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Anesthetic Management in Machado-Joseph Disease: A Case Report and Literature Review

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Abstract

Machado-Joseph disease (MJD), a type of spinocerebellar ataxia (SCA), is a rare genetic neurodegenerative disease with variable presentation. Symptoms may include ataxia, bulbar involvement, amyotrophy, extrapyramidal symptoms, dysautonomia, and peripheral neuropathy. As such, these patients often present with characteristics that convey a higher anesthetic risk and may be challenging to manage. As MJD is a rare condition, its manifestations and the specificities of the anesthetic approach to these patients may not be well known to most anesthesiologists. This article presents a 69-year-old female patient with a severe presentation of MJD who underwent a total hysterectomy with general anesthesia. A review of the available literature on the anesthetic management of this condition was also conducted. Anesthesiologists should be familiar with the possible complications of this type of SCA, the associated risks, and the existing evidence on how to safely manage this and other neurodegenerative conditions in the perioperative period.

Categories: Anesthesiology
Keywords: anesthesia neurodegenerative disease, anesthetic management, bulbar dysfunction, dysautonomia, machado-joseph disease, spinocerebellar ataxia

Introduction

Machado-Joseph disease (MJD), also known as spinocerebellar ataxia (SCA) type 3, is a rare autosomal-dominant progressive neurodegenerative disease. Its prevalence is estimated to be around one to five per 100,000 globally and higher in people of Portuguese descent, especially in the Azores archipelago. MJD is caused by abnormal cytosine-adenine-guanine (CAG) triplet repeat expansion in the ATXN3 gene, resulting in aberrant proteins that deposit in the neurons of the cerebellum and brainstem with progressive neurological damage and dysfunction [1]. Ataxia is therefore the cornerstone of MJD, frequently with bulbar symptoms, although other manifestations may be present in accordance with other areas involved: extrapyramidal symptoms (basal ganglia), amyotrophy (brainstem, spinal cord), peripheral neuropathy, and autonomic symptoms (bladder dysfunction, abnormal thermoregulation, hypohidrosis, orthostatic hypotension). Fatigue, cognitive impairment, and psychiatric symptoms like depression may also be present [2]. The presentation is heterogeneous, varying in severity with the number of CAG triplet repetitions [2,3].

With these symptoms, MJD patients may require special consideration in their anesthetic management, being understandably at a higher risk for both general anesthesia (GA) and regional anesthesia. However, due to its rare incidence, the literature regarding anesthesia management in MJD is very scarce: after extensive database review, we were able to find only one case of GA and three cases of regional neuraxial anesthesia published to date. Therefore, much of the reasoning used to manage anesthesia in these patients has traditionally been an extrapolation of recommendations from similar pathologies of the nervous system, such as Huntington's or Parkinson's disease.

Considerations regarding GA in MJD include the following: possible brainstem involvement, with increased risk of pulmonary aspiration secondary to dysphagia, vocal cord paralysis, delayed postoperative recovery of cough and of gag reflex (aspiration pneumonia being the most common cause of death in MJD) [4]; respiratory muscle weakness secondary to peripheral denervation limiting the use of respiratory function depressants, such as opiates or benzodiazepines, and neuromuscular blockers (NMB) [5]; increased risk of hemodynamic instability due to dysautonomia, requiring strict control of intravascular volume, blood pressure, and heart rate [6]. As such, distinct forms of presentation and severity of MJD may be associated with different degrees of risk, making the choice of anesthetic technique, airway and ventilation approach, as well as hemodynamic management, highly individualized.

With regard to regional anesthesia, its use in patients with central nervous system (CNS) disorders has been controversial [7]. The double crush phenomenon suggests that patients with preexisting neural compromise may be more susceptible to injury at another site when exposed to a secondary insult such as regional anesthesia techniques [8]. However, more recent studies challenge these past recommendations, reporting no disease progression after neuraxial anesthesia in various CNS pathologies [9]. As such, current data suggest that the decision to perform regional techniques in patients with preexisting CNS disorders should also be based on an individualized risk/benefit analysis.

