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Case Report

Anesthetic Challenges in a Pediatric Patient with GORAB Mutation: A Case of Bilateral Orchiopexy

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Abstract

Geroderma Osteodysplastica (GO), associated with a GORAB gene mutation, is a rare genetic disorder characterized by severe connective tissue abnormalities, including skin laxity, osteopenia, and premature aging features. Anesthetic and perioperative management in these patients presents significant challenges due to skin fragility, skeletal vulnerability, and nutritional deficiencies. Individualized, multidisciplinary care is essential to prevent iatrogenic complications. We report the anesthetic management of an 8 kg, 2-year-old male child with a confirmed GORAB gene mutation (NM_152281.3) scheduled for bilateral orchiopexy. The child presented since birth with skin atrophy, reduced subcutaneous fat, malnutrition, and low BMI. A multidisciplinary preoperative plan was crucial, and nutritional optimization was initiated. Specific measures for tissue protection included adhesive-free monitoring (silicone electrodes) and gel supports at pressure points to prevent injury. Anesthesia was induced with sevoflurane. Airway management utilized a video laryngoscope for intubation, and spinal anesthesia was administered post-intubation for superior analgesia. Intraoperative care focused on maintaining normothermia, neutral positioning, and careful handling to mitigate the risk of iatrogenic fractures due to osteopenia. **Conclusion:** Postoperative recovery was uneventful, and skin integrity was perfectly preserved. Anesthetic management of pediatric patients with GORAB mutations demands meticulous planning and a tailored approach focused on skin and bone protection to ensure a safe perioperative course.

Keywords: Geroderma osteodysplastica; GORAB mutation; pediatric anesthesia; perioperative management; skin fragility; osteopenia; nutritional optimization; orchiopexy; sevoflurane induction.

INTRODUCTION

Cryptorchidism is the most frequent congenital issue with male genitalia, happening in around 3% of full-term male births. About 80% of newborns will experience spontaneous resolution and testicular descent by the time they reach three months old ¹. Surgery is recommended for testicles that haven't descended by 6 months of age or later ². Different diagnostic methods and imaging, along with how to manage bilateral impalpable testes have been proposed but still its diagnosis and treatment present a distinct challenge ³. Recent developments involve laparoscopic orchiopexies, the application of internal abdominal testicular fixation techniques, and updated recommendations about the best timing for corrective surgery.

Anesthesia management in pediatric patients with rare genetic disorders requires careful preoperative planning and multidisciplinary coordination to ensure safety and optimal outcomes ⁴. The GORAB (NM_152281.3) gene mutation, associated with geroderma osteodysplastica,

presents unique anesthetic challenges due to features such as skin laxity, osteopenia, poor subcutaneous fat, and potential airway anomalies ⁵. These children are often underweight and malnourished, further complicating perioperative care ⁶. This case report describes the comprehensive anesthetic management of a 2-year-old child weighing 8 kg with a confirmed GORAB mutation undergoing bilateral orchiopexy. The approach focused on tailored airway strategies, fragile skin precautions, bone protection, and individualized fluid and analgesic plans, highlighting the importance of anticipating systemic manifestations and adapting standard protocols to meet the specific needs of syndromic pediatric patients.

CASE PRESENTATION

A 2-year-old male child, weighing 8 kg, previously diagnosed at the age of 8 months with a GORAB gene mutation (NM_152281.3), was scheduled for bilateral orchiopexy. The patient exhibited characteristic clinical features including skin laxity, poor subcutaneous fat, dysmorphic facial features, and signs of malnutrition,

reflected by a low body mass index (BMI) for his age (Fig. 1).

The diagnosis was initially suspected at 8 months of age based on clinical presentation, including skin laxity, poor weight gain, dysmorphic features, and marked reduction in subcutaneous fat. These findings prompted a comprehensive evaluation. Whole exome sequencing confirmed a pathogenic mutation in the *GORAB* gene, consistent with gerodermia osteodysplastica, a rare autosomal recessive connective tissue disorder. The diagnosis was further supported by metabolic and genetic assessments conducted by a specialized pediatric genetics team.

