BACKGROUND

Spinal Muscular Atrophy in Pregnancy

What is SMA?

Genetic Basis

- Autosomal recessive mutation of the SMN1 (survival motor neuron 1) gene
- Loss of SMN1 causes deficiency of survival motor neuron protein
- Progressive degeneration of anterior horn neurons in the spinal cord

Clinical Manifestations

- Progressive proximal muscle weakness and atrophy
- Respiratory insufficiency — most commonly restrictive lung disease
- Skeletal deformities including progressive scoliosis and joint contractures

SMA Classification

- Type 0: Typically causes mortality in the first month of life
- Type 1: Onset <6 mo; may never sit
- **Type 2: Onset 6–18 mo; sits but never walks independently; wheelchair-dependent**
- Type 3: Onset >18 mo; ambulation possible
- Type 4: Adult onset; mildest form

Nishio H et al. Spinal Muscular Atrophy: The Past, Present, and Future of Diagnosis and Treatment. Int J Mol Sci. 2023 Jul 26;24(15):11939. doi: 10.3390/ijms241511939. PMID: 37569314; PMCID: PMC10418635.

SMA & Pregnancy

Limited Clinical Data

- Limited data exists on pregnancy course and outcomes in patients with SMA
- A retrospective case series (Bencivenga et al., 2023) described pregnancy experience in women with SMA Types 2 & 3, supporting thoughtful neuraxial use in selected patients

Anesthetic Challenges

- Respiratory: baseline insufficiency increases risk of general anesthesia and post-operative respiratory failure
- Neuraxial: scoliosis, prior spinal surgeries, and patient positioning create anatomic challenges for spinal/epidural placement

Obstetric Considerations

- Preterm delivery more common due to respiratory limitations and disease burden
- Cesarean delivery often required given difficulty with bearing down in advanced disease
- Often requires expert consultation and collaboration: Maternal-Fetal Medicine, Anesthesiology, Pulmonology, Neurology

Bencivenga RP et al. Pregnancy experience in women with spinal muscular atrophy: a case series. Acta Myol. 2023;42(2-3):60-64.

CASE PRESENTATION

SMA Type 2 in a Pregnant Patient Undergoing Cesarean Delivery

Patient Profile

27-year-old female, G1P0
Primigravida — primary scheduled c-section at 36w6d gestation

Primary Diagnosis
Spinal Muscular Atrophy, Type 2

Comorbidities
- Wheelchair-dependent functional status
- Severe mixed obstructive & restrictive lung disease
- Severe scoliosis s/p thoracolumbar spinal fusion to L5
- Needle phobia — required inhalational induction of GA in the past
- Significant traumatic stress from lifelong medical interventions

Medical Management

Pre-Delivery Counseling (28 weeks gestation)
- Early planning with MFM, PAT Clinic (Anesthesiology), Pulmonology, and Neurology

Patient-Centered Shared Decision Making
- Thorough discussion of anesthetic options, risks, benefits, and patient goals
- Patient elected to pursue neuraxial anesthesia

Trauma-Informed Care
- Pre-delivery exposure therapy for needle phobia
- Planned early admission for appropriate pre-op IV access under sedation prior to scheduled cesarean section

Neuraxial Ultrasound Guidance
- Pre-procedural ultrasound mapping of spinal anatomy and the only accessible interspace - L5/S1

Procedural Sedation
- Inhaled nitrous oxide and IV dexmedetomidine to facilitate spinal placement in a term pregnant patient

Outcome
- **Successful cesarean delivery under spinal anesthesia**

MFM: Maternal-Fetal Medicine | GA: General Anesthesia | s/p: status post | G1P0: Gravida 1, Para 0 | PAT: Pre-Anesthesia Testing

TEACHING POINTS

Lessons Learned from a Medically Challenging Case

Patient-Centered SDM (Shared Decision Making)

- SDM is supported by high-quality evidence in obstetric anesthesia
- An RCT demonstrated that SDM in choosing labor analgesia significantly improves childbirth outcomes[2]
- Patients who participated in SDM reported significantly higher childbirth experience scores, satisfaction, and perceived control and support compared to routine care[2]

Trauma-Informed Care

- Patients with chronic disease often carry significant medical trauma from a lifetime of interventions
- Pre-delivery exposure therapy was used to successfully address needle phobia prior to neuraxial anesthesia
- Procedural sedation, trauma-informed communication, and patient-physician rapport enabled a safe, patient-centered experience and successful outcome

Ultrasound Guidance for Neuraxial Anesthesia

- GA carries elevated risk in SMA patients with severe decrease in respiratory function
- Studies support neuraxial anesthesia in SMA Types 2 & 3, demonstrating successful deliveries with careful risk assessment and management of respiratory and spinal abnormalities[1]
- Ultrasound-guided neuraxial placement increases success with challenging spinal anatomy, including scoliosis and spinal instrumentation[1]

Multidisciplinary Collaboration

- Early multidisciplinary involvement and collaboration with the obstetrical team is essential for successful care of medically complex obstetric patients
- Thoughtful advocacy and interdisciplinary teamwork are the cornerstones of safe, effective care for medically complex pregnant patients

References

[1] Bencivenga RP, Zoppi D, Russo A, et al. Pregnancy experience in women with spinal muscular atrophy: a case series. Acta Myol. 2023;42(2-3):60-64. doi:10.36185/2532-1900-316

[2] Shahveisi M, Nourizadeh R, Mehrabi E. The effect of shared decision-making in choosing the method of labor analgesia on childbirth experience among primiparous women. PLoS One. 2023;18(2):e0274559. doi:10.1371/journal.pone.0274559