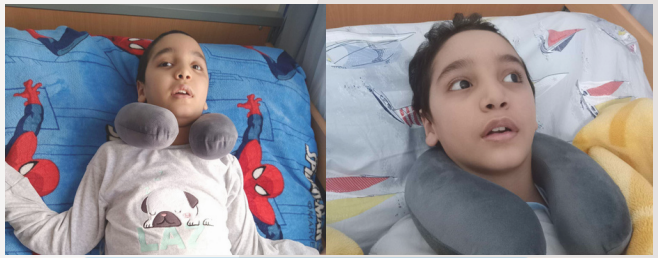
NO CURE!!

There is no cure for ALD in any of its symptomatic forms.
There are lifesaving options available for children with childhood cerebral ALD (the most severe form of ALD). If performed in the very early stages of cerebral ALD, hematopoietic cell transplantation (HSCT) can stop the progression and degradation of myelin in the brain. After intensive chemotherapy, HSCT uses bone marrow stem cell transplantation (BMCT) from another person who "matches" with the patient.
This procedure uses donor-derived cells to make the protein the recipient is unable to make. This procedure is not without risk. Infections, graft-versus-host disease, and other complications could prove to be fatal. Although HSCT may save your life, it is not a cure. Research shows that AMN may be present in patients who have had HSCT as children.
Gene therapy is a promising treatment to stop demyelination's progression with its own risks.



Mohamed Yassin: ALD Warrior

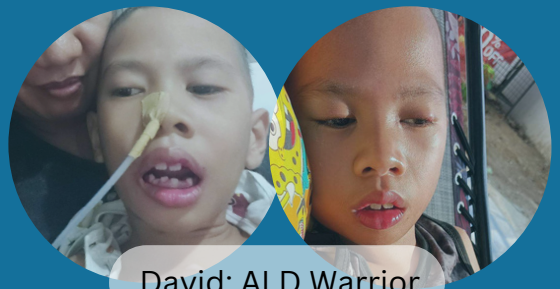
What is Myelin

Also known as the "white matter" in the brain. It is made up of protein & fatty molecules. Myelin provides the protective cover for the nerve cells. It's similar to an insulation surrounding an electrical wire. Myelin allows for precise and rapid transmission of information between neurons in the brain and the spinal cord.

This inhibits nerve conductivity, which can lead to neurological deficits. The brain is destroyed by childhood cerebral ALD.

A mutation in the

ABCD1 gene



David: ALD Warrior

DONATE

ALD Hope
OUR KIDS OUR PRIORITY

FOLLOW US

@ALDHOPE

ALD Hope
OUR KIDS OUR PRIORITY

All you need to know about X-Linked Adrenoleukodystrophy or ALD

WHAT IS ADRENOLEUKODYSTROPHY (ALD)

www.aldhope.org

+1(888)960-0ALD

WHAT IS ALD?

Adrenoleukodystrophy (ALD) is a rare genetic disorder (1 in 17K) that is caused due to a mutation in ABCD1 gene that prevent the body from breaking down very long chain fatty acids (VLCFAs). As a result VLCFAs build up in the brain, nervous system and adrenal glands causing destruction of the myelin sheath around nerve cells. Progressive neurological symptoms such as cognitive decline, vision and hearing loss, seizures and brain damage can result. ALD can also lead to adrenal insufficiency which can prove fatal if it is not treated. There are several types of ALD, including childhood cerebral ALD, adult-onset ALD, and adrenomyeloneuropathy (AMN).

Why boys?

The X chromosome is home to the damaged gene responsible for ALD. The X chromosome is the only one that boys inherit from their mothers. Two X chromosomes are passed to girls from each parent. Female children are protected by the functional copy passed down from their fathers. The mutation is often referred to by females as "carriers". They can pass the abnormally X chromosome to their offspring. The field is still evolving and we are beginning to see that some women may experience neurological symptoms in later adulthood. It is possible for girls to inherit two copies of the mutation, but it is very rare. All ALD men pass it down to 100% of their girls who will be carriers.

