Etiology

- Autosomal recessive inheritance can occur with deletions or mutations in the survivor motor neuron genes located on chr5q13.
- The loss of full-length SMN protein leads to degeneration of anterior spinal motor neurons and, in severe cases, degeneration of brainstem nuclei.
- Degeneration of spinal anterior neurons and brainstem nuclei correspond to a range of clinical characteristics, including global hypotonia, pulmonary insufficiency, and autonomic and bulbar dysfunction.

Usual Treatment

- There is no cure for SMA.
- Supportive treatment as required, including physiotherapy and orthopedic intervention, to prevent contractures and maximize respiratory function.
- Low threshold for antibiotic use during acute illnesses due to the risk of pneumonia.

Classification of Spinal Muscular Atrophy

Type	Age at Onset	Highest Motor Milestone Achieved	Lifespan Without Treatment	Symptoms	Affected Organ
Type I Werdnig-Hoffman disease	Birth–6 mo	Never sits unsupported	<2 y	Progressive muscle weakness, respiratory failure, hypotonia, reduced bulbar function	Muscular: Respiratory
Type II Dubowitz disease	6–12 mo	Sits independently, never stands or walks	70% reach adulthood	Progressive onset of proximal limb weakness in infancy Legs > arms Scoliosis Joint contractures	Muscular: Kyphoscoliosis Joint contractures
Type III Kugelberg-Welander disease	>18 mo	Stands and walks	Normal lifespan	Onset of proximal weakness during childhood Legs > arms Scoliosis Increased risk of fractures	Muscular: Joint problems
Type IV Adult SMA	>5 y to mostly >30 y	Normal	Normal	Onset of proximal leg weakness in adulthood	Muscular

Key References: Islander G: Anesthesia and spinal muscle atrophy, *Paediatr Anaesth* 23(9):804–816, 2013; Darras BT: Spinal muscular atrophies, *Pediatr Clin North Am* 62(3):743–766, 2015.

Perioperative Implications

Preoperative Preparation

- Preop pulm evaluation and pulm function testing.
- Evaluate intubation conditions.
- Start air-stacking techniques preop.
- Make a preop and postop plan. Pt may require postop ventilator support.

Monitoring

- When nondepolarizing muscle relaxants are used, the effect should be monitored carefully both clinically and with a monitor of neuromuscular transmission and muscular contraction.
- Consider ABG.

Airway

- Difficult intubation can occur due to limited mobility of the cervical spine and reduced mouth opening.
- Pt may present with artificial ventilation (NIV).
- Awake fiberoptic intubation could be the technique for intubation in pts with restricted neck movements.

Preinduction/Induction

- No specific anesthetic drug is recommended.
- Laryngeal mask may be appropriate in superficial surgery.
- Peripheral neural blockade may be useful.
- Avoid succinylcholine due to the risk of inducing rhabdomyolysis and hyperkalemia.
- Nondepolarizing muscle relaxants are suitable but should be titrated carefully since sensitivity to these drugs appears to vary.
- Approach when choosing anesthetic techniques and agents:
  - Minimize modifications of chest wall dynamics due to residual muscle relaxants effect or high level of neural axis blockade.
  - Avoid excessive depression of central respiratory drive.

Maintenance

- Both TIVA and inhalation agents may be used.
- Pts with SMA are not at increased risk for malignant hyperthermia.
- Short-acting opioids are suitable for intraop use.
- Continuous infusion of local anesthetic solutions via peripheral nerve block cath should be considered as safer alternatives to systemic opioids.
- Wound infiltration anesthesia is recommended whenever possible.

Extubation

- Muscle strength must be evaluated before extubation, not only with train-of-four stimulation but also clinically.
- Reverse neuromuscular blockade with sugammadex.

Postoperative Period

- Pts with SMA I need postop ventilator support.
- Some pts with SMA II and III will require respiratory support during acute illness or in advanced disease; NIV for bridging from intubation to spontaneous breathing.
- Use oxygen with caution because too much oxygen can mask hypoventilation due to muscle weakness.
- Postop pain management must be individualized and multimodal. Acetaminophen and ibuprofen are useful.

