Neurofibromatosis

Genetic disorder in which multiple organs, including skin and CNS, are site of tumors and hamartomas; hallmark is café-au-lait spots (more than 6 that are > 1.5 cm in diameter) and multiple neurofibromas; laryngeal and tracheal compression may occur secondary to tumors and there is an association with pheochromocytoma; compression of peripheral nerves and intracranial / -spinal mass lesions should also be considered.

ANESTHETIC CONSIDERATIONS:

- Complicated multi-system disease
- Potential **difficult airway** with laryngeal obstruction, and **C-spine abnormalities**
  - Difficulty positioning due to spine abnormalities
- Respiratory compromise with **hypoxemia**
  - Restrictive lung disease (kyphoscoliosis, interstitial lung disease)
- Cardiovascular
  - **HTN** due to **pheochromocytoma** or renal artery stenosis
  - Dysrhythmias
- ↑ **ICP** (tumors, hydrocephalus), **seizures**, spinal cord and peripheral nerve compression
- Abnormal response (prolonged or resistance) to NMBDs

ANESTHETIC GOALS:

- Anticipate difficult airway – assess for pharyngeal / laryngeal tumors & consider AFOI / tracheotomy
- Anticipate possible pheochromocytoma
- Anticipate and evaluate for increased ICP
- Careful positioning

HISTORY

- Airway - airway obstruction, laryngeal stenosis / compression, pharyngeal compression
- Respiratory – exercise tolerance, hypoxemia, restrictive lung disease, kyphoscoliosis, pulmonary fibrosis, interstitial lung disease
- CVS - HTN (renal artery dysplasia, pheochromocytoma), dysrhythmias, RV outflow obstruction, cor pulmonale
- Neurological - intracranial tumors (neurofibromas), pituitary involvement, ↑ ICP, hydrocephalus, seizures, spinal cord and peripheral nerve compression, glaucoma, mental retardation, headache
- Endo - pituitary involvement, thyroid tumors, pheochromocytoma
- GU - urinary obstruction and uremia
- MSK - kyphoscoliosis, pathologic fractures, macrocephaly, craniofacial / vertebral dysplasias

PHYSICAL

- **HEENT** - hoarseness, dysphonia, stridor, difficulty of intubation
- **RESP** - dyspnea, exercise tolerance, cyanosis, hypoxemia, clubbing, kyphoscoliosis
- **CVS** - HTN, headache, perspiration, JVP, pedal edema
- **CNS** - focal nerve deficits

INVESTIGATIONS

- **Labs**
  - CBC
  - BUN, Cr, Urine Catecholamines
  - ABGs
- **Imaging**
  - C-spine, CT Neck, indirect laryngoscopy
  - CXR, PFTs
  - ECG, (ECHO)
  - CT head, Spine films

OPTIMIZATION

- Evaluation of airway for possible laryngeal and pharyngeal tumors

ANESTHETIC OPTIONS

- Common surgical procedures include nerve decompression, pheochromocytoma excision, repair of renal artery stenosis, obstructive uropathy, pathologic fractures or unrelated surgeries
- Local, regional, general
  - Dependent on systems involved and degree of compromise
- Asymptomatic intraspinal neurofibromas can make identification and entry into epidural and subarachnoid spaces very difficult
  - A careful examination of the back is indicated before any regional technique is considered

ANESTHETIC SETUP

- **Drugs**
  - Routine
- **Equipment**
  - Standard CAS
  - Consider a. line depending on respiratory status and presence of pheochromocytoma

MANAGEMENT OF ANESTHESIA

- **Induction**

- If emergency case, abbreviated work-up and maintain high index of suspicion for laryngeal obstruction, pheochromocytoma, RV outflow obstruction, hydrocephalus
- Consider awake FOI or tracheotomy if laryngeal and pharyngeal involvement
- No particular drugs or technique recommended – maintain goals
- Consider potential for spinal cord neurofibromas if regional considered
- Consider potential for ↑ ICP during induction
- Some have shown prolonged response or resistance to SCh and pancuronium (recent evidence suggests no or minimal effect)
- Positioning problems

- **Maintenance**
  - CV instability if pheochromocytoma present
  - Several reported cases of prolonged NMB in response to administration of SCh, pancuronium, d-tubocurarine
- **Emergence**
  - Routine

DISPOSITION & MONITORING

- Pain management critical

COMPLICATIONS

- Presence of pheochromocytoma
- Potential for increased ICP if expanding intracranial tumor

PATHOPHYSIOLOGY

- Genetic disorder; autosomal dominant (although 50% represent new mutations)
- Tumors and hamartomas develop in multiple organs
- Hallmark is café-au-lait spots (> 6 that are > 1.5 cm in diameter) and neurofibromas
- Two forms are best delineated:
  - NF-1
    - 1 in 3000; autosomal dominant neurocutaneous syndrome; mapped to chromosome 17
    - Multiple diagnostic criteria of which 2 or more must be present
    - Café-au-lait spots and neurofibromas common findings
    - Manifestations range in severity and progress with time
    - Usually identified under 6 y of age
    - 50% have neurologic manifestations, also pheochromocytoma, meningioma, glioma, renal artery dysplasia with HTN and hypoglycemia, and 10% have diffuse interstitial pulmonary fibrosis
  - NF-2
    - 1 in 50,000; mapped to chromosome 22
    - Bilateral acoustic neuromas, meningiomas, schwannomas of the dorsal roots of the spinal cord, and gliomas
    - Café-au-lait spots and neurofibromas much less common

REFERENCES

- Roizen & Fleisher – Essence of Anesthesia Practice – p 241
- Stoeltings anesthesia and coexisiting disease 5th ed