ALDConnect

PURPOSE AND *Possibilities*

ANNUAL REPORT 2022

Mission

We aim to improve health outcomes for patients with X-linked adrenoleukodystrophy (ALD) by empowering patients, raising awareness and accelerating the translation of scientific advances into better clinical care.

Vision

We strive to revolutionize care, end suffering from ALD, and find a cure.

DEAR ALD *Community,*

We are honored to present ALD Connect's impact report and are excited to highlight our important work from this last fiscal year January 1, 2022–December 31, 2022.

In 2022, our goal was to help patients with ALD live their best lives. We hit major milestones for our community and organization in working toward this goal.

In July, we hosted an Externally-Led Patient-Focused Drug Development meeting, which focused on adult ALD and AMN patients. This was an opportunity to educate FDA staff about ALD and amplify patient perspectives. This is critical to help provide context when FDA makes regulatory decisions for new drugs. A **Voice of the Patient Report** is published on our website and reflects the main take-aways from this meeting. Our hope is that it will influence every stage of drug development and will be a resource for the ALD community.

In September, the U.S. Food and Drug Administration (FDA) granted the first FDA-approved gene therapy for cerebral ALD.

In October, we announced our three **Emerging Investigator** awardees, who received grants from ALD Connect to further their novel ALD-related research projects. We look forward to hearing updates from these promising researchers in 2023. We also approved funding for "The Grey Zone" project, which is a collaborative, multi-site research project aimed at identifying which genetic variants in the *ABCD1* gene put patients at the most risk for developing ALD symptoms.

In November, we were thrilled to be back in person for our **2022 Annual Meeting** and **Patient Learning Academy** in Baltimore, Maryland, where we saw many familiar faces and welcomed new members to the community.

We continued our monthly **Community Calls** and expanded our **Peer Mentor Program**. In addition, we fundraised, collaborated with other organizations, hosted webinars on new topics and met with our Industry Advisory Council. Our donors enabled us to provide financial support to several ALD families through **The Myelin Program Patient and Family Support Program** and the **Michael Benton Travel Scholarship Program**.

While we are proud of the work we have done this year, there is more to be done. We must continue to advocate for unmet needs within the ALD community and provide support for patients and families.

With Gratitude,

Florian Eichler, MD
Co-Founder, President

Kathleen O'Sullivan-Fortin
Co-Founder, Treasurer, General Counsel

SHARING OUR *Stories*

JANA'S STORY:
HOPE FOR TOMORROW

Progressive and incurable. Those are the words I'll never forget that followed the diagnosis of Adrenoleukodystrophy that had taken three long years to find. Part of me was happy to finally put a name to the horrible symptoms I had been experiencing. Not to mention putting an end to the countless sleepless nights, wondering if this was "all in my head," after running into dead ends at too many specialists to count. The other part of me was scared to realize what the "progressive" adjective would bring, but I knew I would do everything in my power to slow the progression. I was told that I needed to come to grips with the reality that I would be bound to a wheelchair within 3–5 years because of the debilitating muscle weakness that was to come. But she also told me that I was a unicorn...a unique, beautiful, one of a kind masterpiece and I should never forget that.

Being asked to verbalize how ALD impacts my daily life is a very difficult and emotional question, as I make it a part of my daily life to not focus on what I can't do, but instead count the blessings I have. I consider myself to be a very positive, upbeat person, but no one knows the struggles that I face internally each day.

Because of the stiffness, I start every day with a session of stretching which helps greatly. In addition, I exercise 5–6 times per week. I've found I get the best results when using a personal trainer 2–3 times per week. Believe me, I would rather not pay for those personal training sessions and it would be way more fun to do group training, but my physical limitations prohibit me from doing so. My best advice to anyone diagnosed with ALD is to keep moving. The biggest challenge I faced when going to a trainer was simply getting started. Along with so many other burdens, ALD has severely limited my physical abilities, so every day I remind myself that what's most important is doing my personal best, and never comparing my abilities to anyone else's.

