



Foundation for Ichthyosis & Related Skin Types, Inc.®

Ichthyosis Summary

Foundation for Ichthyosis & Related Skin Types

The ichthyoses (plural) are a family of genetic diseases characterized by dry, thickened, scaling skin. Because it is genetic, ichthyosis is not contagious. Because each form of ichthyosis is rare and there is an overlap of clinical features among disease types, the medical community disagrees about clear definitions and classifications of its many forms. Very rare forms of ichthyosis may also exist which are not fully described in the medical literature. Regardless of these difficulties, there are approximately 28 recognized forms of ichthyosis and related skin types.

Most varieties of ichthyosis are relatively rare, affecting only one person in several tens of thousands. However, ichthyosis vulgaris is one exception; this form is estimated to affect one in every 250 people. Ichthyosis vulgaris is also the mildest form of ichthyosis and frequently goes undiagnosed or misdiagnosed as simply “dry skin.”

Ichthyosis can be a disfiguring disease, and as such has numerous social and psychological implications. The more severe forms of ichthyosis can cause many other problems. When the skin loses moisture, it becomes dry, tight, and rigid. This rigidity makes moving uncomfortable as it can cause the skin to crack and break open. Extreme thickening on the skin on the soles of the feet makes walking difficult for many individuals and cracking around the fingers can make even simple tasks difficult or painful. In some types of ichthyosis, the skin is very fragile and will rub off with the slightest abrasion. Cracks and abrasions then leave the skin open to infections.

Severe scaling on the scalp may interfere with normal hair growth. Thick scales elsewhere can block pores, making sweating difficult and increasing the risk of overheating. Although the outer skin is thicker in ichthyosis, it is less effective in preventing water (and calorie) loss by diffusion across the surface of the skin. The rapid turnover of the outer layers of skin, in some forms of ichthyosis, also requires additional energy. Because of these greater energy needs, some children with severe ichthyosis may have trouble taking in enough calories to grow normally.

Some people with ichthyosis have trouble closing their eyes completely because the surrounding skin is so tight. This condition, called ectropion, causes the eyelids to turn outward, exposing the red inner lid and causing continuing irritation. If it is left untreated, damage to the cornea can develop, leading to impaired vision.

Types of Ichthyosis

Ichthyosis Vulgaris

Ichthyosis vulgaris is one of the more commonly seen types of ichthyosis. Sometimes called common ichthyosis (“vulgar” means “common” in Latin), it appears in approximately 1 in 250 individuals. Ichthyosis vulgaris often goes undiagnosed because people who have it think they have simple “dry skin” and never seek treatment.

In ichthyosis vulgaris, the skin cells are produced at a normal rate, but they do not separate normally at the surface of the stratum corneum (the outermost layer of skin) and are not shed as quickly as they should be. The result is a buildup of scales. Usually only a portion of the body may be involved (scaling is most common and most severe over the lower legs), and the scale is usually fine and white. Scaling on the torso is usually less severe and the face is usually unaffected. When the face is affected, the scaling is usually limited to the forehead and cheeks.

Babies with ichthyosis vulgaris often appear normal when they are born, but then the skin abnormalities will almost always show up by their first birthday. Ichthyosis vulgaris may improve in certain climates, during the summer and with age.

Ichthyosis vulgaris is treated topically with moisturizers, and keratolytics. It is not considered severe enough to warrant use of oral synthetic retinoids.

X-Linked Ichthyosis

X-linked ichthyosis is one of the more commonly seen forms of ichthyosis. It occurs in 1 in approximately 6,000 births, can range from mild to severe, and occurs only in males. In X-linked ichthyosis, the skin cells are produced at a normal rate, but they do not separate normally at the surface of the stratum corneum (the outermost layer of the skin) and are not shed as quickly as they should be. The result is a buildup of scales. The scales of X-linked are often dark and usually cover only a portion of the body. Typically the face, scalp, palms of the hands and soles of the feet are unaffected, while the back of the neck is almost always affected. X-linked ichthyosis frequently improves in the summer.

Babies with X-linked often appear normal when they are born, but the skin abnormalities will almost always begin to show up by their first birthday. X-linked ichthyosis is treated topically with moisturizers and keratolytics. Cholesterol containing ingredients may also improve scaling. X-linked ichthyosis is not considered severe enough to warrant use of oral synthetic retinoids.

Autosomal Recessive Congenital Ichthyosis - lamellar type

Autosomal recessive congenital ichthyosis (ARCI) – lamellar type (or classical lamellar ichthyosis) is one of the more commonly seen types of ichthyosis. It is one of the most severe forms, and it occurs in approximately 1 in 300,000 births. Recessive genes cause lamellar ichthyosis, similar to blue eyes. Only when a person receives two recessive genes for lamellar ichthyosis will he or she actually have the disorder.

In lamellar ichthyosis, the skin cells are produced at a normal rate, but they do not separate normally at the surface of the stratum corneum (the outermost layer of the skin) and are not shed as quickly as they should be. The result is a build-up of scales. The entire body is covered with broad, dark, plate-like scales separated by deep cracks. People with lamellar ichthyosis may experience a condition called ectropion (a turning out of the eyelids to expose the red inner lid). People with lamellar ichthyosis may also have thickened nails and hair loss due to the thickness of the scales on their scalp. They may also have reddened skin (erythroderma), thickened skin on the palms of the hands and soles of the feet, and decreased sweating with heat intolerance.

