

Cracks That Speak Of Survival; Challenges In Management And Care Of Harlequin Ichthyosis- A Case Report

Bijaya Nayak, Sheeba Anitha Rani*, Yerni Jyothi Kolli, Prathima Prakasam

Child Health Nursing Department, Driems University, School of Nursing, Kotasahi, Tangi, Cuttack, Odisha 754022

ABSTRACT

Harlequin Ichthyosis is a rare, severe, autosomal recessive congenital disorder of keratinization caused by mutations in the **ABCA12 gene**, leading to defective lipid transport and abnormal skin barrier formation. Affected neonates present with thick, armor-like plates of skin separated by deep fissures, resulting in high risk of dehydration, hypothermia, infection, respiratory compromise, and electrolyte imbalance. Early diagnosis through genetic testing and clinical evaluation is crucial. Management requires intensive neonatal care, including maintenance of a warm humidified environment, strict infection control, fluid and electrolyte monitoring, nutritional support, and specialized skin care with emollients. Systemic retinoids may be considered under expert supervision to improve skin flexibility. Despite high mortality in the past, advances in neonatal intensive care and multidisciplinary interventions have improved survival rates. This study highlights the importance of comprehensive nursing care in optimizing outcomes for neonates with Harlequin Ichthyosis.

Keywords: Autosomal recessive disorder, Keratinization abnormality, Skin barrier dysfunction.

INTRODUCTION

Case Scenario:

A male neonate was delivered at 38 weeks' gestation via cesarean section to consanguineous parents with a history of one previous child affected by Harlequin Ichthyosis. At birth, the infant presented with thick, shiny, plate-like scales covering the entire body, deep fissures, ectropion, eclabium, and restricted limb movement. The newborn was immediately admitted to the NICU.

1. Introduction

Harlequin Ichthyosis is a rare and severe inherited disorder of Keratinization in which the newborn's skin becomes extremely thick, hard, and dry. The skin develops large, diamond- or plate-like scales separated by deep cracks. Because of severe damage to the skin barrier, affected newborns are at high risk of Dehydration, Infection, Hypothermia, Respiratory problems, and Electrolyte imbalance.^{1,2,5}

2. Definition

Harlequin Ichthyosis is a rare, severe autosomal recessive congenital disorder caused by abnormal formation of the skin barrier, characterized at birth by thick, armor-like plates of skin with deep Fissures over the body.^{1,2}

3. Etiology

The major cause of Harlequin Ichthyosis is a mutation in the ABCA12 gene, which plays an important role in the transport of lipids within skin cells. It is inherited in an autosomal recessive pattern, meaning that the child usually inherits one abnormal gene from each parent.^{4,5,6}

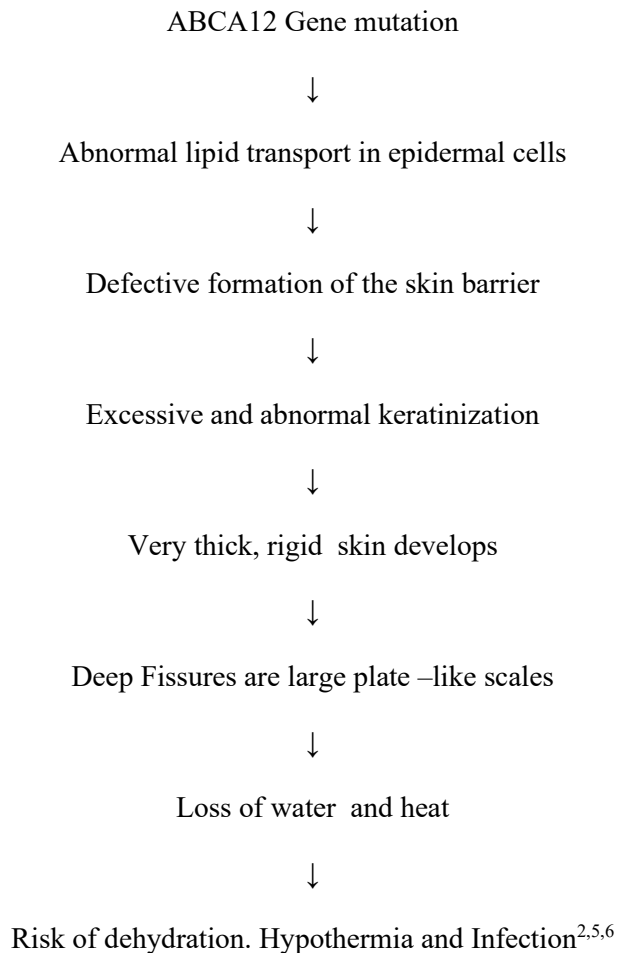
4. Risk factors

- Family history of inherited ichthyosis.
- Parents carrying a Pathogenic ABCA12 variant.
- Previous child affected with Harlequin Ichthyosis.