> **...she also told me that I was a unicorn... a unique, beautiful, one of a kind masterpiece and I should never forget that"**
>
> **— JANA HALE**

I was recently given the honor of receiving the 2022 "Most Inspiring" award at my gym. I feel so fortunate to be a part of a gym family where I can come as I am, be cheered on and celebrated for my progress, no matter how big or small it might be.

I also see a physical therapist 1–2 times weekly who is amazing at recommending exercises for me that work with my abilities and help me maintain the strength I have. I feel so blessed to have found a village of providers who are helping me navigate all of the unknowns associated with ALD.

My daily outings have been severely limited by ALD. I no longer go anywhere by myself that's unfamiliar or where I can't push a cart. Just the thought of walking from my car into a store to grab onto a cart gives me anxiety. Thankfully, I am married to an amazing man and have the most understanding and encouraging daughter and they are both fighting right alongside me. They help me unconditionally to live my best life. One thing that ALD has forced me to do is to give myself grace and realize that some days will be better than others. I've learned to delight in those good days and never take them for granted.

It is my hope to see a therapy developed that would improve my quality of life and enable me to unburden to my loved ones. My six-year-old nephew hasn't the slightest clue of the disorder I have been diagnosed with and it is never a discussion, but every time he tells me goodbye he says, "Bye, Auntie Jana...I hope your legs feel better soon." It is my dream that, as a result of new treatments or therapies, I can finally tell Callen my legs are feeling better.

BRANDON TURNER:
FINDING THE SILVER LINING

My history with this disease is a short one. I was diagnosed with AMN in August of 2021, followed shortly thereafter with a conclusive genetic test.

Without a known history of ALD in my family, there was an additional year-long period of uncertainty prior to my diagnosis. First came the routine physical exam: "Perfectly fit, but I'm happy to refer you to a neurologist." Then came the MRI and confirming there was definitely something going on. I remember sitting in the neurologist's office as he's going over the images. Not being a medical professional, it wasn't quite hitting home until I asked what a normal spinal cord looked like. Oh, THAT's normal? Ok, even my untrained eye can see that one of these is not like the other.

With the advantage of hindsight, I can say that my symptoms started in my early 20s, where a full sprint didn't feel quite right for some reason. In my late 20s, jogging became stiff and kind of springy. "You're getting a little older and have let your conditioning slip," I thought. At the time, both were true and more logical than a rare disease I'd never heard of.

Now in my mid-30s, my symptoms are fairly mild; mostly consisting of gait and balance issues. I've found the slow progression of the disease to be a double-edged sword. On the one hand, I am able to constantly adapt to my new, ever-diminishing abilities before I'm even aware of them.

On the other hand, it can be cruel because I'm constantly reminded of this progression, and there's nothing I can do to stop it.

Despite that and not fully knowing at what pace and with what symptoms the disease will progress, I know that in many ways I'm one of the lucky ones in the AMN/ALD community.

I was incredibly lucky to have lived my childhood completely unaware of the disease. Blissful ignorance in its purest form.

I'm lucky I chose a career that is almost exclusively done from behind a desk. So long as I am of sound mind (fingers crossed), my ability to perform at work is more or less uncompromised.

I'm lucky that most of my hobbies happen to be intellectual in nature. I can't say that I ever had a desire to run a marathon or compete in an Ironman. Now I'll never have to worry about getting pressured into a 5k.

I'm lucky that I only had the one year before my correct diagnosis of swimming through misdiagnoses, where doctors had no idea what or if anything was wrong with me or were so convinced that I had something else that I was on the doorstep of expensive and potentially unnecessary genetic testing.

I'm lucky to have world class medical care all around me in Dallas, Texas. Once the diagnosis did look more serious, I had no trouble seeking out a second (better) opinion.

I'm lucky to have a neurosurgery resident in the family who spotted my odd gait on a hike and recommended that maybe I should see a doctor about it in the first place.

