

# Harlequin Ichthyosis is a Congenital Skin Condition that Affects Harlequins

Uppal S\*

Department of Medicine, Albany Medical College, New York, United States

## Corresponding Author\*

Uppal S  
Department of Medicine,  
Albany Medical College,  
New York,  
United States  
kibret\_m@yahoo.com

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## Editorial

A new treatment regimen is said to have significant benefits in treating eczema, a bothersome and unpleasant skin condition. Additionally, this could lead to the development of medicines to treat other atopic illnesses that are associated to eczema, such as asthma, which is strongly linked to eczema. Harlequin Ichthyosis (HI) is a hereditary skin disorder that affects just a few people. The skin of an infant is affected by HI, which poses a high risk of neonatal death due to a severe and life-threatening infection.

A mutation in the ABCA 12 gene causes HI. This gene controls the creation of the ABCA 12 protein, which transports lipids through the epidermis and is required for normal skin formation. Various mutations in ABCA 12 can occur, some of which can prevent the protein from being manufactured, while others can cause the protein to be produced in an aberrant form.

HI is an autosomal recessive disorder, which means that people who have it have two copies of the faulty genes, one from each parent. If two persons with the mutation conceive a child, the child has a 25% chance of inheriting the mutation. Parents of HI children are carriers of the mutant gene but do not show symptoms of the condition because they have one normal copy to compensate for the defective one.

The skin is the body's biggest organ. Normal skin works as a waterproof and protective barrier between the body and the environment, but it is also incredibly flexible. The skin is responsible for the sense of touch and helps to control many of the body's activities. The natural characteristics of the skin are challenged in HI when taken combined.

Premature birth is prevalent in Hawaii. Instead of a continuous sheet of thin and elastic skin, the infant is born covered in extraordinarily thick

skin that hardens into enormous plates, somewhat like armour. The presence of an excessively thicker integument that is badly broken or fissured gives this appearance. The fissures are so deep that subcutaneous tissues can be exposed.

Preterm birth, as well as the development of numerous and deep skin fissures that result in the loss of the skin's regulatory and protective activities, provide life-threatening threats to a newborn with HI.

Because of these factors, the newborn is frequently admitted to the Neonatal Intensive Care Unit (NICU) shortly after birth.

A youngster with HI usually has a deformed appearance due to the hard skin. Because the defective skin pulls the ears tightly inwards, some infants may have no visible external ears or malformed ears.

The eyes and eyelids are frequently impacted in a variety of ways. The eyelids of some people are unable to close correctly. Others have bulging eyelids that cover their eyes.

Due to such irregularities, the mucous membranes of the eyes may be permanently exposed (ectropion), resulting in dry eyes.

Because the taut skin pushes the lips outwards in a fixed grimace, some infants' eclabium, or red oral mucosa of the eye, may be revealed.

Respiratory difficulties or even respiratory collapse are common in people with HI. This is due to the tight, thick skin restricting chest movement.

Because their capacity to suck and swallow is limited, infants born with this illness may need to be fed via the nose with a tube in the early stages of life.

The typical skin features at birth or in infancy are used to diagnose HI. It may resemble other disorders, such as eczema, in milder cases.

A 3D-ultrasonography can sometimes detect HI in the womb. Neonates must be born in a moist environment and handled carefully in an isolated incubator.

Consistent application of petrolatum-based creams should keep the skin supple. Long daily baths with bath oils added to the water should be given to older children.

It is necessary to use lubricant ointments. Keratolytics are also used to peel off and reduce the thickness of the skin as a result of excessive superficial stratum corneum growth. This will improve the skin's smoothness and suppleness. In severe situations, oral retinoids can help, but they should be used with caution.

Over the course of several weeks, the heavy skin plates progressively peel away. Because of the peeling, the skin appears highly red and glossy.

Some people with limited mobility benefit from physiotherapy. Other therapies may be required; for example, counselling can assist patients in coping with the disease's psychological issues.