

Harlequin ichthyosis

What is Harlequin ichthyosis?

Harlequin ichthyosis (HI) is an extremely rare inherited skin condition, present from birth and lasting for the whole of a patient's life. The condition is considered to be the most serious form of all the inherited ichthyoses at present and is characterised by very dry, red skin. Infants with HI are usually born prematurely and present with thick, armour-like scales covering their skin, with deep, red cracks in between. Over time, this thick, armour-like shell will start to peel off but the skin tends to remain very red and scaly throughout life. It is possible that some babies may not survive if their symptoms are too severe. Most fatalities take place in the first few days of a baby's life, but survival rates for HI are continually improving as better treatments become available.

What are the signs?

The skin normally acts as a type of barrier, maintaining a constant body temperature, while keeping moisture in and infection out. In HI, however, this function is impaired due to the cracks in the scales and the skin becomes "leaky." This leads to a loss of heat, important proteins and moisture and makes the individual highly prone to infection.

The newborn's skin will also be very tight and this may distort facial features and cause the baby's eyes and mouth to be pulled back and turn inside out (everted). This is known as ectropion in the eyes and eclabium in the lips, making it difficult for the infant to feed properly. Their nose often appears compressed or misshapen and their hands and feet tend to be small and contracted. The rigid scales associated with HI are likely to cause the ears to fuse to the side of the head and restrict an infant's normal breathing so they may need to be put on respiratory support. Newborns must be kept in a humidified incubator and treated in the neonatal intensive care unit until they are healthy enough to go home, can feed properly and are free from infection.

Another key feature of HI is a failure to put on adequate weight and slowed growth are also signs of the condition.

How is the condition diagnosed?

Symptoms vary from person to person but there are a number of common signs that doctors will check to differentiate HI from other skin conditions.

- At or soon after birth, babies are usually red all over (erythroderma) and may require incubation to maintain their temperature and keep them hydrated.
- Babies are often premature and are covered in a thick, armour-like shell at birth.
- A small piece of skin may be taken to do a skin biopsy to check for the ABCA12 protein, which can be abnormal or sometimes absent in someone with HI. A skin biopsy is usually done by a dermatologist rather than a GP or paediatrician.
- A blood test can also be taken (and sent to a national reference laboratory) to check for a fault in the ABCA12 gene. You should speak to a geneticist about this. It is anticipated that this service will become more widely available over the next few years.
- Prenatal diagnosis of HI is now possible. Abnormalities in the ABCA12 gene can be detected molecularly or a 3D-ultrasonography (a scan of the foetus) can be used to look for features common in HI. These procedures are not routinely provided, however and patients should let their doctor know in advance if they have a history of HI in the family.

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Why is Harlequin ichthyosis sometimes misdiagnosed?

Occasionally patients are not diagnosed for months or even years as red skin can be a symptom of many other skin conditions, such as severe eczema or an immune deficiency disease. It is important that any persistently red baby is considered to have HI to avoid being misdiagnosed or incorrectly treated.

Managing Harlequin Ichthyosis

As yet, there is no cure for NS but continual lifelong care with moisturisers and anti-infection treatments should help keep the skin as healthy as possible. Success of the treatments will depend on individuals, and what works for one person may not work for another. Patients will need to consider different options to find the best for them. Nutritional advice may also be helpful.

Moisturising creams and skin treatment

It is very important that the skin is kept moisturised at all times and that greasy, moisturising creams and ointments are applied frequently throughout the day. Moisturising agents should be perfume-free and without additives to avoid any allergic reactions. Bath oils are important so that bathing does not cause drying or irritation of the skin. Bathing tends to be more beneficial than showering.

The use of systemic retinoid medicines has helped increase survival rates in newborns with HI and is often used alongside an intensive care regime while the infant is in hospital. Careful monitoring of liver function during retinoid therapy is vital.

Emollients and keratolytic creams which contain urea, and/or salicylic acid (see product's ingredients label for these) should be avoided in infants as these do not help the skin and can even make the condition worse.

Antibiotics are also important to treat secondary skin infections and gentle antiseptics can provide the skin with additional protection.

It is important to always seek advice from a specialist dermatologist before using any new cream in HI to check that it is safe.

