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Anaesthesia recommendations for patients suffering from

Neurofibromatosis Type 2

Disease name: Neurofibromatosis Type 2

ICD 10: Q85.02

Synonyms: NF2

Neurofibromatosis Type 2 (NF2) is an autosomal dominant disorder characterized by central nervous system (CNS) tumors. A mutated allele of the *NF2* gene on chromosome 22 accounts for this disorder. Although characterized as autosomal dominant, greater than 50% of cases are new, sporadic mutations. The incidence of NF2 in the general population is 1 in 33,000-40,000, while the prevalence is 1 in 57,000. The average age of onset of symptoms is typically in the early twenties, with the diagnosis being made on average of seven years from onset. The tumors, usually schwannomas, are classically located at the superior vestibular branch of cranial nerve VIII bilaterally. However, up to 40% may occur in the inferior vestibule. Symptoms of vestibular schwannomas include hearing loss, imbalance, and tinnitus, while late symptoms include headache, mastoid ache, facial twitching, facial numbness, and raised intracranial pressure (ICP). Tumors are often found throughout the CNS, including the brain and spinal cord, although peripheral nerves may also be involved. The types of tumors involved include schwannomas, meningiomas, ependymomas, and neurofibromas. Schwannomas are benign tumors composed of Schwann cells located outside the nerve, and less than 1% become malignant. Unilateral deafness, with or without tinnitus, is the most common presenting symptom. Other presenting symptoms include tinnitus, muscle weakness or wasting, seizure, vertigo, numbness and tingling, and blindness. Approximately 70% of patients will have cutaneous lesions as well.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Disease summary

NF2 is genetically and phenotypically different than Neurofibromatosis Type 1(NF1). NF1, or von Recklinghausen's disease, is an autosomal dominant disorder caused by mutations in the NF1 gene on chromosome 17, and is characterized by café-au-lait spots and freckles, which increase with age, as well as benign tumors and Lisch nodules, which are iris hamartomas. It is much more common that NF2, having a prevalence of 1 in 2,500-3,500 worldwide. The benign tumors of NF1 include peripheral nerofibromas and optic gliomas. Peripheral neurofibromas are tumors of nerve sheaths, and are composed mostly of Schwann cells, along with fibroblasts, perineurial cells, axons, and mast cells.

Typical surgery

Vestibular schwannoma, meningioma, and spinal tumor resections; cochlear implants; stereotactic radiosurgery (e.g. gamma-knife), which may or may not require general anaesthesia.

Type of anaesthesia

There is no definite recommendation for general or regional anaesthesia, however the presence and location of tumors should be considered when determining the anaesthetic plan.

If CNS tumors are suspected by radiographic exam, or suspected clinically, neuraxial anaesthesia may be contraindicated. Potential consequences include a patchy block due to the presence of spinal cord tumors, raised intracranial pressure via mass effect of fluid in the epidural space, bleeding and epidural hematoma formation due to the possibility of tumors being highly vascular, and the risk of spreading mutated cells by incidental puncture.

Peripheral regional anaesthesia is not contraindicated. However, the anaesthesiologist should be aware of the location of known or suspected tumors, and any associated neurological deficit. If peripheral or neuraxial blocks are planned in an area of a known tumor, careful pre-procedural neurological exam should be done and documented to assess for changes post-procedure. A thorough discussion of the risks with documentation in the medical record is warranted.

If a general anaesthetic is planned, one should avoid drugs that can raise ICP, such as ketamine and vasodilating drugs, especially in patients when elevated ICP is known. If CNS tumors are known or suspected and the patient requires an endotracheal tube for the surgery or procedure, adequate blunting of laryngeal reflexes with intravenous lidocaine and opioids should be ensured so that coughing and bucking, which raises ICP, can be avoided. However, use caution with opioids in the spontaneously breathing patient as hypoventilation can result in high levels of CO_2 and increased ICP.

Necessary additional diagnostic procedures (preoperative)

If a patient is suspected of having NF2, genetic testing can confirm the diagnosis. If neuraxial anaesthesia is being considered, radiographic imaging may be useful in determining if the anaesthetic plan is reasonable. Computed tomography (CT) and magnetic

resonance imaging (MRI) can be used to locate CNS tumors, although MRI is the gold standard.

Particular preparation for airway management

Nerve sheath tumors can be located in laryngeal, cervical, and mediastinal regions in NF2. Such tumors can make endotracheal intubation difficult. If one is aware of or suspects such lesions, one should be prepared for a difficult airway. Thus, preoperative evaluation should include inquiring about hoarseness and dysphagia.

Particular preparation for transfusion or administration of blood products

Not reported.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transport or mobilisation

Positioning of a patient with a CNS tumor or mass effect lesion that will increase ICP, such as trendelenburg, should be avoided, if possible. Care must be taken to assess for spinal cord compressing cervical tumors prior to surgery.

