males, molar agenesis, high-arched narrow palate, broad cheeks, shallow supraorbital ridges, hypertelorism, and genu valgum

Etiology

- Currently not elucidated
- Autosomal recessive
- Mutation in RAB 23 gene on chromosome 6P12.1-q12 shown in most cases; at least five different mutations in RAB 23 gene
- Some cases of mutation in MEGF8 gene

Usual Treatment

- Surgery is the only option to correct cranial or cardiac abnormalities

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Craniosynostosis Mandibular or maxillary hypoplasia		Acrocephaly Abnormal facies Overbite/underbite Restricted mouth opening	CT head
RESP	Obstructive sleep apnea	Daytime somnolence Hx snoring	Macroglossia, tonsillar hypertrophy, redundant soft tissue	Polysomnography
CV	Congenital cardiac defects		Varies with type of defect and Hx or repair	ECG, TTE, CBC
GI	None			
CNS	Increased ICP Cognitive delay		Acrocephaly	CT head
HEME	None			
METAB	Obesity	Highest percentile on pediatric growth charts	Central adipose deposits	BMP, lipid panel

Key References: Bissonnette B, Luginbuehl I, Marciniak B, et al.: Acrocephalopolysyndactyly syndromes. In Bissonnette B, Luginbuehl I (eds): *Syndro mes: rapid recognition and perioperative implications.* New York, NY, 2006, McGraw-Hill; Kadakia S, Helman SN, Healy NJ, et al.: Carpenter syndrome: a review for the craniofacial surgeon. *J Craniofac Surg* 25(5):1653–1657, 2014.

Perioperative Implications

Preoperative Preparation

- Equipment available for difficult airway
- Congenital heart disease: may warrant prophylactic antimicrobial therapy
- Increased ICP: may warrant avoiding preop sedation

Monitoring

- Arterial line if indicated
- CVP/PA catheter: Consider as indicated

Airway

- Equipment available for difficult airway
- Surgeon available if surgical airway required

Preinduction/Induction

- Risk for obstruction and difficult mask ventilation is increased.
- Potential for agitation in pt with cognitive impairment.
- Maintain spontaneous ventilation while securing airway if possible.

Maintenance

- Adjustments required if pt Hx of repaired cardiac defects.
- Adjustments required if ICP is increased.
- Judicious opioid use in setting of increased risk for postop airway obstruction.

Extubation

- Difficult airway precautions

Postoperative Period

- Increased risk for upper-airway obstruction

Anticipated Problems/Concerns

- Difficult airway in periop period
- Complications from increased ICP
- Complications from congenital heart defects

Central Neurogenic Hyperventilation

Sarah C. Fausel | Kirk Lalwani

Risk

- True CNH is exceedingly rare; the exact incidence is unknown.
- In pts with neurologic injury, it is most often associated with pulm dysfunction or shunting (aspiration, pneumonia, pulm edema, and baseline disease).
- Primarily seen in comatose pts.
- No association with age or gender.

Overview

- A diagnosis of exclusion in neurologic disorders and in cases of hyperventilation; life-threatening causes of hyperventilation (hypoxemia, ischemic bowel, and acidosis) must be ruled out.
- Primary diagnostic criteria are hyperventilation that persists during sleep; low $PaCO_2$, high PaO_2, and absence of drug or metastatic causes.
- Associated primarily with brainstem inflammation and brainstem tumors with inconsistent involvement of midbrain, pons, and/or medulla.
- CNS lymphomas and astrocytomas are the most common tumor types with gliomas, lymphomatoid granulomatosis, and medulloblastoma; also reported in metastatic tumors.
- May result from seizure activity that stimulates the ventilatory response.
- May be associated with acute intermittent porphyria.
- Effects of GA unknown.

Etiology

- Exact etiology and level of brainstem dysfunction not known.
- Probable etiology:
  - Uninhibited stimulation of inspiration and expiration centers in the medulla and/or loss of descending inhibitory control of ventilation by cerebral cortex with brainstem lesion.
  - Ultimate control of respiration, which may lie in the medulla (dorsal and ventral respiratory groups) with fine control from the pneumotaxic center of the pons with input from cerebral cortex, hypothalamus, chemoreceptors and mechanoreceptors, and vagal nerve.
  - Stimulation of most areas of cerebral cortex except motor/premotor areas, which inhibit respiration.
  - Has been associated with brainstem infarction and malignancy.
  - Tumor, which may reduce local pH in the brainstem and activating respiratory chemoreceptors located in the ventral brainstem at the junction of the pons and the medulla.
  - Destructive lesions of midbrain or pons in animal studies, which do not produce CNH. (It is unclear if animal models serve as an adequate model of the human brain in this instance.)

Assessment Points

System	Effect	Assessment by Hx	PE	Test
RESP	Tachypnea	Tachypnea that persists during sleep and is unpleasant to the conscious patient	Resp rate Normal inspiratory and expiratory excursion	ABG (all must be present to diagnose): PCO_2 (low) pH (alkalotic) PaO_2 (increased for age) Decreased bicarbonate Alveolar-to-arterial gradient not larger than normal
CNS		Pt cannot volitionally inhibit hyperventilation	Focal or nonfocal CNS findings	CSF pH may be normal CT/MRI

Key Reference: Tarulli AW, Lim C, Bui JD, et al.: Central neurogenic hyperventilation. A case report and discussion of pathophysiology, *Arch Neurol* 62:1632–1634, 2005; Kramer CL, Wijdicks EF: Central neurogenic hyperventilation, *Neurology* 83:376, 2014.

