Treacher Collins Syndrome

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Risk

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

Perioperative Risks

- Difficult airway management

Worry About

- Difficult mask ventilation and intubation
- Acute airway obstruction
- Hx of obstructive sleep apnea and cor pulmonale

Overview

- Also known as mandibulofacial dysostosis and Franceschetti-Zwahlen-Klein syndrome.
- Clinical features include various degrees of mandibular hypoplasia, high arched or cleft palate, malar hypoplasia, ophthalmic abn (downward slant of palpebral fissures, lower lid coloboma, partial to total absence of lower eyelashes, visual loss), microtia, atresia of external ear canal, and middle ear hypoplasia.
- Choanal atresia may be present.
- Conductive hearing loss due to ear abn is universal with varying degrees of severity.
- Normal intelligence.
- Airway compromise may occur due to maxillary hypoplasia (narrow nasal passages resulting in choanal stenosis or atresia) and mandibular hypoplasia (tongue base is retropositioned thereby obstructing oropharyngeal and hypopharyngeal spaces).
- Limited oropharyngeal and hypopharyngeal space may lead to obstructive sleep apnea, pulm Htn, and in severe cases, cor pulmonale.
- Affected newborns and infants often have feeding difficulties.
- May have congenital heart disease.

Etiology

- Abnormal bilateral first and second branchial arch development due to mutation in TCOF1 (78–93%) and POLR1C or POLR1D genes (8%).
- When inherited, shows autosomal dominance with variable penetrance and expression.
- TCOF1 mutation results in a deficiency of neural crest cells leading to failed development of cartilage, bone, and connective tissues, particularly in the head and neck region.

Usual Treatment

- Prenatal detection of micrognathia and dysmorphic facial features on fetal US may prompt genetic testing and counseling if TCS is suspected.
- Evaluate airway and assess swallowing and feeding difficulties at birth. Some pts require ET intubation in delivery room.
- Severe airway compromise and feeding issues may require tracheostomy and gastrostomy tube placement. Mandibular distraction procedures can be used to relieve airway obstruction and facilitate tracheal decannulation or avoid tracheostomy.
- Evaluate and correct any hearing and visual impairment. Early use of hearing aids (bone conduction) allows for proper development of speech. Surgery for bone anchored hearing aids placement may improve the quality of sound transmission.
- Oral-motor physical and speech therapy for speech clarity.
- Detailed assessment and imaging to determine the extent of craniofacial involvement during the first year of life. Repeated imaging may be needed prior to reconstructive procedures.
- Staged zygomatic, orbital, maxillomandibular, and nasal reconstruction.
- Surgical repair for cleft palate and choanal atresia.
- Staged external ear reconstruction. Very few TCS pts are candidates for external ear canalplasty to restore hearing.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Limited airway	Stridor, dyspnea, snoring, obstructive sleep apnea	Micrognathia, retrognathia, limited pharyngeal area	Facial X-rays, CT scan, nasal fiberoptic endoscopy
	Hearing loss	Hearing difficulties	External/middle ear atresia	Hearing evaluation
CV	Cor pulmonale Pulm Htn	Easy fatigability	S_3, hepatomegaly Increased jugular venous pulsations Heart murmur	ECG: Right axis deviation P waves in II, IIIa, VF ECHO/cardiac cath
GI	Difficulty feeding GERD, especially in pts with tracheostomy	Difficulty swallowing, chewing, poor PO intake Frequent regurgitation, discomfort after meals	Poor weight gain	Videofluoroscopic swallowing study Upper endoscopy
RESP	Obstructive sleep apnea	Loud snoring, intermittent complete obstruction, frequent arousal, daytime hypersomnolence or hyperactivity		Polysomnography

Key Reference: Plomp RG, van Lieshout MJ, Joosten KF, et al: Treacher Collins syndrome: a systematic review of evidence-based treatment and recommendations. *Plast Reconstr Surg* 137(1):191–204, 2016; Posnick JC, Tiwana PS, Costello BJ: Treacher Collins syndrome: comprehensive evaluation and treatment, *Oral Maxillofac Surg Clin North Am* 16(4):503–523, 2004.

Perioperative Implications

Preoperative Preparation

- Thorough airway assessment and review of previous anesthetics.
- Review of pertinent labs, studies, and imaging.
- Medical Hx, inquiring about obstructive sleep apnea or cor pulmonale.
- Antisialagogues for airway preparation.
- Antibiotic prophylaxis for pts with congenital heart disease as needed.

Monitoring

- Standard monitors.
- Invasive monitoring for lengthy reconstructive procedures with anticipated blood loss.

Airway

- Assume difficult intubation and prepare anesthetic plan in a case-by-case situation (ease of intubation with previous anesthetics may not guarantee ease of intubation with current anesthetic).
- Back up airway devices with fiberoptic bronchoscope, video laryngoscope, and surgical airway preparation. LMA may be bulky due to a small hypopharynx, particularly in younger pts.

Preinduction/Induction

- Avoid sedatives if Hx of severe OSA is present.
- Inhaled sevoflurane induction with maintenance of spontaneous ventilation during laryngoscopy.

Maintenance

- Avoid excessive opioids to minimize risk of postop respiratory depression.

Extubation

- Strict extubation criteria.
- Airway devices and staff support in case pt requires reintubation.

Postoperative Period

- Acute airway obstruction.
- Consider steroids, racemic epinephrine to decrease airway swelling.

Anticipated Problems/Concerns

- Obstructive sleep apnea, pulm Htn, and cor pulmonale
- Difficult airway