How about girls?

ALD can also affect women, but it is much less common in them than in men. The ABCD1 gene can be found on the X-chromosome. This means that males with only one X-chromosome will be more likely to be affected. Mothers with a mutation in the ABCD1 gene have 50% of passing it to their children.

In some rare cases women may show ALD symptoms and progression or adrenal insufficiency. those are known as "**heterozygotes**"

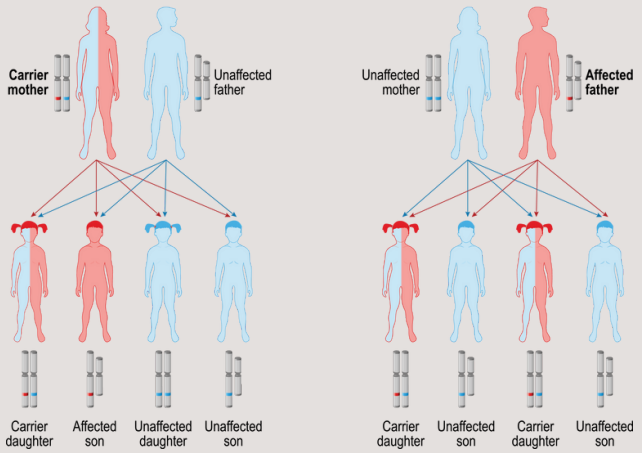
HOW TO DISCOVER ALD?

Two steps are required to test males for ALD. The first step is to measure the plasma concentration of VLCFA. If it is abnormal, the second step will take place by doing ABCD1 molecular genetic testing to assure the diagnosis. For females, only the genetic testing to be completed since VLCFAs exam is not accurate for females. A doctor or genetic counsellor can recommend commercial companies that can do the necessary genetic testing.

HOW DO YOU GET ALD?

Adrenoleukodystrophy (ALD) is an inherited genetic disorder caused by mutations in the ABCD1 gene. This gene provides instructions for making a protein that helps break down certain fatty acids. When the ABCD1 gene is mutated, the protein is not produced correctly, which leads to the buildup of fatty acids in the body, particularly in the brain and adrenal glands. This buildup can cause damage to the nervous system and result in the symptoms associated with ALD. ALD is inherited in an X-linked pattern, which means that it primarily affects males, who only have one X chromosome. Females can also be carriers of the mutated gene and may pass it on to their children.

X-linked recessive inheritance



ALD TYPES

There are several types of Adrenoleukodystrophy (ALD) which can affect individuals in different ways:

1. Childhood cerebral ALD: This is the most severe and common form of ALD, which primarily affects boys. It involves the degeneration of the myelin sheath surrounding nerve cells in the brain, leading to progressive neurological symptoms such as cognitive and behavioral changes, vision and hearing loss, and motor function impairment.
2. Adrenomyeloneuropathy (AMN): This type of ALD usually affects males in their late teens or adulthood. It involves damage to the myelin sheath surrounding nerve cells in the spinal cord and peripheral nervous system, leading to progressive neurological symptoms such as muscle weakness, stiffness, and difficulty walking.
3. Addison disease: This type of ALD affects the adrenal glands, leading to adrenal insufficiency, which means the glands do not produce enough hormones. Symptoms include fatigue, weakness, weight loss, and low blood pressure.
4. Asymptomatic ALD: it occurs in people who have a mutation in the ABCD1 gene but no symptoms.

It's important to note that not everyone with ALD will experience the same symptoms, and the severity of symptoms can vary widely even among individuals with the same subtype of ALD.

ALD Newborn screening

Every 36 hours a baby is born with ALD. Early diagnosis of adrenoleukodystrophy is the key to saving lives, because newborn screening allows prospective monitoring for adrenal function and the onset of cerebral ALD. A newborn screening test has been developed. It detects elevated VLCFA levels (as C26:0-lysoPC) in bloodspots.

ALD newborn screening has been successfully implemented in 37 states in the United States of America.