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- Consanguineous marriage may increase the risk of autosomal recessive disorders.^{1,2,3}

5. Pathophysiology



6. Clinical Manifestations

- Thick, Hard, Shiny skin covering most of the body.
- Large diamond-shaped or Polygonal plates.
- Deep cracks and Fissures between skin plates.
- Ectropion – turning outward of the eyelids.
- Eclabium – outward turning of the lips.
- Restricted movement of the limbs.
- Tight skin around the Fingers and toes.
- Difficulty feeding due to tightness around the mouth.
- Respiratory difficulty.
- Dehydration.
- Hypothermia.
- Increased risk of bacterial infection.
- Poor weight gain.
- Electrolyte disturbance.
- Difficulty regulating body temperature.^{1,2,3}

7. Diagnostic Evaluation

- Family and genetic history.
- Detailed physical examination.
- Genetic testing for ABCA12 mutations.

➤ Laboratory tests to monitor.

- Serum electrolytes.
- Blood glucose.
- Renal Function.
- Hydration status.
- Infection markers.

➤ Microbiological cultures when infection is suspected.^{4,5}

8. Management

A. Immediate neonatal care

- Admit the newborn to a neonatal intensive care unit (nicu).
- Maintain a warm and humidified environment.
- Monitor temperature regularly.
- Monitor heart rate, respiratory rate, oxygen saturation, and blood pressure.
- Assess hydration and urine output.

B. Skin care

- Maintain adequate humidity to reduce water loss.
- Apply appropriate bland emollients regularly to keep the skin soft and flexible.
- Avoid vigorous removal during bathing and cleansing.
- Observe for skin breakdown and infection.

C. Fluid and electrolyte management

- Monitor fluid balance carefully.
- Provide adequate oral or intravenous fluids as required.
- Monitor serum electrolytes and renal function.
- Record intake and output.

D. Respiratory care

- Assess for respiratory distress.
- Provide oxygen or respiratory support when required.
- Monitor chest movement and oxygen saturation.

E. Nutritional management

- Assess sucking and swallowing ability.
- Provide best milk when possible.
- Use assisted feeding when oral feeding is difficult.
- Monitor weight regularly.
- Provide adequate calories and fluids.

F. Infection prevention and treatment

- Maintain strict infection-control measures.
- Monitor for signs of local and systemic infection.
- Obtain cultures when infection is suspected.
- Give antibiotics when clinically indicated.

G. Systemic retinoids

Oral retinoids, particularly acitretin or isotretinoin, may be considered by specialists in severe cases. They can help reduce abnormal scaling and improve skin flexibility. Because these medicines have significant adverse effects, their use requires supervision.

9. Nursing management

I. Assessment

- Assess skin condition and extent of fissures.
- Monitor vital signs frequently.
- Assess respiratory status.
- Monitor urine output.
- Assess feeding ability.
- Monitor daily weight.

- Observe for signs of infection.
- Assess eye and mouth complications.

II. Nursing interventions

- Maintain the newborn in a warm, humidified environment as prescribed.
- Maintain strict hand hygiene and infection-control precautions.
- Apply prescribed emollients gently.
- Avoid unnecessary friction and trauma to the skin.
- Maintain adequate fluid and electrolyte balance.
- Monitor intake and output.
- Provide appropriate nutritional support.
- Monitor oxygen saturation and respiratory status.
- Provide eye care when ectropion is present according to medical orders.
- Observe for fever, lethargy, poor feeding, respiratory distress or other signs of infection.
- Administer prescribed medications safely.
- Maintain accurate documentation of all assessments and interventions.
- Provide emotional support and education to the parents.

III. Parent education

- Parents should be taught:-
- The condition is genetic and not caused by poor hygiene.
- The baby requires long-term skin care.
- Emollients should be applied as prescribed.
- Signs of infection should be reported immediately.

- Adequate feeding and hydration are important.
- Regular follow-up with dermatology and pediatrics is necessary.
- Genetic counseling may be recommended for future pregnancies.
- Parents should receive psychological support because the appearance and prolonged care needs can be distressing.

10. Prognosis

Harlequin ichthyosis was historically associated with very high neonatal mortality; with modern neonatal intensive care, humidified environments, careful fluid and nutritional management, infection prevention, skin care, and appropriate systemic treatment, survival has improved considerably. However, affected children generally require lifelong management of ichthyosis and skin-barrier problems.^{5,6}

11. Prevention

Because Harlequin ichthyosis is an inherited disorder, prevention focuses mainly on genetic counseling and reproductive options for families at risk.

- Genetic Counselling.
- Identification of the pathogenic familial variant.
- Carrier testing when appropriate.
- Prenatal diagnosis in selected pregnancies.
- Preimplantation genetic testing may be discussed with specialists.^{4,5,6,7}

12. Role of nurse

Assess the newborn: Monitor skin condition, vital signs, hydration, feeding, respiratory status, and temperature regularly.

Maintain body temperature: Keep the newborn in a warm, temperature-controlled, and appropriately humidified environment.

Provide skin care: Apply prescribed emollients gently and regularly to prevent excessive dryness and cracking.

Prevent skin injury: Handle the baby gently and avoid rubbing, peeling, or forcibly removing skin scales.

Maintain hydration: Monitor intake and output, daily weight, urine output, and electrolyte levels as ordered.

