Foundation for Ichthyosis & Related Skin Types, Inc.®

Refsum Disease

What is Refsum Disease?

Refsum disease is one of a family of genetic disorders known as the leukodystrophies in which, as a consequence of the disruption of lipid metabolism, the myelin sheath that insulates and protects the nerves of the brain fails to grow. It is inherited as an autosomal recessive trait. It is characterized by progressive loss of vision (retinitis pigmentosa); degenerative nerve disease (peripheral neuropathy); failure of muscle coordination (ataxia); and dry, rough, scaly skin (ichthyosis). The disorder is caused by the accumulation of a particular fatty acid (phytanic acid) in blood plasma and tissues. This occurs because of a malfunction of the gene that makes the enzyme that breaks down (metabolizes) this acid. The essential enzyme is absent.

Synonyms of Refsum Disease?

- Disorder of Cornification 11 (Phytanic Acid Type)
- DOC 11 (Phytanic Acid Type)
- Heredopathia Atactica Polyneuritiformis
- Hypertrophic Neuropathy of Refsum
- Phytanic Acid Storage Disease
- Refsum Disease

What are the Signs & Symptoms

Symptoms of Refsum disease may include vision impairment, degenerative nerve disease, failure of muscle coordination, and bone and skin changes. Symptoms may include night blindness, loss of peripheral vision, and numbness and weakness associated with failure of muscle coordination.

Affected individuals may experience an unusual burning or prickling sensation (paresthesia) of arms and legs. Neurological symptoms include unsteady walking with frequent falls (ataxia), and peripheral neuropathy (characterized by sensory, motor, and reflex changes). Skin changes may include dryness, itchiness, and scaliness.

An infantile form of Refsum disease exists that usually becomes apparent during the first year of life. In addition to early onset, it is characterized by developmental delay, vision and hearing impairment, enlargement of the liver and defective metabolism of bile acid.

How is it Diagnosed?

Refsum disease is inherited as a recessive trait. The genes responsible for the failure in the metabolism of phytanic acid have been traced to the short arm of chromosome 10 (10pter-p11.2) and the long arm of chromosome 6 (6q22-q24). Diagnosis can be made by the presence of phytanic acid in blood or urine samples is diagnostic.

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?

Treatment of Refsum disease involves following a strict diet low in phytanic acid (found in dairy products, beef, lamb and some seafoods) and high in calories. The removal and reinfusion of blood plasma (plasmapheresis) may also be required. Other treatment is symptomatic and supportive.

REF. National Organization for Rare Disorders (NORD) http://rarediseases.org/rare-diseases/refsum-disease/

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

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