I'm lucky that on the eve of my first routine physical exam in years, the one that kicked off this whole journey, I went on a first date with a woman who turned out to be the love of my life. I'm beyond lucky that she's leaned into this diagnosis with me and has supported me emotionally (and who am I kidding, increasingly more often, physically). She's the most amazing, supportive partner I could ever ask for. I'm lucky.

My approach to living with AMN mirrors my approach to many things in life: worry about the things you can control. That's the productive side of worry. So what can I control?

At first it was having a regular schedule for seeing my doctors, taking medication on time, and getting that annual MRI. Then I found staying on top of stretching and practicing what I learned in physical therapy made the disease's impact on my day-to-day life more manageable.

It was during one of my regular doctor's visits that I learned about ALD Connect. Staying plugged into their community has provided me with access and opportunities to interact with the doctors, clinical researchers and pharmaceutical professionals who have dedicated their careers to this disease. And most importantly, their AMN Men's group has given me a sense of connection and a community where this disease doesn't feel so unfortunately unique.

Beyond that, what's there to do? Just worry? No thanks.

AMN's physical limitations will only increase, so I'm determined not to add to them. I'll continue to live as much of an active life as I can while I still can, seeking out new destinations and adventures. MusicFest in the mountains? Let's go. The rodeo in Cheyenne, WY? Saddle up. Wanna elope in the mountains of Colorado? I do. Just bring my hiking poles.

"Staying plugged into ALD Connect's community has provided me with access and opportunities to interact with the doctors, clinical researchers and pharmaceutical professionals who have dedicated their careers to this disease."

— BRANDON TURNER

IN MEMORIAM *Justin*

MOURNING A COMMUNITY LEADER

Justin D'Agostino was an activist, educator, and community leader. He worked as a teacher and recruiter. He wished to pursue a degree in naturopathic medicine. He owned his own home and was very independent.

At the age of 28, he began to have issues related to ALD, especially with his gait and walking. First came the cane, then the walker, soon the wheelchair. Justin was very intelligent and he began his intensive research on ALD.

ALD continued to claim more and more. The results of his MRI revealed extensive brain lesions, which increased dramatically within the course of a year. Justin continued to feel he could beat the disease, unfortunately, this was not going to happen. He continued with his studies and kept in touch with other AMN men, leading virtual support sessions.

Justin died on January 22, 2022, at 3am, peacefully, at the age of 37.

NICKI & GANESH'S STORY:
DISCOVERING JOY IN TODAY

My name is Nicki Bhandaru, and my son Ganesh has X-ALD. A little over 21 years ago, we were pregnant with our son. The son we had always wanted. We could not wait for him to be born and could not wait to see the person he would become. He was born in 2001 and was perfect in every way. He grew to be an adorable, sweet, kind, and smart boy who was gifted, both academically and athletically. He was the point guard and MVP of his basketball team, and moved up quickly in martial arts. He had a great sense of humor and a lot of friends. It was always a joy to go to parent-teacher conferences because his teachers loved him. He was bright, gentle, and extremely kind. We felt like he could be anything he wanted to be and that he would have a successful, happy life.

When Ganesh was in the 5th grade, I started noticing that he was having some mild reading comprehension issues. By 6th grade, it became clear that something was really changing. His academic performance started to decline, despite intense effort on his part. His handwriting suddenly became very sloppy. He became clumsy and started dropping things and falling. His skin was unnaturally darkening and his body was looking emaciated. He was exhausted and had increased moodiness, which was very unlike him. I noticed that he started to get a lazy eye that had gotten much worse over the past two years. After years of trying to narrow down the cause, we finally took him to a different optometry practice. That doctor sent us to a neuro-optometrist and we were eventually referred to Raleigh Neurology who ordered an MRI. At this point, we had seen several different doctors over 2 years. We finally got an MRI and found out that our sweet little 14-year-old boy had X-Linked Childhood Cerebral Adrenoleukodystrophy.

After consulting with more doctors at Duke, we were told it was too late for him to have a bone marrow transplant and there was effectively no treatment available. The only thing they were able to do was give him steroids for the Adrenal Insufficiency. We also found out that both of our daughters, my mother and I also have ALD. If only I had changed eye doctors earlier or had enough medical knowledge to push for an MRI at an earlier age, maybe things would be different. But, unfortunately, I did not.

