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CASE REPORT

ANESTHETIC CONSIDERATIONS IN DUODENAL GASTROINTESTINAL STROMAL TUMOR RESECTION IN A PATIENT WITH NEUROFIBROMATOSIS TYPE 1: A CASE REPORT

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Abstract

Introduction: Neurofibromatosis type 1 (NF1), also known as Von Recklinghausen disease, is a rare autosomal dominant neurocutaneous disorder. It is clinically characterized by multiple café au lait macules, intertriginous freckling, multiple cutaneous neurofibromas and learning disability or behavior problems. About half of the people with Neurofibromatosis type 1 (NF1) develop internal plexiform neurofibromas and may remain undiagnosed. Other organ systems can also be affected. Choosing the appropriate anesthesia for patients with neurofibromatosis is a unique challenge.

Case Presentation: This case report presents the perioperative management of a 54-years-old male diagnosed with the challenging condition of neurofibromatosis type 1 (NF1) who underwent surgery for resection of a duodenal gastrointestinal stromal tumor (GIST). To mitigate the risks associated with a possible difficult airway and neuraxial anesthesia, an awake video-laryngoscopy before induction was done, and a bilateral Transversus Abdominis Plane (TAP) block was performed for pain management. The operation proceeded uneventfully, recovery was without complications and with satisfactory postoperative pain control. The patient reported minimal discomfort and did not require additional analgesics during the recovery period. He was discharged home in good general condition on the fifth postoperative day.

Conclusion: Optimal anesthetic care in NF1 is inherently patient-specific, driven by systematic assessment of organ involvement and anticipation of perioperative complications.

***Key Words**: Anesthesia challenging, Neurofibromatosis type 1; Transversus Abdominis Plane (TAP) block; video-laryngoscopy.*

Introduction

Neurofibromatosis type 1 (NF1), also known as Von Recklinghausen disease, is a rare autosomal dominant neurocutaneous disorder. It is characterized by multiple café au lait macules, intertriginous freckling, multiple cutaneous neurofibromas and learning disability or behavior problems. About half of the people with NF1 have plexiform neurofibromas, most of which are internal and may not be clinically suspected. Plexiform neurofibromas can cause pain, neurologic deficits and abnormalities of involved or adjacent structures. Less common but potentially more serious manifestations include optic nerve and other central nervous system gliomas, malignant peripheral nerve sheath tumors, osteoporosis, scoliosis, tibial dysplasia, vasculopathy, and gastrointestinal, endocrine, or pulmonary disease (1).
The incidence of NF1 is approximately 1 in 2,600 to 3,000 live births. There is no predilection for the male or female gender (2).
The mutation or deletion of the NF-1 gene results in the phenotypic and genotypic manifestations of the disorder. The NF-1 gene encodes a protein called neurofibromin, which is expressed in various tissues (3). Neurofibromin functions as a GTPase-activating protein that inhibits the rat sarcoma (RAS) signaling pathway (4). Mutations in the NF-1 gene result in a lack of neurofibromin expression, thereby promoting tumorigenesis. Neurofibromas develop when both alleles of the NF-1 gene are mutated. Neurofibromas embody Schwann cells, perineural cells, mast cells and fibroblasts. Cutaneous neurofibromas (cNF) involve dermal nerve terminals, whereas plexiform neurofibromas (pNF) characteristically affect the nerve plexuses and fascicles.
The diagnosis of NF1 is established in a proband with two or more of the characteristic clinical features or one characteristic clinical feature and a heterozygous NF-1 pathogenic variant.
There is no definite therapy for this genetic disorder, so the treatment for NF1 is primarily symptomatic, whereas, for plexiform neurofibromas, surgical removal is the only treatment option but may be associated with damage to involved nerves or adjacent tissues. Complete surgical excision, when possible, of malignant peripheral nerve sheath tumors is the treatment of choice; chemotherapy may be beneficial in some individuals.

