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CASE REPORTS

Case report

Anaesthetic implications in Von Recklinghausen disease: A case report☆

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ABSTRACT

Von Recklinghausen disease or neurofibromatosis Type I (NF1) is an autosomal dominant disease with a wide spectrum of clinical manifestations. Neurofibromas are the characteristic lesions. This disorder is associated with important anaesthetic considerations, mainly when neurofibromas occur in the oropharynx and larynx, leading to difficult laryngoscopy and tracheal intubation. We describe the anaesthetic management of a patient with NF1 under general anaesthesia for facial neurofibroma excision. We performed a brief review of the literature with the aim of optimizing the anaesthetic management and reducing the number of complications associated with the systemic manifestations of this syndrome.

Implicaciones anestésicas en la enfermedad de Von Recklinghausen

Palabras clave:
Neurofibroma
Neurofibromatosis
Anestesia
Manejo de la vía Aérea
Manchas Café Con Leche

RESUMEN

La enfermedad de Von Recklinghausen (EVR) o neurofibromatosis tipo I (NF1) es una enfermedad con herencia autosómica dominante con un amplio espectro de manifestaciones clínicas. Los neurofibromas son las lesiones características. Este trastorno se asocia con importantes consideraciones anestésicas, principalmente cuando los neurofibromas aparecen en la orofaringe y laringe, produciendo dificultades en la laringoscopia y en la intubación endotraqueal. Describimos el manejo anestésico de un paciente con NF1 bajo anestesia general para extirpación de neurofibromas faciales. Hemos realizado un breve repaso de la literatura existente para optimizar el manejo anestésico y reducir el número de complicaciones asociadas con las manifestaciones sistémicas de este síndrome.

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Introduction

Von Recklinghausen disease (VR) or neurofibromatosis type I (NF1) is an autosomal dominant disorder characterized by the propensity to form ectodermal and mesodermal tissue tumours,[1] affecting primarily the nervous system and the skin.[2] Friedrich Recklinghausen identified the origin of the tumours in the nervous tissue in 1882.[1,3] The pathophysiology is characterized by a mutation of the NF1 gene located on chromosome 17q11.2, responsible for secreting neurofibromin, a protein that inhibits abnormal cell growth.[1,2] The clinical spectrum of this disorder is quite broad, characterized mainly by skin neurofibromata and café-au-lait spots.[2]

Clinical case

We present a case of a 14-year-old patient with a personal history of VR and a surgical history of excision of a right hemicranial plexiform neurofibroma. No fibromas of the oral cavity or predictors of a difficult airway were found during airway exploration. The patient did not report dyspnoea, dysphagia or changes in voice tone that could suggest the presence of laryngeal fibromas. Additional tests included biochemistry, whole blood count, coagulation tests and a chest radiograph, all of which came back normal.

The patient presented important facial asymmetry as a result of multiple retroauricular neurofibromas that disfigured the face and made it impossible for him to use reading glasses. It was decided to excise the neurofibromas and attempt facial remodelling.

After setting up the usual non-invasive monitoring using blood pressure, electrocardiogram, pulse oximetry (SpO_2) and neuromuscular blockade with TOF Watch® SX monitoring, a peripheral venous access was established on the back of the left hand. The anaesthetic induction was done using propofol 130 mg, fentanyl 120 μg, and rocuronium 30 mg. There was no airway obstruction after induction during manual ventilation with a facial mask. Endotracheal intubation proceeded uneventfully and mechanical ventilation was instituted. Laryngoscopy did not reveal gross laryngeal fibromas. Sevoflurane at 1 MAC and fentanyl according to analgesic needs were used during anaesthetic maintenance. The procedure lasted 160 min (Fig. 1) and proceeded uneventfully.

At the end of the procedure, the patient received metamizol 2 g, ondansentron 4 mg, atropine 0.6 mg and neostigmine 2 mg, and was extubated upon reaching a train-of-four value of 0.9. The patient was then taken to the post-anaesthetic care unit and had a favourable course.

Discussion

Neurofibromatosis (NF) is a congenital disease of the group of autosomal dominant neurocutaneous phakomatoses including also the tuberous sclerosis complex, the Hippel–Lindau syndrome and the basal-cell nevi syndrome.[4] It is possible to distinguish two types of neurofibromatosis on the basis of the phenotypical and genetic characteristics: NF1 or VR and neurofibromatosis type 2 (NF2).

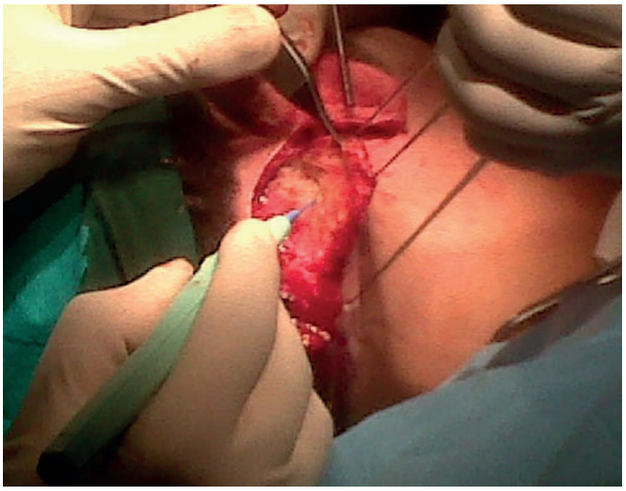

Fig. 1 – Resection of multiple retroauricular neurofibromas. Source: authors.

