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%)

- III: Very rare, extreme malformation (<1%) in which cerebellum and brainstem herniate into posterior encephalocele; associated with poor prognosis, severe neurologic deficits, respiratory insufficiency, developmental delay, and hydrocephalus
- IV: Cerebellar hypoplasia or aplasia without associated herniation (extremely rare)
- 0: Syringomyelia without tonsillar herniation that resolves with posterior fossa decompression
- 1.5: Similar to type I but other brainstem components are herniated, in addition to cerebellar tonsils

Etiology

- No unifying pathophysiologic mechanism between different types
- Multiple hypothesis to explain various malformations
- Syringomyelia may have a common origin between different Chiari malformations related to altered CSF dynamics
- Slightly more prevalent in female gender and European ethnicity

Usual Treatment

- No known medical treatment
- Posterior fossa decompression via suboccipital craniotomy with or without duroplasty to re-establish normal CSF flow from posterior fossa to cervical subarachnoid space
- Associated abnormalities, including hydrocephalus, syringomyelia, or scoliosis, which might dictate varied surgical treatment pathways
- Syringomyelia rarely needs direct surgical drainage and typically collapses following successful posterior fossa decompression

Assessment Points

System	Effect	Assessment by Hx	PE	Test
		Chiari I Malformation		
CNS	Brainstem compression	Headache and nonradicular occipital/cervical worsens with activity or Valsalva Dysphagia Severe snoring and OSA Respiratory dysrhythmias	C2 dysthesia Downbeat nystagmus Hoarse voice Tongue atrophy, fasciculations Dysarthria Facial numbness	MRI/cine-MRI CT X-ray (total spine, flex/ex)
	Spinal cord dysfunction (Syringomyelia)	Spasticity Urinary incontinence Arm/hand weakness	Scoliosis Loss of pain/temp sensation Extremity/trunk dysesthesia Arm/hand wasting	Urodynamic testing
	Cerebellar compression	Ataxia	Nystagmus	
		Chiari II Malformation		
CNS	Brainstem compression	Dysphagia, poor suck Aspiration pneumonia Gastroesophageal reflux Opisthotonus Sleep apnea Breath holding Weak cry Prolonged hiccups	Nasal vocalization Palatal weakness Tongue fasciculation/atrophy Inspiratory wheeze Cranial nerve VI palsy Lack of response to inspired CO_2 Tracheal anesthesia Depressed/absent gag	MRI CT
	Spinal cord dysfunction(syringomyelia)	Upper-extremity spasticity Upper-extremity weakness	Persistent cortical thumbs Loss of pain and temperature Upper-extremity weakness and muscle wasting Scoliosis	
	Cerebellar compression	Truncal ataxia	Nystagmus	

Key References: Cesmebasi A, Loukas M, Hogan E, et al.: The Chiari malformations: a review with emphasis on anatomical traits, *Clin Anat* 28(2):184–194, 2015; Tubbs RS, Hankinson TC, Wellons JC: The Chiari malformations and syringohydromyelia. In Ellenbogen RG, Abdulrauf SI, Sekhar LN, editors: *Principles of neurological surgery,* ed 3, Philadelphia, PA, 2012, Saunders, pp 157–168.

Perioperative Implications

Preoperative Preparation

- Assess for increased ICP, brainstem compression, and spinal cord dysfunction.
- Assess any other associated neurologic abnormalities and comorbidities.
- If history of recurrent aspirations, consider chest plain films and ABG analysis.

Monitoring

- Dictated by intended case, ranging from posterior fossa decompression to labor analgesia.
- For posterior fossa decompression, routine ASA monitors with invasive arterial monitoring.
- Central venous access not typically indicated.
- Foley catheter.
- Prepare for neurophysiologic monitoring.

Airway

- Endotracheal intubation while minimizing neck extension; use of in-line traction, fiberoptic bronchoscopy, Bullard laryngoscopy, and video laryngoscopy
- Special precautions in pts with myelomeningocele to avoid undue pressure on the defect

Preinduction/Induction

- Routine IV induction unless dictated by coexisting conditions.
- Consider rapid sequence induction if full stomach, dysphagia, uncontrolled gastroesophageal reflux, or recurrent aspiration are present.

Maintenance

- For posterior fossa decompression, total IV anesthesia may facilitate certain neurophysiologic monitoring modalities if employed.
- Prone positioning.
- Maintain rigid head fixation using Mayfield pin system (in pts >5 y old).
- In neonates, maintain strict temperature and glucose control.
- For obstetric analgesia and anesthesia, weigh risks and benefits of the anesthesia type provided for cesarean and vaginal delivery in parturients with Chiari malformations. Assess for signs and symptoms of increased ICP or neurologic deficits. In asymptomatic pts, spinal and epidural techniques appear safe in pts with Chiari I malformations. Imaging can help target levels that do not have syringomyelia for spinal or epidural placement.

Extubation

- Routine extubation with the goal of rapid emergence for neurologic evaluation and protection of airway reflexes
- Prepare for postop mechanical ventilation when indicated based on neuro deficits

Postoperative Period

- ICU for postop monitoring.
- Possible need for transport to remote locations to obtain postop imaging studies (CT scan).
- Prognosis: 90% of pts with Chiari type I have improvements or stabilization of symptoms. Syrinx typically resolves within 3 mo.

Anticipated Problems/Concerns

- Surgical complications include vascular or neuro injury, pseudomeningocele, CSF leak, meningitis, postop hemorrhage, occipital-cervical instability, hydrocephalus, brainstem compression, cranial nerve palsy, stroke, or persistent syringomyelia.
- Increased ICP management including mild hyperventilation, mannitol, or IV anesthesia.
- Brainstem compression leading to cardiovascular collapse, resistant hypotension, and dysrhythmias.
- Venous air embolism.