



ICHTHYOSIS OVERVIEW

Ichthyosis is a family of disorders characterized by dry or scaly and thickened skin. “Ichthy” comes from the Greek word for fish. This condition is called “ichthyosis” because the thickened skin sometimes has the appearance of fish scales.

Ichthyosis may be either inherited or acquired. Inherited ichthyosis is usually apparent during the first year of life, often at birth, and continues to affect a person throughout life. Acquired ichthyosis may occur as the result of medical problems including hormonal, inflammatory, or malignant disorders. This fact sheet focuses on inherited ichthyosis.

WHAT ARE THE DIFFERENT TYPES OF ICHTHYOSIS?

There are many different types of inherited ichthyosis. Most types involve only the skin, and some regions of the skin may be more severely affected than others. Other types involve internal organs as well as the skin, and are referred to as syndromic types of ichthyosis. Although there is an increase in the tendency to categorize inherited ichthyosis by the gene that causes it, the clinical appearance usually falls into one of five categories (see box on right).

WHAT IS THE CAUSE?

The inherited forms of ichthyosis are caused by mutations in genes passed from one or both parents to a child. In some cases, the parents themselves do not have the condition, but carry the genetic mutations. In other cases, mutations occur spontaneously during the formation of the egg or sperm or at the time of, or shortly after, conception.

DIFFERENT TYPES OF ICHTHYOSIS

- **Ichthyosis vulgaris.** Characterized by mild skin scaling and dryness. Ichthyosis vulgaris and another form, recessive X-linked ichthyosis, are relatively common and appear similar.
- **Epidermolytic ichthyosis (previously called epidermolytic hyperkeratosis).** Characterized by thick, often spiny dark scales and skin that may blister easily following trauma.
- **Lamellar ichthyosis.** Characterized by large, platelike scales and thickening of the skin.
- **Congenital ichthyosiform erythroderma.** Characterized by red skin and fine scales.
- **Localized ichthyosis.** Characterized by thick or scaly skin that is localized to particular regions such as the palms of the hands and soles of the feet.

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and Skin Diseases (NIAMS)

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The mission of the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), a part of the U.S. Department of Health and Human Services' National Institutes of Health (NIH), is to support research into the causes, treatment, and prevention of arthritis and musculoskeletal and skin diseases; the training of basic and clinical

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Mutations in more than 40 genes are now known to cause inherited ichthyosis. In general, mutations in more than one gene cause each clinical type of ichthyosis.

WHAT ARE THE SIGNS AND SYMPTOMS?

Specific signs and symptoms depend on the particular form. The most common signs include dry scaly skin, redness, blistering, or excessive skin shedding. Symptoms include itching, overheating, and pain.

HOW IS ICHTHYOSIS DIAGNOSED?

The diagnosis is based on the results of a medical history and physical exam. In some cases, doctors use blood tests or skin biopsies to diagnose the condition or identify the particular form.

HOW IS ICHTHYOSIS TREATED?

Because there is no cure for ichthyosis, treatment is targeted at managing the signs and symptoms. Treatment may include creams, lotions, or ointments to relieve dryness. Lengthy bathing in salt water or preparations containing salicylic acid (aspirin) or urea may also ease scaling. For more severe cases, doctors may prescribe vitamin A derivatives called retinoids.

WHAT TYPES OF DOCTORS DIAGNOSE AND TREAT ICHTHYOSIS?

Ichthyosis is treated by dermatologists, who are doctors specializing in diagnosing and treating diseases involving the skin, hair, and nails.

WHAT RESEARCH IS BEING DONE?

Ichthyosis research is focused on understanding better the causes of the disorder, as well as finding more effective treatments. Although the genes responsible for the majority of ichthyosis cases have been identified, there are still some people with the disease who don't have a mutation in one of the known genes. For that reason, researchers continue

to search for genes involved in the disease. They also are looking for ways to manipulate the responsible genes to stop the disease.

Other scientists are working to develop pharmaceutical treatments, including new emollients or preparations that soften the outer layer of skin and cause it to hold in more moisture. New formulations of topical keratolytics also are being developed. Keratolytics soften the keratin, which is the key structural material of the outer layer of the skin.

In 1994, the National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS) funded the establishment of a National Registry for Ichthyosis and Related Disorders at the University of Washington. The goal of the registry is to identify people with ichthyosis and other related disorders, and to collect information about their skin disorder and how it has affected them. The registry established a large database with historical, clinical, and quality-of-life information for 600 people with ichthyosis and related disorders. Most of them are interested in participating in related research. Although the registry is now closed to new enrollment, it is maintained to provide information to scientists studying ichthyosis, and has already served as a source of crucial information for numerous studies.

RESOURCES

National Institute of Arthritis and Musculoskeletal and Skin Diseases

Website: www.nih.gov/niams

American Academy of Dermatology

Website: www.aad.org

Foundation for Ichthyosis and Related Skin Types, Inc. (FIRST)

Website: www.firstskinfoundation.org

National Registry for Ichthyosis and Related Disorders

Website: <http://depts.washington.edu/ichreg/ichthyosis.registry>

ACKNOWLEDGMENT

The NIAMS gratefully acknowledges the assistance of Leonard Milstone, M.D., in the development and review of this publication.

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NIH Publication No. 12–7934

FOR YOUR INFORMATION

This publication contains information about medications used to treat the health condition discussed here. When this publication was printed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

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