All of our lives changed the day we received the phone call from Raleigh Neurology. I was told to call Make-A-Wish, as we did not know how much time we had before Ganesh's condition would progress even more. We were also told that usually after diagnosis, life expectancy is 6 months to 2 years. Ganesh started high school shortly after his diagnosis and he graduated in a power wheelchair with the assistance of a personal aide. He has since lost much of his vision, his ability to walk and his ability to use his arms and legs. He needs support with all of his daily living activities. We have hired several care-taking aides over the years to help me and my husband with moving, transferring, feeding, dressing him and more.

Life is very different from what we expected, but Ganesh has somehow managed to keep his amazing sense of humor and positive attitude. He hates being dependent on us and others for his daily care, but he makes the best of a bad situation. He wants to live and is just waiting for a cure or treatment. Until that time arrives, he spends time with his two dogs that bring him much joy and happiness. He wants to accomplish a lot in his life. For now, he is working on getting his college degree with the help of his aides for reading and writing, taking it one class at a time.

ALEINES' STORY:
PEACE IN COMMUNITY

My name is Aleines Valentin and I am from Puerto Rico. In August of 2013, two of my children were diagnosed with ALD. It was certainly unexpected because there was no family history with this disease. I think that like most families, this news was somewhat devastating, in our case doubly when we discovered that there were no experts on ALD on our island.

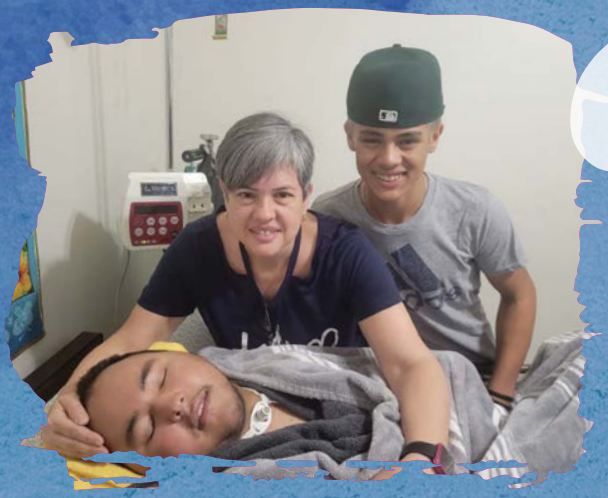
In the summer of 2012, my middle son Iván Andrés, suffered an adrenal crisis and was admitted to the intensive care unit, unfortunately his episode was mistaken for viral encephalitis. The MRI findings that were performed on him indicated a number of possible conditions and one of them was ALD, however the attending physicians did not take this into account. The findings of that study were not in our possession until 13 months later when a neurologist requested it. I am sharing this because it seems very important to encourage parents to seek information from specialists.

In our case, the limited number of specialists and the lack of knowledge about the disease did not give us that opportunity.

Once our children (middle and younger) were diagnosed by an excellent neurologist (in fact, the first cases of ALD for her), we began the search for information in a "lonely" way and facing the reality of the disease is something still impossible to describe to this day.

Going back to my story, my diagnosis arose 9 months after my children's. The VLCFA test was originally ordered for me, but the result was negative. Given the haste with which we had to attend to the emergency transplant for Iván Andrés, everything related to my follow-up was put on hold until May 2014, when they then performed a genetic mutation test on me. This test confirmed what the specialists already knew, I was the carrier and apparently it was due to a spontaneous mutation.

I thought that my constant spasms, muscle and extremity pain, among other things, were related to prolonged time sitting working, however, everything began to worsen and what worried me the most was that work was no longer the reason. Participating in the ALD Connect meetings in Spanish raised a red flag, because all the symptoms that the other women described made me feel like I was in front of a mirror.



My experience with ALD Connect was extraordinary, we kept in touch at all times, they were very supportive throughout the process and from an emotional point of view, I have no words to describe the peace I felt."