A preoperative evaluation was conducted to assess anesthetic and surgical fitness. The child was alert, responsive, and free of active infections or systemic illnesses. Despite marked skin laxity and minimal soft tissue coverage, cardiopulmonary and neurological examinations were within normal limits. Laboratory investigations, including complete blood count (CBC), serum electrolytes, and renal and hepatic function tests, were all within normal ranges.

Although his weight remained significantly below the normal percentile for age, nutritional status had shown improvement following targeted feeding interventions since diagnosis.

Preoperative evaluation involved a multidisciplinary approach. A consultation with the genetics team confirmed the *GORAB* mutation and its associated features, including skin laxity and osteopenia. Nutritional assessment revealed malnutrition, prompting the initiation of high-calorie supplements in preparation for surgery. The pediatric team evaluated the child's developmental milestones and ruled out the presence of any cardiac or respiratory comorbidities. The patient observed a fasting period of 6 hours for food and 2 hours for clear liquids prior to surgery. Airway evaluation was conducted thoroughly due to the anticipated risk of a difficult airway. However, micrognathia and cervical spine instability were excluded through physical examination and imaging. Clinical assessment of the airway was performed, and no imaging was deemed necessary for this purpose. As a precautionary measure, a video laryngoscope was kept ready for airway management.



Fig.1: 2-Year-Old Child with *GORAB* presenting with characteristics consistent with Gerodermia Osteodysplastica (*GORAB* Mutation). Note the appearance of thin, loose skin and reduced subcutaneous fat on the limbs, particularly around the hands which are key features of the gerodermia (skin laxity) associated with this rare connective tissue disorder.

Given the patient's skin fragility, extra care was taken to avoid injury. Silicone-based electrodes (such as Kendall™ ARBO) were used for ECG monitoring to reduce the risk of skin damage from adhesives. Gel padding was applied to pressure points including the sacrum, heels, and occiput for additional protection during the procedure.

Anesthesia was induced with inhalational sevoflurane at 8% in a 50% oxygen/air mixture to minimize airway irritation. Intravenous access was established post-induction using a 24G catheter to reduce procedural distress. Airway management was successfully achieved

with the insertion of an uncuffed 3.5 mm endotracheal tube (ETT), using a C-MAC® video laryngoscope fitted with a Miller blade size 1. The ETT was lubricated with a water-soluble gel to prevent mucosal trauma.

Following intubation, spinal anesthesia was administered in the lateral position using a 23G spinal needle at the L5-S1 level, with 0.8 ml of 0.5% heavy bupivacaine. General anesthesia was maintained with 2% sevoflurane in 50% air, allowing for spontaneous respiration throughout the procedure. The child was positioned supine with foam supports under the thighs and knees to prevent hip hyperextension. Neutral spine

alignment was ensured using a gel head ring and vacuum mattress for safe stabilization.

Monitoring during surgery included standard ASA monitors (ECG, SpO₂, EtCO₂, and non-invasive blood pressure) along with advanced monitoring such as esophageal core temperature and hourly blood glucose measurements. Blood glucose levels were monitored hourly during and after the surgery. A multimodal analgesia approach was employed, with intravenous acetaminophen at a dose of 15 mg/kg administered intraoperatively. NSAIDs were avoided due to the patient's nutritional status and associated risk of renal vulnerability.

Intraoperative care involved meticulous skin protection. Hydrogel ECG electrodes and cloth tape were used to secure lines without adhesives. A forced-air warmer set to 38°C was employed to maintain normothermia. Due to the child's osteopenia and potential for bone fragility, all transfers were performed using a two-person lift technique to minimize fracture risk. Fluid therapy included a balanced crystalloid solution administered at 4 mL/kg/h, supplemented with 5% dextrose to prevent hypoglycemia.

Extubation was performed while the child was deeply anesthetized to prevent coughing and airway irritation. The patient was transferred to the post-anesthesia care unit (PACU) in a lateral position. The spinal anesthesia provided effective postoperative analgesia for approximately eight hours. The child remained free of pain and did not require any additional analgesic doses. Rescue analgesia with intravenous fentanyl at 0.5 mcg/kg was available but was not required. Postoperative monitoring revealed intact skin integrity with no evidence of blisters or erythema. The hospital stay was uneventful, lasting one day. The child was discharged on the second day post-surgery. Respiratory function remained stable with SpO₂ maintained at 98% on room air and no signs of stridor. The child's hospital course was uneventful, leading to discharge on the second postoperative day, further highlighting the success of the tailored approach.