More specific information on caring for the skin, eyes and ears in HI and other forms of ichthyosis can be found on the Ichthyosis Support Group website at www.ichthyosis.org.uk.

Diet

Due to their slowed growth, some babies with HI may need to be fed a special high protein diet to help them gain weight. This is usually given "artificially" via a tube as the baby cannot feed well enough and should be managed by a children's dietician. The feeding tube is either passed through the baby's nose into their stomach (nasogastric feeding) or through their abdominal wall, known as gastrostomy. Some babies with HI may need to be given special feeds into a vein.

Patients with HI will also be prone to dehydration and should therefore drink plenty of water. To minimise the risk of dehydration, make sure teachers and colleagues are aware of the need for additional hydration throughout the day while at school or in the workplace.

Eye treatment

It is extremely important that the eyes are kept sufficiently lubricated in babies with HI if their eyelids are everted (ectropion) and do not close properly. Some patients find that using moisturising eye drops can help prevent the eyes from becoming too dry. A Corneal ophthalmologist (eye doctor) should be contacted and minor eye surgery may be considered to try and ameliorate ectropion.

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Physiotherapy

Patients with HI often have slightly less mobility than other individuals, especially when newborn due to the armour-like membrane characteristically seen at birth. This will likely improve as the thick-skin peels but physiotherapy may be useful to help improve general mobility throughout a patient's life.

Herbal remedies

Some patients find that over-the-counter herbal remedies and dietary supplements can help but check with your doctor beforehand. Protein based supplements are most likely the best.

Personal care

Apart from keeping the skin hydrated and moisturised at all times, patients may get tired and overheated quickly when doing active sports (e.g. running, football etc). They may need to rest more than other individuals and teachers should be aware of this so they do not push children to continue participating.

Red and peeling skin, particularly on the face can be difficult for building self-confidence so individuals may need extra support. General practitioners need to understand the psychological impact of the condition and provide adequate support to the family, which may require the involvement of other health professionals.

Support

Although abnormal mental development has not been reported in children with HI, additional teaching support may be needed. Some children have difficulties with speech due to ear problems or eclabium (everted lips) and a child's progress in school may be affected due to frequent hospital visits, eye appointments or hearing checks during school hours.

For employed adults with HI, it is important to ensure their place of work knows about the condition and any additional support required. If patients are unable to work because of the condition, they may be entitled to financial support. Visit the DirectGov website Gov.uk (<https://www.gov.uk/>)

How is the condition inherited?

HI is an inherited (i.e genetic) disorder so it runs in families. However, as it is an autosomal recessive condition, HI often occurs out of the blue. Each parent will have one copy of the faulty gene. They will not have HI but they will be a carrier of the condition and may pass on their faulty gene. A child inheriting one faulty gene will be a carrier like their parent, but a child inheriting a faulty gene from each parent will have HI.

An individual with HI will always pass on a faulty copy of the gene to their children. However, unless the partner is a carrier, or has HI themselves (which is more likely to occur within the same family), their children will only be carriers and will not have HI.

It is advisable for families affected by HI to seek genetic counselling before expanding their family.

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People who care about ichthyosis

What genes are affected?

The faulty gene in HI has recently been confirmed as the ABCA12 gene. This gene codes for an important protein in the skin, responsible for transporting lipids to the skin surface and supporting the skin's protective function. Though still in the early stages, researchers are now investigating ways of replacing this missing protein, offering real hope for the future.

Further help

There are a number of online forums about HI where individuals can share their experiences and detail their own treatment recommendations. Not everything will work for everyone but they can be a good place both to receive and offer support.

Contact the Ichthyosis Support Group for information, advice, details on useful products, and to be connected with other people to share experiences and helpful advice.

To find out more about the ISG or become a member please get in touch in one of the following ways:

By Phone or Fax:

Tel: **0845 602 9202** Fax: **0560 343 8046** (on request)

By Email:

isg@ichthyosis.org.uk

By Post:

Ichthyosis Support Group
PO Box 1242, Yateley GU47 7FL

Facebook:

facebook.com/ichthyosissupportgroup

Twitter:

twitter.com/ISG_Charity