Foundation for Ichthyosis & Related Skin Types, Inc.®

Epidermolytic Ichthyosis (EI)

Bullous Congenital Ichthyosiform Erythroderma

What is Epidermolytic Ichthyosis?

Epidermolytic ichthyosis (EI) (formerly epidermolytic hyperkeratosis (EHK) is considered one of the five main types of ichthyosis (the others being lamellar ichthyosis, congenital ichthyosiform erythroderma, ichthyosis vulgaris, and X-linked ichthyosis). EI is rare, occurring in approximately 1 in 300,000 Americans.

What are the Signs & Symptoms?

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 are usually born with red, blistering, and denuded skin with visible areas of skin thickening. Over time there is a gradual decrease in blistering, but an increase in the severity of the scaling and thickening. A generalized erythroderma (redness of the skin) is present in some individuals. Skin infections with commonly found bacteria (Staphylococcus and Streptococcus) can be a chronic problem. The teeth, hair and nails are normal, but scalp involvement can be severe, resulting in hair loss. Heat intolerance is common.

Possibly fifty percent of known cases of EI can be traced to an affected parent, while the other fifty percent are thought to be caused by a spontaneous mutation, which can then be passed on to their children. The gene for EI is dominant over the gene for normal skin so there are no invisible carriers for EI. Anyone who carries the EI gene will show the disease. If a partner of the person with EI does not also have the disease, each of their children will have a fifty percent chance (the same for each pregnancy) of inheriting the disease. (For more information on the genetics of EI, refer to FIRST's publication, Ichthyosis: The Genetics of Its Inheritance.)

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

Treating EI is a challenge. The medications that are used to help remove the excess thickened skin layers (topical keratolytics or oral retinoids) often remove too much scale, leaving a very fragile epidermis (underlying living cell layers) exposed. Skin infections are also a particular problem.

Although no currently available treatments are ideal, a combination of these strategies may help:

- Application of a barrier repair formula containing ceramides or cholesterol.
- Application of a barrier repair formula containing petrolatum or lanolin.
- Topical or systemic anti-bacterial agents.
- Keratolytics (lotions containing alpha-hydroxy acids) and oral retinoids.
- Both of these should be used with caution.

(For more information on which products contain these ingredients, request FIRST's Skin Care Products List.)

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

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