International Research Journal of Multidisciplinary Scope (IRJMS), 2024; 5(2): 535-539

Case Study | ISSN (O): 2582-631X DOI:10.47857/irjms.2024.v05i02.0559

Anesthetic Challenges and Goals in a 17-Year-Old Patient with Nail Patella Syndrome Undergoing Emergency Open Appendectomy: A Case Report

Bharat Krishna M*, Niranjni S, Navaneetha Krishnan M, Lakshmi R

Department of Anesthesiology, Saveetha Medical College, SIMATS, India. *Corresponding Author's Email: malar_udt@yahoo.co.in

Abstract

This case report delves into the intricate anesthetic management of a 17-year-old male presenting Nail Patella Syndrome (NPS) and end-stage renal failure necessitating emergency open appendectomy. NPS, an uncommon hereditary disorder, manifests with distinct abnormalities affecting the nails, patellae, elbows, and iliac crests, thereby complicating perioperative care. The patient's extensive medical history, notably including hypertension and chronic renal disease, posed formidable challenges during anesthesia administration. Addressing these complexities required a meticulous approach, tailoring anesthetic goals to mitigate risks while striving for optimal outcomes. Despite the inherent intricacies associated with NPS, successful perioperative management was realized through careful planning and execution of anesthesia protocols. This case underscores the critical role of individualized care and interdisciplinary collaboration in navigating the multifaceted challenges posed by rare genetic conditions, especially in emergency surgical scenarios. By shedding light on the nuanced considerations and strategies employed in this instance, this report contributes valuable insights to the broader discourse surrounding the management of NPS patients undergoing surgical interventions, enriching our understanding of effective approaches in emergency settings.

Keywords: Anesthetic management, Emergency appendectomy, Hypertension, Nail Patella Syndrome, Renal failure.

Introduction

Nail Patella Syndrome (NPS), also known as hereditary onycho-osteodysplasia, stands as a rare autosomal dominant genetic disorder characterized by a constellation of musculoskeletal and renal abnormalities (1). It is a clinically heterogeneous disorder primarily attributed to genetic alterations in the LMX1B gene on chromosome 9q34.1, which plays a pivotal role in limb development (2, 3) The syndrome's hallmark features include dystrophic or absent nails, hypoplastic or absent patellae, posterior iliac horns (Fong prongs), and a spectrum of renal anomalies (4, 5). Beyond the musculoskeletal manifestations, patients with NPS may also encounter complications such as renal osteodystrophy, glaucoma, and nephropathy, significantly impacting their quality of life (6, 7)

This case report endeavors to illuminate the intricate interplay among Nail Patella Syndrome (NPS), advanced renal disease, and the imperative for surgical intervention. While the existing literature acknowledges the musculoskeletal and renal dimensions of NPS, a noticeable dearth exists in comprehensive accounts addressing anesthetic considerations (8, 9), especially within the context of emergency appendectomy. The rarity of this presentation is underscored by the infrequency of Nail Patella Syndrome, rendering the scenario of a patient with NPS necessitating an emergency open appendectomy an uncommon occurrence. This case offers valuable insights into the specific challenges encountered during anesthesia in emergency surgical scenarios involving individuals with complex genetic disorders.

The complexity of the patient's medical history, encompassing hypertension, CKD stage 5, and musculoskeletal anomalies, mandates a collaborative, multidisciplinary approach. Anesthesiologists, nephrologists, and orthopedic surgeons must work in tandem to comprehend the nuances of perioperative management, thereby

(Received 29th January 2024; Accepted 21st April 2024; Published 30th April 2024)

optimizing patient outcomes in the face of multifaceted medical challenges. The contribution of this case report lies in its response to the limited literature surrounding Nail Patella Syndrome, particularly in the realm of emergency appendectomy. By presenting a comprehensive account of the challenges and strategies entailed in the perioperative care of these patients, this report augments the existing body of knowledge.

