Assessment Points

System	Effect	Assessment by Hx	PE	Test
RESP	Congenital hypoventilation ("Ondine's curse")	Apnea		
CV	Hypovolemia, septic shock, 2-5% cardiac anomalies (tetralogy of Fallot)	IV replacement Extent of vomiting Cyanosis	Mucous membranes Vital signs/UO Murmur, cyanosis Capillary refill	BUN, Cr BUN/Cr ratio ECHO
GI	Intestinal obstruction	Presence of meconium Constipation Diarrhea Vomiting	No feces in rectum, Abdominal distention Malnutrition	Lyte panel Abdominal films Barium enema

Key References: Butler Tjaden NE, Trainor PA: The developmental etiology and pathogenesis of Hirschsprung disease, *Transl Res* 162(1):1–15, 2013; McKeown SJ, Stamp L, Hao MM, et al.: Hirschsprung disease: a developmental disorder of the enteric nervous system, *Wiley Interdiscip Rev Dev Biol* 2(1):113–129, 2013.

Perioperative Implications

Preoperative Preparation

- Consider associated congenital anomalies or syndromes and the possible need for further cardiac evaluation and genetic testing.
- Thoroughly assess volume status. Assess for bowel preparation, diarrhea, and vomiting and ensure adequate preop fluid resuscitation.
- Review preop labs to assess for lyte abnormalities.
- Consider cardiac evaluation with associated cardiac anomalies.

Monitoring

- Standard ASA monitors
- Urinary cath

Airway

- Consider associated syndromes affecting airway anatomy.

Induction

- Rapid sequence induction necessary in the presence of bowel obstruction to avoid pulmonary aspiration.
- In the setting of hypovolemia or sepsis (HAEC), IV and volatile anesthetics may be poorly tolerated.

Maintenance

- Use neuromuscular blocking drugs for maintenance of muscle relaxation.
- Maintenance IV fluids with balanced, isotonic solution.
- Consider checking intraop blood glucose level.
- Monitor urine output.
- Avoid nitrous oxide.
- Maintain normothermia with warming devices (full access warming blankets and radiant warmers), as radiant heat loss may be excessive. Keep forced warm air blankets dry. They cool the pt if they become wet.
- Carefully position pt; use added care with lithotomy position.

Extubation

- Reverse neuromuscular blockade.
- Routinely extubate when pt is awake and meets extubation criteria.

Postoperative Period

- Consider regional technique with epidural/caudal anesthesia for postop pain management (which may need to be performed postop if lower body antibacterial preparation performed).
- Postop apnea in newborns more likely following narcotic administration.

Anticipated Problems/Concerns

- Early postop complications include prolonged ileus, anastomotic leak, and wound infection/dehiscence.
- Late complications include anastomotic strictures, constipation, fecal incontinence, bowel obstruction, and enterocolitis.
- Postop HAEC, with an incidence between 5-42%, is a major cause of increased morbidity and mortality after definitive pull-through procedure. This is hypothesized to involve intestinal stasis and immature mucosal immunity, allowing for proliferation and mucosal invasion by luminal pathogens.
- Mild obstructive symptoms are treated with dietary changes, laxatives, enemas, or repeated botulinum toxin injections. Myectomy procedure may be required.
- For residual aganglionosis, strictures, or dysfunctional proximal bowel, repeat pull-through procedure can be done, although this is challenging due to scarring.
- In individuals with extensive intestinal aganglionosis and irreversible intestinal failure, intestinal transplantation may be considered.

Histiocytosis

Tyler J. Paradis | Jeffrey R. Kirsch

Risk

- LCH is the most commonly known form.
- Incidence: 1:250,000 in children, with about a third of this incidence in adults.
- Seen in all ages, but peak incidence is at 0–3 y of age.
- Male:female ratio: 1.5:1.
- Sporadic development with no established genetic predisposition.

Perioperative Risks

- Dependent on organ systems involved and extent of dysfunction

Worry About

- Specific organ dysfunction caused by infiltration with histiocytes, including liver, lungs, hematopoietic system, pituitary, spleen, and bone
- Can involve single or multiple sites and organs
- Treated with steroids and chemotherapy, which may cause adrenal insufficiency and result in the pt requiring stress steroids in the periop period
- Central diabetes insipidus due to posterior pituitary involvement
- Cervical instability if lesions present in cervical vertebrae
- Severe pulm dysfunction possible; pulm Htn without overt right heart failure

Overview

- A broad group of disorders involving infiltration of affected organs with monocytes, macrophages, and dendritic cells.
- The most commonly discussed disorder is LCH, previously known separately as eosinophilic granuloma, Hand-Schüller-Christian disease, and Letterer-Siwe disease.
- Severity of clinical symptoms varies markedly and can involve primarily skin and/or bone or liver, lung, or brain.
- Can be limited or progressive and fatal. Younger children with multiple or severe organ involvement of "risk organs" (liver, lungs, spleen, hematopoietic system) have a high mortality.
- Usual clinical presentation is in the first decade of life.
- Pathophysiology is unclear and treatment is nonspecific.

Etiology

- Unknown; suggested factors include immune dysfunction, viral infections, neoplastic processes, and genetic predisposition.
- Isolated pulm LCH is strongly associated with cigarette smoking.

