CASE REPORT

Anaesthetic Management of An Adult Patient with Homocystinuria and Dystonia Undergoing Orthopaedic Surgery: A Case Report

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ABSTRACT

Homocystinuria is a rare inherited metabolic disorder associated with increased perioperative risk due to hypercoagulability and metabolic stress. Anaesthetic management in adult patients undergoing non-ocular surgery has rarely been reported. We describe the anaesthetic management of a 29-year-old man with homocystinuria, marfanoid features, and dystonia who underwent elective orthopaedic foot surgery. Perioperative care focused on minimizing thromboembolic and metabolic risks through reduced fasting duration, maintenance of normothermia and adequate hydration, avoidance of nitrous oxide, and use of mechanical thromboprophylaxis. General anaesthesia was administered using a laryngeal mask airway with sevoflurane and atracurium. The intraoperative and postoperative courses were uneventful, and the patient was discharged on the same day of the surgery.

This case highlights that with careful, physiologically guided planning, safe anaesthesia can be achieved in adult patients with homocystinuria, even in resource-limited settings.

Key Words*: Anaesthetic Management; Case Report; Homocystinuria; Hypercoagulability; Orthopaedic Surgery.*

Introduction

Homocystinuria is a rare autosomal recessive disorder of methionine metabolism associated with multisystem involvement, including ocular, skeletal, neurological, and vascular abnormalities, and an increased perioperative anaesthetic risk due to hypercoagulability and endothelial dysfunction[1,2]. From an anaesthetic perspective, elevated homocysteine levels are linked to an increased risk of thromboembolism, metabolic instability, and adverse neurological outcomes, particularly during general anesthesia[3]. Anaesthetic management is further complicated by skeletal abnormalities, neurological manifestations, and the need to avoid nitrous oxide, which can exacerbate hyperhomocysteinemia[3]. The published literature on anaesthesia in homocystinuria is limited and largely confined to paediatric ophthalmic procedures, with few reports addressing adult patients undergoing non-ocular surgery. We report the anaesthetic management of a 29-year-old man with homocystinuria and dystonia who underwent elective orthopaedic foot surgery.

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Case Presentation

A 29-year-old man with a known diagnosis of homocystinuria presented for the elective correction of left foot hammer toes (Figure 1). His medical history was notable for dystonia with intermittent left-sided limb tremors, resulting in a progressive functional impairment. He had previously undergone nasal surgery under general anaesthesia in adulthood and ophthalmic surgery in childhood, both without any perioperative complications. There was no history of thromboembolic events, seizures, fractures, or allergies to drugs.

On examination, the patient had a marfanoid habitus and mild intellectual developmental delay, with intermittent tremors in the left upper and lower limbs. Airway assessment revealed a Mallampati class III view with upper dental cap. Cardiovascular and respiratory examinations were unremarkable, and the baseline vital signs were within normal limits.

Laboratory investigations were within normal limits, except for a markedly elevated plasma homocysteine level of 31.7 µmol/L (5.46-16.2

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µmol/L). The patient was under regular metabolic follow-up for homocystinuria and was receiving vitamin supplementation as part of his long-term management. Standard metabolic therapy includes pyridoxine (vitamin B6), folate supplementation, and dietary modification to reduce homocysteine levels. Perioperatively, continuation of metabolic therapy and avoidance of factors that could increase homocysteine levels were emphasized. Adequate hydration and minimization of fasting were also implemented to reduce metabolic stress. Electrocardiography and chest radiography showed no abnormalities. (Table I)

The patient was scheduled for a left second-to-fourth metatarsal tenotomy with distal interphalangeal joint excision and K-wire arthrodesis. To minimize fasting-related metabolic stress, the procedure was scheduled first on the operating list and planned as a day-case surgery. Preoperative fasting followed ASA guidelines, permitting clear fluids up to 2 h and solids up to 6 h before anaesthesia. Intravenous access was established upon admission, and maintenance hydration was initiated with Ringer's lactate at 70 mL/h.

Standard ASA monitoring was performed before anaesthesia induction. Following preoxygenation, general anaesthesia was induced with nalbuphine, midazolam, propofol, and atracurium. A laryngeal mask airway was used for airway management, and anaesthesia was maintained with sevoflurane in an oxygen–air mixture, avoiding nitrous oxide. Normothermia was maintained using forced air warming, and mechanical thromboprophylaxis was applied throughout the procedure. The hemodynamic parameters remained stable, and blood loss was minimal. Warmed intravenous fluid was administered intraoperatively.

