



Foundation for Ichthyosis & Related Skin Types, Inc.[®] Chanarin Dorfman Syndrome

What is Chanarin Dorfman Syndrome?

Synonyms of Ichthyosis, Chanarin Dorfman Syndrome:

- Chanarin Dorfman Disease
- Disorder of Cornification 12 (Neutral Lipid Storage Type)
- DOC 12 (Neutral Lipid Storage Type)
- Dorfman Chanarin Syndrome
- Ichthyosiform Erythroderma with Leukocyte Vacuolation
- Ichthyotic Neutral Lipid Storage Disease
- Neutral Lipid Storage Disease
- Triglyceride Storage Disease Impaired Long-Chain Fatty Acid Oxidation

Chanarin Dorfman syndrome is a rare hereditary disorder of fat (lipid) metabolism. It is characterized by scaly skin (ichthyosis), degeneration of the muscles (myopathy), and abnormal white blood cells with small spaces (vacuoles) filled with fat (lipids).

What are the Signs & Symptoms?

Chanarin Dorfman syndrome is characterized by moderately red, itchy, dry skin and other skin changes (eczematous dermatitis). Prominent fat (lipid) droplets appear in most circulating white blood cells. Lipid droplets are also present in numerous other cells, including those of the skin and the ducts of sweat glands.

Degeneration of the muscles may be identified through neurologic testing, and muscle enzyme levels in the blood are elevated. The liver biopsy samples of all patients have shown severe fatty change, but this may not be reflected in liver function studies. Both nerve deafness and cataracts are present in some patients with Chanarin Dorfman syndrome. Abnormally slow development and growth may also be symptoms of this disorder.

How is it Diagnosed?

Chanarin-Dorfman syndrome is the result of an inborn error of metabolism and is transmitted as an autosomal recessive trait. It is associated with a gene on the short arm of chromosome 3 (3p21).

Chromosomes, which are present in the nucleus of human cells, carry the genetic information for each individual. Human body cells normally have 46 chromosomes. Pairs of human chromosomes are numbered from 1 through 22 and the sex chromosomes are designated X and Y. Males have one X and one Y chromosome and females have two X chromosomes. Each chromosome has a short arm designated "p" and a long arm designated "q". Chromosomes are further sub-divided into many bands that are numbered. For example, "chromosome 3p21" refers to band 21 on the short arm of chromosome 3. The numbered bands specify the location of the thousands of genes that are present on each chromosome.

Genetic diseases are determined by the combination of genes for a particular trait that are on the chromosomes received from the father and the mother.

Recessive genetic disorders occur when an individual inherits the same abnormal gene for the same trait from each parent. If an individual receives one normal gene and one gene for the disease, the person will be a carrier for the disease, but usually will not show symptoms. The risk for two carrier parents to both pass the defective gene and, therefore, have an affected

child is 25% with each pregnancy. The risk to have a child who is a carrier like the parents is 50% with each pregnancy. The chance for a child to receive normal genes from both parents and be genetically normal for that particular trait is 25%. The risk is the same for males and females.

All individuals carry a few abnormal genes. Parents who are close relatives (consanguineous) have a higher chance than unrelated parents to both carry the same abnormal gene, which increases the risk to have children with a recessive genetic disorder.

Diagnosis of Chanarin Dorfman syndrome can be made when blood taken from a finger, toe, heel, or ear shows fat droplets in certain white blood cells. 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?

Dermatologic symptoms are treated by applying skin softening (emollient) ointments, preferably plain petroleum jelly. This can be especially effective after bathing while the skin is still moist. Salicylic acid gel is another particularly effective ointment. The skin should be covered at night with an airtight, waterproof dressing when this ointment is used. Lactate lotion can also be an effective treatment for this disorder.

Drugs derived from Vitamin A (retinoids) such as tretinoin, motretinide, and etretinate can be effective against skin symptoms of Chanarin Dorfman syndrome, but can cause toxic effects on the bones in some cases, especially if taken by pregnant women. A synthetic derivative of Vitamin A, isotretinoin, when taken by pregnant women, can cause severe birth defects to the fetus. These Vitamin A compounds have not yet been approved by the Food and Drug Administration (FDA) for treatment of ichthyosis.

Treatment of other features of Chanarin Dorfman syndrome is symptomatic and supportive.

REF. National Organization for Rare Disorders (NORD) <http://rarediseases.org/rare-diseases/ichthyosis-chanarin-dorfman-syndrome/>

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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