WHAT ARE THE TREATMENTS?

Now that your baby has been identified to have the ALD gene you will be advised about an appropriate care plan for your son that will include regular MRIs and blood work to monitor any changes. You have an advantage many families before you did not have the benefit of. If his blood work comes back abnormal for adrenal insufficiency it is easily treated with a corticosteroid in either liquid or tablet form. If there is evidence of changes in the MRI a bone marrow(BMT), stem cell or cord blood transplant is used to stop the progression of ALD. There is a 100% survival rate with a sibling match. With the opportunity to have a transplant before symptoms are evident you can be assured he will have the best possible outcome if it comes down to that. It is too early to say if an experimental study using gene replacement therapy may be a preferred treatment in the future.

What Happens Now?

Educate yourself and your family so that you can make informed decisions for all of you. Get Genetic counseling so you can identify any other family members who may also have the ALD gene.

For your son, follow the recommended protocol with his medical team to do blood work and MRIs as well as determine if there is a donor match in the family in case transplant becomes necessary. We also encourage you to sign up to be a donor through BAHBAD.org. If you're not a match for your child you may be for someone else's.

Follow the progress of experimental studies and know your options.

Get your support system in place so if the need arises and your son would need a life saving bone marrow, cord blood or stem cell transplant you can dedicate your time to him and his hospital stay which can last up to 100 days.

Get your finances in order including determining what your insurance coverage is should transplant be needed.

And Most Importantly...BREATHE!

Take care of yourself. Don't be afraid to ask for help. Reach out to the many organizations who are here for you. Many of us have been on this journey and can offer advice, emotional support, answer questions or just lend an ear.

And when the dust settles and the initial fear subsides, and it will, offer your support to the many organizations that have made it possible for your children to be screened at birth for ALD.

RESOURCES

The Myelin Project
www.myelin.org

Be A Hero, Become A Donor.
www.bahbad.org

Fight ALD-Fighting Illness Through Education
www.fightald.org

The United Leukodystrophy Foundation
www.ulf.org

ALD Connect
www.aldconnect.org

Starbeam Study
www.starbeamstudy.com

California Dept. of Health
www.cdph.ca.gov

This brochure has been put together with love by parents affected by ALD.

My newborn has ALD... What Happens Now?

First and foremost know that You Are Not Alone!

The fact that you have an early diagnosis gives you an advantage that many family's never got. Although this may feel devastating now, you will learn that it truly is a blessing. Late diagnosis often leaves no option for treatment to halt the progression of the disease.

Try and focus on the positive. With the advancement of newborn screening and treatments you will find that what may seem like a curse today, will turn out to be a blessing in the long run. You now have the tools and time to get your son the care he needs.

KNOWLEDGE IS POWER!

The information in this brochure is intended to help you understand what ALD is. It lays out a health monitoring protocol that will ensure your baby has the best chance to thrive, help you put together a list of things you can do now to prepare for challenges that may arise in the future and connect you with other affected families and organizations that can offer emotional and financial support.

In Honor of our Heroes!

What Is ALD?

Adrenoleukodystrophy is an X-linked genetic disorder that affects approximately 1 in 17,000 newborns worldwide. It is caused by a mutation on the X chromosome which has been identified as the ABCD1 gene. This mutation prevents the body from producing a working ALD protein that aids in the degradation of Very Long Chain Fatty Acids (VLCFAs). Because it is an X- linked disease it is more dominant in males because they carry only one copy of the chromosome.

Females carry two copies so they have an advantage in that only one X is affected and the other one makes enough of the ALD protein to prevent the childhood onset of the disease.

The accumulation of VLCFAs can cause adrenal insufficiency and deterioration of the myelin sheath in the brain. The most severe form of the disease is the childhood cerebral onset (cALD). However, there are four different phenotypes and only 30-40% of affected males will develop this form.

In cALD symptoms typically present in boys between the ages of four and eleven. These can include but are not limited to vision disturbances, hearing problems, attention difficulty, moodiness and changes in gait to name a few. These usually aren't big red flags for a serious illness so they often are dismissed as normal childhood problems and the disease may have progressed too far for treatment by the time ALD is diagnosed.

Early diagnosis is key!

How did my baby get ALD?

ALD is an inheritable disease caused by a mutation on the X-chromosome. Females have 2 X's and males have one X and one Y. The sex of a baby is determined by their chromosomes. Mothers can only pass an X and will have a 50% chance of passing the affected one to any of her children. A girl will get one X from each parent. Either parent could have passed the affected X. A boy will get his X from his mother and his Y from his father, therefore the mother would have passed the affected chromosome. Rarely a baby will mutate the gene on their own but it has happened. Once it is determined which parent passed the affected X other family members on that side should be screened.

Understanding Phenotypes of ALD

In the world of genetic disorders doctors group collections of symptoms into "phenotypes" based on the cells and tissues that are most severely affected by a gene abnormality. Individuals with the ALD gene may have one of 4 different phenotypes which can overlap in one's lifetime.

- **Asymptomatic**
- **Adrenomyeloneuropathy (AMN)**
- **Adrenal Insufficiency (AI)**
- **Cerebral ALD**

The onset and severity of symptoms can vary from patient to patient.

Asymptomatic ~

Meaning symptom free. Although some with the ALD gene may have no symptoms until later in life it has been determined that anyone with the gene will develop some form of the disease in their lifetime.

Adrenomyeloneuropathy ~

AMN is the adult onset. Males who did not develop cALD can present with symptoms similar to Multiple Sclerosis (MS) as early as late teens and as late as their 60's or anywhere in-between.

Women typically develop symptoms after the age of 45, but that isn't always the case.

Adrenal Insufficiency ~

AI, also known as Addison's disease, is the result of the adrenal glands not producing enough stress hormone. This is easily treated with a pennies a day cortisol tablet. Without treatment an adrenal crisis could cause death. Symptoms are hypoglycemia, lethargy, high fevers, vomiting and bronzing of the skin.

Cerebral ALD ~

The myelin sheath works as the protective covering of nerves similar to the plastic coating on electrical wiring. Lack of the ALD protein causes the myelin to deteriorate and short circuit messages to other organs which results in loss of vision, hearing, motor skills, and causes more serious problems that often result in death.

AMN causes problems with the myelin and neurons in the spinal column. Approximately 30% of men with AMN will also develop cerebral involvement.