In light of the increasing awareness of genetic disorders and their implications for perioperative care, this case report assumes clinical relevance. It serves as a pertinent exploration of the unique considerations inherent in managing emergencies in patients with Nail Patella Syndrome. The insights derived from this case have the potential to inform future clinical practices and contribute to the formulation of guidelines for anesthetic management in similar scenarios.

Case Presentation

A 17-year-old male presented to the Emergency Room with complaints of dull aching abdominal pain, vomiting (4-5 episodes), and fever. Due to the severity of symptoms, the patient was scheduled for an Emergency Open Appendicectomy. The patient's past medical history revealed a diagnosis of Nail Patella Syndrome (NPS) with bilateral medical renal disease, specifically focal segmental glomerulosclerosis (FSGS), at CKD-Stage 5. The patient had been on hemodialysis three times per week since the age of five, and despite ongoing renal care, he had been anuric for the past 12 years. An arteriovenous (AV) fistula in the left forearm facilitated hemodialysis, and the last session was completed the day prior to the appendectomy.

Upon examination, the patient, with a height of 151 cm and weight of 38 kg, demonstrated vital signs showing a heart rate of 96/min, blood pressure (BP) of 220/130 mmHg measured in the right upper limb in a sitting posture, and oxygen saturation (SpO2) of 96% in room air. The patient was under a medication regimen that included T.Amlodipine 5mg BD, T.Clonidine 0.1mg BD, T.Carvedilol 6.25mg BD, T.Sodium bicarbonate 500mg TDS, and T.UbideCarenone HS.

During the immediate preoperative period, vital signs changed, with a heart rate of 105/min, BP of 215/150 mmHg (MAP: 171 mmHg), and SpO2 of 97% in room air. Premedication included Inj.Midazolam 1mg and Inj.Fentanyl 120mcg, leading to induction with Inj.Propofol 50mg and Inj.Atracurium 20mg. Peripheral access was established using a wide-bore IV cannula, and the patient was intubated with a size 6.5 endotracheal tube. Intraoperatively, Inj.Dexa 4mg, Inj.Paracetamol 500mg, and Inj.Emeset 4mg were administered. The patient received a total fluid input of 200ml, experienced a blood loss of 150ml, and received a USG-guided right-side Ilioinguinal Iliohypogastric block with 10ml of 0.25% Bupivacaine and 1% lignocaine for postoperative analgesia. The reversal of anesthesia was achieved using Inj.Neostigmine 2.5mg and Inj.Glycopyrrolate 0.5mg, and the patient was successfully extubated and later discharged.

This case presents challenges in anesthetic management due to the patient's underlying hypertensive state. Hemodynamic volatility in hypertensive patients during induction and intubation necessitates careful consideration. A suggested approach involves intra-arterial catheter placement followed by a multimodal induction, incorporating transient β-blockade with esmolol. Strategies include a loading dose of 500–1,000 mcg/kg/min esmolol over 1 min, followed by a 50 mcg/kg/min infusion rate. Additionally, other agents such as Glyceryl Trinitrate, Sodium nitroprusside infusion, and clevidipine can be employed to achieve rapid arterial dilation and blood pressure reduction. These considerations are crucial to ensure hemodynamic stability during the perioperative period, especially in the context of emergency surgery for patients with complex medical histories like Nail Patella Syndrome and advanced renal disease (Figure 1 and 2).

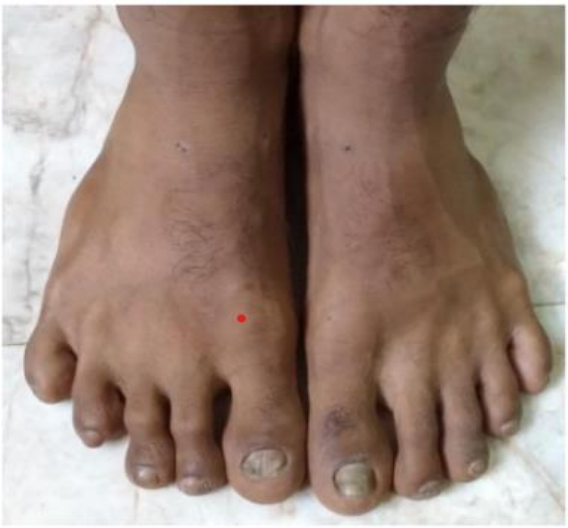

Figure 1: Clinical Presentation of Nail Patella Syndrome (Fong's Disease / Turner-Kieser Syndrome)

