Neurofibromatosis and Debulking Surgery: A Case Report

Shveta Goyal[1], Kunal Khera[1], Rajvir Kaur[2], Seema Jindal[3]

ABSTRACT

Introduction: Neurofibromatosis type 1 is an autosomal dominant disorder characterised by propensity to form ectodermal and mesodermal tissue tumours. It exhibits a variety of symptoms, ranging from simple skin neurofibromas to devastating plexiform neurofibromas that cause skin disfiguration, nerve compression and airway obstruction.

Case report: A 25 yr old male, diagnosed case of neurofibromatosis type1, presented with complaint of enlarging lower limb with hanging masses for last 9-10 years. Patient was known case of hypertension for past one year and was on treatment with tab. amlodipine 5 mg OD. Patient was posted for debulking surgery of neurofibroma under general anaesthesia. General physical examination showed multiple giant café au lait macule on trunk, gross enlargement of both lower limbs, lisch nodules on iris. Patient was induced with inj. propofol, inj. fentanyl and cisatracurium and intubated with 7.5mm ID endotracheal tube under c-MAC guidance. Patient was extubated uneventfully.

Discussion: Anaesthetic considerations include presence of intracranial or spinal neuromas, neurofibromas of tongue, larynx, cervical spine involvement, hypertension, phaeochromocytoma, hypertrophic cardiomyopathy. Thus, a careful systemic assessment is required before selecting the anaesthetic technique.

Conclusion: Anaesthesiologist needs to perform a thorough preoperative evaluation of every patient and prepare a comprehensive anaesthetic plan for any complications that may occur. Close cooperation between the surgeon, the anesthesiologist and the cardiologist is required especially for patients with undiagnosed hypertension.

KEYWORDS: Neurofibromatosis Type 1, Plexiform Neurofibromas, General Anaesthesia, Airway Obstruction, Hypertension, Phaeochromocytoma.

INTRODUCTION

Neurofibromatosis (NF) is an inherited autosomal dominant disorder and is classified as type 1 and type 2. It is caused by a mutation of chromosome 17q11.2 (NF1 gene) in type 1 and chromosome 22q12.1 in type 2. Neurofibromatosis type 1 (NF 1) or Von Recklinghausen is characterised by propensity to form ectodermal and mesodermal tissue tumours affecting primarily the nervous system and the skin.[1]. The incidence of NF1 lies between 1 in every 2500–3300 births and its prevalence is 1 in every 5000 inhabitants. Although, it has 100% penetrance, expression varies with 50% of the patients having no family history.[2] NF1 gene encodes for protein neurofibromin, which appears to have a tumour suppressive role. Mutations at the NF1 gene result in diminished levels of neurofibromin with resultant development of the wide variety of tumours seen in the disease.[3] NF 1 frequently involves the head and neck region and exhibits a variety of symptoms, ranging from simple skin neurofibromas to devastating plexiform neurofibromas (PNs) that cause skin disfiguration, blindness, nerve compression and airway obstruction. It may also involve the cardiovascular, respiratory, genitourinary and gastrointestinal system. It is associated with variety of conditions that often require surgical treatment including painful neurofibromas, severe kyphoscoliosis, pseudoarthroses, hydrocephalus, intracranial tumours and other malignancies.[2,4] Therefore it becomes important for an anaesthetist to pay attention to proper evaluation of such patients.

CASE REPORT

A 25 yr old male, weighing 80 kg, diagnosed case of NF 1, was planned to be taken up for elective debulking surgery. The patient presented with chief complaint of abnormal growth of lower limbs (Right >> left) with hanging masses for last 9-10 years (Figure 1). Patient had history of dyspnoea (NYHA II) and was a known case of hypertension for past one year and was on regular treatment with tab. amlodipine 5 mg OD. There were no symptoms of peripheral nerve involvement. Patient had positive family history with cutaneous lesions seen in both parents. Patient underwent deformity correction of right foot under general anaesthesia (GA) 13 yrs back and the recovery was uneventful. On general physical examination, multiple giant café au lait macules (CALM) were present on trunk, about 6-7 in number (Figure 2). Small neurofibromas were present on medial side of elbow. Right inguinal lymph nodes were enlarged with bag of worms feeling. There was gross enlargement of both lower limbs with skin thrown in folds with hyperpigmentation (Figure 1). There was Lisch nodules seen on slit lamp examination (Figure 3). Airway examination was within normal limits with no fibromas of the oral cavity found during airway exploration. All routine investigations were within normal limits including chest xray and ECG. X-ray of long bones showed decreased bone density (Figure 4). GA was planned for the procedure. The involving risks and benefits were

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Figure-1: Abnormal growth of lower limbs with hanging masses

