AdrenoLeukoDystrophy

ALD is a genetic disease that is caused by a mutation on the X-Chromosome which prevents the production of the ALD protein that allows Very Long Chain Fatty Acids (VLCFA's) to enter into the peroxisome where they would normally be broken down into smaller chains. These VLCFA's accumulate and over time may affect the adrenal gland, the myelin sheath and other organs. Of those affected, 80% of boys and 50% of men will have adrenal involvement as do some women. 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.

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.

Newborn screening would provide early detection to all newborn children.

50% of Males with Addison's disease will also have ALD or AMN

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, perhaps we should test all boys if they present with any one of these common symptoms, especially 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.

For more information visit: www.fightald.org

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 ADHD. This disorder does not come on suddenly in the second grade. Many boys are experiencing cognitive difficulties which may include vision and hearing disturbances that translate to problems in school. 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 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 an extra boost. 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.

Bone Marrow or Umbilical Cord Blood transplant is the only known cure at this time, 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 for a possible cure. Bluebird bio, a biotechnology company is currently conducting a clinical trial for ALD boys. You can learn more about this study at www.starbeamstudy.com.

A SIMPLE INEXPENSIVE BLOOD TEST IS USED TO DIAGNOSE ALD

A family history should be conducted to determine if there is evidence of any undetermined illnesses or deaths, MS, debilitating arthritis, degenerative disc disease or unknown brain disease. This research should include all family members..

The test used to identify patients with 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 gene for testing other family members. Many males are diagnosed with Addison's disease, an illness that causes adrenal insufficiency. A few symptoms of this is bronzing of the skin, vomiting, dehydration, hypoglycemia, fatigue and recurring viral infections.

AMN

AMN is the adult onset of ALD, short for Adrenomyeloneuropathy, and is more prevalent among males than ALD. There are similarities in that there is a missing protein and an accumulation of VLCFA's, however, the myelin sheath of the spinal column is affected first. Approximately one third of males will also have brain involvement. 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. Bone marrow transplants are now seen as a viable treatment for some AMN men. Most people learn to live with their disabilities and get some relief through pain medications and other therapies. Once believed to only affect males, it is now known that any one with the ALD gene will have some form of the disease in their life time. It is believed to begin later in life for women and progress slower, alhtough new evidence may show differently. Men commonly develop symptoms between the ages of 18 and 35. The progression and severity of the disease can vary greatly from patient to patient.

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*

I founded Fight ALD after Sawyer lost his fight to this disease, to spread awareness to medical professionals and our communities with the hope that at risk boys will get their diagnosis in a timely manner. After visiting 3200 medical facilities across 48 states over the last 5 years, I have now turned my attention to screen all newborn babies for ALD. It has recently been added to the Recommended Uniform Screening Panel (RUSP) by the Secretary of Health. This is merely a recommendation and is up to each state to determine whether or not they add it to the panel of diseases they currently screen for. As of this printing California, New Jersey, Connecticut, Illinois and Florida have either added it or are in the process of doing so. New York passed legislation and has been screening for 26 months, identifying 44 babies and numerous other family members.

I hope you benefit from this information.

You can find a link to a printable copy of this brochure at www.fightald.org/links.htm.

I welcome your questions and comments. Please e-mail me at janis@fightald.org.

Join The Fight

Deductible donations (Tax ID #56-2467099) to help spread awareness can be made on-line at:

www.FightALD.org

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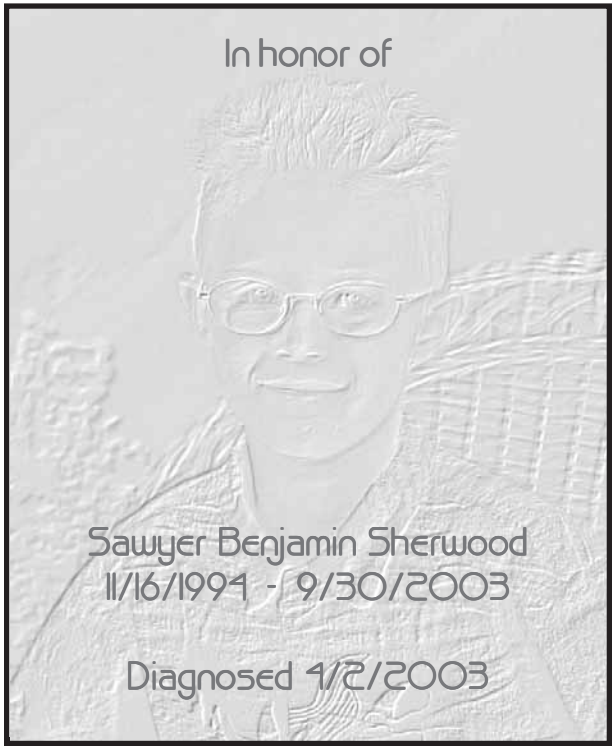
Adrenoleukodystrophy

Fighting Illness Through Education



X-Linked Adrenoleukodystrophy (ALD) and Adrenomyeloneuropathy (AMN) are genetic diseases that affect

1 in 15,000 people.

ALD, also known as Lorenzo's oil disease, affects boys in childhood and is the most severe form of the disease.

AMN affects both men and women.

ADD/ADHD is the primary misdiagnosis in boys while many adults are often presumed to have MS (Multiple Schleoris).

A simple inexpensive blood test is available to diagnose affected persons.

Unfortunately, many boys will not survive because of late diagnosis.

www.fightald.org