— ALEINES VALENTIN

One day through Messenger, I communicated with Kathleen O'Sullivan-Fortin and I shared what was happening to me, she advised me to contact Dr. Florian Eichler. I faithfully followed her advice and made an appointment even without having any financial resources to cover the expenses. This represented travel and lodging expenses, but my faith was bigger than that limitation.

I contacted staff at Massachusetts General Hospital to request information about organizations that might consider helping me make my trip to Boston and the name ALD Connect came up. Honestly, the only thing I knew about the organization was the monthly meetings they offered that I participated in.

I applied for help, thanks to God and to them, I was able to receive my evaluation on December 13, 2022. From my evaluation I obtained a series of recommended studies and laboratories. In addition, I was considered to be part of a research program on ALD in women.

My experience with ALD Connect was extraordinary, we kept in touch at all times, they were very supportive throughout the process and from an emotional point of view, I have no words to describe the peace I felt. My visit to the Massachusetts General Hospital, treatment and care by Dr. Florian Eichler, his team and hospital staff, was first class.

ALD Connect is certainly much more than an organization, staff, meetings or specialists–they are angels who are somehow on a mission to help, support and reach out to those of us who have been touched by ALD disease. Last but not least, I describe them as that oxygen tank when we no longer have any strength left. Their empathy, sensitivity and love make a difference.

THE MYELIN PROJECT
PATIENT AND FAMILY SUPPORT PROGRAM

ALD Connect is proud to carry on the legacy of The Myelin Project by offering grants to ALD patients and families who need financial assistance. In 2022, we have funded 10 grant applications for help with travel to medical appointments, mortgage payments and car payments. Your donation will help ALD patients, like our friend Michael, who received a new wheelchair through our program!

JANUARY 2022

Michael Benton Travel Scholarship for a family to fly from Florida to Minnesota

FEBRUARY 2022

Michael Benton Travel Scholarship for a family to fly from Oklahoma to California

Mortgage payment for a symptomatic woman with ALD in Missouri

MARCH 2022

Mortgage payment for a family in Michigan that has a son with cerebral ALD

MAY 2022

Rent and car payment for a family in Georgia that has a son with cerebral ALD

JULY 2022

Michael Benton Travel Scholarship for a family in Virginia to travel to a fertility clinic in New York

Michael Benton Travel Scholarship for a family in Georgia to travel to Minnesota

Michael Benton Travel Scholarship for a family in New York to travel to attend ALD Family Weekend in California

SEPTEMBER 2022

Custom wheelchair for a patient with cerebral ALD in Indiana

OCTOBER 2022

Michael Benton Travel Scholarship for a family in Mexico City to travel to Pennsylvania for an appointment

DECEMBER 2022

Mortgage payment and electric bill payment for a family in California that has a son with cerebral ALD

Rent payment for a mother in Florida whose son was hospitalized due to his ALD

MEET OUR *Team Members*

KATIE DOW

Project Manager

Katie joined ALD Connect in 2022. Katie is an experienced administrative professional and helps with a variety of projects. She enjoys learning more about the ALD community.

FELICITY EMERSON

Project Manager

Felicity joined ALD Connect as a Volunteer in 2020 and was hired as a Project Manager in 2021. She is currently finishing her PhD in Biomedical and Biological Sciences at Cornell University, and is looking forward to graduating in 2023. She plans to become a genetic counselor. She values every opportunity she has to learn from the ALD community about the experiences of families with ALD.

EMMA HAYES

Project Manager

Emma was ALD Connect's first summer Intern in 2019 and joined ALD Connect again in the spring of 2022. She has a BA in Chemistry from Saint Anselm College and an MS in Science, Technology and Entrepreneurship from the University of Notre Dame. Emma also works for Alexion Pharmaceuticals as a Patient Liaison in Neurology and is a Board Member and Treasurer for Remember the Girls. Emma is a dedicated and fearless advocate for ALD which stems from a family history with the disease.

