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ANESTHETIC CONSIDERATIONS IN A PATIENT WITH VON RECKLINGHAUSEN'S DISEASE: A CASE REPORT

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ABSTRACT Neurofibromatosis type-1 (NF-1) is a rare and complex multisystem genetic disorder, inherited in an autosomal dominant fashion with penetrance close to 100%, involving chromosome 17, with incidence being one in 3000-4000 people. The different features include development of multiple benign tumours called neurofibromas all over the body, café-au-lait spots, axillary or inguinal freckling, eye lesions like Lisch nodules in iris, optic nerve glioma, osseous lesions like sphenoid bone dysplasia, pseudoarthrosis, brain tumours like gliomas etc. The patients can also have neurofibromas in the airway, hypertension secondary to pheochromocytoma or renal artery stenosis, chest wall deformities like kyphosis and scoliosis etc. Hereby, we report a case of a 73 years old female patient diagnosed with Von Recklinghausen's disease, having a large plexiform neurofibroma over abdomen, multiple neurofibromas all over the body, café-au-lait spots and a positive family history. The patient was scheduled for excision of the plexiform neurofibroma under general anesthesia.

KEYWORDS : Neurofibromatosis, café-au-lait spots, pheochromocytoma, endotracheal intubation.

INTRODUCTION :

Neurofibromatosis type-1 (also called as Von Recklinghausen's disease) is an autosomal dominant condition, due to mutation in NF-1 gene on chromosome 17, with incidence being one in 3000-4000 people.[1] It is characterized by multiple neurofibromas, plexiform neurofibroma(30%), café-au-lait spots(95%), axillary or inguinal freckling(70%), optic nerve glioma, Lisch nodules in iris(95%), osseous lesions etc. The patients can also have neurofibromas in the airway, secondary hypertension(6%), chest wall deformities etc.[2] Hereby we present a case of Von Recklinghausen's disease having large, irregular plexiform neurofibroma over abdomen scheduled for excision of the same under general anesthesia.

CASE REPORT:

A 73 years old female patient, weighing 40 kg, known case of neurofibromatosis type-1 developed plexiform neurofibroma on abdomen over three years. She also had multiple neurofibromas all over the body. Numerous café-au-lait spots of different sizes were present over the trunk and limbs. She also showed skeletal involvement in the form of scoliosis. There was a positive history of hypertension since one year for which she took medications on and off. Her daughter also had similar neurofibromas all over the body. She denied history of diminished vision. (Figure 1)

Fig.1A: Multiple neurofibromas over face
Fig.1B: Skeletal involvement- scoliosis

PRE-OPERATIVE EVALUATION:

On physical examination, she was 130 cm tall. Baseline heart rate was 90 bpm, blood pressure was 138/90 mmHg and SpO2 was 98% on room air.

No neurofibromas were seen in the oral cavity and oropharynx as seen on indirect laryngoscopy and airway assessment was normal. Spine examination showed scoliosis at lower thoracic level. Her haemoglobin was 11.6 gm%, platelet count was 2.19 lakhs/mm3, PT/INR was 18.5/1.19, her liver function test, renal function test and serum electrolytes were within normal limits. Her electrocardiogram showed normal sinus rhythm. Chest x-ray was also normal.

The patient was kept fasted for eight hours pre-operatively. One night before surgery tab. alprazolam 0.5 mg was given. In the morning chlorhexidine mouth wash was done and then two hours before the surgery capsule omeprazole 20 mg was given with a sip of water. Fully informed written consent was taken. After applying all the standard monitoring (heart rate = 84 bpm, blood pressure = 142/92 mmHg, SpO2 = 99%), patient was premedicated with inj. midazolam 1 mg, inj. glycopyrrolate 0.2 mg, inj. fentanyl 80 mcg and simultaneously preoxygenated with 100% oxygen. Then patient was induced with inj. propofol 80 mg (with prior administration of inj. lignocaine 60 mg IV) and inj. atracurium 20 mg, and then intubated with seven mm cuffed endotracheal tube under videolaryngoscopic vision with minimal neck extension. Ringer's lactate was the fluid of choice intra-operatively. Anesthesia was maintained with inj. atracurium (4+4 mg) and a mixture of air + O2 + sevoflurane.

Intra-operatively, her blood pressure shot up to 209/145 mmHg. Inj. dexmedetomidine 40 mcg in 100 ml NS was infused over 10 min. But blood pressure still remained high at 167/113 mmHg, when Inj. nitroglycerin 25 mg in 50 ml NS infusion was started @ 2 ml/hr and continued throughout the surgery. A heart rate in the range of 75-85 bpm and a blood pressure of 130-150/100-120 mmHg was maintained throughout the procedure. (Figure 2)

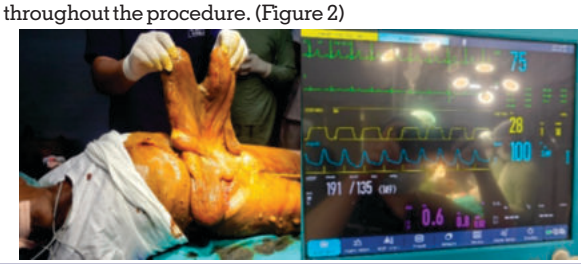

Fig. 2A: Plexiform neurofibroma over abdomen
Fig. 2B: Intra-operative hemodynamic changes

After completion of surgery, when spontaneous respiration was regained, neuromuscular blockade was reversed with inj. neostigmine 2 mg and inj. glycopyrrolate 0.2 mg I/V. Patient was smoothly extubated once adequate muscle power was achieved. Post-extubation vitals were- heart rate= 73 bpm, blood pressure= 145/109 mmHg, SpO2= 100% on room air. The patient was then shifted to post-anesthesia care unit.