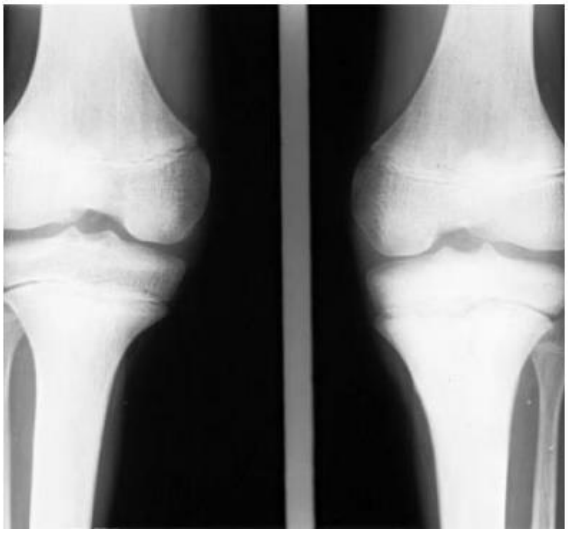

Figure 2: Radiological Manifestations of Nail Patella Syndrome: Exploring Hypoplastic/Absent Patella and Posterior Iliac Horns

Discussion

The intricate case of a 17-year-old male with Nail Patella Syndrome (NPS) undergoing emergency open appendectomy amid advanced renal disease presented multifaceted challenges, necessitating a meticulous approach to perioperative planning. The dynamic interplay of hypertensive crises, chronic kidney disease (CKD) stage 5, and the distinctive features of NPS mandated a thoughtful and multidisciplinary strategy.

The preoperative blood pressure of 220/130 mmHg indicated severe hypertension, prevalent in NPS with renal involvement. To mitigate hemodynamic instability risks during induction and intubation, an intra-arterial catheter facilitated real-time blood pressure monitoring. Esmolol, a short-acting β-blocker, was administered during induction to temper the hypertensive response, averting potential complications like cardiovascular strain, increased bleeding, and cerebrovascular events.

Standard anesthesia practice employed Inj.Propofol and Inj.Atracurium for smooth induction and intubation, aligning with established protocols. Careful titration of anesthetics, particularly in hypertensive patients, ensured a seamless process without significant vital sign fluctuations. Intraoperative medications, including Inj.Dexa, Inj.Paracetamol, and Inj.Emeset, targeted pain management, analgesia, and prevention of postoperative nausea, respectively.

Judicious fluid use, limited to 200ml, factored in the patient's anuric state and CKD. A balanced fluid approach aimed to maintain hemodynamic stability without compromising renal function. Appropriate management of the recorded blood loss (150ml) prevented excessive bleeding, averting complications such as anemia and postoperative issues.

The decision to administer a USG-guided right-side Ilioinguinal Iliohypogastric block with 0.25% Bupivacaine and 1% lignocaine played a pivotal role in postoperative pain management. This regional anesthesia approach minimized systemic opioid use, mitigating the risk of respiratory depression and opioid-related side effects, crucial in the context of the patient's renal dysfunction.

Utilizing Inj.Neostigmine and Inj.Glycopyrrolate for neuromuscular blockade reversal facilitated smooth extubation. Swift reversal is paramount to prevent postoperative residual paralysis, especially critical in patients with compromised respiratory function due to conditions like CKD.

