- Directed pharmacologic approach to increase chloride conductance of skeletal muscle includes taurine and clofibric acid; however, the effect is modest.
- Other interventions include optimizing pt's emotional state and relaxation techniques. Some subjects have shown improvements with alcohol use. Exercises that improve flexibility and decrease muscle strains can be helpful.
- Gene therapy to introduce a functional copy of the normal gene has been considered; however, this may not be effective in disorders caused by a dominant-negative mechanism.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
No effects on CNS, CV, RESP, GI, GU, ENDO, HEME/ID systems because BD only affects skeletal muscle membranes.				
HEENT	Blepharospasm (myotonia of the eyelids)			
MS	Delayed muscle relaxation that is resistant to NDMR and DMR	Obtain full Hx regarding symptoms and aggravating factors	Upper-extremity atrophy Muscular hypertrophy in other areas of the body Reflex hammer test	EMG

Key References: Bandschapp O, Iaizzo PA: Pathophysiologic and anesthetic considerations for patients with myotonia congenital or periodic paralyses, *Pediatr Anesth* 23(9):824–833, 2013; Dunø M, Colding-Jørgensen E: Myotonia congenita. In Pagon RA, editor: *GeneReviews*. Available at <http://www.ncbi.nlm.nih.gov/books/NBK1355/>. (Accessed 23.02.16.)

Perioperative Implications

Preoperative Preparation

- Keep pt normothermic throughout pre-/intra-/and postop periods because shivering can trigger myotonic episode.
- DMR, NDMR, and regional anesthesia are ineffective in minimizing myotonic contractions because the defect lies within the muscle membrane.
- Regional anesthesia with peripheral nerve stimulation in combination with fentanyl and midazolam sedation for shoulder surgery has been used successfully without complication.

Monitoring

- Neuromuscular monitoring is mandatory.
- Core temperature monitoring is recommended.

Airway

- Can be difficult to ventilate/intubate if myotonia was elicited during induction/extubation.

Preinduction/Induction

- Consider administering IV lidocaine prior to propofol induction as pain associated with propofol injection can lead myotonia.
- Avoid depolarizing muscle relaxant (succinylcholine), as it has been shown to provoke severe generalized muscle stiffness, including masseter spasm and decerebrate posturing, making intubation and ventilation difficult to impossible.
- Response to NDMR appears to be normal; however, consider reducing the dose of NDMR in pts with associated muscle wasting.

Maintenance

- Consider short-acting NDMR and allow pt to recover fully from muscle relaxant without reversal because anticholinesterase can precipitate myotonia.
- Currently, there are no data on reversal of rocuronium by sugammadex for MC pts.

Extubation

- Consider avoiding anticholinesterase use.
- Avoid coughing on extubation.

Postoperative Period

- Continue to maintain normothermia and adequate pain control.

Anticipated Problems/Concerns

- No association with malignant hyperthermia as previously suggested.
- In rare cases, epinephrine or selective beta-adrenergic agonists in high doses may aggravate myotonia. Beta-antagonist propranolol has also been reported to worsen myotonia.

Beckwith-Wiedemann Syndrome

Arlyne K. Thung | Lee A. Fleisher

Risk

- 1 per 13,700 individuals.
- No gender predilection, although with monozygotic twins it is seen more in females than males.
- Conceptions from IVF have a 3–5 times increased risk of BWS.

Perioperative Risks

- Acute airway obstruction; difficult mask ventilation and intubation secondary to macroglossia
- Hypoglycemia due to islet cell hyperplasia and hyperinsulinemia
- Cardiac malformations

Worry About

- Persistent hypoglycemia, which may cause CNS damage; therefore intraop infusion of a glucose-containing solution and frequent glucose checks are required.
- Difficult airway management.

Overview

- Commonly known for the triad of EMG.
- Other clinical features include anterior earlobe creases, posterior helical pits, facial nevus flammeus, hemihyperplasia, renal anomalies, embryonal tumors, cardiac malformations, and hypoglycemia.
- 7.5% estimated risk for embryonal tumor development, which occurs in the first 10 y of life. Most common tumors are Wilms tumor and hepatoblastoma but may also include rhabdomyosarcoma, adrenocortical carcinoma, and neuroblastoma.
- Cardiac involvement often limited to mild cardiomegaly, although other cardiac defects have been reported (atrial and ventricular septal defects, tetralogy of Fallot, hypoplastic left ventricle, cardiomyopathy, cardiac tumors, and valvular disease).
- Hypoglycemia due to islet cell hyperplasia and hyperinsulinemia occurs in 50% of BWS pts, is often responsive to medical therapy, and usually regresses during the first 4 mo of life. Persistent hypoglycemia refractory to medical management may require pancreatectomy.

Etiology

- Clinically and genetically heterogeneous.
- May be genetically transmitted (15%) or occur sporadically (85%).
- Variety of mutations in chromosome 11p15.5 region.
- Mutation near gene for IGF-II.

