Opitz-Frias Syndrome (The G Syndrome)

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Risk

- Overall incidence not reported
- Very rare congenital disorder

Preoperative Risks

- Very high risk of recurrent pulm aspiration; hypoplasia of both pulm and vascular components of one lung (pulm hypoplasia)
- High mortality rate in infancy

Worry About

- NM dysfunction of laryngoesophageal apparatus
- Laryngotracheoesophageal cleft or fistula
- Difficult tracheal intubation due to assoc craniofacial deformity
- Assoc congenital anomalies (Htn, hypospadias, wide eyes, cleft lip, cleft palate, cryptorchidism, imperforate anus, cardiac deficits)

Overview

- Also known as the hypospadias-dysphagia syndrome.
- Emergency presentations are for cardiopulmonary resuscitation, upper respiratory obstruction, severe respiratory stridor, regurgitation, aspiration.
- Presence of one hypoplastic lung.
- Laryngeal hypoplasia.
- Laryngotracheoesophageal cleft or fistula.
- Anticipate very difficult tracheal intubation.
- Thorough preop cardiac evaluation; need to assess for cardiac abnormalities (possible ECHO).
- Any male infant presenting for tracheoesophageal fistula with genital defect should be suspected.
- Classically: Weak, hoarse cry

Etiology

- X-linked recessive inheritance.
- Autosomal dominant inheritance or new mutation.
- Partial male sex limitation.
- Autosomal recessive inheritance, high parenteral consanguinity.
- Females can be equally or nearly as severely affected as males.

Usual Treatment

- Prophylactic gastrostomy
- Feeding jejunostomy
- Cervical esophagostomy if infant is unable to swallow
- Prophylactic antibiotics (for pulm infection)

Assessment Points

System	Effect	Assessment by Hx	PE	Tests
HEENT	Cleft lip/palate (35%) Ankyloglossia Micrognathia	Feeding difficulties, speech anomalies	Short lingual frenulum	
CNS	Dolichocephaly (20%) Large metopic sagittal suture and anterior fontanel	Mental dysfunction, prominent forehead	"Cone-head" Palpation	CT (if indicated)
FACIES	Hypertelorism/telecanthus (90%) Mongoloid palpebral fissures Strabismus	Mother-related disease	Large nasal bridge downslanting	Facial x-ray
CV	Congenital heart defects (40%) (ASD, VSD, PDA, coarctation of aorta)	Failure to thrive	Auscultation	ECG, TEE, ABG
RESP	Agenesis, hypoplasia of one lung Tracheoesophageal cleft, fissure Hypoplasia of vocal cord Tracheomalacia Short trachea, high carina	Polyhydramnios on delivery Coughing, choking, cyanosis Hoarse, weak cry Stridorous respirations	Auscultation Tracheal stenosis	CXR Bronchogram Esophagogram
GI	Achalasia of the cardia (70%) NM dysfunction of esophagus	Dysphagia		Esophagogram (if indicated) Cinefluoroscopy of swallowing (if indicated)
GU	Hypospadias with descended testis Ureteral stenosis or duplication		Perineal or penoscrotal	Nephrogram

Key Reference: Bershof JF, Guyuron B, Olsen MM: G syndrome: a review of the literature and a case report. *J Craniomaxillofac Surg* 20(1):24–27, 1992.

Perioperative Implications

Preoperative Preparation

- Evacuation of the stomach with NG tube (if pt has gastrectomy open to air)
- Feeding: Clear water or apple juice (standard NPO guidelines)
- Consideration of H_2 blocker
- No atropine IM or metoclopramide preop
- Sodium citrate through NG tube
- Complete cardiac evaluation
- Assessment of renal function
- Not appropriate for outpatient or same-day process
- IV access 24 h before surgery to reduce stomach content

Monitoring

- All standard monitors
- Invasive arterial pressure if indicated owing to procedure or unstable hemodynamics

Airway

- Tubes smaller than normal secondary (as assessed by age) to laryngeal hypoplasia

Preinduction

- Warm OR.
- Decompress stomach with suction.
- Atropine and succinylcholine backup.

Induction

- Maintain spontaneous respiration.
- Danger of regurgitation and aspiration requires careful inhalation induction.
- Cricoid pressure should be applied.
- Atropine 20 mcg/kg at induction to prevent bradycardia during intubation.

Maintenance

- Hand ventilation (low PPV).
- Avoid hypothermia.

Extubation

- Based on pt's lung condition and preop assessment and/or lung function

Adjuvants

- All medications can be used (no contraindication to IV or inhaled anesthesia).

Anticipated Problems/Concerns

- Regurgitation and pulm aspiration. Difficult tracheal intubation. Increased incidence of pneumothorax. High mortality rate in infancy.
- Unanticipated cardiac issues.
- Difficult to assess recovery from anesthesia owing to associated mental conditions.