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With this publication, we aim to provide new insight into the anesthetic management of patients with MJD by presenting the perioperative approach to a successful case of major gynecological surgery and summarizing the available evidence on the anesthetic and perioperative care of this rare disease, including the risks, benefits, limitations, and gaps in knowledge.

Case Presentation

A 69-year-old female patient presented for total hysterectomy due to an endometrial carcinoma. She had a diagnosis of MJD, established 10 years prior, that manifested at this time with severe ataxia, difficulty in locomotion (wide-based gait and decreased strength in the lower limbs, with frequent falls), bulbar compromise (dysarthria and dysphagia, with no ocular involvement), dysmetria, urinary incontinence, and respiratory weakness with the need for overnight continuous positive airway pressure. The patient had a positive family history for MJD (mother, maternal grandfather, and two maternal uncles), and confirmatory genetic testing had been performed.

Preoperative assessment focused on a Neurology consultation, which documented the symptoms present at the time, and a Pneumology evaluation with pulmonary function tests. Lung gas exchange was found to be normal on arterial blood gas, but other data were unobtainable due to poor patient collaboration. Despite this, the patient was considered fit for surgery due to the time-sensitive nature of the oncological procedure, and an intensive care unit (ICU) bed was procured for the postoperative recovery, foreseeing a need for delayed extubation. At the time, the patient weighed 52 kg, with a height of 161 cm (body mass index 20.1 kg/m^2) and was considered to have an American Society of Anesthesiologists III physical status. The preoperative laboratory results are shown in Table *1*; no alterations are expected with MJD, but a mild anemia and thrombocytosis, as well as slightly prolonged activated thromboplastin time, most likely reflect the malignancy process.

Parameter	Value	Reference range
Hemoglobin	11.4 g/dL	12.0-15.0 g/dL
Leukocytes	6.780 × 10^3/uL	4.000-10.000 × 10^3/uL
Platelets	475.000 × 10^3/uL	150-400 × 10^3/uL
Prothrombin time	11.3 seconds	12.0 seconds
Activated thromboplastin time	37 seconds	24-36 seconds
Fibrinogen	4.13 g/dL	1.5-4.5 g/dL
Creatinine	0.62 mg/dL	0.50-1.20 mg/dL
Sodium	143 mmol/L	136-145 mmol/L
Potassium	4.4 mmol/L	3-5-5.1 mmol/L
Total bilirubin	0.22 mg/dL	0.30-1.20 mg/dL
Arterial pH	7.45	7.35-7.45
Arterial partial pressure of oxygen	83.6 mmHg	81.0-108.0 mmHg
Arterial partial pressure of carbon dioxide	41.4 mmHg	32.0-48.0 mmHg
Arterial bicarbonate	30.24 mEq/L	21.0-27.0 mEq/L

TABLE 1: Preoperative laboratory results including complete blood count, coagulation, and renal and hepatic function tests

After multidisciplinary discussion between the anesthesiologist, the surgical and intensive care team, and the patient, the chosen anesthetic technique was GA, specifically total intravenous anesthesia (TIVA). No sedative premedication was administered to avoid negative impact on airway reflexes and ventilation. Induction of anesthesia was achieved with the administration of propofol in a target-controlled infusion (Schnider model, which was maintained during the procedure), fentanyl 100 mcg, and rocuronium 30 mg. Endotracheal intubation was achieved with videolaryngoscopy without difficulty, and mechanical ventilation (maintained on volume control) was unremarkable throughout the procedure, with peripheral oxygen saturation remaining at 100%. Hemodynamically, the patient presented some fluctuation, with

hypotension after induction of anesthesia controlled with titrated boluses of ephedrine (15 mg total) and fluid infusion. The heart rate remained stable throughout the procedure, and no severe manifestations of dysautonomia were found (blood pressure and heart rate data are presented in Figure *1*). Quantitative NMB monitoring was maintained throughout the procedure, revealing 0 or 1 response in train-of-four stimulation.