KELLY MIETTUNEN

Executive Director

Kelly joined ALD Connect as the Executive Director in 2019. Kelly has worked in administration and with the ALD community for more than 12 years. She has a Master of Healthcare Administration degree from the University of Minnesota. She enjoys working with such an incredible network of patients, families, advocates, physicians, scientists and industry partners.

PATRICIA MUSOLINO

Board of Directors Member

Dr. Patricia Musolino joined the ALD Connect Board of Directors in February 2022. Dr. Musolino is a critical care and vascular neurologist at Massachusetts General Hospital/Harvard Medical School. She is an expert in neurogenetics and vascular neurology. Her laboratory work focuses on leveraging insights from neuroimaging and gene-editing tools to understand how single gene mutations alter blood brain barrier and cerebrovascular function. Dr. Musolino has been involved with ALD Connect for many years, especially with our Spanish-speaking community. We are grateful for her willingness to make further contributions as a member of the Board.

> "Educating medical professionals about Addison's Disease and Adrenoleukodystrophy as well as having correct information accessible in addition to finding treatments and dare I say cures, is needed for both male and females."
>
> **— BARBARA MACHADO**

2022 Annual Meeting and Patient Learning Academy

INSIGHTS AND *Possibilities*

2022 ANNUAL MEETING AND PATIENT LEARNING ACADEMY

We are proud to have hosted the largest ALD-specific meeting in the world! Our **2022 Annual Meeting and Patient Learning Academy** was held on November 11–12, 2022 in Baltimore, Maryland. Our theme was, "Live Your Best Life with ALD". At our meeting, we heard exciting updates from experts on ALD research and important recommendations for ALD monitoring, care, and mental health. With our patient panels, we ensured that doctors, researchers, and industry members hear the patient perspective. We are already looking forward to our 2023 Annual Meeting and Patient Learning Academy. You can check out the recordings from the Annual Meeting on our website or YouTube channel. Your donation will help make it possible!

Thank you to our sponsors:

bluebirdbio · minoryx therapeutics · SwanBio THERAPEUTICS · (vigil) NEURO

GENE THERAPY

ALD Connect is proud to advocate for treatment options for our community. bluebird bio's Cellular, Tissue and Gene Therapies Advisory Committee meeting was held on June 9, 2022, where committee members voted on whether to allow bluebird bio's gene therapy product for childhood cerebral ALD to move forward. ALD Connect worked with the third-party consultants and other advocates to help coordinate the patient testimonies for the Open Public Hearing. ALD Connect played the role we thought was appropriate—amplifying the patient perspective.

When the vote passed, we were told that it is uncommon for so many committee members to refer to the Open Public Hearing speakers as having an impact on their vote. Many stakeholders reached out to say how moved they were by the testimonies and how grateful they were for families sharing their experiences with ALD so openly!

EXTERNALLY-LED PATIENT-FOCUSED DRUG DEVELOPMENT MEETING

On July 22, 2022, ALD Connect hosted an **Externally-Led Patient-Focused** Drug Development meeting focused on ALD in adulthood: men with AMN or cerebral ALD and symptomatic women with ALD.

The goal was to educate the FDA on what it is like to live with the adult forms of ALD: what symptoms patients experience, how their lives are impacted, what kinds of treatments they need and what kinds of risks they are willing to accept to see improvements.

In 2022, we have proudly expanded our programming to the Spanish-speaking community. We offered live Spanish-translation of our Externally-Led Patient-Focused Drug Development meeting and have recently partnered with Fundación Lautaro te Necesita to Co-Host our Spanish Community Call.

Scan to view our Voice of the Patient Report, covering the main take-aways from the Externally-Led Patient-Focused Drug Development Meeting.

Thank you to our sponsors:

Autobahn Therapeutics

minoryx therapeutics

SwanBio THERAPEUTICS

COMMUNITY CALLS

ALD Connect coordinates periodic virtual meeting calls to facilitate community building and provide support. Whether it's men with AMN, women with ALD, cerebral ALD caregivers, or newborn screening parents, our Community Calls continue to bring support and deepen our community's connections with each other. These calls are not recorded.

