

## **Congenital Ichthyosiform Erythroderma (CIE)**

### **What is ichthyosis?**

Ichthyosis describes dry, thickened, scaly or flaky skin. There are at least 28 different ichthyosis subtypes, which are mainly inherited (have a genetic cause).

### **What is CIE?**

This is an extremely rare skin disorder characterised by abnormal scaling of the skin with underlying redness. It is estimated to occur in 1/300,000 births. Most affected individuals will be born as a “collodion baby”. See separate leaflet for further information about Collodion baby.

### **What does it look like?**

The skin appears bright red (erythroderma) and has accompanying fine white or flesh coloured scales. The scaling often involves all of the skin although the skin on the face may show milder changes. In severe cases the lower eyelids are normally pulled down and outwards giving a red appearance. The condition appears in the first few days of life and usually lasts lifelong. The severity of CIE varies quite considerably.

### **What is it caused by?**

It is caused by a genetic or inherited abnormality that affects normal skin shedding. There are a number of genes that cause CIE.

### **Are there different types of CIE?**

Yes there are. Some individuals have a very persistent erythroderma whereas in others it fades significantly as they get older. Similarly the scaling varies in severity. It may be that the “diagnosis” of CIE actually covers a number of different distinct conditions known collectively as ARCI (Autosomal Recessive Congenital Ichthyosis). This may be clarified by a blood and/or genetic diagnosis.

### **Classification of CIE.**

CIE is part of the group of ichthyoses known as Autosomal Recessive Congenital Ichthyoses, known by the acronym ARCI. This is a varied group and includes other ichthyoses such as Lamellar ichthyosis (LI) and the more severe Harlequin Ichthyosis (HI). This group is classified not only by its skin features, but also by the underlying genetic causes and the appearance of the skin cells under the microscope. LI is the most common clinical type and over half of ARCI patients have this form of ichthyosis. However, many patients do not exactly fit either the LI or CIE description as they may have features of both conditions. Therefore, it can be useful to think of these two conditions as different ends of a spectrum.

### **What are the signs?**

Under normal circumstances, the skin acts as a type of barrier, maintaining a constant body temperature, while keeping moisture in and infection out. In CIE, however, this function is impaired and the skin becomes “leaky.” This leads to a loss of heat and moisture and makes the individual prone to infection. Sometimes babies with this condition have to stay in a hospital incubator for a while to prevent dehydration or infection and allow their skin to be kept moist at all times. The first year of life is the critical period and some children are often admitted to hospital with repeated infections and dehydration.

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Whether or not they are born with a “collodion membrane”, babies with CIE can appear “scalded” as their skin can be red (erythema). When the membrane is shed, the scaling associated with CIE is generally whiteish in colour and may resemble “fish-scales.” The scale is often large or “plate-like”, can affect the whole body, but may be larger on the legs. Patients with CIE may have thickened skin on the palms and soles of their feet (keratoderma) which may be mild but can be very thickened with painful cracks. Nails may often be thickened and in very severe cases, the rate at which nails grow may be affected.

It is possible for CIE to cause problems in other areas of the body because the skin is so tight. The eyelids are often pulled outwards (everted), known as ectropion, which can cause the patient’s eyes to dry out. This may also make it hard for patients to sleep as the eyelids do not close properly. Reduced flexibility in the fingers and some hair loss (alopecia) has also been seen in CIE. As the thickened scaling can affect the way the sweat glands under the skin work, some patients with CIE may not sweat easily so can suffer from heat intolerance. They may overheat easily in hot weather or whilst doing sports. Cracking or splitting of the skin may lead to bacterial skin infections which could become generalised if not treated.

### **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 CIE from other skin conditions.

- For the first few days or weeks following birth, babies with CIE have a “collodion membrane”, a clear but tight film which covers the baby’s skin. (This condition is also seen in other forms of ichthyosis).
- The skin will have large, whiteish “scales” all over the body which have a “plate-like” or “fish-scale” appearance. Thickening of the palms and soles of the feet is possible. There is likely to be obvious redness of the skin.
- A small piece of skin (a skin biopsy) may be taken to check for certain proteins commonly associated with CIE, such as transglutaminase 1 (TGM1). However, problems with these proteins can also be seen in other forms of ichthyosis. 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 certain genes. Mutations in 11 genes are currently known to cause ARCI with TGM1 and ALOX12B being the most common. However, some people with ichthyosis will have faults in genes that are not yet known about, and this test may be negative.

