

# Anaesthetic Challenges of a Paediatric Patient with Harlequin Ichthyosis

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## Abstract

Harlequin Ichthyosis (HI) is a rare form of congenital ichthyosis and these patients are at an increased risk of sepsis, respiratory failure, dehydration, and thermoregulatory dysfunction. We present a case of a 2-year-old child with active upper respiratory tract infection and background of harlequin ichthyosis for emergency upper limb surgery under general anaesthesia at our tertiary paediatric hospital. This rare case highlights the importance of a multi-disciplinary approach to perioperative management of harlequin patients undergoing surgery. Key learning points with regards to temperature monitoring, eye care, securing intravenous access and airway management are demonstrated.

**Keywords:** Harlequin Ichthyosis, paediatrics, anaesthesia

### Introduction

Harlequin Ichthyosis (HI) is a rare autosomal recessive form of congenital ichthyosis associated with a mutation in the gene ABCA12. This mutation leads to a defect in intracellular lipid deposition, resulting in disruption of the skin barrier [1]. Patients present at birth with thick plates of stratum corneum separated by deep fissures covering the entire skin surface, with complications of restricted chest wall excursion and digital necrosis [2]. These patients are at an increased risk of sepsis, respiratory failure, dehydration and thermoregulatory dysfunction [3]. We present a case of a child with HI who underwent an emergency upper limb surgery under general anaesthesia at our tertiary paediatric hospital, the anaesthetic challenges faced and the steps are undertaken to overcome them.

### Case Report

The patient was a 2-year-old female toddler with a known history of HI, who was admitted for a right Type IV Monteggia radio-ulnar mid-shaft fracture with radial head dislocation secondary to a mechanical fall. She was listed for an emergency

intramedullary pinning of the right radius and closed reduction of the right radial head under general anaesthesia.

Her clinical diagnosis of HI was made at birth. Routine antenatal checks were unremarkable, and she was delivered at 40 weeks gestation via elective caesarean section for breech position. At birth, she had severe ectropion, eclabium, alopecia and thick plate-like scales covering her body and limbs. Her initial stay in the Neonatal Intensive Care Unit (NICU) was complicated by difficult venous access, skin infection, dry gangrene of the digits with subsequent auto-amputation of both hands. The medical management of her HI comprised intensive skin emollients, oral retinoids (acitretin 1.25mg/kg/day) and regular eye drop for lubrication. Confirmatory gene testing revealed the presence of a mutation in the ABCA12 gene. She had been on regular follow-up with a dermatologist since her discharge from NICU, with satisfactory control of her ichthyosis.

During the preoperative assessment, it was noted that she had an acute upper respiratory tract infection (URTI) with a 7-day history of clear rhinorrhoea, productive cough, and positive sick contacts. She was afebrile and haemodynamically stable with normal saturation on room air. Her cardiorespiratory examination was unremarkable with no lower respiratory tract signs. Bilateral ectropion, eclabium and generalized erythematous skin with thick scaling

and fissuring over the face, neck, trunk, and extremities were noted. Mouth opening was limited at 2 fingers-breadth due to bilateral deep perioral fissures. Her Mallampati score was III with adequate thyromental distance and neck mobility.

She had been fasted for 7 hours prior to surgery and was deemed to have low aspiration risk. As such, general anaesthesia was induced with sevoflurane 8% in an air-oxygen mixture and intravenous access achieved thereafter. A lubricated size 2 Proseal Laryngeal Mask Airway (LMA)<sup>©</sup> was inserted and anaesthesia maintained with 2-3% Sevoflurane with pressure support ventilation via the circle circuit.

Due to the presence of bilateral eye ectropion and eyelid contractures, her eyes remained exposed in sleep. Vidisic<sup>®</sup> gel and a cotton gauze covering were applied for lubrication and eye protection. Passive and active warming techniques were employed including an underbody Bair Hugger<sup>®</sup> blanket and occlusive plastic coverings. A nasopharyngeal temperature probe was inserted for intra-operative temperature monitoring.

Dermatology input was sought on wound dressings, with a choice of Urgotul<sup>©</sup> dressing after application of tetracycline 3% ointment to her wounds.

At the conclusion of the surgery, special care was taken when removing the diathermy pads and ECG leads. White soft paraffin was applied over her body and lips for further lubrication. Deep extubation was performed

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**Figure 1:** From clockwise top left to right: (LMA)© secured with ties, saphenous vein cannulation under ultrasound guidance which was later secured with occlusive dressings and xray of patient's right arm.