DISCUSSION :

Von Recklinghausen's disease is an autosomal dominant disorder and the most common neurocutaneous syndrome, characterized by mutation in NF1 gene on chromosome 17q 11.2, causing decreased production of neurofibromin protein. This protein normally has a role in tumour suppression and therefore its reduced production causes development of multiple tumours (neurofibromas) all over the body.[2]

There are two types of neurofibromatosis. The diagnostic criteria is as following[2] :

<table>
<tr><td>Diagnostic criteria of NF-1 (Given by National Institute of Health)</td><td>Diagnostic criteria of NF-2 (Manchester criteria)</td></tr>
<tr><td>Presence of two or more of the following features :</td><td>The following features confirm NF-2 :</td></tr>
<tr><td>1.Six or more café-au-lait spots 5 mm or larger in prepubertal individuals 15 mm or larger in post-pubertal individuals</td><td>1.Bilateral vestibular schwannomas or</td></tr>
<tr><td>2. Two or more neurofibromas of any type or one or more plexiform neurofibroma</td><td rowspan="7">2.Family history of NF-2 (first degree relative) + unilateral vestibular schwannoma in a patient less than 30 years old or any two of the following meningioma, glioma, schwannoma, juvenile posterior subcapsular cataracts juvenile cataracts</td></tr>
<tr><td>3. Axillary or inguinal freckling</td></tr>
<tr><td>4. Optic glioma</td></tr>
<tr><td>5.Two or more Lisch nodules</td></tr>
<tr><td>6.Distinctive bony lesions like sphenoid dysplasia or dysplasia/ thinning of long bone cortex</td></tr>
<tr><td>7.A first degree relative with NF-1</td></tr>
</table>

Von Recklinghausen's disease is characterized by multiple benign neurofibromas dispersed all over the body including oral cavity, larynx, arytenoids, aryepiglottic folds (areas rich in terminal nerve plexuses) which may pose a difficulty in endotracheal intubation. Cervical neurofibromas can be present causing painless cervical dislocations. Our patient did not have such lesions in the oral cavity and larynx. A difficult airway has to be diagnosed in pre-operative assessment by an anesthesiologist or otolaryngologist and an indirect laryngoscopic examination is highly recommended to avoid any complication while intubating the patient. Awake fibreoptic intubation can be done for those with neurofibromas in the oral cavity.[2,3]

We preferred to give general anesthesia to this patient for better control of haemodynamics especially blood pressure and our patient's airway examination did not reveal any anticipated difficulty. Spinal deformities like kyphosis, scoliosis may be associated with NF-1 which can become a challenge if central neuraxial blockade is chosen as a technique of anesthesia. If the deformities are severe, spine x-rays or computed tomography scans can be obtained pre-operatively.[2]

These patients can have cardiovascular involvement in the form of hypertension which might be secondary to pheochromocytoma or renal artery stenosis. Our patient had a blood pressure of 138/90 mmHg pre-operatively which could be normal for an elderly patient. Also her electrocardiogram was normal which made the possibility of the secondary causes of hypertension less likely. But such patients should be assessed for possible causes of secondary hypertension like pheochromocytoma and renal artery stenosis. As hypertension is the most consistent clinical sign of pheochromocytoma, it should be ruled out by absence of symptoms like sweating, palpitations, headache; negative plasma and urine analysis for fractionated metanephrines and a negative abdominal magnetic resonance imaging to avoid intra-operative hypertensive crisis. Intra-operative blood pressure in our patient shot up to 209/145 mmHg which we managed with nitroglycerin infusion. It could be because of an undiagnosed pheochromocytoma, but we could not follow it up with urine analysis for fractionated metanephrines. Hypertension could also be because of renal artery stenosis and the same has to be ruled out by renal artery doppler imaging especially in young patients.[4,5]

Optic nerve gliomas can also be an association which are low grade tumours, presenting with decreased visual acuity and visual field defects. Hence if the patient complains of any symptom related to vision, a detailed ophthalmological examination with magnetic resonance imaging to rule out optic nerve glioma should be done.[6]

These patients may have increased sensitivity to neuromuscular blockers and therefore non-depolarizing muscle relaxants were used in low doses. We used inj. atracurium (20+4+4 mg) in this patient. Neuro-muscular monitoring can be used intra-operatively to assess the exact requirement of muscle relaxants. Such patients might also have residual neuro-muscular blockade and require mechanical ventilation in post-anesthesia care unit (PACU).[7]

CONCLUSION :

Hereby we report a case of successful management of excision of large plexiform neurofibroma over the abdomen under general anesthesia in a patient of Von Recklinghausen's disease. As this disease can have multisystem involvement like airway, cardiovascular system, skeletal system, eyes, respiratory system etc, a detailed evaluation is highly recommended to anticipate respective anesthetic challenges so that proper optimization can be done pre-operatively and necessary measures can be taken to reduce peri-operative morbidity and mortality.

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