National Institute of Arthritis and Musculoskeletal and Skin Diseases

ENGLISH | ESPAÑOL

Ichthyosis

Overview of Ichthyosis

Ichthyosis refers to a group of skin disorders that lead to dry, itchy skin that appears scaly, rough, and red. The symptoms can range from mild to severe. Ichthyosis can affect only the skin, but some forms of the disease can affect internal organs as well.

Most people have a genetic form of ichthyosis that results from a changed gene, often inherited from their parents. However, some people develop a form of acquired (nongenetic) ichthyosis from another medical disorder or certain medications. While there is currently no cure for ichthyosis, research is ongoing and treatments are available to help manage the symptoms.

The outlook for people with ichthyosis varies depending on the type of the disease and how severe it is. Most people with ichthyosis need treatment for life to help make the disease more manageable.

Who Gets Ichthyosis?

Anyone can get ichthyosis. The disease is usually passed down from your parents; however, some people can be the first in a family to develop ichthyosis due to a new gene mutation. Other people develop an acquired (nongenetic) form of ichthyosis, which results from another medical condition or a side effect of a medication.

Types of Ichthyosis

There are more than 30 distinct types of ichthyosis, including those that occur as part of another syndrome or condition. Doctors may determine the type of ichthyosis by identifying the:

- Gene mutation.
- Inheritance pattern through analyzing family trees.
- Symptoms, including their severity and which organs they affect.
- Age when symptoms first appeared.

Some types of the disease, which are inherited and are not part of a syndrome, include the following:

- Ichthyosis vulgaris is the most common type. It is usually mild and appears in the first year of life with dry, flaky skin.
- Harlequin ichthyosis is usually seen at birth and causes thick scaly plates of skin that cover the entire body. This form of the disorder can affect the shape of facial features and may limit joint movement.
- Epidermolytic ichthyosis is present at birth. Most infants are born with fragile skin and blisters covering their body. Over time, the blisters disappear, and scaling of the skin develops. This can have a ridged appearance over areas of the body that bend.
- Lamellar ichthyosis is present at birth. The infant is born with a tight clear covering the entire body, called a collodion membrane. Over several weeks, the membrane peels away, and large, dark, plate-like scales develop over most of the body.
- Congenital ichthyosiform erythroderma is present at birth. Infants also often present with a collodion membrane.
- X-linked ichthyosis usually develops in males and begins at about 3 to 6 months of life. Scaling is usually present on the neck, lower face, trunk, and legs, and symptoms can worsen over time.
- Erythrokeratodermia variabilis usually develops a few months after birth and progresses during childhood. The skin can develop rough, thick or reddened areas

of skin, usually on the face, buttocks, or limbs. The affected areas can spread on the skin over time.

- Progressive symmetric erythrokeratoderma usually appears in childhood with dry, red, scaly skin primarily on the limbs, buttocks, face, ankles, and wrists.

Symptoms of Ichthyosis

The symptoms of ichthyosis can range from mild to severe. The most common symptoms include:

- Dry skin.
- Itching.
- Redness of the skin.
- Cracking of the skin.
- Scales on the skin that are white, gray, or brown and have the following appearance:
  - Small and flaky.
  - Large, dark, plate-like scales.
  - Hard, armor-like scales.

Depending on the type of ichthyosis, other symptoms may include:

- Blisters that can break, leading to wounds.
- Hair loss or fragile hair.
- Dry eyes and difficulty closing eyelids. Inability to perspire (sweat) because skin scales clog the sweat glands.
- Difficulty hearing.
- Thickening of the skin on the palms of the hands and soles of the feet.
- Tightening of the skin.
- Difficulty flexing some joints.

Cause of Ichthyosis

Gene mutations (changes) cause all of the inherited types of ichthyosis. Many gene mutations have been identified and the inheritance pattern depends on the type of ichthyosis. People continually grow new skin and shed old skin throughout their lives. For people with ichthyosis, the mutated genes change the normal skin growth and shedding cycle, causing skin cells to do one of the following:

- Grow faster than they are shed.
- Grow at a normal rate, but shed at a slow rate.
- Shed faster than they grow.

There are different types of inheritance patterns of ichthyosis, including:

- Dominant, which means you inherit one normal copy and one mutated copy of the gene that causes ichthyosis. The abnormal copy of the gene is stronger or "dominant" over the normal copy of the gene, causing the disease. A person with a dominant mutation has a 50% chance (1 in 2) of passing the disorder to each of his or her children.
- Recessive, which means that your parents do not have signs of ichthyosis, but both parents carry only one abnormal gene, which is not enough to cause the disease. When both parents carry one recessive gene, there is a 25% chance (1 out of 4) per pregnancy of having a child who inherits both of these mutated genes and develops the disorder. There is a 50% chance (2 out of 4) per pregnancy of having a child who inherits only one mutated recessive gene, making them a carrier of the disease gene without noticeable signs. If one parent has a recessive form of ichthyosis with two mutated genes, all their children will carry one abnormal gene, but will not usually have noticeable signs of ichthyosis.
- X-linked, which means the gene mutations are located on the X sex chromosome. Each person has two sex chromosomes: Females generally have two X chromosomes (XX), and males generally have one X chromosome and one Y chromosome (XY). The mother always passes on an X chromosome, but the father can pass on either an X or Y chromosome. The inheritance pattern for X-linked ichthyosis is usually recessive; this means that males, who only have one X chromosome to begin with, pass on the mutated X chromosome. Because of this pattern, females are affected more often, and typically they have one mutated and one normal X chromosome.

- Spontaneous, which means the gene mutation occurs randomly without a family history of the disorder. This is most common in dominant and X-linked ichthyoses.