Usual Treatment

- Prenatal detection of polyhydramnios, omphalocele, placentomegaly, macrosomia, macroglossia, and renal anomalies on fetal US may prompt genetic testing and counseling if BWS is suspected.
- Screening for hypoglycemia in the first few days of life if BWS is suspected. Surgical intervention if hypoglycemia persists despite medical management.
- Surgical repair of omphalocele.
- Possible reduction of macroglossia in the first year of life to avoid complications of airway obstruction, feeding, and speech difficulties.
- Infants with hypoglycemia and severe oral intolerance due to macroglossia may require gastrostomy tube placement as a temporizing measure until regular feeds become possible after glossal resection.
- Orthopedic follow-up to monitor leg-length discrepancies due to hemihyperplasia.
- Tumor surveillance (abdominal US, alpha-fetoprotein).
- Surgical resection of operative tumors.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Macroglossia	Hx of difficult mask ventilation and intubation	Determine extent by physical inspection and oral palpation, previous anesthesia Hx	No testing
CV	VSD, ASD, TOF, valvular disease, hypoplastic LV, cardiac tumor and cardiomegaly (most common) possible	SOB, DOE	Cardiac exam for murmurs	ECHO CXR
ENDO	Hypoglycemia Hypothyroidism	Shaking, lethargy		Glucose Thyroid function tests
RENAL	Renal medullary dysplasia Nephrolithiasis	Hx of renal tumors/previous resections, chronic UTIs	Palpate for masses, Flank pain	US BUN/Cr

Key References: Weksberg R, Shuman C, Beckwith JB: Beckwith–Wiedemann syndrome, *Eur J Hum Genet* 18(1):8–14, 2010; Eaton J, Atiles R, Tuchman JB: GlideScope for management of the difficult airway in a child with Beckwith–Wiedemann syndrome, *Paediatr Anaesth* 19(7):696–698, 2009.

Perioperative Implications

Preoperative Preparation

- Coordinated care with endocrinology and an ENT specialist to assist in the management of hypoglycemia and difficult airway.
- Discussion with ENT for planned tracheostomy if significant airway edema and swelling is anticipated following glossal resection.
- Review of lab results (hypothyroidism, polycythemia, hypocalcemia, and hyperlipidemia have been reported in pts with BWS in addition to hypoglycemia).
- Review cardiac workup if available.
- Pretreatment with antisialagogue (glycopyrrolate or atropine) if intubation is planned.

Monitoring

- Standard monitoring appropriate for surgical procedure
- Frequent glucose checks

Airway

- Assume difficult mask ventilation due to macroglossia.
- Nasal intubation may be more easily performed than oral intubation in pts with significant macroglossia. Pretreat with a nasal decongestant and dilate with nasal trumpets if nasal intubation is considered.
- Assistance with glossal manipulation if direct laryngoscopy is performed.
- Backup airway devices (e.g., fiberoptic, glidescope, LMA) and surgical support (ENT) if conventional laryngoscopy fails.
- Age-appropriate ETT.

Induction

- Inhaled induction with sevoflurane versus awake intubation with sedation/topicalization.
- Clinicians should be aware that administration of IV anesthetics and muscle relaxants may cause pt's tongue to fall backward, causing acute airway obstruction.

Postoperative Period

- After meeting strict extubation criteria, pts should be monitored in ICU or recovery area with immediate backup for management of airway issues and hypoglycemia.

Anticipated Problems/Concerns

- Difficult airway
- Hypoglycemia

Behçet Disease

Anurag Gupta

Risk

- Affects age group between 20–40 y
- Nations along Silk Route have higher incidence
- Males and females are equally affected

Perioperative Risks

- Increase in IOP during intubation in pts with uveitis complicated by glaucoma
- Pulmonary embolism
- Difficult airway due to oral inflammation

Worry About

- Difficult airway
- Hyperreactive skin
- Pulmonary aneurysm
- Intracranial Htn
- Concurrent anti-inflammatory medications

Overview

- Multisystem inflammatory disorder of unknown etiology characterized by relapsing episodes of oral aphthous ulcers, genital ulcers, other skin lesions, and ocular lesions.
- For diagnosis of BD, an international study group proposed the presence of oral aphthous ulcers and any two other manifestation among the following:
  - Recurrent genital ulceration.
  - Skin lesion.
  - Papulopustular lesions.
  - Ocular involvement.
  - Positive pathergy test: Hyperreactivity of skin leading to sterile pustule and erythematous papule formation after intracutaneous injection or needle prick.
- Ocular and vascular involvement increases morbidity.
- Major vessel disease and neurologic involvement are the major cause of death.
- Newer drugs have shown good improvement in resistant cases of BD, but further studies are needed to reinstate their efficacy.
- Extracutaneous ulcers, which heal by scarring, may be found in children.
- Erythematous nodosum-like lesions may occur mostly in females; the lesions are more erythematous and edematous, and they heal within a week, leaving hyperpigmentation after healing.

Etiology

- Although exact etiology unknown, BD is found to be mostly associated with HLA-B51.
- Other genes implicated are HLA-26, PSOR1C1, HLA-Cw1602, GIMAP, UBAC2, IL-10, and IL-23.

Treatment

- Mucocutaneous BD: Thalidomide, dapsone, TNF-α inhibitor (Etanercept), IFN-α, and colchicine
- Ocular involvement: Azathioprine, cyclosporine, IFN-α, and methotrexate
- Vascular involvement: Azathioprine, cyclophosphamide, and cyclosporine
- Joint involvement: Colchicine and NSAIDs
- GIT: Azathioprine
- CNS: Anticoagulants, infliximab, IFN-α, and adalimumab
- Corticosteroids: Severe, life-threatening disease in ocular, vascular, GI, and neuro BD (as advised by an expert committee report)