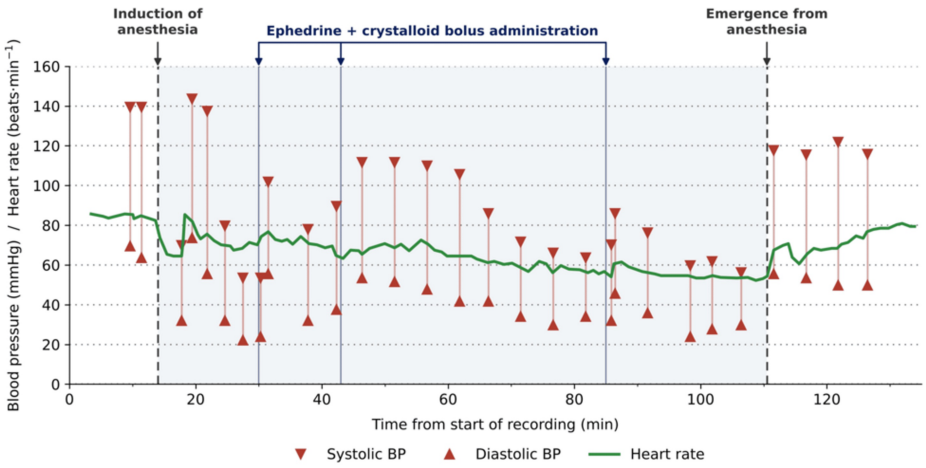


FIGURE 1: Blood pressure and heart rate values throughout the procedure

The green line represents continuous heart rate measurement (in beats per minute, on the vertical axis); the red arrows represent continuous noninvasive blood pressure monitoring (in mmHg, on the vertical axis). Downward arrows represent systolic blood pressure and upward arrows represent diastolic blood pressure

Key moments in the case are annotated in the figure

BP: blood pressure

The analgesic approach aimed to avoid further opioid administration (to reduce the risk of respiratory depression and impaired airway reflexes), so analgesic infusions of ketamine and dexmedetomidine were administered during surgery. Paracetamol 1 g and ketorolac 30 mg were also administered, and a bilateral rectus sheath block was performed with 60 mg of ropivacaine 0.2%. The surgery lasted approximately two hours, and at the end of the procedure, the patient was extubated after complete NMB reversal with sugammadex confirmed by quantitative monitoring (train-of-four ratio greater than 0.9), without adverse events. She was transferred to the ICU for observation in the immediate postoperative care, where she maintained spontaneous ventilation on room air and was transferred to the ward the following day. No exacerbation of neurological symptoms was identified, and she was discharged home after four days. Postoperative pain control was adequate. The patient maintained follow-up with her neurologist, showing no difference in MJD manifestations up to one year after the procedure.

Discussion

Previous case reports

We performed a comprehensive database search for all publications, of any time period, referencing MJD, SCA, and anesthesia management keywords. From this research, we were able to find only four different cases, from diverse institutions, of perioperative management in MJD.

One available case report [10] describes a 38-year-old female patient with a two-year history of MJD who underwent an elective vaginal hysterectomy under combined spinal and epidural anesthesia. Neurologic preanesthetic evaluation showed bulbar symptoms (dysphagia and dysarthria) as well as diplopia, with preserved gag reflex. Motor strength was diminished on the left side (4/5) when compared to the right. Nonuniform decreased sensation to touch and pain in dermatomes L3-S1 on the left side were also present. There was no evidence of autonomic involvement. A combined spinal-epidural was performed at the L2-L3 intervertebral space, administering 12 mg of hyperbaric bupivacaine, 100 mcg of morphine, and 10 mcg of fentanyl in the subarachnoid space. An epidural catheter was placed for block augmentation if needed during surgery but was not used and was removed after the surgery. Peak sensory level was T6, with blood pressure decreased to 90/60 mm Hg but controlled after titrated boluses of ephedrine and fluid challenge. After six hours, the patient returned to her neurological state, without worsening of symptoms at two days post-op follow-up.

Another case [11] describes a 65-year-old woman with known MJD, manifested as severe weakness in the arms and legs and polymyalgia, apparently without other alterations. Comorbidities included atrial fibrillation, an implanted pacemaker due to bradycardia-tachycardia syndrome, obesity, hypertension, and dyslipidemia. She underwent bimalleolar fracture repair under a subarachnoid block at the L3-L4 intervertebral space using 10 mg of hyperbaric bupivacaine. The peak sensory block level was T10, and no information is provided on hemodynamic changes. The patient was discharged from the hospital after two days without complications, and one month after spinal anesthesia, no new neurological changes had been identified.

