Cherubism

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Risk

- >250 cases in world literature
- Cherubs have a 40% chance of having a cherub offspring

Perioperative Risks

- Swelling of lower face causing airway obstruction
- Displacement of ocular orbit and lower eyelid, causing visual changes
- Excessive blood loss from curettage of vascular lesions
- Association with Noonan syndrome

Worry About

- Pulm valve stenosis (Noonan syndrome)
- Undiagnosed hyperparathyroidism
- Convex, V-shaped hypertrophied hard palate
- Small mouth opening and mild trismus

Overview

- Progressive symmetric fullness of cheeks and jaw, with retraction of lower eyelids exposing an inferior rim of sclera.
- Onset age: 2–12 y.
- These round-faced, upwardly gazing infants look like Renaissance art cherubs.
- Diagnostic biopsy of mandible shows multinucleated giant cells.
- Associated problems with speaking, breathing, swallowing, chewing.
- Pathognomonic x-ray of jaw demonstrates radiolucent lesions.

Etiology

- Mutations in the *SH3BP2* gene cause cherubism.
- Familial: Autosomal dominant.
- Penetrance: 100% for boys, 50% for girls.
- Etiology unknown, but alternative names include familial fibrous dysplasia, bilateral giant cell tumors, and familial multilocular cystic disease.
- Multilocular cystic malformation of mandible and maxilla with painless submandibular lymphadenopathy.

Usual Treatment

- Operative curettage, removal of displaced teeth, cortical reshaping of mandible
- Selective embolization with operative excision of vascular lesions
- Bone grafts
- For hyperparathyroidism: Normalization of 25(OH) D, Ca, K, and iPTH

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Orbits shifted Enlargement	Loss of binocular vision Photo review by age	Upward gaze Painless jaw swelling Lymphadenopathy	Jaw series
	Poor opening	Moderate trismus	Soft tissue swelling Concave palate	
	Malocclusion	Absence of third molar	Loose teeth	X-ray
CV	If associated with Noonan syndrome	Pulmonic valve disease	Pulm valve stenosis	ECHO
RESP	Generally unaffected	Obstructive airway	Sleep study	
ENDO	Rule out hyperparathyroidism	Onset at older age	Normal Ca^{2+}, K^+	
CNS	Midparental intelligence	No developmental delay except with Noonan syndrome		
MS	Long bone lesions		Humerus, anterior ribs, femoral neck	

Key References: Monclus E, Garcés A, Artés D, et al.: Oral to nasal tube exchange under fibroscopic view: a new technique for nasal intubation in a predicted difficult airway, *Paediatr Anaesth* 18(7):663–666, 2008; Papadaki ME, Lietman SA, Levine MA, et al.: Cherubism: best clinical practice, *Orphanet J Rare Dis* 7(Suppl 1):S6, 2012.

Perioperative Implications

Preoperative Preparation

- Rule out parathyroid disease.
- Ensure available blood for curettage replacement.

Monitoring

- Routine

Airway

- Difficult airway protocol.
- Oral intubation using a laryngeal mask technique has been reported. Fiberscopic control of the exchange and the introduction of a Cook exchange catheter into the trachea through the oral tube before withdrawal permits oxygenation of the pt and acts as a guide for oral tube reintroduction if required.

Preinduction/Induction

- Spontaneous ventilation
- Laryngeal mask airway

Maintenance

- Consider hypotensive technique for minimizing blood loss.

Extubation

- May require ICU admission for prolonged intubation.

Adjuvants

- Routine

Postoperative Period

- Extubation awake with confirmation of no bleeding

Anticipated Problems/Concerns

- Nasal intubation for oral procedures may be problematic, similar to Pierre Robin, Goldenhar, and Treacher Collins syndromes. As mandibular rami approach midline, no space for visualization of airway.

Chiari Malformations

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Risk

- 1:1000 live births
- Increased use of MRI leading to greater diagnosis
- Chiari malformation types I and II: Most common pediatric hindbrain abnormality
- Chiari type II always accompanied by myelomeningocele (which occurs in 0.6 of 1000 live births)

Perioperative Risks

- Respiratory and gastrointestinal dysfunction
- OSA, which has been targeted as an independent risk factor in perioperative morbidity, regardless of type of procedure
- Herniation

Worry About

- Increased ICP
- Herniation
- Hydrocephalus
- Syringomyelia
- Respiratory and cardiac center dysfunction
- Neurogenic dysphagia
- Rapid neuro deterioration in Chiari type II

Overview

- Group of hindbrain abnormalities ranging from herniation of cerebellar tonsils to cerebellar agenesis
- Often complicated by syringomyelia, a cavity fluid collection of the spinal cord
- Classification of Chiari malformations: Types I to IV (also type 0 and 1.5):
  - I: Herniation of cerebellar tonsils through foramen magnum into upper cervical spinal canal, disrupting normal CSF flow; not typically associated with hydrocephalus, but often complicated by syringomyelia (30–70%); referred to as adult-type secondary to delayed diagnosis
  - II (Arnold-Chiari): Herniation of cerebellar vermis, brainstem, and fourth ventricle through foramen magnum in the setting of myelomeningocele; commonly associated with both syringomyelia (20–95%) and hydrocephalus (90%)