Lamellar ichthyosis is present at birth. Many babies born with lamellar ichthyosis are also “collodion babies” because a clear membrane (the collodion) may cover their bodies. The collodion is then shed within a few days to a few weeks. Sometimes described as having a shellacked appearance, these newborns have skin which is taut, dark and split. Often the eyelids and lips are forced open by the tightness of the skin, and there may be contractures around the fingers. Problems with temperature regulation, water loss, secondary infections, and systemic infection can occur in the newborn with lamellar ichthyosis.

Lamellar ichthyosis is typically treated topically with moisturizers and keratolytics. Creams with high concentrations of alpha-hydroxy acids are commonly used. Lamellar ichthyosis may be treated systemically with oral synthetic retinoids (Accutane or Soriatane). Retinoids are used only in severe cases due to their known bone toxicity and other complications.

Netherton Syndrome

Netherton syndrome is a rare syndrome characterized by red, scaly skin, short brittle hair and a predisposition to allergies, asthma and eczema. Newborns with Netherton syndrome have reddened skin (erythroderma) and, occasionally, a thick shell-like covering of skin (collodion membrane). They may also be born prematurely. Trouble gaining weight during infancy and childhood is common. Atopic dermatitis (red, itchy patches of skin) may be present, and a cradle cap-like scale and redness may appear on the face, scalp and eyebrows.

Unlike many of the ichthyoses, Netherton syndrome produces too few layers of the outer skin, instead of two many layers. Therefore, agents that remove scale, such as the alpha-hydroxy acids and oral retinoids, are **not** helpful in the management of this disorder and may aggravate

the symptoms. Current treatment options are limited to use of mild moisturizers containing petrolatum or lanolin and/or a barrier repair formula containing ceramides or cholesterol.

Autosomal Recessive Congenital Ichthyosis – CIE type

Autosomal recessive congenital ichthyosis (ARCI) – CIE type (nonbullous congenital ichthyosiform erythroderma) is considered one of the more commonly seen types of ichthyosis. Like lamellar ichthyosis, CIE is rare, occurring in 1 in 300,000 births. Recessive genes cause it.

In CIE, there is an overproduction of skin cells in the epidermis. These cells reach the stratum corneum (the outermost layer of skin) in as few as four days, compared to the normal fourteen. New skin cells are made faster than old cells are shed and build up in the stratum corneum and underlying layers. The severity and scaling of CIE varies. The scales on the face, scalp and torso are usually fine and white, but the scales on the legs can be large and plate-like (like the scales of lamellar ichthyosis). The skin is often quite reddish beneath the scales.

CIE is present at birth. Many babies with CIE were born as “collodion babies,” so called because a clear membrane (the collodion) covers their bodies. The collodion is then shed within a few days to a few weeks. Sometimes described as having a shellacked appearance, these newborns have skin that is taut, dark and split. After the membrane is shed, dry red skin is revealed. Often the eyelids and lips are already forced open by the tightness of the skin, and there may be contractures around the fingers.

CIE is treated topically with moisturizers and keratolytics. Creams with high concentrations of alpha-hydroxy acids are commonly used. CIE can be treated systemically with oral synthetic retinoids (Accutane, Soriatane). Retinoids are only used in severe cases due to their known bone toxicity and other complications.

Epidermolytic Ichthyosis (EI)

Epidermolytic ichthyosis (formerly called epidermolytic hyperkeratosis or EHK) (bullous congenital ichthyosiform erythroderma) is rare, occurring in approximately 1 in 300,000 births.

EI is characterized by thick, blistering, warty hardening of the skin over most of the body, particularly in the skin creases over the joints. Scales tend to form in parallel rows of spines or ridges. The skin may be fragile and may blister easily following injury. Babies with EI are usually born with red, blistering, and denuded skin with visible skin thickening. Over time there is a gradual decrease in the blistering, but an increase in the severity of the thickness and scaling. A generalized redness of the skin (erythroderma) is present in some individuals. Skin infections and heat intolerance can be common problems.

Treating EI can be a challenge. The medications that are used to help remove the excess thickened skin (topical keratolytics or oral retinoids) often remove too much scale, leaving very fragile underlayers exposed. Barrier repair creams, containing ceramides, cholesterol, petrolatum or lanolin, can help along with topical or systemic anti-bacterial agents. Keratolytics and oral retinoids should be used with caution.

This information is provided as a service to patients and parents of patients who have ichthyosis. It is not intended to supplement appropriate medical care, but instead to complement that care with guidance in practical issues facing patients and parents. Neither FIRST, its Board of Directors, Medical & Scientific Advisory Board, Board of Medical Editors, nor Foundation staff and officials endorse any treatments or products reported here. All issues pertaining to the care of patients with ichthyosis should be discussed with a dermatologist experienced in the treatment of their skin disorder.



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.

February 2021