Inadequate hypertensive management during induction and intubation posed severe risks, including myocardial infarction, cerebrovascular accidents, or hemorrhage. The absence of real-time

blood pressure monitoring heightened the risk of missing critical changes, potentially leading to cardiovascular collapse or organ damage. Inadequate fluid management could contribute to fluid overload, exacerbating hypertension and compromising renal function. The absence of appropriate postoperative analgesia might intensify pain, leading to increased sympathetic activity and aggravating hypertension.

According to the paper titled "Nail-Patella syndrome: a case report and anaesthetic implications, a 64-year-old guy with NPS, renal impairment, and coronary artery disease had asystole under combination epidural and general anaesthesia. The research reviewed the several abnormalities related with NPS and emphasised the significance of knowledge in anaesthesia treatment for these patients" (10).

Another paper titled "Nail-patella syndrome." The article "Overview of Clinical and Molecular Findings delves into the uncommon autosomal dominant condition that causes glaucoma and progressive nephropathy, among other defects. The discovery of the causal gene, LMX1B, sparked more research into the molecular pathophysiology of kidney disease in NPS. Despite the absence of compelling evidence for a relationship between LMX1B genotype and the severity of renal and extrarenal abnormalities, the study adds useful insights into the clinical and molecular aspects of NPS" (11).

The study titled "Kidney disease in nail-patella syndrome identified NPS as a pleiotropic autosomal-dominant illness caused by mutations in the gene LMX1B. Emphasizing the many forms of NPS nephropathy, the study indicated that worldwide podocyte dysfunction, driven by LMX1B mutations and other genetic variables, might lead to end-stage kidney failure" (12).

The research "Nail-patella syndrome and its association with glaucoma: a review of eight families found an elevated risk of glaucoma in Australian families with NPS. This highlights the importance of frequent glaucoma tests for NPS patients" (13).

A research titled "Knee-Ding a Diagnosis: A Case of Nail Patella Syndrome" examined a 54-year-old lady with NPS who was identified later in life, emphasising the necessity of early detection through signs and symptoms. The case report emphasised the importance of taking proactive steps to avoid recurring subluxations or dislocations and to preserve patellar integrity" (14).

Another research, "Nail patella syndrome: a rare cause of renal failure in a young adult, described a 22-year-old man who had NPS and developed renal failure. Emphasizing the importance of thorough information regarding NPS components for effective diagnosis and early identification of additional system involvements" (15).

The comprehensive approach to hypertensive management, anesthetic techniques, fluid and blood loss management, and regional anesthesia collectively contributed to a successful outcome in our complex case. The multidisciplinary strategy, tailored to the patient's unique medical history, prevented potential complications, optimizing postoperative recovery. The integration of our findings with insights from these reference studies enriches our understanding of the challenges posed by NPS, providing valuable knowledge for future perioperative care considerations in similar patients.

Conclusion

In conclusion, the case of a 17-year-old male with Nail Patella Syndrome (NPS) undergoing emergency open appendectomy emphasizes the need for a meticulous, individualized approach to perioperative care. Managing the intricate interplay of NPS, advanced renal disease, and urgent surgery demanded careful planning. Key strategies included controlling severe hypertension during induction and intubation, using an intra-arterial catheter and administering esmolol to mitigate hypertensive crises. Anesthetic techniques, fluid management, and regional anesthesia contributed to a smooth intraoperative course, minimizing the impact on renal function and ensuring hemodynamic stability. Collaboration among a multidisciplinary team was crucial in navigating the patient's complex medical history. This case underscores the importance of tailored perioperative strategies for patients with rare genetic disorders and advanced renal disease, highlighting the efficacy of a collaborative, patient-centric approach in optimizing outcomes during emergency surgical interventions.

Abbreviation

NPS: Nail Patella Syndrome
CKD: Chronic Kidney Disease
BP: Blood Pressure
SpO2: Oxygen Saturation
IV: Intravenous

Acknowledgement

We would like to express our gratitude to the patient and their family for their cooperation and consent for this study. We also acknowledge the healthcare professionals involved in the care of the patient.

Author Contributions

Nil

Conflict of Interest

The authors declare no conflict of interest.