Anticipated Problems/Concerns

- Opioid-induced respiratory depression is dangerous in SMA pts with weak muscles. Careful monitoring is mandatory.
- The major concern related to the response from anesthesia is prolonged impairment of neuromuscular function and suppression of central respiratory drive, which can compromise the limited pulm reserve leading to acute respiratory failure.
- Neuraxial anesthesia can be difficult or unreliable due to altered spine anatomy (severe scoliosis).

Stevens-Johnson Syndrome

Lauren M. Nakazawa | Lee A. Fleisher

Risk

- Incidence of SJS and TEN, a more severe variant of SJS, is 2–7 cases per million per y.
- Incidence around 100 times higher in the HIV-positive population.
- More common in women.
- Affects all age groups.

Perioperative Risks

- High risk for infection
- Hypovolemia
- Cutaneous, mucosal, and ocular injury
- Respiratory failure requiring mechanical ventilation in around 25% of pts

Worry About

- Sepsis and septic shock
- Fluid and lyte imbalances
- Development of multiorgan failure
- Disease recurrence if culprit drug is readministered

Overview

- Severe cutaneous reaction with epidermal necrosis and detachment in conjunction with mucosal and conjunctival involvement.
- SJS and TEN fall along a disease continuum. SJS is less severe, involving <10% total BSA. TEN involves >30% BSA, and SJS-TEN overlap involves 10–30%.
- Clinical presentation:
  - Prodrome: Fever, flu-like symptoms (malaise, myalgia, arthralgia), skin pain/tenderness, oral pain, photophobia, and conjunctival burning can be early signs of mucosal involvement.
  - Cutaneous lesions: Diffuse erythema or erythematous macules starting on trunk and face and developing central necrosis and bullae formation with eventual sloughing off of epidermis and exposed dermis.
  - Mucosal involvement in 90% of pts.

- Mortality from SJS is around 5–10% and increases to 30% or more for TEN.
- Mortality primarily from sepsis, respiratory failure, and multiorgan dysfunction.
  - Prognosis worse with advanced age and greater BSA involvement.
  - Prognostic scoring system, called SCORTEN, can estimate pt survival.

Etiology

- Leading causes of disease are medications, followed by infections
- Medications most commonly implicated include allopurinol, anticonvulsants (lamotrigine, phenytoin, carbamazepine, phenobarbital), sulfonamide antibiotics, and oxicam NSAIDs.
- Reactions to medications occur in early treatment, typically occurring within the first 2 mo of initiation.
- Infectious etiologies: *Mycoplasma pneumoniae*, cytomegalovirus.
- Pathogenesis not completely understood; keratinocyte apoptosis attributed to cytotoxic T cells and natural killer cells through release of cytokines and cytotoxic proteins (granulysin, Fas-ligand, perforin, TNF-alpha).

Usual Treatment

- Depending on severity of disease and pt comorbidities, consider transfer to burn unit or ICU.
- If medication is suspected trigger of disease, attempt to identify and discontinue culprit drug.
- Similar to pts with major burns, treatment mainly consists of supportive care:
  - Wound care and eye care.
  - Pain management.
  - Fluid resuscitation, thermoregulation, and correction of electrolyte imbalances.
  - Nutritional support.
  - Monitoring for and treatment of superinfections.

Assessment Points

System	Effect	Assessment by Hx	PE	Test
HEENT	Conjunctivitis, corneal ulceration Stomatitis, mucositis, pharyngeal erosions	Eye pain, photophobia Oral pain, odynophagia, impaired oral intake	Purulent discharge, corneal ulceration Oral/mucosal friability	Obtain baseline ophthalmologic exam
RESP	Erosions of trachea and bronchi Respiratory failure (pulm edema, pneumonia, infiltrates)	Dyspnea, cough, hemoptysis, hypoxemia	Tachypnea, pulm. consolidation, rales	CXR, CT scan ABG, bronchoscopy
CV	Hypovolemia Sepsis and septic shock	Dizziness, decreased urine output Lethargy, confusion	Tachycardia, hypotension, oliguria Fever	BP, CBC/BMP, lactate, ECG Blood culture—bacteremia (especially with *Staphylococcus aureus, Pseudomonas aeruginosa*)
GU	Urethritis Genital erosions	Difficulty voiding, dysuria Genital pain	Urinary retention Vulvar/vaginal bullae	Bladder scan, UA/culture Early gynecologic exam
HEME	Anemia Leukocytosis	Fatigue	Pallor, tachycardia Fever	CBC, differential Blood culture, CXR, urine culture, thorough dermatologic exam
METAB	Electrolyte abnormalities Insulin resistance Hypoalbuminemia			CMP Albumin

