

Adrenoleukodystrophy

Definition

X-linked Adrenoleukodystrophy (ALD) is one of a group of genetic disorders called the leukodystrophies that cause damage to the myelin sheath, an insulating membrane that surrounds nerve cells in the brain. Women have two X chromosomes and are the carriers of the disease, but since men only have one X chromosome and lack the protective effect of the extra X chromosome, they are more severely affected. People with X-ALD accumulate high levels of saturated, very long chain fatty acids (VLCFA) in the brain and adrenal cortex. The loss of myelin and the progressive dysfunction of the adrenal gland are the primary characteristics of X-ALD. While nearly all patients with X-ALD suffer from adrenal insufficiency, also known as Addison's disease, the neurological symptoms can begin either in childhood or in adulthood. The childhood cerebral form is the most severe, with onset between ages 4 and 10. The most common symptoms are usually behavioral changes such as abnormal withdrawal or aggression, poor memory, and poor school performance. Other symptoms include visual loss, learning disabilities, seizures, poorly articulated speech, difficulty swallowing, deafness, disturbances of gait and coordination, fatigue, intermittent vomiting, increased skin pigmentation, and progressive dementia. The milder adult-onset form is also known as adrenomyeloneuropathy (AMN), which typically begins between ages 21 and 35. Symptoms may include progressive stiffness, weakness or paralysis of the lower limbs, and ataxia. Although adult-onset ALD progresses more slowly than the classic childhood form, it can also result in deterioration of brain function. Almost half the women who are carriers of X-ALS will develop a milder form of AMN but almost never will develop symptoms seen in boys the X-ALD. X-ALD should not be confused with neonatal adrenoleukodystrophy, which is a disease of newborns and young infants and belongs to the group of peroxisomal biogenesis disorders.

Treatment

Adrenal function must be tested periodically in all patients with ALD. Treatment with adrenal hormones can be lifesaving. Symptomatic and supportive treatments for ALD include physical therapy, psychological support, and special education. Recent evidence suggests that a mixture of oleic acid and erucic acid, known as "Lorenzo's Oil," administered to boys with X-ALD prior to symptom onset can prevent or delay the appearance of the childhood cerebral form. It is not known whether Lorenzo's Oil will have any beneficial effects in AMN. Furthermore, Lorenzo's Oil has no beneficial effect in symptomatic boys with X-ALD. Bone marrow transplantations can provide long-term benefit to boys who have early evidence of the childhood cerebral form of X-ALD, but the procedure carries risk of mortality and morbidity and is not recommended for those whose symptoms are already severe or who have the adult-onset or neonatal forms.

Prognosis

Prognosis for patients with childhood cerebral X-ALD is generally poor due to progressive neurological deterioration unless bone marrow transplantation is performed early. Death usually occurs within 1 to 10 years after the onset of symptoms. Adult-onset AMN will progress over decades.

Clinical Trial

Select this link to view a list of studies currently seeking patients.

Organizations

United Leukodystrophy Foundation

224 North 2nd Street, Suite 2

DeKalb, IL 60115

<https://www.ulf.org/>

(815) 895-3211

(800) 728-5483

National Tay-Sachs and Allied Diseases Association

2001 Beacon Street, Suite 204
Boston, MA 02135
<https://www.ntsad.org>
(800) 90-NTSAD

National Organization for Rare Disorders (NORD)

55 Kenosia Ave
Danbury, CT 06810
<https://www.rarediseases.org>
(203) 744-0100
(800) 999-NORD (6673)

Myelin Project

P.O. Box 39
Pacific Palisades, CA 90272
<https://myelin.org/>
(800)-869-3546

Global Foundation for Peroxisomal Disorders

5147 S. Harvard Avenue, Suite 181
Tulsa, OK 74135
<https://www.thegfpd.org/>

This information was developed by the Office of National Institute of Neurological Disorders and Stroke, National Institute of Health.

National Institute of Neurological Disorders and Stroke. NINDS Adrenoleukodystrophy Information Page. Available at: <https://www.ninds.nih.gov/Disorders/All-Disorders/Adrenoleukodystrophy-Information-Page>. Last accessed January 31, 2017.

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