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Adrenoleukodystrophy

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). *Adrenoleukodystrophy Information Page*.

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; such carriers almost never develop symptoms that are 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.