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Anesthetic Nuances Of A Patient With Acyanotic Congenital Heart Disease And Smith-Lemli-Opitz Syndrome Scheduled For Hypospadias Repair: A Case Report

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Abstract

Introduction: *Smith-Lemli-Opitz Syndrome (SLOS) is a rare autosomal recessive disorder characterized by defective cholesterol biosynthesis leading to multiple congenital anomalies including central nervous, cardiovascular, and genitourinary malformations. Patients with SLOS pose unique anesthetic challenges including susceptibility to neuromuscular hyperexcitability and malignant hyperthermia-like events.*

Main symptoms: *A 16-year-old male with SLOS and acyanotic congenital heart disease presented with dribbling of urine diagnosed as hypospadias requiring surgical repair.*

Diagnosis, intervention, outcome: *He had a small patent ductus arteriosus (PDA) with a left-to-right shunt and stable biventricular function. Anesthesia was successfully managed with total intravenous anesthesia (TIVA) using target-controlled infusion (TCI) of propofol, vecuronium muscle relaxation, and caudal block without complications. The perioperative course was uneventful, and the patient was discharged on post-operative day one.*

Conclusion: *This case highlights the importance of individualized anesthetic management in SLOS patients with congenital heart disease, avoiding volatile anesthetics and depolarizing muscle relaxants while maintaining cardiovascular stability.*

Key words :

- *Smith-Lemli-Opitz Syndrome*
- *Acyanotic Congenital Heart Disease*
- *Patent Ductus Arteriosus*
- *Hypospadias Repair*
- *Anesthetic Management*
- *Total Intravenous Anesthesia (TIVA)*

INTRODUCTION :

Smith-Lemli-Opitz Syndrome (SLOS) is a rare genetic disorder caused by mutations in the DHCR7 gene resulting in deficiency of 3β-hydroxysterol Δ7-reductase, disrupting cholesterol biosynthesis and embryonic development.(1) Clinical manifestations include multiple congenital anomalies—craniofacial abnormalities, intellectual disability, and malformations of the cardiac and genitourinary systems. Anesthetic challenges involve potential malignant hyperthermia-like neuromuscular hyperexcitability, difficult airway due to craniofacial anomalies, and cardiovascular compromise related to congenital heart defects (CHD). This report presents the anesthetic management of a 16-year-old male with SLOS and acyanotic CHD (PDA) undergoing hypospadias repair.

PATIENT INFORMATION

- De-identified specifics: 16-year-old male, 44 kg, known SLOS patient.
- Primary symptoms: Dribbling of urine for 1 month, diagnosed hypospadias.
- Medical history: Uneventful birth history, delayed motor milestones, no ICU admissions, cooperative with normal airway examination, no aggressive behavior.
- Cardiac history: Congenital acyanotic heart disease with PDA and left-to-right shunt.

CLINICAL FINDINGS

On examination, apart from hypospadias, systemic and airway assessments were unremarkable without signs of difficult airway. Echocardiography confirmed a restrictive PDA of 2.0 mm with left-to-right shunt (pressure gradient 70 mmHg), pulmonary artery and left ventricular dilation, and good biventricular function.

TIMELINE

Event	Details
Birth and developmental history	Uneventful birth; delayed motor milestones
Presentation	Dribbling urine, diagnosis of hypospadias
Preoperative evaluation	Normal labs, echocardiogram revealing PDA
Surgery date	Scheduled for hypospadias repair
Anesthesia	TIVA with TCI propofol, vecuronium, caudal block
Postoperative care	Stable vitals, extubated, PACU monitoring
Discharge	POD-1

DIAGNOSTIC ASSESSMENT

- Routine blood tests: Within normal limits
- Echocardiography: PDA with left-to-right shunting (PG=70 mmHg) and cardiac chamber dilation
- Airway exam: Normal parameters, no anomalies

THERAPEUTIC INTERVENTION :

- Anesthesia induction: Fentanyl 2 mcg/kg, lignocaine 1.5 mg/kg, propofol 2 mg/kg, vecuronium 0.1 mg/kg
- Airway management: Endotracheal intubation with 6.5 mm cuffed tube
- Maintenance: Target-controlled infusion of propofol titrated to entropy (40-60)
- Regional anesthesia: Caudal block with 22 ml of 0.25% bupivacaine for analgesia
- Monitoring: ASA standard monitors, entropy, temperature vigilance; avoided volatile agents and depolarizing muscle relaxants to reduce risk of hypermetabolic events