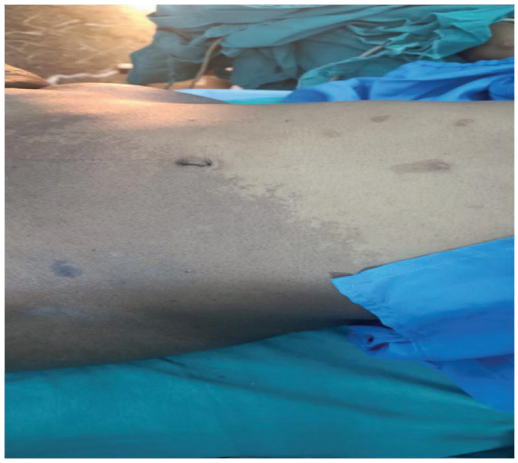

Figure-2: Café au lait macules on trunk

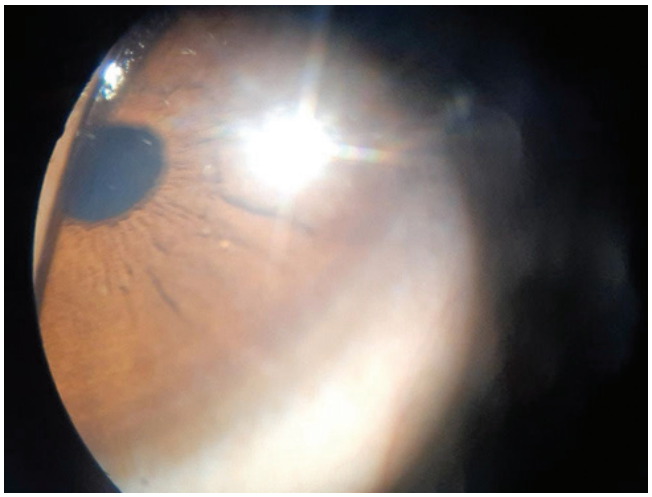

Figure-3: Lisch nodules on slit lamp examination

explained to the patient and written informed consent was obtained. Patient's nil per oral status was confirmed. Upon arrival at the operation room, standard ASA monitors were connected and peripheral venous access was secured with two wide bore cannulas on upper limb. Pre-operative vitals were within normal limits. Premedication was given with inj. glycopyrrolate 0.005 mg/kg i.v., inj. dexamethasone 0.1 mg/kg i.v. Patient was preoxygenated with 100% oxygen for 5 minutes. Induction was done with inj. propofol 2.5 mg/kg i.v. till loss of responsivenes and inj. fentanyl 2 mcg/kg i.v. Patient was given loading dose of inj. cis-atracurium 0.15 mg /kg iv after confirming adequacy of bag and mask ventilation. Patient was intubated orotracheally with cuffed endotracheal tube (ETT) 7.5 mm ID under c-MAC guidance (video laryngoscope) and ET tube secured after confirming bilateral equal air entry. Anaesthesia was maintained with maintenance doses of inj. cis-atracurium 0.03 mg/kg i.v. and 50:50 mixture of N_2O & O_2 + isoflurane @ 0.2-1%. Intraoperatively, there was an episode of hypotension because of massive blood loss (around 1-1.2 litres) which was managed by rushing 1.5 litres of i.v. crystalloids, 500 ml of colloid and a unit of packed red blood cells after cross match. The whole procedure lasted for 2 hours and 6 minutes. At the end, patient was successfully reversed with inj. neostigmine 0.05 mg/kg i.v. and inj. glycopyrrolate 0.01 mg/kg i.v. and was extubated upon achieving good muscle tone.