Usual Treatment

- 10–20% spontaneous regression rate, almost exclusively in pts with single system disease.
- Chemotherapy with steroids for multisystem disease with local or constitutional symptoms (vinblastine, etoposide, mercaptopurine, doxorubicin, cyclophosphamide, methotrexate, others).
- Surgery is required for biopsy and Dx, isolated bone lesions, and occasionally splenectomy.
- Orthotopic liver or lung transplantation has been performed for end-stage disease.
- Radiation therapy (bone lesions, pituitary disease).
- Bone marrow or stem cell transplant.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Soft tissue distortion of airway, loose teeth, mucosal ulceration	Stridor	Airway and dental evaluation	
RESP	Spontaneous pneumothorax, reactive airways, infiltrates, fibrosis, pulm Htn	Tachypnea, dyspnea, cough, smoking history		CXR, ABG, PFTs, CT with cysts or nodular infiltrate
GI	Ulceration, obstruction, hepatic dysfunction		Jaundice Hepatomegaly	Bilirubin, albumin AST, ALT, INR
CNS	Diabetes insipidus, neuropathy, exophthalmos	Polyuria, polydipsia	Neuro exam	Urine and serum Osm, lytes
HEME	Thrombocytopenia, anemia, leukopenia	Bruising or bleeding	Splenomegaly	CBC

Key References: Morimoto A, Oh Y, Shioda Y, et al.: Recent advances in Langerhans cell histiocytosis, *Pediatr Int* 56(4):451–461, 2014; Broscheit J, Eichelbroenner O, Greim C, et al.: Anesthetic management of a patient with histiocytosis X and pulmonary complications during Caesarean section, *Eur J Anaesthesiol* 21(11):919–921, 2004.

Perioperative Implications

Monitoring

- Routine
- Foley in pt with DI
- Arterial cath for ABG in those with pulm involvement, frequent Na checks in those with DI, as well as pulse pressure variation as a marker of volume status in those with DI

Preinduction/Induction

- Airway soft tissue or mandibular involvement may distort anatomy.
- Cervical vertebrae lesions may cause cervical instability.
- Ensure adequate preoxygenation, especially if there is significant pulm involvement.
- Usual precautions, depending on severity of organ involvement.

Maintenance

- For pts with DI, consider aqueous ADH infusion and isotonic crystalloid fluids.
- Stress dose steroids if pt has had steroid therapy.
- Usual precautions, depending on severity of organ involvement.

Extubation

- Awake extubation if anatomy is distorted and airway was difficult for mask ventilation or intubation.
- Severe pulm involvement may delay extubation.

Regional Anesthesia

- Follow ASRA precautions if thrombocytopenic or elevated INR.
- Use caution with interscalene and supraclavicular blocks in pts with pulm disease.

Postoperative Period

- May need continued stress dose steroid coverage for several days postop.
- Closely monitor oxygenation and ventilation when pulm disease present, and evaluate for pneumothorax.

Anticipated Problems/Concerns

- Organ dysfunction (hepatic, pulm, hematologic, hypothalamic, or bone).
- DI.
- Adrenal suppression due to chronic steroid therapy; may experience intraop hypotension without stress steroids.
- Severe pulm involvement may increase risk of pneumothorax and complicate extubation.

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Huntington Disease

David A. Wyler

Risk

- General prevalence: 5-7:100,000
- Highest prevalence in Caucasians of western European descent

Perioperative Risks

- Increased risk of respiratory complications secondary to bulbar muscle incoordination
- Autonomic dysfunction

Worry About

- Microaspiration, bronchospasm, chemical pneumonitis, and aspiration pneumonia
- Drug-drug interactions with anesthetic drugs and psychotropic medications
- Prolonged effects with succinylcholine
- Dysautonomia, gastroparesis, and fluctuating HR and BP

Overview

- Inherited progressive neurodegenerative disease of the CNS, primarily the basal ganglia.
- More common adult-onset variant begins in the fifth decade and leads to complete disability and death within 20 y.
- Heterogeneous presentation of dysregulation of motor coordination, cognitive decline, and psychiatric manifestations.
- Classically known for choreiform (repetitive, rapid, jerky, involuntary) movements from degeneration of GABAergic neurons of the basal ganglia specifically of the striatum (caudate and putamen).
- Chorea, early motor sign along spectrum; progresses to parkinsonian-like movements (bradykinesia, rigidity, and postural instability) late in the adult-onset disease.
- Worsening subcortical dementia (declining executive function and cognition without amnesia) and severe depression accompany disease progression.
- Juvenile variant presents with parkinsonian signs at onset, lacks choreiform movements, and has least favorable prognosis along spectrum.
- Skeletal muscle incoordination of the laryngeal and pharyngeal muscles leads to devastating respiratory sequelae and death.
- See also Parkinson Disease.

Etiology

- Autosomal dominant inheritance.
- Trinucleotide repeat expansion of CAG codon on the IT15 gene on chromosome 4 results in the overproduction and aggregation of the protein Huntingtin.
- Length of repeat correlates well with extent of Huntingtin production, disease severity, and age of onset.
- Huntingtin accumulates in the nuclei and cytoplasm of all CNS neurons; degeneration occurs most notably in vulnerable neurons of the caudate and putamen.
- Striatal cell death occurs by glutamate- and dopamine-induced excitotoxicity, oxidative stress, impaired energy metabolism, and apoptosis.

Usual Treatment

- No definitive cure; treatment is supportive, focusing on alleviation of symptoms.
- Early symptoms of chorea are treated with neuroleptics, dopamine-depleting medication. Surgical implantation of deep brain stimulators may be helpful.
- Gene-modifying therapy is currently under investigation.