A third case [12] reports on a 58-year-old man with known MJD who was proposed for inguinal hernia repair. The specific manifestations of MJD in this patient are not mentioned. This team opted for epidural anesthesia, and a catheter was placed at the L3-L4 intervertebral space. A bolus of 13 mL of epidural ropivacaine 0.75% was administered, and peak sensory block was identified at the T10 level. Procedural sedation was also performed with 2 mg of midazolam and 50 mcg of fentanyl intravenously. Recovery from motor block occurred three hours after epidural bolus without complications or abnormal alterations. Discharge from the hospital was given at 24 hours postoperative with the patient in his basal neurological state.

The only known case describing use of GA [6] follows a 48-year-old woman with breast cancer who presented for lumpectomy with axillary node dissection. She had a presumptive diagnosis of MJD with severe manifestations: motor weakness making her wheelchair bound and significant bulbar symptoms (poor cough and dysarthria, but without major dysphagia). Regional anesthesia techniques (considered by the team to be a safer option) were proposed to the patient, but she refused, opting for GA instead. As such, a TIVA was performed using propofol. No sedative premedication was used to avoid blunting airway reflexes, and no opioid was given throughout the case due to the risk of hypoventilation. A laryngeal mask was placed instead of an endotracheal tube, allowing the avoidance of neuromuscular blocking agents. Considering the risk of autonomic dysfunction, a preventive strategy was used, administering high-volume intravenous fluids (1,500 mL), frequent doses of phenylephrine, and one dose of glycopyrrolate. Emergence from GA and removal of the laryngeal mask in the operating room were uneventful. She was under observation in the postanesthesia care unit and was discharged home after 1.5 hours of recovery without complications. Table *2* summarizes the four previously published cases on the anesthetic management of MJD and the one reported in this publication.

Case	Patient	MJD manifestations	Surgical procedure	Anesthetic technique	Anesthetic drugs	Intraoperative events	Outcome
This study	69-year-old woman	Severe bulbar symptoms, decreased limb motor strength, and respiratory weakness	Total hysterectomy	General anesthesia (TIVA)	Intravenous propofol, fentanyl, rocuronium, ketamine, and dexmedetomidine. Perineural (fascial) ropivacaine	No significant hemodynamic changes, nor issues with airway or ventilation	No change in neurological symptoms 4 days postoperatively or at 1-year follow-up in Neurology
Teo et al. [10]	38-year-old woman	Bulbar symptoms, decreased motor strength, and touch sensation	Vaginal hysterectomy	Regional anesthesia (combined spinal and epidural)	Intrathecal bupivacaine, morphine, and fentanyl	Adequate block, no significant hemodynamic changes	No change in neurological symptoms 2 days postoperatively
Tavares [11]	65-year-old woman	Decreased motor strength (severe limb weakness) and polymyalgia	Bimalleolar fracture repair	Regional anesthesia (spinal)	Intrathecal bupivacaine	Adequate block, no mention of hemodynamic changes	No change in neurological symptoms 1 month post-operatively
Almeida and Teixeira [12]	58-year-old man	Not mentioned in the publication	Inguinal hernia repair	Regional anesthesia (epidural)	Epidural ropivacaine, intravenous midazolam, and fentanyl	Adequate block, no significant hemodynamic changes	No change in neurological symptoms 1 day postoperatively
Aldawoodi et al. [6]	48-year-old woman	Bulbar symptoms and severely decreased motor strength	Breast lumpectomy with axillary node dissection	General anesthesia (TIVA)	Intravenous propofol. No NMB drugs	No significant hemodynamic changes (preventive strategy), nor issues with airway or ventilation	No change in neurological symptoms 1.5 hours postoperatively