Case Presentation

A 54-years-old male patient, 185cm tall and weighing 107kg (BMI: 31.2kg/m^2), with neurofibromatosis type 1 (NF1), was admitted to the University Clinic for Abdominal Surgery in Skopje for an elective resection of a duodenal gastrointestinal stromal tumor (GIST). This tumor was diagnosed with a computed tomography scan and by a gastroscopic biopsy. The patient had

a significant surgical history related to Neurofibromatosis type 1 (NF1), including a right crural amputation twenty years ago due to neurofibroma infiltration and resection of a jejunal gastrointestinal stromal tumor (GIST) sixteen years ago. He also had surgery for a pancreatic cyst and chronic pancreatitis, requiring lifelong pancreatic enzyme replacement therapy. Comorbidities included well-controlled epilepsy managed with antiepileptic medication and osteoporosis treated regularly. Preoperative assessments showed normal cardiovascular and respiratory findings. Airway examination revealed adequate mouth opening, Mallampati class II, normal neck mobility and a few small oral mucosal neurofibromas. Laboratory investigations, coagulation profile and chest radiograph were within normal limits.

The planned anesthetic technique involved general endotracheal anesthesia (GETA) in combination with a bilateral transversus abdominis plane (TAP) block for perioperative pain management. The associated risks and procedures were thoroughly discussed, and written informed consent was obtained.

On the day of surgery, the patient continued taking his regular medications and received antibiotic prophylaxis. At the operating room, standard ASA monitoring was established, which included ECG, non-invasive blood pressure (NIBP), and oxygen saturation (SpO_2). The baseline parameters recorded were the following: blood pressure of 160/80mmHg, heart rate of 80 beats per minute, and SpO_2 at 98%.

After preoxygenation with 100% oxygen via a tight-fitting non-rebreathing facial mask with a fresh gas flow of 20L/min for 3 minutes and administering premedication with 2mg of midazolam and 50μg of fentanyl, video-laryngoscopy was performed, revealing several small neurofibromas around the epiglottis, none of which obstructed the airway. Anesthesia induction was then achieved with the following medications: propofol (2mg/kg/BW), fentanyl (1mcg/kg/BW), lidocaine (0.6mg/kg/BW), and rocuronium (0.6mg/k/BW). Tracheal intubation was successfully performed using a video-laryngoscope.

An ultrasound-guided bilateral transversus abdominis plane (TAP) block was performed in the midaxillary line between the costal margin and the iliac crest. After confirming negative aspiration, a total of 20mL of 1% lidocaine and 20mL of 0.25% bupivacaine was injected.