Probable interaction between anaesthetic agents and patient's long term medication

Many patients are on antiepileptic drugs, and interactions with planned anaesthetic agents should be checked before administering.

Anaesthesiologic procedure

If a difficult airway is anticipated due to the presence of laryngeal, cervical, or mediastinal tumors, short acting neuromuscular blocking agents, such as succinylcholine, should be considered. As fasciculations associated with the use of succinylcholine can increase ICP, a defasciculating dose of a non-depolarizing neuromuscular blocking agent can be used to negate this.

During induction of a general aneasthetic, drugs that increase ICP, such as ketamine, should be avoided. Safer alternatives include propofol, etomidate, barbiturates, benzodiazepines, and opioids. Also, if an endotracheal tube will be used, adequate blunting of laryngeal reflexes with intravenous lidocaine, LTA, and intravenous opioids should be accomplished.

During the termination of anaesthesia, a deep extubation can be considered in a patient without a laryngeal or mediastinal tumor to prevent coughing and bucking on the endotracheal tube, and the administration of lidociane prior to an awake extubation may be beneficial as well.

Particular or additional monitoring

Not reported.

Possible complications

If one suspects laryngeal, cervical, or mediastinal tumors, a difficult airway may be encountered. Hence, the difficult airway cart should be readily available.

If CNS tumors are present, increased ICP and brain herniation are possible.

Postoperative care

If neuraxial anaesthesia is administered, serial surveillance for epidural hematoma should be carried out, including neurological assessment of motor and sensory function, until the patient returns to baseline neurologic function.

If peripheral regional anaesthesia is used in a region of known or suspected tumor, a neurological workup to document preexisting pathology should be performed. If clinically indicated, radiological studies may be useful in documenting sites of peripheral tumors. After completion of the procedure, a postprocedural neurological exam should be completed and documented.

Information about emergency-like situations / Differential diagnostics

caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the disease

Disease triggered emergency-like situations can include epidural hematoma in neuraxial anaesthetics. In the presence of brain tumors, a postictal state should always be considered in the differential for delayed emergence.

Ambulatory anaesthesia

Not reported.

Neuraxial anaesthesia for laboring patients and patients in need of cesarean section involves potential risks of bleeding and epidural hematoma formation, patchy block, raised ICP, and puncture of Schwannomas. We recommend that in high risk patients, an MRI prior to labor may be prudent to assess for lesions, which tend to enlarge during pregnancy. Pushing during the second stage of labor also can increase ICP. Due to these risks, patients with known or suspected CNS lesions may undergo general anaesthesia to avoid the potential complications described above. In general, we would still recommend neuraxial over general anaesthesia unless imaging showed severe disease, especially considering airway risks as noted above.

Literature and internet links

1. Evans, D G R, et al. Neurofibromatosis type 2. J Med Genet 2000; 37: 897-904
2. Evans, D G, et al. A genetic study of type 2 neurofibromatosis in the United Kingdon. I. Prevalence, mutation rate, fitness, and confirmation of maternal transmission effect on severity. J Med Genet 1992; 29: 841-846
3. Evans, D G R, et al. A Clinical Study of Type 2 Neurofibromatosis. QJM 1992; 84 (1): 603-618
4. Evans DG, et al. Birth incidence and prevalence of tumor prone syndromes: estimates from a UK genetic family register service. Am J Med Genet 2010; 152A (2): 327-332
5. Moffat, David, et al. Management strategies in neurofibromatosis type 2. European Archives of Oto-Rhino-Laryngology 2003; 260 (1): 12-18
6. Armbardekar AP, et al. The value of ultrasound in the safe care of a patient with neurofibromatosis. Anesthesiology 2013; 118 (5): 1206
7. Mautner, V F, et al. Spinal tumors in patients with neurofibromatosis type 2: MR imaging study of frequency, multiplicity, and variety. American Journal of Roentgenology 1995; 165 (4): 951-955
8. Phi, Ji Hoon , et al. Radiosurgical treatment of vestibular schwannomas in patients with neurofibromatosis type 2. Cancer 2009; 115 (2): 390-398
9. Cihangiroglu, Mutlu, et al. Laryngeal Neurofibroma Associated with Neurofibromatosis Type 2. AJNR 2002; 23: 1637-1639
10. Gong, S, et al. Neurofibromatosis type 2. Journal of Clinical Otorhinolaryngology 2006; 20 (16): 721-723
11. Sakai, T, et al. A parturient with neurofibromatosis type 2: anesthetic and obstetric considerations for delivery. International Journal of Obstetric Anesthesia 2005; 14: 332-335
12. Upadhyaya, Meena, et al. Neurofibromatosis Type 1. Methods of Molecular Medicine – Molecular Diagnosis of Genetic Diseases 2004; 92: 285-310
13. McLaughlin, Margaret E. and Tyler Jacks. Neurofibromatosis Type 1. Methods of Molecular Biology – Tumor Suppressor Genes 2003; 222: 223-237.

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