Neuromuscular blockade was reversed at the conclusion of the surgery, and the patient was transferred to the recovery area in a stable condition. Postoperatively, early mobilization and oral hydration were encouraged in this patient. The postoperative course was uneventful, and the patient was discharged on the same day with analgesics, vitamin supplementation, and follow-up instructions provided. Postoperative radiographs confirmed satisfactory alignment following K-wire fixation of the lesser toes (Figure 1)

Table I: Baseline Laboratory Investigations

Investigation	Result	Reference range
Hemoglobin	14.8 g/dL	13.0–17.0 g/dL
Total leukocyte count	11,290 /µL	4,000–11,000 /µL
Platelet count	214,000 /µL	150,000–450,000 /µL
Serum sodium	137 mEq/L	135–145 mEq/L
Serum potassium	4.3 mEq/L	3.5–5.0 mEq/L
Serum chloride	99 mEq/L	98–107 mEq/L
Serum bicarbonate	20 mEq/L	22–29 mEq/L
Blood urea nitrogen	12 mg/dL	6–20 mg/dL
Serum creatinine	0.73 mg/dL	0.72–1.25 mg/dL
Estimated GFR (CKD-EPI)	143 mL/min/1.73 m²	>60 mL/min/1.73 m²
Prothrombin time	10.2 s	9.5–11.7 s
INR	0.95	0.8–1.3
Activated partial thromboplastin time	25.0 s	24.8–36.2 s
AST (SGOT)	33 U/L	≤50 U/L
ALT (SGPT)	19 U/L	≤50 U/L
Alkaline phosphatase	64 U/L	40–130 U/L
Total bilirubin	0.56 mg/dL	0.2–1.2 mg/dL
Direct bilirubin	0.23 mg/dL	≤0.30 mg/dL
Gamma-GT	13 U/L	≤60 U/L
Plasma homocysteine (fasting)	31.7 µmol/L	5.46–16.2 µmol/L

Figure 1: Postoperative radiographs of the left foot (lateral and dorsoplantarviews) demonstrating K-wire fixation following corrective surgery for hammer toe deformities.

Discussion

Homocystinuria poses significant anaesthetic challenges due to hypercoagulability, metabolic vulnerability, and multisystem involvement.[4,5] Most published anaesthetic reports involve paediatric patients undergoing ophthalmic procedures, while adult patients requiring non-ocular surgery, particularly orthopaedic procedures associated with venous stasis, are rarely described.[6] This case highlights the safe perioperative management of an

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adult patient with homocystinuria who underwent elective foot surgery.

In addition to metabolic considerations, our patient had dystonia and limb tremors, which influenced perioperative communication, positioning, and postoperative mobilization efforts. Neurological manifestations are less frequently emphasized in anaesthetic reports of homocystinuria but are clinically relevant, particularly in adult patients, and warrant individualized perioperative planning.

Thromboembolism remains the principal cause of perioperative morbidity in patients with homocystinuria.[5] In this case, risk mitigation focused on nonpharmacological strategies, including minimization of fasting, maintenance of adequate hydration and normothermia, application of mechanical thromboprophylaxis, and early postoperative mobilization. These measures have been consistently recommended in previous reports and were effective in our patient despite the absence of pharmacological anticoagulation.[7] Given the patient's lack of prior thrombotic events, normal cardiac evaluation, and moderate duration of surgery, a non-pharmacological approach was considered appropriate. In addition to thromboembolic complications, several other systemic features of homocystinuria may influence anaesthetic management. Connective-tissue abnormalities such as marfanoid habitus, osteoporosis, and joint laxity increase the risk of fractures and positioning injuries during surgery, necessitating careful handling and padding[8]. Skeletal abnormalities and dental issues may also contribute to airway management challenges, particularly in patients with kyphoscoliosis or limited cervical mobility. Neurological manifestations including developmental delay, tremors, dystonia, and seizures can complicate perioperative communication, positioning, and postoperative recovery. Awareness of these multisystem features is therefore essential for anaesthesiologists when planning individualized perioperative care in patients with homocystinuria.

Avoidance of nitrous oxide was a key anaesthetic decision, as nitrous oxide inhibits methionine synthase, a vitamin B12 dependent enzyme involved in homocysteine metabolism, leading to further accumulation of homocysteine and potential neurological and cardiovascular complications.[3] The sevoflurane-based anaesthetic technique allowed for stable intraoperative conditions without metabolic disturbance.

Airway management can be challenging in patients with marfanoid features; however, for this short orthopaedic procedure, a laryngeal mask airway provided a safe and effective alternative to endotracheal intubation. Overall, this case demonstrates that with careful physiologically guided planning, anaesthesia can be safely administered to adult patients with homocystinuria, even in resource-limited settings such as ours.

Conclusion

This case demonstrates that adult patients with homocystinuria can safely undergo non-ocular orthopaedic surgery when perioperative management is guided by the physiological principles. Key strategies include the avoidance of nitrous oxide, minimization of fasting, maintenance of normothermia and hydration, and use of mechanical thromboprophylaxis. Careful anaesthetic planning is essential to reduce thromboembolic and metabolic risks in this population.

Consent statement: Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

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CONFLICT OF INTEREST

Authors declared no conflicts of Interest.

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DATA SHARING STATEMENT

The data that support the findings of this study are available from the corresponding author upon request.