

AdrenoLeukoDystrophy

X-ALD is a genetic disease that is caused by a mutation on the X-Chromosome which prevents the production of a transporter protein that binds with Very Long Chain Fatty Acids (VLCFA's). Without this protein they can not enter the peroxisome where they would normally be broken down into smaller chains. These VLCFA's accumulate in tissue and blood and over time may affect the adrenal gland and the myelin sheath in the brain. Of those affected 80% of boys and 50% of men will have adrenal insufficiency. Once there is Myelin involvement the disease can progress very rapidly causing neurodegeneration and cognitive problems. Females have a 50% chance of passing it on to their children. Males cannot pass it to their sons, but will always pass it to their daughters. In many cases, a family history of ALD is not discovered until a boy presents with the severe childhood onset, even though some relatives may have been symptomatic for some time.

OF NEARLY 1600 BOYS DIAGNOSED THROUGH KENNEDY KRIEGER INSTITUTE, ALD AND AMN WAS TRACED TO OVER 20,000 OTHER FAMILY MEMBERS.

There have been 1024 mutations reported in the ABCD1 gene of which 505 are unique mutations. This wide variation makes it nearly impossible to determine how the disease will progress from one child to another or among adults. Many boys develop visual, hearing and speech problems and as the disease destroys the brain may be prone to psychotic episodes. Medications used to calm them have adverse side affects increasing stiffness and spasticity of the joints.

Although considered rare, approximately 150 boys are diagnosed each year in the United States, most because of neurological symptoms. We must diagnose boys who are at risk of developing cerebral ALD before they exhibit cognitive problems associated with demyelination. A newborn screening test has been developed by Kennedy Krieger Institute and recently approved in Maryland. We need to be testing all babies.

COMMON CHILDHOOD AILMENTS MAY NOT BE NORMAL.

There are early warning signs that are often dismissed as common childhood ailments, but now with the development of a simple diagnostic blood test, we should be testing all boys who present with any one of these symptoms, especially ADD/ADHD.

Many boys between 4 and 10 years of age are clumsy, have difficulty staying focused, wear eyeglasses and are born with undescended or underdeveloped testicles. But sudden changes should be of major concern. Childhood onset migraines, vomiting, failure to heal or recover from a minor illness and declining performance in school, especially noted in their hand writing skills, are often noted by parents prior to getting the diagnosis of ALD.

Many boys don't get diagnosed until they experience an Addisonian crisis which can cause the disease to progress more rapidly.

COMMON SYMPTOMS

- ADD/ADHD, losing information
- Visual disturbances, lazy eye, strabismus
- Behavioral problems
- Declining writing skills
- Seizures
- Eye pain/Childhood onset migraines
- Recurring viral infections
- Lethargy, tires easily
- Clumsiness
- Hypoglycemia
- Undescended or underdeveloped testicles
- Tanning or bronzing of the skin

EMERGENCY SYMPTOMS

- Acute adrenal insufficiency; vomiting, headache, unconsciousness or coma
- Mimics Spinal Meningitis

Without early detection
ALD usually leads to a vegetative state
within 6 months to 2 years
after diagnosis, followed by death.

Early Diagnosis is KEY!

Misdiagnosis plays a major roll in the delay of identifying ALD in a timely manner. Teachers are often the first ones to note changes in the boys and refer the child to the school nurse who informs the parents that they suspect ADD/ADHD, although this does not come on suddenly in 2nd grade. Many boys are experiencing cognitive difficulties which may include vision and hearing disturbances that are causing ADD like symptoms. If it is ALD these changes indicate that the boy is already in the early stages of cerebral involvement and his options for treatment are good. But at this stage the disease can also progress quite rapidly. White blood cells programmed to fix the problem create the opposite effect by causing inflammation which in turn causes more damage. It is an autoimmune response to the disease.

Treatments

Steroids support the Adrenal gland function. An emergency injection is prescribed in case of accident or injury which could be life threatening without the shot. The adrenal hormone helps the body deal with stress. Emotional stress and illness may present the need to temporarily increase the dosage. Many boys may experience adrenal crisis even though they are taking medication.

Lorenzo's oil has proven to lower the VLCFA's to a normal level and slow, or even stop the progression of the disease as reported July 11, 2005 by KKI (www.kennedykrieger.org)

Bone Marrow or Umbilical Cord Blood transplant is the only treatment once there is cerebral involvement, but all too often the disease is too far advanced for a successful outcome. Additionally, identifying a donor can prolong the time between diagnosis and transplant eating up valuable time. There is also the risk of infection or rejection post transplant, although survival rates have increased with the introduction of new BMT drugs. Some boys end up in a complete vegetative state, although the transplant was considered to be a success by medical standards.