**Table 1: Intraoperative Temperature trend**

Time (Min)	0	15	30	45
Temperature (°C)	34.4	34.5	34.5	34.6

and the patient was transferred to the Post Anaesthesia Care Unit (PACU). The temperature in the PACU was 36.1 degrees Celsius. The patient was subsequently discharged well to the general ward.

### Discussion

Patients with HI have distinctive features which pose multiple unique anaesthetic challenges. In our case, this included (i) potentially difficult venous access, (ii) difficulty in monitoring with electrodes and securing of tapes due to the oily nature of applied emollients, (iii) thickened, fragile, non-elastic skin resulting in ectropion and eclabium and (iv) thermoregulatory dysfunction. In addition, further risks were posed by the active URTI, which predisposed her to a higher risk of intra and postoperative respiratory complications, and the emergency status of her condition, which incurred the risk of aspiration.

With an ongoing URTI, minimal airway intervention and an intravenous induction technique were chosen in order to reduce the risk of laryngospasm or bronchospasm [4]. Furthermore, as with any trauma situation, there is a risk of aspiration due to inadequate fasting and delayed gastric emptying, for which there exists recommendations for a rapid sequence induction which requires intravenous access [5].

In her case, the decision for an inhalational induction technique was carefully balanced. She was deemed to have low aspiration risk clinically as she was adequately fasted and

sustained minimal trauma. Her father reported easy venous access by the phlebotomist at her regular outpatient follow-up visits, despite her history of bilateral hand auto-amputation and thick keratotic skin which obscured visibility of any superficial veins. Hence, we opted to conduct an inhalational induction technique with Sevoflurane 8% in 100% oxygen. To facilitate rapid venous access, ultrasound-guided venous access was planned and made ready prior to induction. The choice of the venepuncture site was also carefully selected, given her bilateral upper limbs auto-amputation. Her father had reported the use of brachiocephalic veins by the clinic phlebotomist, but in this author's experience, the saphenous vein anterior to the medial malleolus is often readily accessible and palpable. This was our first choice [6].

We achieved venous access on our first attempt under ultrasound guidance and was able to secure it with Tegaderm® as per usual practice, and further wrapped it with gauze circumferentially to prevent it from slipping. The hyperkeratosis in HI results in the epidermis being prone to skin fissuring, increasing the risk of infection from the compromised skin barrier. To mitigate this, we wrapped cotton gauze loosely under the non-invasive blood pressure cuff to minimise skin trauma induced by the 5-minute cycling. The liberal use of emollients, the fragility of the skin and constant flaking of the skin in such children make securing the adhesive tapes and ECG electrodes difficult. It might

be a challenge to monitor the ECG in these patients due to the application of protective ointments interfering with ECG discs' adherence to the skin. In this situation, one may consider the use of staple electrode over dressing material or esophageal ECG similar to that used in burns patients [7,8].

The tapes for securing the (LMA)© were poorly adherent in her case, hence the use of ties. White soft paraffin was used to lubricate her lips to reduce the risk of skin trauma.

The abnormal skin also resulted in eye ectropion. This child required regular nightly application of eye-drops. Intra-operatively, Vidisic® eye gel was used to lubricate the eyes to reduce the risk of corneal abrasions and ocular injury.

Due to the presence of skin contractures, transfer of such patients should be undertaken with care and minimised as far as possible. Steps were taken intra-operatively to ensure adequate padding and positioning of the child to reduce skin abrasions and any strain on the joints.

The compromised skin barrier also compromises thermoregulatory dysfunction and necessitates keeping the child warm pre-operatively while in the holding area, as well as intra and postoperatively [9]. In her case, we opted to use an underbody Bair® Hugger blanket and occlusive plastic coverings in order to conserve body heat, while maintaining operating theatre ambient temperature at 22 degrees Celsius and monitoring core temperature with a nasopharyngeal temperature probe. Despite the initial thermoregulatory disturbances due to the induction of anaesthesia, she was able to quickly resume normothermia and remained so in recovery after surgery.

Restricted chest excursion and lung compliance may be an issue in such patients and puts them at an increased risk of respiratory failure and postoperative respiratory complications. In our case, she had adequate lung compliance with good ventilation while on minimal pressure support ventilation and PEEP +5.

### Conclusion

In conclusion, this report highlighted the multiple anaesthetic challenges faced when a paediatric patient with Harlequin Ichthyosis required emergency limb surgery and outlined how these challenges were circumvented.

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