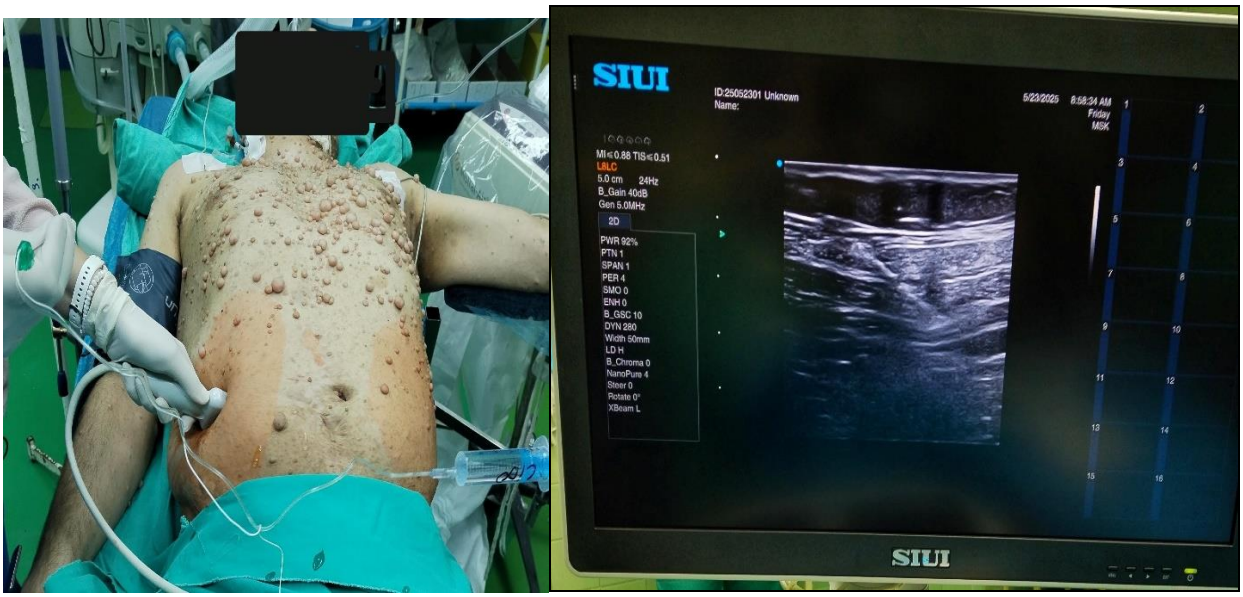

Figure 1. Ultrasound-guided TAP block performed in the patient with neurofibromatosis type 1: (A) The anesthesiologist performing the block with the ultrasound probe.
(b) Ultrasound view showing the needle advancing through the fascial plane between the internal oblique and transversus abdominis muscle.

Additional analgesia included 100µg of fentanyl, 1g of paracetamol and 1.5g of magnesium sulfate. Anesthesia was maintained with sevoflurane at 1.0 MAC. During the surgery, the patient remained hemodynamically stable. The estimated blood loss was minimal. The total duration of the surgery was 180 minutes. At the end of the procedure, after reversing the residual neuromuscular blockade, the patient was extubated uneventfully in the operating room. Postoperatively, the patient received antibiotics, anticoagulation and multimodal analgesia with paracetamol and metamizole. His regular antiepileptic therapy was resumed immediately. The postoperative course was without any complications, and the patient was discharged home on the fifth postoperative day in good general condition.

Discussion

Patients with neurofibromatosis type 1 (NF1) pose unique challenges for anesthesiologists due to the multisystem involvement of the disease. Airway, neurological, cardiovascular and skeletal manifestations must all be carefully considered when planning anesthetic management. Intraoral manifestations occur in approximately 5% of the patients with Neurofibromatosis Type 1 (NF1) (5). Sharma and Fisher have described cases where discrete neurofibromas were found on the tongue or larynx, with the aryepiglottic folds and arytenoids being the most commonly affected areas. This is likely due to the dense presence of terminal nerve plexuses in these regions (6,7). Involvement of these structures may lead to airway obstruction, with symptoms such as shortness of breath, stridor, difficulty swallowing or changes in voice serving as critical

clinical indicators of a potentially difficult airway. In our patient, video-laryngoscopy revealed small neurofibromas around the epiglottis that did not obstruct the airway. Nevertheless, the possibility of sudden airway compromise underscores the importance of preparedness for a difficult airway scenario.

The patient's prior abdominal surgeries - jejunal GIST resection and pancreatic cystectomy for chronic pancreatitis - further complicated anesthetic planning by increasing the likelihood of adhesions and unpredictable intra-abdominal dynamics. Chronic pancreatitis might also impair absorption and alter the pharmacokinetics of fat-soluble drugs, while nutritional deficiencies could influence anesthetic drug dosing. Consequently, meticulous hemodynamic monitoring and individualized fluid management were essential.

Neurofibromatosis type 1 (NF1) is also associated with central nervous system tumors, vasculopathies and seizures (8). Epilepsy, often treated with antiepileptic drugs (AEDs), may alter anesthetic metabolism through hepatic enzyme induction. Ensuring perioperative continuation of AEDs is critical to avoid breakthrough seizures. Although our patient did not exhibit any active neurological symptoms, continuous vigilance remained paramount. From a cardiovascular perspective, patients with Neurofibromatosis type 1 (NF1) have a higher risk of developing secondary hypertension, most commonly due to pheochromocytoma or renal artery stenosis (9). Although our patient was normotensive and did not exhibit catecholamine-related symptoms preoperatively, the literature emphasizes maintaining a high index of suspicion, as an unrecognized pathology may precipitate catastrophic intraoperative hemodynamic instability.

