Saethre-Chotzen Syndrome

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Risk

- Incidence: 1:25,000–50,000 live births.

Perioperative Risks

- Difficult airway.
- Seizures.
- Oculocardiac reflex.
- The severity of cardiac lesion may affect the hemodynamic stability of the pt during anesthesia.

Overview

- A type of acro-cephalo-syndactyly syndrome, characterized by premature fusion of the coronal sutures, facial dysmorphism, syndactyly, skeletal deformity, and congenital heart malformations.
- Named after two physicians who independently reported it in the early 1930s—Haakon Saethre, a Norwegian psychiatrist, and F. Chotzen, a German psychiatrist.
- It may lead to brachycephaly and plagiocephaly, late closure of fontanels, and raised ICP, eventually provoking seizures.
- Midfacial hypoplasia leads to small maxilla and relative mandibular prognathia, as well as high arched palate.
- These pts can have a beaked nose; deviated nasal septum; narrow palate; cleft palate; super numerary teeth; small, low-set, unusually shaped ears; and enamel hypoplasia.
- Facial appearance tends to improve with age throughout childhood.
- It may involve multiple organs. The predominating involved systems are the cardiac system, the skeletal system, as well as the sensory and motor systems.
- Less common signs and symptoms include congenital heart defects (ASD, VSD, pulm stenosis, PDA, TOF), renal anomalies, cryptorchidism, and anorectal malformations.

Worry About

- Ruling out increased ICP is important, either by using CT scan or fundus examination.
- High arched palates make placement of the tube difficult, because of limited lateral space availability.
- Facial features can lead to difficulty in bag and mask ventilation, intubation, and LMA insertion.
- Vertebral fusion is progressive and hence may present at a more advanced stage, leading to cervical instability.

Etiology

- Autosomal dominant inherited syndrome.
- Results from a mutation of the TWIST1 gene on chromosome 7, which plays a key role in the early development of the skull, face, and limbs.
- Early fusion of the coronal or lambdoidal suture.

Usual Treatment

- These pts can present with multiple surgical and medical complaints.
- Most of them will undergo surgery and anesthesia, at least once in their lifetime, for correction of craniofacial, orthopedic, ophthalmic, or cardiac lesions, apart from incidental surgical conditions.

Assessment Points

System	Assessment by Hx, PE	Anesthetic Concerns and Management	Test
HEENT	Progressive cervical spine fusion, cleft palate, high arch palate Midfacial hypoplasia, small maxilla, relative mandibular prognathia asymmetry, flat-looking face due to underdeveloped cheekbones, deviated nasal septum, narrow palate, cleft palate, super numerary teeth, and enamel hypoplasia Eye: Shallow orbits with orbital asymmetry, orbital hypertelorism, ptosis, strabismus, blepharophimosis, down-slanting palpebral fissures, sparse eyebrows medially, epicanthal folds, optic atrophy, and corneal opacity Ear: Small, low-set, unusually shaped ears, sensorineural hearing loss	Limited neck extension, difficult intubation, cervical spine instability Difficulty in bag and mask ventilation, difficulty in oral and nasal intubation Increased incidence of OCR and postop N/V during eye surgery. Difficulty in communication with child	X-ray, CT scan X-ray, CT scan
MS	Short stature, cutaneous syndactyly, small distal phalanges, clinodactyly of fifth finger, digitalization of thumb, limited elbow extension, contracture of elbow and knee Short clavicles	Difficulty in positioning on OT table, difficult IV access Placement of subclavian venous catheter more difficult	
NEURO	Brachycephaly, plagiocephaly, late closure of fontanelles, ossification defects, hyperostosis of skull	Increase in ICP, seizure disorders.	CT scan
CV	ASD, VSD, pulm stenosis, PDA, TOF	Anesthetic implications as per the cardiac lesion	ECHO, cardiac cath
OTHER	Renal anomalies, cryptorchidism, anorectal malformations	Deranged renal parameters, lyte imbalance	Cr, BUN

Key References: Niemann-Seyde SC, Eber SW, Zoll B: Saethre-Chotzen syndrome (ACS III) in four generations, *Clin Genet* 40(4):271–276, 1991; Netke M, Carver E: Saethre-Chotzen syndrome and anesthesia, *Paediatr Anaesth* 18(11):1128, 2008.

Perioperative Implications

Preoperative Preparation

- Genetic counseling.
- Detailed airway examination using indirect laryngoscopy is recommended prior to anesthesia, whenever possible, to prepare airway management and map any upper airway deformity.
- Cervical instability can be seen on standard neck x-ray.
- If child is taking anticonvulsants, continue on the day of surgery.
- Excessive sedative premedications should be avoided.

Monitoring

- Routine.
- Cutaneous syndactyly may cause difficult IV access.
- Precautions to prevent cervical spine movement during airway management at the time of induction and recovery from anesthesia.
- Meticulous positioning and padding of pressure points should be done during surgery.
- Short clavicles may make the placement of subclavian venous catheter more difficult if indicated.

Airway

- Plan for airway management (a supra-glottic device for short duration surgery and, if required, fiberoptic guided intubation of the trachea).

Induction (General Anesthesia)

- Inhalational induction

Maintenance

- Hypoventilation should be avoided because it may exacerbate preexisting elevated ICP.
- These pts may have shallow orbits with orbital asymmetry. It may predispose to exacerbated OCRs during extra-ocular muscles handling.
- Interaction between neuromuscular blockers and anticonvulsants.

Postoperative Period

- Postop seizures
- Postop N/V during eye surgery
- Difficulty in communication with child

Anticipated Problems/Concerns

- Difficult intubation and cervical instability
- Seizure on induction or emergence