

X-LINKED ICHTHYOSIS

What is X-linked ichthyosis?

There are lots of causes of dry skin. X-linked ichthyosis (XLI) is one of the causes that has an inherited component. It is passed on through the mother's side of the family and typically affects males only. Patients with X-linked ichthyosis are missing an enzyme called "steroid sulfatase" and this enzyme is important for normal skin shedding.



What does X-linked ichthyosis look like?

Patients with X-linked ichthyosis usually have scaling over the body that is usually not itchy. The palms, soles and "folds" of the skin (like the elbow creases and the back of the knees) tend to be clear. Many patients have dry skin that starts in the first year of life and about 20% have it at birth.

Many patients these days are diagnosed when a prenatal blood test shows an abnormal result (which is used for screening a different condition in the fetus). Genetic testing can be done to confirm the diagnosis of X-linked ichthyosis. Often, there are other family members with this condition.

If an eye exam is done, there can be harmless spots in the eye (cornea) that can be found. Patients with X-linked ichthyosis do not need to see an eye doctor for these, as the spots do not need treatment.



What can be done for the skin?

Most patients have fairly mild dry skin with scales. This is usually managed with regular bathing and using moisturizers (with or without peeling agents). Some peeling agents include 10% to 20% urea cream, lactic acid, and/or salicylic acid-containing moisturizers. These shouldn't be used in young infants (under 6 months of age). In severe cases, special vitamin A creams or pills can be used (but these need to be prescribed by a physician).