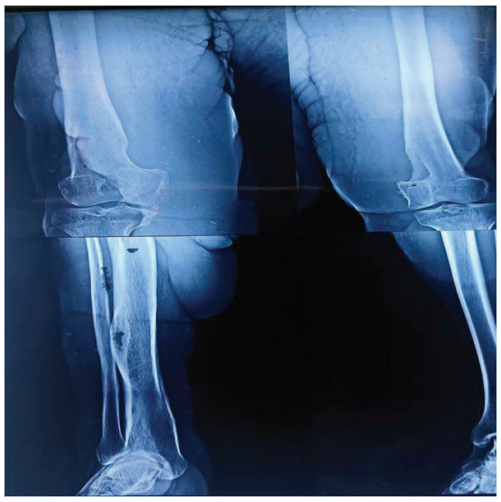

Figure-4: X-ray of long bones showing decreased bone density

DISCUSSION

Neurofibromatosis type 1 is an autosomal dominant disorder caused by heterozygous mutation in NF 1 gene located on chromosome 17q11.2. About 95% of NF1 patients have café-au-lait spots. Neurofibromas are the major feature of NF1 and fall into three distinct types: Cutaneous neurofibromas (found in more than 95% of patients), *nodular* neurofibromas (arise in peripheral nerves) and paraspinal neurofibromas (may grow to give rise to the classic thoracic 'dumbbell' tumour). *Plexiform* neurofibromas are the hallmark lesion of NF1, are usually congenital and affect long portions of the nerve; infiltration of the nerve and the surrounding tissue may occur, giving rise to extensive disfiguration. About 95% of individuals with NF1 will develop Lisch nodules. There may be associated bony abnormalities, pheochromocytoma, carcinoid tumours, vertebral deformities.[2] As the neurofibromatoses affect both ectodermal and mesodermal tissue, all systems of the body may be involved that may give rise to multiple perioperative adverse events. Thus, a careful systemic assessment is required before selecting the anaesthetic technique. General anaesthesia has been

considered safer as there can be presence of intracranial neuromas or unknown spinal neuromas which may worsen the neurological picture when regional anaesthetic techniques are used with devastating consequences such as haematomas and paralysis. Gliomas, meningiomas, hydrocephalus, spinal tumours if present also discourage the use of locoregional anaesthesia.[5] NF1 patients may have airway involvement as neurofibromas of tongue, pharynx or larynx that can cause upper airway obstruction and thus, difficulty in endotracheal intubation. Symptoms of dyspnoea, stridor, loss or change of voice or dysphagia if present should warn the anaesthetist of potential airway problems and specialist examination with indirect laryngoscopy and CT or MR imaging should be done. The presence of macroglossia, macrocephaly, specific mandibular abnormalities and cervical spine involvement may also contribute to difficulties of airway management. In a patient with multiple cervical neurofibroma, painless dislocation of cervical vertebra has been reported, thus radiographic examination of the neck should be performed to avoid spinal cord damage during laryngoscopy and tracheal intubation. Facial malformations due to intraosseous involvement may be present and can contribute to difficult ventilation when face mask and orotracheal intubation are used. Thus, a thorough assessment should be done to identify difficult airway predictors. If a difficult airway is expected, intubation under video laryngoscopic guidance should be considered as the technique of choice.[1,6,7] There can be respiratory compromise with neurofibromatosis affecting the conducting airways, lung parenchyma, the thoracic cage and the chest wall. Mediastinal neurofibromas can cause tracheal and bronchial compression. In some cases, there is bilateral upper lobe pulmonary fibrosis, which can result in restrictive defect, pulmonary hypertension and right ventricular failure. Scoliosis if present can produce a reduction in lung volume, which if severe, may result in respiratory failure. Hence, evaluation of the respiratory system is an essential part of the pre-operative management of patients with NF1 and may include CT evaluation of the thorax and detailed lung function testing.[2,8] Hypertension is the most commonly occurring cardiovascular manifestation of NF. Hypertension presenting in the young NF1 sufferer is usually because of renal artery stenosis, which may be bilateral.[9] Regular arterial pressure measurement is thus mandatory in the pre-operative assessment of these patients. Phaeochromocytoma affects between 0.1–5.7% of patients with NF1. Sustained or paradoxical hypertension that is resistant to treatment should raise the suspicion of phaeochromocytoma and appropriate investigations such as urinary catecholamine estimation and abdominal CT scanning should be performed to exclude phaeochromocytoma as anaesthesia, surgery and pregnancy are particularly hazardous if it is undiagnosed pre-operatively. A careful questioning regarding cardiovascular disease with appropriate investigations is important in the assessment of the patient with neurofibromatosis.[10] Some cases have altered sensitivity to neuromuscular blockers giving rise to prolonged episodes of apnea of unexplained mechanism. So, in patients with neurofibromatosis, neuromuscular monitoring should be done when neuromuscular blocking drugs are used, especially in patients with renal impairment or those on concurrent medication (e.g. anticonvulsant drugs), which may interfere with the normal pharmacokinetics or pharmacodynamics of neuromuscular blocking drugs. Other anaesthetic considerations include hypertrophic cardiomyopathy, epilepsy, carcinoid tumours, gastrointestinal haemorrhage and obstructive ureteral stenosis due to neurofibromas. Hence, we suggest that NF1 patients should undergo thorough investigation prior to elective surgery due to diverse clinical manifestations of the disease that may give rise to multiple perioperative adverse events.

CONCLUSION

Anaesthesiologist needs to perform a thorough preoperative evaluation of every patient and prepare a well-formed and comprehensive anaesthetic plan for any complications that may occur. Airway obstruction after induction of anaesthesia has been reported therefore plan of emergency tracheostomy should always be kept. Close cooperation between the surgeon, the anesthesiologist, and the cardiologist is required especially for patients with undiagnosed hypertension.

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