Harlequin ichthyosis

Description

Harlequin ichthyosis is a severe genetic disorder that affects the skin. Infants with this condition are born prematurely 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 in babies with harlequin ichthyosis. Affected infants also have feeding problems.

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 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.

Following the newborn period, the hard, skin plates are shed and the skin develops widespread scales and redness.

It used to be very rare for affected infants to survive the newborn period. However, with intensive medical support and improved treatment, babies with this disorder now have a better chance of living into childhood and early adulthood.

Frequency

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

Causes

Variants (also known as 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) and enzymes in the outermost layer of skin (the epidermis).

Some variants in the *ABCA12* gene prevent the cell from making any ABCA12 protein. Other variants 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 before and after birth, resulting in the severe skin abnormalities characteristic of harlequin ichthyosis.

Learn more about the gene associated with Harlequin ichthyosis

- ABCA12

Inheritance

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

Other Names for This Condition

- Autosomal recessive congenital ichthyosis 4B
- Harlequin baby syndrome
- HI
- Ichthyosis congenita, harlequin fetus type

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 4B (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0598226/)

Genetic and Rare Diseases Information Center

- Harlequin ichthyosis (https://rarediseases.info.nih.gov/diseases/6568/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Harlequin ichthyosis%22)

Catalog of Genes and Diseases from OMIM

- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 4B; ARCI4B (https://omim.org/entry/242500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28harlequin+ichthyosis%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D)

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