



Foundation for Ichthyosis & Related Skin Types

Foundation for Ichthyosis & Related Skin Types, Inc.[®] **Ichthyosis Vulgaris**

What is Ichthyosis Vulgaris?

Ichthyosis Vulgaris is considered one of the five main types of ichthyosis (the others being lamellar ichthyosis, congenital ichthyosiform erythroderma, X-linked ichthyosis and epidermolytic ichthyosis). Most varieties of ichthyosis affect only one person in several thousand or tens of thousands. Ichthyosis vulgaris, sometimes called common ichthyosis (“vulgar” means “common” in Latin), is the exception. It appears in approximately one person in every 250 to 300. The name is rarer than the disorder itself, which often goes undiagnosed because people who have it think that they have simple “dry skin” and never seek treatment.

What are the Signs & Symptoms?

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 build-up of scale. Only a portion of the body may be involved, but scaling is most common and most severe over the lower legs. The scale is usually fine and white. Scaling on the torso is less severe and the face is usually unaffected. If the face is affected, the scaling is usually limited to the forehead and cheeks. Often, the skin on the palms of the hands and the soles of the feet is thickened and may have exaggerated lines. Babies with ichthyosis vulgaris often appear normal when they are born, but the skin abnormalities will almost always begin to show up by their first birthday.

Ichthyosis vulgaris sometimes improves in certain climates or during the summer. Ichthyosis vulgaris also tends to improve with age.

How is it Diagnosed?

Ichthyosis vulgaris was thought to be caused by an autosomal dominant gene. If a person inherited the gene for ichthyosis vulgaris, the gene overshadowed the gene for normal skin and the person displayed the disease. However, researchers recently discovered that the disorder is semi-dominant. Profilaggrin (a protein that makes up most of the granules in the granular layer in the epidermis) was reduced or absent from the skin of people with vulgaris. Severely affected individuals have mutations in both copies of the profilaggrin gene, while people with only one copy of the mutated gene usually have a very mild skin problem.

Doctors frequently use genetic testing to help define which ichthyosis a person actually has. This may help them to treat and manage the patient. Another reason to have a genetic test is if you or a family member wants to have children. Genetic testing, which would ideally be performed first on the person with ichthyosis, is often helpful in determining a person's, and their relative's, chances to have a baby with ichthyosis. Genetic testing may be recommended if the inheritance pattern is unclear or if you or a family member is interested in reproductive options such as genetic diagnosis before implantation or prenatal diagnosis.

Results of genetic tests, even when they identify a specific mutation, can rarely tell 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?

Ichthyosis vulgaris is treated topically with moisturizers containing urea or glycerol. Lotions containing alpha-hydroxy acids may help. (For more information on which products contain these ingredients, refer to the Foundation's Skin Care Products.) However, some individuals with ichthyosis vulgaris also experience atopic dermatitis (red, itchy patches of skin) and the alpha-hydroxy acids may irritate their skin.

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.



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Improve lives and seek cures for those affected by ichthyosis and related skin types.

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