Gene Therapy is the new hope on the horizon. A clinical trial is being conducted in France and is expected to expand to the U.S. by 2014. (www.businesswire.com/news/home/20091106005331/en)

For more information visit www.fightald.org.

**A SIMPLE INEXPENSIVE BLOOD TEST
IS USED TO DIAGNOSE ALD**

The test used to identify ALD is called a **Plasma Total Lipid Very Long Chain and Branched Chain Fatty Acids** test and can be drawn at any lab and sent to a number of facilities including Kennedy Krieger Institute and the Mayo Clinic. The cost of the test is under \$200. There is no need to do a DNA analysis unless it comes back positive and would then be needed to identify the mutation for testing other family members. A family history should be conducted to determine if there is evidence of any undetermined illnesses or deaths, MS, debilitating arthritis, or degenerative disc disease. This research should include all family members. Many males are diagnosed with Addison's disease, an illness that affects the Adrenal gland. 50% of males with Addison's will also have ALD or AMN so endocrinologists need to be routinely testing for these diseases upon diagnosing Addison's.

AMN

AMN or Adrenomyeloneuropathy is the adult onset of Adrenoleukodystrophy and is more prevalent among males than ALD. There are similarities in that there is a missing protein and an accumulation of VLCFAs, but only approximately 15% of men will have brain involvement. This accumulation instead leads to a mixture of axonal loss and multifocal demyelination in the spine. Symptoms can be progressive with stiffness and weakness in the legs, abnormalities of sphincter control, sexual dysfunction, incontinence and depression. Many adults are misdiagnosed with MS, debilitating arthritis and degenerative disc disease.

Once believed to only affect males, it is now known that AMN also affects women, who until recently were identified only as carriers. Now they are considered patients too, as statistics show they will develop some symptoms during their lifetime. It is believed to begin later in life for most women. Approximately 50% will show signs by age 45. Most men commonly develop symptoms between the ages of 18 and 35. There are no treatments for AMN at this time. Most people learn to live with their disabilities and get some relief through medications and other therapies. Bone marrow transplants are being evaluated as a possible treatment for some AMN men with cerebral involvement.

**STATISTICS SHOW THAT ANYONE WHO HAS THE
ALD GENE WILL BE AFFECTED AT SOME POINT.**

Although a mere 2% show symptoms by age four, that figure increases to 33% by age ten, 50% by age twenty and 98% by the age of fifty-two.

Please read the MediView report titled
**“AVOIDING THE MISDIAGNOSIS OF
ADRENOLEUKODYSTROPHY: DISTINGUISHING
ALD FROM ADD/ADHD”**
at www.stopald.org

FIGHTING ILLNESS THROUGH EDUCATION

Sawyer's courage set an example for us all.
One that will lead us forever forward
in our own goals and desires to persevere.
We will remember him fondly
and continue our fight against
Adrenoleukodystrophy in his honor.
Janis Sherwood/Founder

*Personal note**

After losing Sawyer to ALD I became intent on spreading awareness with the hope that at risk boys would get their diagnosis in a timely manner. Since then, it became apparent that there is a lack of education in many medical schools and CME programs so in 2009 I took my mission on the road. I travel in an RV and visit medical schools, hospitals, and clinics to distribute this information and as of this printing have been to nearly 2000 facilities across 43 states.

I am a member of the ALD AMN Global Alliance, a coalition of ALD organizations from around the world who have united to spread awareness and raise money for research and a cure. These include The Myelin Foundation, Stop ALD, ALD Life, Cure ALD, the Australian Leukodystrophy Support Group and the Be A Hero, Be A Donor foundation.

I hope you are inspired to help us spread awareness. Your questions and comments are appreciated. Please e-mail me at janis@fightald.org

JOIN THE FIGHT

Tax deductible donations to help spread awareness can be mailed to: Fight ALD,
P.O. Box 3318, Vista, CA. 92085
(Tax ID #56-2467099)
or made on-line at:

www.FightALD.org

www.FightALD.org
Fight ALD.org
Adrenoleukodystrophy
Fighting Illness Through Education

In honor of



Sawyer Benjamin Sherwood
11/16/1994 - 9/30/2003

Diagnosed 1/2/2003

**X-Linked Adrenoleukodystrophy,
is a genetic disease that affects**

1 in 15,000 boys

**in childhood but is usually misdiagnosed or
goes undetected until the disease is too far
advanced for treatment and chances
for survival are minimal.**

**It is easy to diagnose and it is treatable
if detected in the early stages.**

ADD/ADHD is the #1 misdiagnosis.

www.fightald.org