The incidence of VR is 1 in every 2500–3300 births[5] and the prevalence is 1 in every 5000 inhabitants.[5] Although it has a 100% penetrance,[5] expression varies[5,6] with 50% of the patients having no family history,[5,7] which implies a spontaneous mutation.[6]

Despite significant advances in molecular genetics[8,9] the diagnosis of VR is made when a series of clinical criteria are met (Table 1).[5]

Café-au-lait spots are found in 95% of adults with VR.[5] Neurofibromas are the most characteristic lesion[10] representative of this disorder and they can be divided into three types depending on their clinical and histopathological characteristics[5]: cutaneous (found in 95% of patients), nodular and plexiform neurofibromas. Plexiform neurofibromas are found in 30% of cases, causing severe body deformities.[5] They may become malignant in 2–16% of cases[9] and they are the primary cause of morbidity and mortality.[5]

Lisch nodules are present in 95% of cases. They may be associated with bone abnormalities, pheochromocytoma,[11,12] gut tumours,[13] carcinoid tumours, spinal or cerebral,[14]

Table 1 – Diagnostic criteria for Von Recklinghausen disease (VR) or neurofibromatosis Type I (NF1).

Diagnostic criteria for VR (NF1). It must include the following manifestations:
6 or more café-au-lait spots: 1.5 cm or larger in post-pubescent age, or 0.5 or larger in pre-pubescent age.
Two or more neurofibromas of any type, or 1 or more plexiform neurofibromas.
Axillary (Cowe's signs) and/or inguinal freckling.
Optic nerve glioma.
Two or more Lisch nodules (iris hamartomas).
Distinctive bone lesion: sphenoidal dysplasia, or dysplasia or cortical thinning of the long bones.
First-degree relative with NF1.

Source: authors.

vertebral deformities, juvenile chronic myelogenous leukaemia,[5] and growth and mental retardation.[1,15]

Neurofibromatosis Type 2 is diagnosed on the basis of a series of clinical criteria, defined by the presence of bilateral vestibular schwannomas leading to hearing loss,[5] cataracts, and central nervous system involvement, such as meningioma.

VR poses a challenge to anaesthetists, including a potentially difficult airway, abnormalities of the spinal anatomy and peripheral neurofibromas;[16] hence the need for a careful systemic assessment before selecting the anaesthetic technique.[17]

Classically, general anaesthesia has been considered safer since the presence of intracranial neuromas or the existence of unknown spinal neuromas (up to 40%[16] of cases) may worsen the neurological picture when locoregional anaesthetic techniques are used,[17,18] with devastating consequences such as haematomas and paralysis.[16] Gliomas, meningiomas, hydrocephalus, spinal tumours and spina bifida have been described in VR, discouraging the use of locoregional anaesthesia when these findings are present.[19]

Macroglossia, abnormal formations in the tongue, pharynx, larynx[2] and even supraglottic plexiform fibromas[5] may prevent endotracheal intubation[2,19] and determine upper airway obstruction during anaesthetic induction. These lesions must be suspected after a detailed history and patient report of dysphagia, dysarthria, stridor and voice changes.[5] Facial malformations may result in facial asymmetry due to intraosseous involvement[6] and contribute to difficult ventilation when face mask and orotracheal intubation are used.

Consequently, as anaesthetists we are required to perform a thorough assessment to identify difficult airway predictors, as well as an adequate interview designed to detect intraoral lesions.[2] If a difficult airway is expected, awake fibre optic bronchoscopy intubation must be considered as the technique of choice.[2]

Multisystem involvement in VR requires special attention to other potential intra-operative findings such as hypertension, which could be related with an unknown pheochromocytoma (up to 20%[19] of patients with VR) or renal artery stenosis.

Other causes of pheochromocytoma that need to be ruled out include von Hippel-Lindau syndrome, multiple endocrine neoplasia Type 2B (MEN 2B) and paraganglioma syndromes.[20] The finding of a pheochromocytoma mandates individualized pre-operative management in order to avoid life-threatening intra-operative hypertensive crises. Pre-operative blood pressure control is based on alpha receptor blockade using prazosin or phenoxybenzamine to replenish plasma volume and counteract the vasopressor effects of high catecholamine levels, followed by beta blockade.

Other considerations include respiratory compromise with intrapulmonary fibromas and pulmonary fibrosis, and cardiovascular compromise with hypertrophic cardiomyopathy or mediastinal tumours compressing the superior vena cava, beside hypertension.

The presence of scoliosis compromises cardiopulmonary function, leading to right ventricular failure.[5] Other anaesthetic considerations include epilepsy, carcinoid tumours and obstructive ureteral stenosis due to neurofibromas.[5]

Exceptionally, some cases have been described of altered sensitivity to neuromuscular blockers,[1] giving rise to prolonged episodes of apnea of unexplained mechanism.[19,21]

In summary, we reviewed the existing literature in order to avoid deleterious effects from our clinical anaesthesia practice because of the multi-organ involvement in a disease that may give rise to multiple perioperative adverse events.

Patient perspective

The patient perceived the anaesthetic management as the most beneficial given the surgical intervention and the associated anaesthetic risks.

Informed consent

The informed consent was obtained.

Information disclosure

Patient information has remained confidential.

Ethics committee

The study was approved by the ethics committee.

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Conflict of interest

There is no conflict of interest.

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