DISCUSSION

Undescended testicles are a common condition, happening in around 3% of full-term baby boys, and can occur in up to 30% of premature male infants. The right side is more frequently involved in unilateral cryptorchidism⁷. The 28th week of pregnancy is often when testicular descent into the inguinal canal begins. 90% of undescended testes can be identified in the inguinal canal, and about 70% can be felt⁸. Indirect inguinal hernias or patent processus vaginalis occur concurrently in over 90% of patients with cryptorchidism. Reduced male fertility (particularly in cases of bilateral cryptorchidism), testis atrophy, testicular torsion, elevated risk of traumatic injury, and an increased chance of testicular cancer are among the long-term consequences of untreated cryptorchidism⁹. Our patient presented with bilateral cryptorchidism.

Patients who have undescended testes face a higher risk of infertility, testicular cancer in both testes, and

testicular torsion, particularly if they do not receive treatment¹⁰. The likelihood of developing testicular cancer is 40 times higher than in the general population, but it is still considered rare¹¹. The testis on the opposite side, even when it's in the right place in the scrotum, has around a 20% higher risk of cancer compared to the general population¹¹. Our patient was diagnosed with GORAB (NM_152281.3) gene mutation.

This gene encodes a protein belonging to the golgin family, a group of coiled-coil proteins primarily associated with the Golgi apparatus¹². The protein is believed to play a role in the secretory pathway and may also participate in mitosis, as it interacts with the N-terminal kinase-like protein and localizes to both the Golgi and cytoplasm¹³. Mutations in this gene have been linked to geroderma osteodysplastica, a rare connective tissue disorder¹³. Additionally, multiple alternatively spliced transcript variants of this gene have been identified.

GORAB gene mutations cause geroderma osteodysplastica, a condition that commonly shows up in children with considerable skin laxity and fragility. This increases their risk of iatrogenic damage including blistering during perioperative care, pressure ulcers, and epidermal tears. Strongly advised are adhesive-free or low-trauma monitoring devices such as silicone-backed tapes and hydrogel-based ECG electrodes in order to reduce this danger¹⁴. Soft gel cushioning at pressure sites including the sacrum, heels, and occiput and silicone-based electrodes helped to greatly reduce skin damage¹⁴. These techniques are compatible with skin-protective procedures applied in other fragile-skin diseases, such as epidermolysis bullosa, where even little adhesive stress can cause severe blistering and delayed healing¹⁴.

Because of inadequate fat distribution and feeding difficulties, malnutrition is often linked with connective tissue diseases and progeroid syndromes¹⁵. The low body mass index and lowered subcutaneous fat in this youngster revealed a catabolic condition that might impact immunological competency, anaesthetic medication metabolism, and wound healing¹⁵. High-calorie preoperative dietary supplements targeted at stabilising the child's metabolic reserve and lowering perioperative morbidity. Particularly in undernourished children undergoing major surgery, studies in paediatric surgical populations reveal that focused nutritional care improves postoperative outcomes¹⁵. Moreover, keeping euglycemia intraoperatively using glucose-containing fluids helped to meet metabolic needs and lower the hypoglycemia risk.

Because of defects in connective tissue, patients with GORAB mutations commonly show osteopenia and increased bone fragility. This calls for cautious placement, little manipulation, and fracture prevention techniques used under anaesthesia and during surgery. To reduce mechanical strain on delicate bones in the reported case, neutral spine alignment, hoover mattress use, and a two-person transfer approach were applied. Research on perioperative treatment of patients with cryptorchidism and related disorders underlines the

need of supportive positioning devices and atraumatic handling to prevent pathological fractures ¹⁶. For children with bone sensitivity resulting from rare genetic abnormalities, these ideas are absolutely essential for guaranteeing safe anaesthetic and surgical care.

CONCLUSION

Anesthetic management of pediatric patients with GORAB mutations demands meticulous planning, attention to skin and bone protection, and nutritional optimization. This case underscores the importance of a tailored, multidisciplinary approach to ensure a safe perioperative course.

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