Harlequin Ichthyosis

Subjects: Genetics & Heredity
Contributor: Camila Xu

Harlequin ichthyosis is a severe genetic disorder that mainly affects the skin.

Keywords: genetic conditions

1. Introduction

Infants with this condition are born with very hard, thick skin covering most of their bodies. The skin forms large, diamond-shaped plates that are separated by deep cracks (fissures). These skin abnormalities affect the shape of the eyelids, nose, mouth, and ears, and limit movement of the arms and legs. Restricted movement of the chest can lead to breathing difficulties and respiratory failure.

The skin normally forms a protective barrier between the body and its surrounding environment. The skin abnormalities associated with harlequin ichthyosis disrupt this barrier, making it more difficult for affected infants to control water loss, regulate their body temperature, and fight infections. Infants with harlequin ichthyosis often experience an excessive loss of fluids (dehydration) and develop life-threatening infections in the first few weeks of life. It used to be very rare for affected infants to survive the newborn period. However, with intensive medical support and improved treatment, people with this disorder now have a better chance of living into childhood and adolescence.

2. Frequency

Harlequin ichthyosis is very rare; its exact incidence is unknown.

3. Causes

Mutations in the *ABCA12* gene cause harlequin ichthyosis. The *ABCA12* gene provides instructions for making a protein that is essential for the normal development of skin cells. This protein plays a major role in the transport of fats (lipids) in the outermost layer of skin (the epidermis). Some mutations in the *ABCA12* gene prevent the cell from making any ABCA12 protein. Other mutations lead to the production of an abnormally small version of the protein that cannot transport lipids properly. A loss of functional ABCA12 protein disrupts the normal development of the epidermis, resulting in the hard, thick scales characteristic of harlequin ichthyosis.

3.1. The gene associated with Harlequin ichthyosis

- ABCA12

4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- Harlequin baby syndrome
- HI
- Ichthyosis Congenita, Harlequin Fetus Type

References

1. Akiyama M, Sakai K, Sugiyama-Nakagiri Y, Yamanaka Y, McMillan JR, Sawamura D, Niizeki H, Miyagawa S, Shimizu H. Compound heterozygous mutations including a de novo missense mutation in ABCA12 led to a case of harlequin ichthyosis withmoderate clinical severity. J Invest Dermatol. 2006 Jul;126(7):1518-23.
2. Akiyama M, Sugiyama-Nakagiri Y, Sakai K, McMillan JR, Goto M, Arita K,Tsuji-Abe Y, Tabata N, Matsuoka K, Sasaki R, Sawamura D, Shimizu H. Mutations in lipid transporter ABCA12 in harlequin ichthyosis and functional recovery bycorrective gene transfer. J Clin Invest. 2005 Jul;115(7):1777-84.
3. Akiyama M. The pathogenesis of severe congenital ichthyosis of the neonate. J Dermatol Sci. 1999 Sep;21(2):96-104. Review.
4. Hovnanian A. Harlequin ichthyosis unmasked: a defect of lipid transport. JClin Invest. 2005 Jul;115(7):1708-10. Review.
5. Kelsell DP, Norgett EE, Unsworth H, Teh MT, Cullup T, Mein CA,Dopping-Hepenstal PJ, Dale BA, Tadini G, Fleckman P, Stephens KG, Sybert VP,Mallory SB, North BV, Witt DR, Sprecher E, Taylor AE, Ilchyshyn A, Kennedy CT,Goodyear H, Moss C, Paige D, Harper JI, Young BD, Leigh IM, Eady RA, O'Toole EA. Mutations in ABCA12 underlie the severe congenital skin disease harlequinichthyosis. Am J Hum Genet. 2005 May;76(5):794-803.
6. Moskowitz DG, Fowler AJ, Heyman MB, Cohen SP, Crumrine D, Elias PM, WilliamsML. Pathophysiologic basis for growth failure in children with ichthyosis: anevaluation of cutaneous ultrastructure, epidermal permeability barrier function, and energy expenditure. J Pediatr. 2004 Jul;145(1):82-92.
7. Richard G. Autosomal Recessive Congenital Ichthyosis. 2001 Jan 10 [updated2017 May 18]. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJH, Stephens K, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University ofWashington, Seattle; 1993-2020. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK1420/
8. Thomas AC, Cullup T, Norgett EE, Hill T, Barton S, Dale BA, Sprecher E,Sheridan E, Taylor AE, Wilroy RS, DeLozier C, Burrows N, Goodyear H, Fleckman P, Stephens KG, Mehta L, Watson RM, Graham R, Wolf R, Slavotinek A, Martin M, Bourn D, Mein CA, O'Toole EA, Kelsell DP. ABCA12 is the major harlequin ichthyosisgene. J Invest Dermatol. 2006 Nov;126(11):2408-13.

Retrieved from https://encyclopedia.pub/entry/history/show/11527