Differential Diagnosis for Hyperventilation

- Metabolic acidosis
- Bowel ischemia with acidosis
- Pulm pathology with hypoxemia (pneumonia, pulm embolus, pulm edema, and restrictive or obstructive lung disease)
- Drug toxicity (salicylates, theophylline, cyanide, and topiramate)
- Sepsis
- Encephalopathy/CNS lesions (glioblastoma, encephalitis, MS, brainstem lymphoma, brainstem glioma, brainstem infarction, and liver dysfunction)
- Anxiety
- Psychogenic
- Cardiac (CHF and valvular disease)
- High altitudes
- Hyperthyroidism
- Pregnancy
- Must exclude other etiologies for respiratory alkalosis with appropriate lab/Dx testing

Adverse Effects

- Respiratory alkalosis shifts oxyhemoglobin curve to the left.
- Hypocapnia is a potent cerebral vasoconstrictor, subsequently decreasing cerebral blood flow and volume, which may result in ischemic insults.
- Effect of severe hypocapnia in normal brains is less clear and may produce ischemia when combined with Bohr effect. (Hemoglobin's oxygen affinity is inversely related to the concentration of CO_2.)

Treatment

- No completely effective or consistent treatment currently known
- Narcotics may attenuate resp rate and improve blood gases but will not correct rate or alkalosis.
- Mechanical ventilation with neuromuscular blockade and sedation during treatment of tumor has been attempted.
- Treatment of tumor with steroids, chemotherapy, or radiation therapy is sometimes effective.
- Increasing dead space ventilation and administration of supplemental oxygen and benzodiazepines are not effective.

Outcome

- Death from progressive neurologic deterioration or other complications (aspiration, pneumonia) is likely.
- Pt may have improvement with treatment of tumor or long-term narcotics.

Cephalopelvic Disproportion

John Kissko III

Risk

- Occurs in 1–3% of the pregnant population

Perioperative Risks

- Increased maternal and fetal morbidity and mortality
- Protracted labor
- Arrested labor
- Uterine rupture
- Increased rate of cesarean section
- Increased rate of forceps- or vacuum-assisted delivery

Worry About

- Increased need for surgical delivery
- Increased incidence of fetal distress and need for emergency intervention

Overview

- Subset of fetopelvic disproportion.
- Methods of Dx include various clinical and radiographic estimations of fetal head size and pelvic capacity, none of which alone is an accurate predictor of increased risk of failed labor.
- Leads to abnormal labor pattern with increased likelihood of operative delivery.
- Operative delivery associated with higher incidence of morbidity and mortality in mother and fetus.
- Anesthesia necessities: Complete preop evaluation for possible emergency C-section, including airway exam and neuraxial anesthesia landmarks.

Etiology

- Maternal causes include contracted pelvis (inlet, midpelvis, or outlet), Hx of scoliosis, previous pelvic trauma (especially fracture with callous formation), and Hx of poliomyelitis.
- Fetal causes include macrosomia (often secondary to gestational diabetes), abnormal presentation, and hydrocephalus.

Usual Treatment

- Obstetric: Proper evaluation and planning before and during labor
- Anesthesia: Neuraxial anesthesia for pain relief during labor or operative delivery

Assessment Points

System	Effect	Assessment by Hx	PE	Test
GYN	CPD	Failure to progress adequately in labor	Pelvic exam	Radiographic cephalopelvimetry

Key References: Cunningham F, Leveno KJ, Bloom SL, et al: Abnormal labor. In Cunningham F, Leveno KJ, Bloom SL eds: *Williams obstetrics*, ed 24, New York, NY, 2013, McGraw-Hill; Hillyard SG, Bate TE, Corcoran TB, et al: Extending epidural analgesia for emergency Caesarean section: a meta-analysis. *Br J Anaesth.* 107(5):668–678, 2011.

Perioperative Implications

- Labor usually more prolonged and painful in pts with CPD
- Epidural or CSE usually adequate to cover pain without significantly prolonging the course of labor
- Increased need for surgical delivery or C-section, many times in emergent fashion

Anesthetic Technique

- Labor epidural (PCEA): A low concentration of bupivacaine 0.125% or ropivacaine 0.10% is supplemented with an opioid (e.g., fentanyl) to help reduce motor block. Continuous infusion at 8–10 mL per h with pt-controlled boluses of 6–8 mL every 10 min can be used as needed. Programmed intermittent bolus with additional patient-controlled boluses is also an option.
- Labor CSE: In early first-stage labor (<4 cm cervical dilation), consider injection of 15 mcg fentanyl combined with 1 mL sterile saline followed by continuous epidural as with a PCEA. In late first-stage labor (>4 cm cervical dilation), intrathecal injection of local anesthetic (e.g., 1 mL of 0.25% bupivacaine) and opioid (e.g., 15 mcg fentanyl) is sometimes sufficient for the remainder of the first and second stages of labor, although a PCEA is usually started.