Provide nutritional support: Assess sucking and swallowing; assist with breast or alternative feeding as required.

Prevent infection: Follow strict hand hygiene and aseptic technique and observe for redness, discharge, fever, or other signs of infection.

Provide respiratory care: Monitor respiratory rate, oxygen saturation, chest movement, and signs of respiratory distress.

Provide eye and oral care: Assess for ectropion and eclabium and provide prescribed eye, lip, and oral care.

Administer medications: Give prescribed medications, including topical treatment and systemic medicines, and observe for adverse effects.

Monitor complications: Watch for dehydration, hypothermia, infection, electrolyte imbalance, feeding difficulty, and respiratory problems.

Maintain accurate records: Document assessment findings, skin care, intake/output, feeding, medication, and response to treatment.

Educate parents: Explain the condition, skin-care techniques, feeding, hydration, infection prevention measures, and warning signs.

Provide emotional support: Encourage parents to express their concerns and participate in appropriate newborn care.

Coordinate multidisciplinary care: Collaborate with pediatrician, dermatologist, nutritionists, and other health care professionals.

Prepare for discharge and follow-up: Teach parents about long-term skin care, medications, follow-up visits, and genetic counselling when appropriate.^{3,5}

Nursing diagnosis

1. Impaired skin integrity related to altered skin barrier and abnormal keratinization as evidenced by thick, dry, fissured, and scaly skin.
2. Risk for deficient fluid volume related to excessive transepidermal water loss secondary to impaired skin barrier.
3. Imbalanced nutrition: less than body requirements related to difficulty with feeding secondary to restricted oral movement, as evidenced by poor sucking reflex and poor feeding tolerance.
4. Ineffective thermoregulation related to impaired skin barrier and increased heat loss as evidenced by low body temperature, cold skin, lethargy, and poor feeding.
5. Impaired oral mucous membrane integrity related to altered skin barrier and dryness as evidenced by dry/cracked lips and restricted oral movement.^{2,3}

Nursing Care Plan – Harlequin Ichthyosis

Assessment	Nursing Diagnosis	Goals / Expected Outcomes	Nursing Interventions	Rationale	Evaluation
Thick, plate-like scales, deep fissures, ectropion, eclabium	Impaired Skin Integrity related to defective keratinization	Neonate will maintain intact skin barrier and reduced risk of infection	- Apply sterile emollients frequently	Emollients soften scales, humid environment prevents cracking, asepsis reduces infection risk	Skin remains moist, fissures reduced, no signs of infection

Assessment	Nursing Diagnosis	Goals / Expected Outcomes	Nursing Interventions	Rationale	Evaluation
			- Maintain warm, humidified incubator - Handle gently to avoid skin trauma - - Use aseptic technique during care		
Restricted limb movement due to thickened skin	Impaired Physical Mobility related to rigid skin plates	Neonate will demonstrate improved flexibility and comfort	- Encourage gentle passive ROM exercises - Collaborate with physiotherapist - Position to prevent contractures	Early mobility prevents stiffness and supports musculoskeletal development	Infant shows improved limb movement
Risk of dehydration, electrolyte imbalance	Risk for Fluid Volume Imbalance related to excessive transepidermal water loss	Neonate will maintain stable hydration and electrolyte balance	- Monitor fluid intake/output - Assess daily weight - Provide IV fluids as prescribed - Monitor electrolytes	Thickened skin increases fluid loss; monitoring prevents imbalance	Hydration and electrolytes remain within normal limits
Poor feeding due to eclabium and restricted mouth opening	Imbalanced Nutrition: Less Than Body Requirements related to feeding difficulties	Neonate will achieve adequate nutrition and weight gain	- Provide specialized feeding techniques - Use NG tube if oral feeding inadequate - Collaborate with dietician for high-calorie feeds	Feeding support ensures growth and prevents malnutrition	Infant maintains weight gain and tolerates feeds
Parents anxious due to previous affected	Anxiety (Family) related to	Parents will verbalize reduced anxiety and	- Provide emotional support - Offer counseling	Family support improves coping and reduces stress	Parents express understanding and

Assessment	Nursing Diagnosis	Goals / Expected Outcomes	Nursing Interventions	Rationale	Evaluation
child and current condition	chronic illness and prognosis	demonstrate coping	- Educate parents about condition and care - Connect with support groups		confidence in care
Risk of respiratory compromise due to restricted chest expansion	Ineffective Breathing Pattern related to rigid thoracic skin	Neonate will maintain adequate oxygenation and ventilation	- Monitor respiratory rate, effort, and O ₂ saturation - Position for optimal breathing - Provide humidified oxygen if needed	Thickened skin may restrict chest expansion; monitoring prevents hypoxia	Infant maintains stable respiratory status ²

CONCLUSION

Harlequin ichthyosis is a rare and severe congenital skin disorder that requires early recognition and intensive neonatal care. Proper management of skin integrity, hydration, body temperature, nutrition, respiratory function, and infection prevention is essential for improving comprehensive care, monitoring complications, administering prescribed treatment and education provided, and supporting the parents with appropriate multidisciplinary care and follow-up; the quality of life and survival of affected children can be improved.

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