

What Is Harlequin Ichthyosis?

Harlequin ichthyosis (HI) is a rare skin condition. Infants with HI are born with their bodies covered in hard, thick skin that forms large diamond-shaped plates separated by deep cracks. HI is the most severe form of autosomal recessive congenital ichthyosis (ARCI).

What causes it?

HI is caused by a change or variant in a gene called ABCA12. This gene usually provides instructions for making a protein that's needed for normal skin development. Without this protein, the epidermis, or outer layer of the skin, doesn't develop normally. People with HI inherit two copies of the altered gene – one from each parent.

How common is it?

HI is very rare. According to the National Organization of Rare Disorder, about one baby in 500,000 is born with HI. Male and female infants are equally affected.

Other names you might hear it called

Autosomal recessive congenital ichthyosis (ARCI) – harlequin ichthyosis type

Congenital ichthyosis

Harlequin baby syndrome

Ichthyosis congenita

Harlequin fetus

Signs & Symptoms

Infants with HI are usually born prematurely with hard, thick, plate-like scales covering their body. Their skin is very tight, which causes the scales to crack and split. Scales on the face and chest can make it difficult for the baby to breathe and eat. Babies born with HI also usually have:

- Eyelids that are turned inside out so that the eyes can't close
- Lips that are pulled tight, so the mouth stays open; this makes it difficult for the baby to nurse or take a bottle
- A flat nose and ears that are fused to the head
- Small, swollen hands and feet
- Arms and legs with limited mobility
- Dry skin that easily becomes infected
- A lower-than-normal body temperature

Babies with HI need immediate intensive care. They may need treatment with antibiotics to prevent skin infections. Over the first few weeks of life, the hard, thick scales covering their body will gradually peel off and be replaced by thinner scales covering dry, reddened skin.

Children with HI will likely have red, scaly skin throughout their lives. They may also have:

- Thin or fragile hair due to scales on the scalp
- Reduced hearing caused by scale buildup in the ears
- Thickened, discolored, or oddly shaped fingernails and toenails
- Difficulty moving hands, arms, feet and legs because of stiff, inflexible, sometimes painful joints
- Frequent skin infections
- Inability to sweat

What to Expect

- Physical growth and development may be delayed in children with HI. Mental development, however, should be normal.
- Every person's experience with HI is unique. Many people may offer advice about skin care products and coping strategies that they have found helpful. Keep in mind that what has worked for someone else may or may not work for you/your child. You may need to try a variety of skin care products and approaches before finding the one that produces the best results for you/your child.
- It's important to find a dermatologist that knows how to treat HI, especially as it is so rare.
- Developing a daily routine to help control skin scales, itching, and skin cracking is important.
- Thick fingers and stiff knuckles can make it difficult for people with HI to grip things with their hands, while stiff knees and ankles can limit walking.

Treatments and Self-Care

Infants with HI are cared for in high-humidity incubators and fed frequently. Nurses apply moisturizers to the babies' skin. A drug called *etretinate* may be given to help remove the thick, plate-like scales covering the skin. After these scales peel off, babies may be able to go home. Thankfully advances in neonatal care and the use of etretinate have helped babies with HI survive.

The main treatments for HI are:

- Bathing frequently using a mild, soap-free cleanser to soften the skin and loosen skin scales
- Rubbing the skin lightly with a loofah, rough-textured sponge, or pumice stone to remove scales
- Applying moisturizers to the skin after bathing to reduce skin dryness and help the skin be more flexible and pliable
- In the neonatal and infantile period, drugs called retinoids can be life-saving (see below)

HI may also be treated with drugs called *retinoids*. These drugs can reduce skin scaling, but if they are discontinued the scales come back. Because long-term use of retinoids can cause serious adverse effects, doctors prescribe them only when HI symptoms are very severe.

Oral retinoids, such as Etretrate, can be lifesaving for infants with severe HI manifestations. Oral retinoids may help to reverse issues such as: constricting fingers or even compromised blood flow, chest that is so tight there is trouble breathing, and/or face that is so tight the baby cannot feed. Oral retinoids are usually temporary and tapered off by 6 months. They can be considered in older children if helpful, but often are not needed because these skin changes at birth are not recurrent.

Things to Keep in Mind

- HI is a chronic condition that needs lifelong, daily care.
- HI is not contagious. No one can catch it by touching or being close to you or to your child with HI.
- Both cold and hot weather can be challenging for people with HI.
 - In cold weather, the air is drier and the skin is likely to dry out and crack more easily. You may need to moisturize your skin even more frequently or use stronger moisturizing products.
 - In hot weather your body will overheat very quickly because it can't expel heat by sweating. To minimize overheating when outdoors, wear sun-protective clothing, apply sunscreen, and seek shade.
- People (some well meaning, others less so) may stare at you, or at your child with HI, and ask questions like "What's wrong with your skin/your child?" or "Does it hurt?"
 - Other children may tease or bully your child with HI because of their appearance.

- Explaining to people that you, or your child, were born with HI may help them to understand that you're not sick, it's not something they can catch, and that you live with the condition every day.
- Although your skin may look different from other people's, but you can still do most things that other people do.
- You are not responsible for other people's reactions to your or your child's appearance. Your skin condition does not define who you are.
- Parents of a child with HI may feel a need to be very protective of them.
 - Do your best to treat your child with HI in the same way as unaffected children in the family, while allowing for the fact that your child with HI needs a special skin care regimen and must take certain precautions (e.g., in hot or cold weather).
 - For example, hold them to the same expectations and encourage them to follow their dreams and ambitions just as you do your other children.
 - Reinforce in your child with HI that their skin condition does not define who they are or what they can achieve in their lives.

Questions to Ask

- What makes Harlequin ichthyosis different from other types of ichthyosis?
- What are the best treatments for HI?
- If it's mainly a skin condition, what makes the eyes so red?
- How can my child live a normal life with HI?
- What tends to make HI worse? Are there triggers?
- Are there activities I should avoid?
- Is sunlight harmful?
- Does HI put someone at risk for other health problems? What should we watch for?
- Where can I find support or other parents to talk to?

Sources

Dahl AA. *Ichthyosis Treatment & Management*. 2018 Aug 28. <https://emedicine.medscape.com/article/1198130-treatment#d2>

FIRST. *Harlequin Ichthyosis: A Patient's Perspective*. <https://www.firstskinfoundation.org/types-of-ichthyosis/harlequin-ichthyosis>

Healthline.com. *A Parent's Guide to Harlequin Ichthyosis*. 2018 Dec 3. <https://www.healthline.com/health/harlequin-ichthyosis>

Medline Plus Genetics. *Harlequin ichthyosis*. <https://medlineplus.gov/genetics/condition/harlequin-ichthyosis/>

National Organization for Rare Disorders. *Harlequin Ichthyosis*. <https://rarediseases.org/rare-diseases/ichthyosis-harlequin-type/>



Foundation for
Ichthyosis & Related
Skin Types

PO Box 1067, Lansdale, PA 19446-0687

Phone: 800-545-3286

Website: www.firstskinfoundation.org

Email: info@firstskinfoundation.org

Improve lives and seek cures for those affected by ichthyosis and related skin types.

January 2023