FOLLOW-UP AND OUTCOMES:

- Intraoperative course stable without hypermetabolic signs or rigidity
- Extubated uneventfully after reversal
- PACU monitoring for 2 hours with stable vitals
- Discharged on postoperative day 1 without complications

DISCUSSION:

Patients with SLOS present with unique anaesthetic challenges. SLOS is a rare genetic disorder of cholesterol metabolism resulting in the deficiency of 3β hydroxy sterol Δ7 - reductase, a key enzyme in the biosynthesis of cholesterol.(1) This enzyme defect leads to reduced cholesterol levels and accumulation of precursor sterols, disrupting cellular membrane stability, myelination, and embryogenesis. As a result, patients frequently present with a range of congenital malformations affecting the central nervous system, cardiovascular system, gastrointestinal tract, and genitourinary tract. In our case, the presence of hypospadias necessitated surgical correction.

The most critical perioperative consideration is the potential susceptibility to malignant hyperthermia. Neuromuscular hyperexcitability, which mimics or predisposes to malignant hyperthermia (MH), can also be

seen in patients with SLOS. This hyperexcitability manifests as increased tone and episodic rigidity. Although the precise process is unclear, it is believed that altered cholesterol content in cellular membranes affects calcium processing and causes muscle cells to become hyperexcitable. There have been published reports in literature describing perioperative complications in SLOS patients, including muscle rigidity, temperature instability (both hyperthermia and hypothermia), and difficult intubation. Most events were associated with the use of volatile anaesthetics and, in some cases, depolarising muscle relaxants. Given this concern, we avoided volatile anesthetics and depolarizing muscle relaxants even though true MH susceptibility is not well established.(2) In our case, we used target-controlled infusion (TCI) of propofol, ensuring precise control of anaesthetic depth, aided by entropy monitoring. The use of vecuronium, a non-depolarising muscle relaxant with minimal cardiovascular effects, further enhanced safety. This approach aligns with the anaesthetic technique described by Govindarajan et al., who successfully managed a 3-year-old child with SLOS undergoing hypospadias repair using total intravenous anaesthesia (TIVA) and vigilant temperature monitoring while avoiding inhalational agents. (1)

The presence of a patent ductus arteriosus (PDA) with a left-to-right shunt (PG = 70 mmHg) and pulmonary artery dilatation introduced additional challenges. In our patient although the PDA was restrictive, the elevated pressure gradient indicated a significant left-to-right shunt, posing a risk of pulmonary overcirculation and left ventricular volume overload. Our management focused on maintaining systemic vascular resistance to limit excessive shunting, while avoiding hypoxia, hypercarbia and acidosis to prevent increases in pulmonary vascular resistance. We also carefully titrated propofol using entropy, which helped us to avoid excessive depth that could lead to vasodilation and a fall in systemic vascular resistance, thereby ensuring stable haemodynamics throughout the procedure.(3)

Regional anaesthesia, administered via a caudal block, offered effective intraoperative and postoperative analgesia, minimized opioid requirements, and reduced the risk of developing respiratory depression.(4)

Airway anomalies such as micrognathia, high-arched palate, or cleft palate are frequently described in SLOS and may contribute to difficult airway management. However, in our patient, airway examination was reassuring, and no difficulty was encountered with mask ventilation or intubation. Intellectual impairment along with aggression, self-injurious behavior and hyperactivity may be seen in some patients with SLOS. Our patient was cooperative and did not show aggressive behavior.

PATIENT PERSPECTIVE

The patient was informed clearly about the anesthetic plan and associated risks preoperatively. He remained cooperative and reported satisfactory perioperative experience with no distress or awareness of any complications.

INFORMED CONSENT

Written informed consent was obtained from the patient's parents after a detailed discussion of anesthetic management and potential risks.

This case contributes to limited clinical data guiding anesthetic care in SLOS patients, highlighting avoidance of triggering agents, depth-monitored TIVA, and vigilant cardiovascular management in the presence of congenital defects. Further studies are warranted to develop standardized protocols.

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