Key References: Rabito SF, Sultana S, Konefal TS, et al: Anesthetic management of toxic epidermal necrolysis: report of three adult cases, *J Clin Anesth* 13(2):133–137, 2001; Saeed H, Mantagos IS, Chodosh J: Complications of Stevens-Johnson syndrome beyond the eye and skin, *Burns* 42(1):20–27, 2016.

Perioperative Implications

Preoperative Preparation

- Correct preexisting electrolyte imbalances.
- Ensure adequate fluid resuscitation, as increased water loss from exfoliated skin can occur.
- Anticipate challenges with vascular access.
- Aim to minimize further cutaneous injury by placing soft foam or gel padding on OR table.
- Maintenance of normothermia is a challenge; transport pts to and from the OR with warm blankets and increase ambient OR temperature.

Airway

- Minimize upper airway instrumentation.
- Use extreme caution with friable oral and pharyngeal mucosal surfaces.
- Avoid nasal airway.

Preinduction/Induction

- Lubricate face and face mask prior to preoxygenation, and apply face mask with gentle pressure.
- Skin trauma can occur from tape, blood pressure cuffs, tourniquets, and adhesives (ECG leads, securing IV catheters).
- Use soft padding under blood pressure cuffs, nonadhesive pulse oximeters, and limit use of tape to secure IV or intra-arterial catheters. (Consider suturing in place or using gauze wrap.)
- Administer prophylactic antibiotics appropriate to surgical procedure; routine systemic antibiotic therapy not recommended in SJS or TEN unless there is evidence of superinfection.
- Similar to burn injured pts, SJS/TEN pts at risk for hyperkalemia if given succinylcholine.
- Meticulous ocular care and lubrication with eye drops or ointments.
- Anticipate difficulty securing and stabilizing endotracheal tube.

Maintenance

- Maintain normothermia; pts prone to heat loss from epidermal loss.
- Minimize conductive, convective, and evaporative heat loss by maintaining warm OR temp, using warming blankets, fluid warmers, and wrapping extremities with thermal insulation.
- Respiratory mucosal sloughing leading to tracheal or bronchial obstruction can occur and be life threatening; consider fiberoptic scope in the OR to aspirate bronchial casts and assess airway involvement.
- Monitor for adequate fluid resuscitation.

Extubation

- Use care with oropharyngeal suctioning to avoid further mucosal damage.
- Decision to extubate or not should be based on degree of airway involvement and intraop course.

Postoperative Period

- Increased susceptibility to infection
- Pain management

Anticipated Problems/Concerns

- Morbidity is worse in pts with tracheal or bronchial epithelial involvement.
- For pts that survive, long-term sequelae are common and primarily involve the skin, eyes, oral cavity, and teeth. Pulm complications occur, in addition to genital and urinary symptoms in female pts.

Sturge-Weber Syndrome

Puneet Khanna | Renu Sinha

Risk

- Incidence: 1 in 5000.
- Prevalence: No racial or sex prediction; sporadically occurring neurocutaneous syndrome.

Perioperative Risks

- Increased risk of seizures, neurologic deficits, bleeding due to presence of angiomas involving the oral cavity, vascular abnormality, and congenital cardiac malformations.

Worry About

- Seizures, mental retardation, neurologic deficits, headache
- Congenital glaucoma, retinal detachment
- Difficult airway
- Intracerebral angiomas

Overview

- Described by Sturge (1879) and Weber (1929).
- Also known as encephalotrigeminal angiomatosis.
- Involves a triad of (1) vascular malformation (port wine stain); (2) leptomeningeal angioma; and (3) vascular malformation of the eye.
- Facial, extrafacial, and bilateral port wine stain, along with hypertrophy of the facial soft tissue and facial bone: