of acute heart failure can be reduced by using restrictive fluid regimen.¹

A multidisciplinary preanesthetic assessment is essential to determine the most appropriate anesthetic technique.

GA in a patient with muscular dystrophy presents some challenges, amongst which the increased risk of MH should be highlighted. MH can have a 70% mortality if not diagnosed and treated quickly, is associated with the use of inhalational anesthetics and depolarizing muscle relaxants, and thus administration of these drugs should be avoided.⁴ This risk could be reduced using intravenous GA. However, GA does not overcome another challenge presented by these patients, i.e., airway management due to contractures of the cervical muscles.

Thus, regional anesthesia provides greater patient safety by avoiding the two major difficulties, the increased risk of MH and the management of a Difficult Airway (DA). Regional techniques have additional advantages, namely providing excellent perioperative analgesia, more hemodynamic and ventilatory stability, preventing opioid side effects, and reducing hospital stay.¹

Choosing a regional anesthesia technique was especially advantageous for our patient that presented restrictive pulmonary disease due to sarcoidosis, as regional anesthesia is associated with lower likelihood of events related to airway management and invasive ventilation, such as hypoventilation, atelectasis, and hypoxia.

Considering the estimated surgery duration was more than 2 hours and the need to avoid sudden and unpredictable hemodynamic changes, we chose epidural anesthesia instead of spinal anesthesia. The epidural catheter also provides safe and effective analgesia and matches patient needs, which would not be delivered by spinal anesthesia.

Although positioning in the right lateral decubitus position with cranial displacement of the knees was difficult and painful for the patient, we chose not to previously administer opioids or anxiolytic drugs, such as fentanyl or benzodiazepines, due to the ventilatory depression risk and consequent management of potentially difficult airway. This condition was explained to the patient, who understood the need to remain immobile throughout the technique.

All anesthesia approaches are suitable; however, orotracheal intubation and neuraxial blockage may be difficult due to muscle contractures, which particularly affect the cervical and lumbar region. Consequently, the anesthesia technique must be customized and adjusted to each patient.

The lack of guidelines for anesthetic management of patients with EDMD, due to the scarcity of published data, significantly limited our intervention as anesthesiologists. However, this limitation inspired the entire medical team to conduct exhaustive research on the disorder and to establish an individualized and adequate anesthetic-surgical approach for our patient. In addition to the obvious enhancement of the medical literature, we hope that this case report will also assist other anesthesiologists to organize the key points in the management of patients with EDMD (Table 1).

Table 1 Main difficulties in anesthetic care of a patient with EDMD.

Airway management difficulties
Reduced cervical mobility and cervical contractures
Neuraxial management difficulties
Lumbar contractures
Osteoarticular abnormalities
Cardiovascular disorders
Bradyarrhythmias
Dilated cardiomyopathy
Consider external pacemaker in the preoperative period
Increased susceptibility to malignant hyperthermia
Avoid halogenated anesthetics and succinylcholine
Strict intraoperative surveillance

Conclusion

Due to the rarity and peculiarities associated with EDMD, there is no established superior anesthetic technique. The approach will always be guided by the type of surgery and clinical status of the patient, with the primary target being greater patient safety.

In our case report, epidural anesthesia helped bypass the two main challenges associated with the anesthetic procedure – the susceptibility to MH and the management of a potential difficult airway. The approach also enabled us to ensure stable perioperative cardiovascular and respiratory parameters, adjust the anesthetic duration according to surgery duration, and provide safe and effective postoperative analgesia.

Informed consent

The authors obtained informed consent from the patient for this case report publication.

Conflicts of interest

The authors declare no conflicts of interest.

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