



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

What is Trichothiodystrophy?

Trichothiodystrophy as known as TTD is a rare recessive DNA repair disorder,

TTD is a rare autosomal recessive disorder that is characterized by brittle, sulfur-deficient hair, short stature, and multisystem abnormalities. Patients may have exaggerated sensitivity to sunlight (photosensitivity), developmental delay, recurrent infections, and ichthyosis.

What are the Signs & Symptoms?

Many babies with trichothiodystrophy are born with ichthyosiform erythroderma or collodion membrane that generally improves during the first year. Children with TTD are frequently sensitive to light and UV, and experience extreme pain and blistering, as well as fatigue, dizziness, headaches and other physical problems when exposed to sunlight.

How is it Diagnosed?

Diagnosis is made by studying the hair mounts, and by amino acid analysis which demonstrates decreased high sulphur matrix proteins. The hair breaks and fractures once it emerges from the skin and is exposed to the environment, and so the result is brittle, short and sparse hair. Analysis of cellular DNA available for some cases. The genes that are abnormal in these cases are transcription factors ERCC2 or ERCC3 (in some).

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?

There is currently no treatment for trichothiodystrophy.

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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