Ethics Approval

Ethical approval for this study was obtained from the Institutional Ethics Committee of Saveetha Medical College.

Funding

No external funding was received for this study. The study was conducted as part of routine clinical practice.

Reference

1. Sweeney E, Fryer A, Mountford R, Green A, McIntosh I. Nail patella syndrome: a review of the phenotype aided by developmental biology. Journal of medical genetics. 2003; 40(3): 153–162.
2. Lee BH, Cho TJ, Choi HJ, Kang HK, Lim IS, Park YH, Ha IS, Choi Y, Cheong HI. Clinico-genetic study of nail-patella syndrome. Journal of Korean medical science. 2009; 24 Suppl:S82-S86.https://doi.org/10.3346/jkms.2009.24.S1.S82.
3. Chaturvedi S, Pulimodd A, Agarwal I. Hypoplastic nails, bowed elbows, and nephrotic syndrome. Nail-patella syndrome hereditary osteo-onychodysplasia, Turner-Keiser syndrome, Fong disease. Quiz page. American journal of kidney diseases: the official journal of the National Kidney Foundation. 2013; 62(6): A25–A27.
4. Price A, Cervantes J, Lindsey S, Aickara D, Hu S. Nail-patella syndrome: clinical clues for making the diagnosis. Cutis. 2018;101(2):126-129. PMID: 29554154.
5. Lo Sicco K, Sadeghpour M, Ferris L. Nail-patella syndrome. J Drugs Dermatol. 2015;14(1):85-6. PMID 25763426.
6. Lovelace PD, May LA. Nail-Patella Syndrome. Treasure Island FL: StatPearls Publishing; 2023 Jan-. Available from: https://www.ncbi.nlm.nih.gov/books/NBK559190/.
7. Dunston JA, Reimschisel T, Ding YQ, Sweeney E, Johnson RL, Chen ZF, McIntosh I. A neurological phenotype in nail patella syndrome NPS patients illuminated by studies of murine Lmx1b expression. European journal of human genetics. EJHG. 2005; 13(3): 330–335.
8. Sherf O, Nashelsky Zolotov L, Liser K, Tilleman H, Jovanovic VM, Zega K, Jukic MM, Brodski C. Otx2 Requires Lmx1b to Control the Development of Mesodiencephalic Dopaminergic Neurons. PloS one. 2015; 10(10): e0139697.
9. Harita Y, Kitanaka S, Isojima T, Ashida A, Hattori M. Spectrum of LMX1B mutations from nail-patella syndrome to isolated nephropathy. Pediatric nephrology Berlin, Germany. 2017; 32(10): 1845–1850.
10. Hennessey TA, Backman SB, Meterissian SH, Schricker T. Nail-Patella syndrome a case report and anesthetic implications. Canadian journal of anaesthesia Journal canadien d'anesthesie. 2007; 54(10): 835–839.
11. Bongers EM, Gubler MC, Knoers NV. Nail-patella syndrome. Overview on clinical and molecular findings. Pediatric nephrology Berlin, Germany. 2002; 17(9): 703–712.
12. Lemley KV. Kidney disease in nail-patella syndrome. Pediatric nephrology Berlin, Germany. 2009;, 24(12): 2345–2354.
13. Mimiwati Z, Mackey DA, Craig JE, Mackinnon JR, Rait JL, Liebelt JE, Ayala-Lugo R, Vollrath D, Richards JE. Nail-patella syndrome and its association with glaucoma: a review of eight families. The British journal of ophthalmology. 2006; 90(12): 1505–1509.
14. Dzhugarian S, Hamamah S, Frugoli A, Shepard A. Knee-Ding a Diagnosis: A Case of Nail Patella Syndrome. Cureus. 2023; 15(11): e48805.
15. Senguttuvan NB, Sivaraman A, Kandasamy D, Marimuthu K. Nail patella syndrome a rare cause of renal failure in a young adult. The Pan African medical journal. 2011; 9: 31.