TABLE 2: Summary of available case reports on the anesthetic management of MJD

MJD: Machado-Joseph disease; TIVA: total intravenous anesthesia

Anesthetic management

MJD is a rare neurodegenerative disease, a type of SCA, with several neurological manifestations that may increase patient risk in the perioperative period. Currently, very limited literature is available on the anesthetic management of these patients; adding our case, there are now only five published reports, to the best of our knowledge: two of GA (both propofol-based TIVA) and three of neuraxial anesthesia (epidural and spinal). Notably, these published reports differ not only in the anesthetic technique and pharmacology used, but also in several aspects, from the degree of disease manifestation (severity of bulbar and respiratory impairment, for instance) to the timing of follow-up after the procedure. The size and heterogeneity of this dataset, where no complications following anesthesia were identified, limit its interpretation. Additionally, the reports differed in the information offered and in detail, providing further difficulty in analysis, and do not cover all possible approaches. We were not able to find published descriptions of GA maintenance with volatile anesthetics or of peripheral nerve blocks (besides our own, which was a fascial plane block with inherently smaller risk of neurological injury).

Regarding GA, there is very scant literature on the use of volatile agents not only in MJD but also in other types of SCA. Although not disease-specific, volatiles like sevoflurane and desflurane have been associated with cellular mechanisms behind neurodegenerative disease progression, contrary to propofol [13,14]. Despite the very limited, still incipient data, anesthesia providers may wish to be mindful of these proposed mechanisms in patients with MJD. Propofol-based TIVA offers other benefits as it is easily and rapidly titratable and may offer faster recovery; despite this, there is insufficient evidence as of today to claim one technique is superior to the other. As for NMB, it is not essential for all surgical procedures, but it facilitates airway management, improves surgical exposure, and optimizes mechanical ventilation. Despite this, its use in patients with neuromuscular disorders may increase the risk of residual blockade and postoperative complications, highlighting the importance of quantitative monitoring of NMB and its reversal. However, achieving adequate neuromuscular recovery using a steroid NMB and sugammadex may be more challenging and less predictable in patients with underlying neuromuscular disorders [15].

Regarding peripheral regional anesthesia, available evidence seems to indicate that it should not worsen

neurological symptoms in patients with SCA [16]. Furthermore, fascial plane blocks seem particularly advantageous in patients with neuromuscular disorders, as they avoid direct injection in close proximity to neural structures, minimizing the risk of injury [17]. With these limited data, it seems likely that regional blocks performed according to the leges artis could also be a safe option in MJD patients, especially when they allow avoidance of GA in high-risk patients. Of note, the patient that we describe first-hand in this publication had in her past medical history a recent orthopedic surgery of the lower limb, which was performed under spinal anesthesia with a popliteal sciatic nerve block performed for pain control. Unfortunately, this case was performed at another institution, and available records and information are scarce. However, we were able to ascertain that, in subsequent Neurology consultations at the same institution, the patient did not appear to have any change in MJD manifestations. Nevertheless, these are individual cases that do not allow for generalization, and more evidence is necessary to draw informed conclusions, not only regarding the efficacy and safety of regional anesthesia techniques but also regarding all anesthetic management approaches in the MJD population. Decisions regarding anesthetic and perioperative care should be highly individualized to the specific patient and preferably with multidisciplinary discussion.

Conclusions

Management of patients with rare disorders and increased perioperative risk can be challenging. Information and literature are often scarce, and anesthesia providers may have to rely on small case reports, like our own, or infer from similar, more common disorders. As such, sharing evidence on the care for these patients and making it available for other teams is essential to support our decisions and further improve care for patients. However, it is important to underline that this small sample of case reports is insufficient to draw conclusions that can be generalized for all MJD patients, and one instance of successful anesthetic approach does not signify that it will be acceptable and safe for every case; further data is essential to clarify the aspects discussed in this publication. Nonetheless, we hope to shine a light on MJD, raising awareness and striving for better care.

Additional Information

Author Contributions

All authors have reviewed the final version to be published and agreed to be accountable for all aspects of the work.

Concept and design: Beatriz Soares, Nuno Serrano, Pedro Pinto, Catarina C. Rodrigues

Acquisition, analysis, or interpretation of data: Beatriz Soares, Pedro Pinto

Drafting of the manuscript: Beatriz Soares, Pedro Pinto

Critical review of the manuscript for important intellectual content: Beatriz Soares, Nuno Serrano, Pedro Pinto, Catarina C. Rodrigues

Supervision: Nuno Serrano, Catarina C. Rodrigues

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