You can always find the upcoming call schedule on our Community Calendar at aldconnect.org/calendar.

INDUSTRY ADVISORY COUNCIL

Our Industry Advisory Council (IAC) is composed of representatives from our ALD Connect Board of Directors and several companies that are working on ALD therapies. The purpose of the IAC is to bring together stakeholders for robust discussions about the disease, clinically meaningful outcomes, treatment approaches, and research. The IAC plays an important role for our organization. The recommendation to pursue an Externally-Led Patient-Focused Drug Development meeting came from this committee.

MARCH 2022

Understanding Zebrafish for Modeling ALD and Leukodystrophy Research
Josh Bonkowsky, MD, PhD

SEPTEMBER 2022

The Grey Zone
Stephan Kemp, PhD

DECEMBER 2022

The Neurocognitive And Mental Health Impact Of ALD: Insights For Observational Studies And Clinical Trials
Rene Pierpont, PhD, LP

NEW POSSIBILITIES *Through Research*

Funding research has always been a priority for ALD Connect. We have been very thoughtful about how to invest in ALD research projects while avoiding conflicts of interest, since so many ALD experts and centers are represented on our Board of Directors.

THE GREY ZONE

We are excited to share that in October 2022, we approved funding for a collaborative, multi-site research project, "The Grey Zone". This community-wide initiative is a great fit for ALD Connect as it includes many of our board members! As many know, adrenoleukodystrophy (ALD) is an inherited neurological disease caused by genetic variants of the gene *ABCD1* and is usually accompanied by increased levels of very long-chain fatty acids (VLCFAs). Babies with ALD are neurologically normal at birth. Boys with ALD are at risk of developing adrenal insufficiency and/or inflammatory demyelinating brain lesions (cerebral ALD). Newborn screening has revolutionized the diagnosis and treatment of ALD, as it allows for the early identification of at- risk boys and life-saving interventions. These include initiating timely adrenal steroid replacement therapy following detection of adrenal insufficiency, and providing allogeneic hematopoietic stem cell transplantation (HSCT) as a means of treating cerebral ALD.

However, in the past five years, newborn screening has identified many boys and families with novel variants of *ABCD1* who have no family history of any ALD-related disease symptoms, even with older relatives with the same variant. These novel variants are reported as "variants of uncertain significance (VUS)" on genetic testing reports. If a variant has not yet been linked to a specific disease, it is uncertain whether the variant is benign or pathogenic. However, a positive newborn screening result and referral to a medical specialist starts the process of follow-up and periodic testing for adrenal dysfunction and cerebral ALD by MRI.

Our goal is to understand which genetic variants put a male at risk for disease. This is an important project for our patients, families, and physicians. We are collaborating internationally in a joint project for this work and would appreciate your help to make this happen.

DONATE TODAY!

Please consider donating to "The Grey Zone"
Visit aldconnect.org or scan the QR code.

WEBINARS

ALD Connect hosts live, interactive webinars via Zoom that are free and can be accessed anywhere in the world. Experts from varying fields take questions and discuss a broad range of topics requested by ALD patients and their families.

View past webinars at **youtube.com/c/ALDConnect.**

MARCH 29, 2022

Community Webinar for ALD Connect's EL-PFDD Meeting

Kathleen O'Sullivan-Fortin
James Valentine, JD, MHS
Larry Bauer, RN, MA

MAY 3, 2022

Special Needs Financial Planning

Cynthia Haddad, CFP®, CHSNC

SEPTEMBER 27, 2022

EL-PFDD Debrief

Dr. Al Freedman

OCTOBER 6, 2022

EL-PFDD Debrief

Dr. Al Freedman

DECEMBER 13, 2022

Physical Therapy Considerations for the Management of X-Linked Adrenoleukodystrophy

Nisha Bhandaru, Doctor of Physical Therapy Student

"As a parent of a young boy with cerebral ALD who has received a transplant, I attend both the Cerebral ALD Community Call and the Parent and Caregiver Call. An ALD diagnosis can be lonely and isolating.

