



Foundation for Ichthyosis & Related Skin Types, Inc.® **X-Linked Ichthyosis**

What is X-Linked Ichthyosis?

X-linked ichthyosis is considered one of the five main types of ichthyosis (the others being lamellar ichthyosis, congenital ichthyosiform erythroderma, ichthyosis vulgaris and epidermolytic ichthyosis (formerly epidermolytic hyperkeratosis)). X-linked ichthyosis is less common (occurring in approximately 1 in 6,000 births), can range from mild to severe, and occurs only in males.

What are the Signs & Symptoms?

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 build-up of scales. The scales of X-linked ichthyosis 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 free from scales, while the back of the neck is almost always affected. X-linked ichthyosis frequently improves in the summer. Babies with X-linked ichthyosis often appear normal when they are born, but the skin abnormalities will almost always show up by their first birthday.

How is it Diagnosed?

The genetic defect in X-linked results in a deficiency of the enzyme, steroid sulfatase. Genetic testing can detect the abnormality prenatally using amniocentesis or chorionic villus sampling (CVS). Decreased maternal serum or urine estriol levels and dehydroepiandrosterone levels can suggest X-linked ichthyosis in the fetus. Deficiency of the sulfatase enzyme in the placenta may result in failure of labor to initiate or progress.

X-linked ichthyosis is carried on the X sex chromosome. Women are unaffected but can carry the disease and pass it on to their sons. Men who have X-linked ichthyosis will have unaffected sons (they get their X chromosome from their mother), but their daughters will all be carriers. (For more information on the genetics of X-linked ichthyosis, refer to the Foundation's publication, *Ichthyosis, The Genetics of its Inheritance*).

Approximately 50% of adult males and some female carriers will have asymptomatic comma-shaped corneal opacities (cloudy spots), which do not affect sight. Female carriers of X-linked ichthyosis occasionally report dry skin problems and, rarely, shadows of scales on the skin.

Results of genetic tests, even when they identify a specific mutation, can rarely tell you how mild or how severe a condition will be in any particular individual. There may be a general presentation in a family or consistent findings for a particular diagnosis, but it's important to know that every individual is different. The result of a genetic test may be "negative," meaning no mutation was identified. This may help the doctor exclude certain diagnoses, although sometimes it can be unsatisfying to the patient. "Inconclusive"

results occur occasionally, and this reflects the limitation in our knowledge and techniques for doing the test. But we can be optimistic about understanding more in the future, as science moves quickly and new discoveries are being made all the time. You can receive genetic testing through the Yale University's Disorders of Keratinization study with Dr. Keith Choate or for more information about genetic tests performed you can visit GeneDx, www.genedx.com.

What is the Treatment?

X-linked ichthyosis responds relatively well to topical treatment with alpha-hydroxy acids, which accelerate the shedding of the stratum corneum. Cholesterol containing emollients may also improve the scaling. (For more information on which products contain these ingredients, request the Foundation's Skin Care Products List.) Alpha-hydroxy acids may sting the skin of babies and young children and should be used cautiously or in combination with another mild emollient product. X-linked ichthyosis is usually not considered severe enough to warrant the use of oral synthetic retinoids.

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
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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