Anesthetic Considerations in Neurofibromatosis Type 1

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Introduction:
Neurofibromatosis is an autosomal dominant disorder that involves ectodermal and mesodermal tumor formation. Neurofibromatosis Type 1 (NF1, Von Recklinghausen disease) is caused by a heterozygous mutation in the NF1 gene that leads to uninhibited cell division. NF1 has a wide clinical presentation and can affect every physiological system. Most commonly it presents with numerous neurofibromas, café-au-lait spots, optic nerve glioma, and Lisch nodules. Anesthetic considerations for NF1 originate from the condition’s broad clinical spectrum and possible association with other neuroendocrine tumors.

Case:
A 29 year old presented electively to Hackensack University Medical Center for spine surgery. Upon reviewing his chart, it was noted that he had a past medical history of Neurofibromatosis Type I diagnosed ten years earlier, but the patient had been lost to follow-up. What are the anesthetic considerations for this patient intraoperatively and postoperatively? Additionally, what workup should the patient have done prior to the operation?

Discussion:
The birth incidence of NF1 is 1 in 2500–3300 with a prevalence of 1 in 5000 individuals. The life expectancy of these individuals is shortened by about 15 years and the most common causes of death are CNS tumors and transformation of neurofibromas into malignant peripheral nerve sheath tumors (MPNSTs). NF1 has 100% penetrance with a wide spectrum of clinical presentations. In addition, NF1 is a disorder of neuroendocrine origin and may coexist with other neuroendocrine tumors such as pheochromocytomas and carcinoid tumors. The varied distribution of neurofibromas and other tumors throughout the body can lead to obstacles with regards to anesthetic management. There are no current standard perioperative guidelines for neurofibromatosis patients, however there are certain pre-operative considerations to keep in mind to ensure optimization.

Pre-operative work up:
â€¢ Thorough evaluation of airways. About 5% of NF patients present with manifestations of disease in the oral cavity. Neurofibromas in the tongue, larynx, and trachea can lead to upper airway obstruction and cause difficulty with intubation. Facial asymmetry and spine abnormalities (such as severe scoliosis) may cause further difficulty with ventilation and intubation. A detailed history with additional indirect laryngoscopy may be necessary to assess for any respiratory
obstruction. Further studies such as CT/MRI along with pulmonary function tests may also be indicated. ENT consultation may be necessary for further evaluation. If a difficult airway is expected, plan for an awake fiber optic bronchoscopy intubation and possible tracheostomy, depending on the level of airway compromise.

- Assess for hypertension. The most common cardiovascular concern in NF1 is hypertension; this can be idiopathic or secondary to renal artery stenosis, pheochromocytoma (found in up to 20% of NF1 patients) or carcinoid tumor. Pre-operatively it is important to screen blood pressure and probe for history indicative of pheochromocytoma. This is significant because intraoperative use of beta blockers and ketamine may lead to hypertensive crisis and ultimately death in patients with unknown pheochromocytoma.

- Assess for mediastinal mass due to the risk of severe intraoperative hypotension from SVC obstruction. If suspected, preoperative chest imaging should be completed in order to assess for the extent of obstruction.

- Possible endocrine work up to evaluate for other neuroendocrine tumors such as pheochromocytoma and carcinoid tumors. Gastrointestinal stromal tumors (GISTs) have also been found to coexist in NF patients with pheochromocytoma.

- Neurological exam. NF patients should undergo annual neurological assessments due to their high incidence of intracranial tumors. New neurological signs or symptoms should be followed up by referral to a neurologist and imaging.

- Assess for skeletal defects. NF patients have reduced bone density and are prone to scoliosis, osteoporosis, pseudarthrosis, and bony lesions. NF children should have annual spine assessments and, if appropriate, referral to an orthopedic surgeon. Severe scoliosis can reduce lung volumes, therefore those patients should undergo regular pulmonary function tests and assessed for sleep apnea. Skeletal assessment is also significant for patient positioning, as NF patients are more prone to have fractures and have demonstrated delayed bone healing. Clinical findings suggestive of defects should be followed up with radiographs.

Intraoperative:

- Patients with hypertension should be monitored for fluctuations in blood pressure or arrhythmia. Careful control of arterial pressure is also indicated due to possible vasculopathy-associated stenoses and/or aneurysms. Nephrotoxic drugs should be used with caution in patients with suspected renal artery stenosis.

- Patients with mediastinal masses are prone to intraoperative hypotension and may be less responsive to fluid resuscitation.

- Patients with CNS tumors that involve brainstem structures can have central hypoventilation syndromes and protracted weaning from mechanical ventilation. There is also a higher incidence of epilepsy during brain tumor resection

- Patients with neurofibromatosis-1 were thought to have varied responses to neuromuscular blocking agents, but research has disproven this.
Removal of benign plexiform neurofibromas is frequently very difficult due to their vascular nature and can result in life threatening hemorrhage.

Post operative:

Post brain tumor resection neurological exam is indicated to assess for any deficits.

Conclusion:

Neurofibromatosis Type 1 is a common genetic disorder and can be difficult to manage due to the wide variety of clinical presentations. Therefore it is important for anesthesiologists to maintain a working knowledge of NF1, its associated perioperative risks, and possible multisystem complications. A complete pre-operative assessment including a thorough history with appropriate investigations is essential for creating an anesthetic plan and ensuring proper perioperative management of NF1 patients.

References:


http://orthoinfo.aaos.org/topic.cfm?topic=A00050