Review Article

Management of New Born with Harlequin Ichthyosis: A Rare Disease Condition

Abstract

Harlequin ichthyosis is the most severe form of non-bullous ichthyosis, which is unusual in newborns and is usually marked by deadly excessive keratinization of the skin. Ichthyosis is a family of genetic skin disorders, characterized by dry, thickened, scaly skin with severe morbidity and mortality. The term "harlequin" derives from the facial appearance and the triangular and diamond-shaped pattern of the scaly skin. It occurs in about 1 in 300,000 births and has no known sex predilection. The disorder affects the skin in utero causing thick, horny, armoury-like plates that cover the skin with contraction abnormalities of the eyes, ears, mouth and appendages. There is no specific guideline to manage the newborn of harlequin ichthyosis, in that nursing care is most important in the initial phase. There are required multi-disciplinary teams to take care of harlequin ichthyosis.

Keywords: Care, Harlequin, Nursing Management, Case, Medical Ethics, Newborn.

Introduction

Harlequin ichthyosis is a rare genetic disorder characterized by defective keratinization and desquamation of the epidermis. The genetic mutations underlying HI result in dysfunction of adenosine triphosphate-binding cassette A12, a protein involved in intracellular lipid transport, and cause abnormal lamellar granule secretion in the epidermis. [1]

Consequently, important lipids and proteases are not secreted into the interstitial space between granular layer keratinocytes. The absence of interstitial lipid delivery results in a profoundly abnormal epidermal permeability barrier and increased transcutaneous water losses. [2]

The primary case was accounted for in 1750 by Reverend Oliver Hart. The general occurrence is 1 out of 300,000 births. Around 200 cases have been accounted for all

through the world. In 1984, a newborn child with this issue was brought into the world in Pakistan and the child lived till 2008. One more such birth was recorded in the USA in 1994. As of late, one case was accounted for in June 2016 at Nagpur, India.[3]

Signs & Symptoms

Newborn with Harlequin ichthyosis is covered in thick plate-like scales of skin. The tightness of the skin pulls around the eyes and the mouth, forcing the eyelids and lips to turn inside out, revealing the red inner linings. The chest and abdomen of the infant may be severely restricted by the tightness of the skin, making breathing and eating difficult. The hands and feet may be small and swollen and partially flexed. The ears may appear to be misshapen or missing but are fused to the head by the thick skin. Infants born with harlequin ichthyosis may also have a flat nose (depressed nasal bridge), abnormal hearing, frequent respiratory infections, and decreased joint mobility.[4]

Premature birth is typical, leaving the infants at risk for complications from early delivery. These infants are also at high risk for low body temperature, dehydration, and hypernatremia (elevated levels of sodium in the blood). Constriction and swelling of the mouth may interfere with the suck response and infants may need tube feeding. The baby's corneas need to be lubricated and protected if the eyelids are forced open by the tightness of the skin.[4]

Causes

Harlequin ichthyosis is caused by changes (mutations) in the ABCA12 gene, which gives instructions for making a protein that is necessary for skin cells to develop normally. It plays a key role in the transport of fats (lipids) to the most superficial layer of the skin (epidermis), creating an effective skin barrier. When this gene is mutated, the skin barrier is disrupted. [5]

Harlequin ichthyosis is inherited in an autosomal recessive pattern. Recessive genetic disorders occur when an individual inherits an abnormal gene from each parent. If an individual receives one normal gene and one abnormal gene for the disease, the person will be a carrier for the disease, but usually will not show symptoms. The risk for two carrier parents to both pass the abnormal gene and, therefore, have an affected child is 25% with each pregnancy. The risk to have a child who is a carrier, like the parents, is

50% with each pregnancy. The chance for a child to receive normal genes from both parents is 25%. The risk is the same for males and females. [6]

Affected Populations

Harlequin ichthyosis affects males and females in equal numbers. This condition affects approximately one in 500,000 persons or about seven births annually in the United States. [7]

Nursing Management

There's no cure for Harlequin ichthyosis, so management becomes a crucial part of the equation after initial treatment. And it's all about the skin. The skin protects the body from bacteria, viruses, and other harmful elements in the environment. It also helps to regulate body temperature and fluid loss. The most harlequin newborn will need one-on-one nursing care for the first several weeks of life. Nursing care is based on the condition of the newborn. Initial nursing care given to the newborn is spending time in a heated incubator with high humidity. Tube feeding for the prevention of malnutrition and dehydration. Apply special lubricant apply on the eye can protect to keep eyes healthy. Further management is as:

- Applying retinoids to help shed hard, scaly skin
- Applying topical antibiotics to prevent infection
- Covering the skin in bandages to prevent infection
- Placing a tube in the airway to help with breathing
- Using lubricating eye drops or protective devices on the eyes

Standard Therapies

A multi-disciplinary team is involved in the care of infants with harlequin ichthyosis as soon as they are born. This has been shown to improve outcomes and reduce complications such as respiratory distress, dehydration, electrolyte imbalances, impaired thermoregulation, systemic bacterial infections, and feeding difficulties. Early treatment with oral retinoids is also thought to improve outcomes. However, they are only used in severe cases due to their known toxicity and side effects. [8]

The thick, plate-like skin of harlequin type ichthyosis will gradually split and peel off over several weeks. Antibiotic treatment may be necessary to prevent infection at this time. Administration of oral acitretin may accelerate the shedding of the thick scales. [8]

After the thick plates peel off, the skin is left dry and reddened and may be covered in large thin scales. The skin symptoms are treated by applying skin softening emollients. This can be particularly effective after bathing while the skin is still moist. Many patients with severe ichthyosis exfoliate manually by rubbing off the thick scale with special exfoliating gloves with a rough surface. Skin barrier repair formulas containing ceramides or cholesterol, moisturizers with petrolatum or lanolin, and mild keratolytic (products containing alpha-hydroxy acids or urea) can all work to keep the skin moisturized and pliable, and prevent cracking and fissuring that can lead to infection. Removal of damaged tissue (debridement) from the fingers may be needed if they are constricted by bands of skin to avoid a loss of circulation.[9]

Conclusion

Harlequin ichthyosis, is a congenital disorder are mostly occurring in newborns. There is no cure treatment for harlequin ichthyosis. In the management of harlequin ichthyosis is multi-disciplinary action taken to care for the newborn. Nursing management is a most prime treatment for harlequin ichthyosis. An initial time of care is the most intricate time for newborn and their parents also.

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