Regarding analgesia, the use of neuraxial techniques in patients with Neurofibromatosis Type 1 (NF1) is not contraindicated if spinal imaging (CT or MRI) excludes the presence of neurofibromas along the neuraxis. In our case, because spinal imaging had not been performed, neuraxial anesthesia was avoided (10). Instead, we employed a multimodal analgesic strategy that included a bilateral ultrasound-guided transversus abdominis plane (TAP) block. This approach is well-documented in the literature and has been integrated into Enhanced Recovery After Surgery (ERAS) protocols for both open and laparoscopic abdominal surgery (11, 12). Ultrasound guidance ensured precise administration, minimizing the risk of infiltration into neurofibroma-infiltrated tissues, and effectively reduced opioid consumption while providing stable analgesia.

Conclusion

NF1 is presented with a wide range of clinical manifestations, requiring personalized anesthetic planning driven by a systematic evaluation of organ involvement and the anticipation of potential perioperative complications. A comprehensive preoperative assessment is crucial for managing difficult airways, ensuring safe intubation with video-laryngoscopy. A balanced general anesthesia approach, combined with an ultrasound-guided transversus abdominis plane (TAP) block, provided adequate intraoperative and postoperative analgesia, minimized opioid requirements, and maintained hemodynamic stability.

References:

1. Hirsch N.P., Murphy A., Radcliffe J.J. Neurofibromatosis: clinical presentations and anaesthetic implications. BJA Br J Anaesth. 2015;43:107–110.
2. Le C, Thomas A, Lui F. Neurofibromatosis. [Updated 2025 May 4]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2025 Jan.
3. Ferner RE, Gutmann DH. Neurofibromatosis type 1 (NF1): diagnosis and management. Handb Clin Neurol. 2013; 115:939-55.
4. Jouhilahti EM, Peltonen S, Heape AM, Peltonen J. The pathoetiology of neurofibromatosis 1. Am J Pathol. 2011 May;178(5):1932–1939.
5. Baden, E., Pierce, H. E., & Jackson, W. F. (1955). Multiple neurofibromatosis with oral lesions; review of the literature and report of a case. Oral Surgery, 8, 263.
6. Sharma, SC · Srinivasan, S Isolated plexiform neurofibroma of tongue and oropharnyx: A rare manifestation of von Recklinghausen's disease J Otolaryngol. 1998; 27:81-84.
7. McD Fisher, M Anaesthetic difficulties in neurofibromatosis Anaesthesia. 1975; 30:648-650.
8. Hirsch NP, Murphy A, Radcliffe JJ. Neurofibromatosis: clinical presentations and anaesthetic implications. Br J Anaesth. 2001 Apr;86(4):555–564.
9. Huson, SM Recent developments in the diagnosis and management of neurofibromatosis Arch Dis Child. 1989; 64:745-749.
10. Dounas M, Mercier FJ, Lhuissier C, Benhamou D. Epidural analgesia for labour in a parturient with neurofibromatosis. Can J Anaesth. 1995 May;42((5 Pt 1)):420–422.
11. Peltrini R, Cantoni V, Green R, Greco PA, Calabria M, Bucci L, et al. Efficacy of transversus abdominis plane (TAP) block in colorectal surgery: a systematic review and meta-analysis. Techniques in Coloproctology. 2020;24(8):787–802.
12. Hain E, Maggiori L, Prost À la Denise J, Panis Y. Transversus abdominis plane (TAP) block in laparoscopic colorectal surgery improves postoperative pain management: a meta-analysis. Colorectal Disease. 2018;20(4):279–287.