### **Why is CIE sometimes misdiagnosed?**

Occasionally patients are not diagnosed for months or even years as scaly and red skin can be a symptom of many other skin conditions, such as severe eczema or immune deficiency disease. It is important that CIE is considered in any persistently red baby to avoid misdiagnosis or incorrect treatment. The skin of newborns with CIE can also closely resemble another form of ichthyosis called Lamellar ichthyosis, where redness of the skin tends to be less pronounced and the flaky whiteish scaling more apparent, making it difficult to distinguish between the two until later in life.

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## **Managing CIE**

As yet, there is no cure for CIE 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.

## **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 is considered to be more beneficial than showering.

Emollients and keratolytic (anti-scaling) creams which contain urea, and/or lactic acid and propylene glycol (see product's ingredients label for these) can be used to keep the skin as moist and hydrated as possible, however keratolytics can cause irritation if applied to inflamed and/or broken skin.

Steroid creams should be avoided as these do not help the skin.

Antibiotics are also important to treat secondary skin infections and gentle antiseptics can provide the skin with additional protection. Washing with an emollient containing an antimicrobial agent may help minimise the risk of skin infection.

It is important to check with your doctor or pharmacist before trying any new treatment. It is advisable to patch test any new cream for sensitivity before general use, and even "herbal" and "natural" creams may contain unsuitable ingredients.

Oral treatment (tablets) may be needed in severe ichthyosis. A group of drugs known as retinoids (synthetic vitamin A derivatives) are sometimes used (e.g. acitretin). They can reduce the thickness of the scale and help improve the appearance by reducing the overactive growth of the outer skin layer. Although often effective, they have a number of side-effects which should be fully discussed and considered before starting the tablet. Monitoring blood tests are needed while taking retinoid tablets, and pregnancy must be avoided as the drug can severely damage any unborn baby (teratogenic).

More specific information on caring for the skin, eyes and ears in CIE and other forms of ichthyosis can be found on the Ichthyosis Support Group website at [www.ichthyosis.org.uk](http://www.ichthyosis.org.uk)

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### **Eye treatment**

Some patients find that using moisturising eye drops can help prevent the eyes from becoming too dry. An ophthalmologist (eye doctor) may be contacted if ectropion (everted eyelids) is preventing the eyelids from closing at night but surgery for this condition is not generally recommended.

### **Ear treatment**

Patients may experience a build up of dead skin in their ears and regular appointments with the Ear, Nose and Throat department at the local hospital should help to prevent problems such as potential impaired hearing.

### **Physiotherapy**

Patients with CIE may have problems with flexibility in their fingers, known as contractures. This is when the elastic tissue in these appendages is replaced with non-elastic tissue. Some individuals find that physiotherapy and splinting at night can help relieve this problem.

### **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. Individuals should be given the flexibility to access fluids throughout the day and not just when participating in sporting activities.

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.

Individuals may experience hair loss, or restricted hair growth due to the thick scaling in CIE, and patients should manage their scalp to lessen the scaling where possible. Please see our leaflet for advice about how to look after your scalp.

### **How is the condition inherited?**

CIE is an inherited (i.e genetic) disorder so it runs in families. It is a condition passed on by parents with normal skin who both carry a copy of the faulty gene (autosomal recessive). They will not have CIE 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 CIE. The risk of any further child being affected is 25% or a 1 in 4 chance.

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

Families affected by CIE who would like genetic counselling should ask their GP to refer them to a clinical geneticist.

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People who care about ichthyosis

### **Further help**

There are a number of online forums about LI 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](mailto:isg@ichthyosis.org.uk)

**By Post:**

**Ichthyosis Support Group**  
**PO Box 1242, Yateley GU47 7FL**

**Facebook:**

[facebook.com/ichthyosissupportgroup](https://facebook.com/ichthyosissupportgroup)

**Twitter:**

[twitter.com/ISG\\_Charity](https://twitter.com/ISG_Charity)