Both of these calls are amazing at providing not only healing in connecting with families at varying points on the diagnosis spectrum, but also immense support and encouragement to those new to navigating the world of ALD. Thank you ALD Connect for facilitating these sessions!"

— KIRSTEN FINN

NEW PROGRAM: *Emerging Investigators*

ALD Connect strives to engage the next generation of researchers and accelerate research. In 2022, we launched a new grant program with the goal to introduce graduate students, medical students and post-doctoral researchers to our field and foster their continued-learning and interest in ALD. Each applicant submitted a written proposal and then presented their research plan to a panel of ALD experts. Three awards were granted. Your donation will help support research projects and help us grow this program!

HEMMO YSKA, MD

Mentor: Marc Engelen, MD, PhD

Hemmo Yska is a PhD candidate at the Amsterdam University Medical Center in the Netherlands. After becoming a medical doctor, he worked as a resident in pediatrics. Due to his interest in scientific research and pediatric neurology, he decided to pursue a PhD where he focuses on clinical research in the field of ALD and other leukodystrophies. Currently, together with the other members of the Dutch ALD group, he is involved in a number of projects that focus on the addition of ALD to the newborn screening and the identification of surrogate outcomes for spinal cord disease.

Yska's Project: Attitudes Of Patients Towards Sex–Specific ALD Newborn Screening

Males with X-linked adrenoleukodystrophy (ALD) are at risk for developing adrenal insufficiency and/or progressive leukodystrophy (cerebral ALD) at an early age. Newborn screening (NBS) for ALD enables pre-symptomatic monitoring and therapeutic intervention, thereby preventing irreversible damage and saving lives. ALD has been included in NBS programs in more than 25 U.S. states and several European countries are considering its addition.

As girls are not at risk for developing the childhood cerebral form of ALD, some countries are reluctant to screen them for ALD. Sex-specific screening has not yet been implemented in newborn screening and there is limited scientific data regarding the attitude of patients to screening males only. In order to make a well-informed decision, it is important to understand the attitudes of all groups and stakeholders that are involved in the disease. Therefore, a group that cannot be overlooked is the ALD patients themselves. Patients and their direct relatives can provide valuable insights on matters such as sex-specific screening and the potential burden of knowledge of a late-onset disease.

With our study, we would like to learn whether ALD patients feel that boys only, both boys and girls or neither should be screened for the disease. In order to investigate this, we will send out a questionnaire to members of "the Dutch ALD cohort" and their relatives. The results will be of importance to all countries that are considering the addition of ALD to their NBS.

CHENXU LI, MS

Mentor: Reena Kartha, MS, PhD

Chenxu Li is currently a PhD student from the Department of Experimental and Clinical Pharmacology at the University of Minnesota, Twin Cities. Chenxu earned her BS degree in Pharmaceutical Analysis from Shenyang Pharmaceutical University in 2019. Upon graduation, she continued research as a graduate student and earned her MS degree in Pharmaceutics from the University of Minnesota in 2021. Soon after, she began investigating novel therapies for adrenoleukodystrophy as a PhD student under the supervision of Dr. Reena Kartha.

Li's Project: Nervonic Acid – A Potential Therapeutic Targeting Mitochondrial Dysfunction and Cellular Oxidative Stress

Mutations in the ALD protein result in the accumulation of very long chain fatty acids (VLCFAs), which in turn causes the elevation of harmful substances including reactive oxygen species. This can also further damage other cellular components such as the mitochondria, the energy source of the cells. Chenxu's research mainly focuses on developing novel therapies for ALD, specifically targeting the effect of these toxic chemicals on mitochondria. She will investigate the mechanism of a dietary fatty acid, which she hypothesizes will arrest or delay progression in ALD. By treating cells derived from patients with ALD using NA, this study will investigate how NA can mitigate the toxic chemicals and positively affect energy production in cells. Her hope is to develop NA as a safe and effective dietary intervention that can benefit all individuals with ALD.

APRIL RICKLE

Mentor: Adelheid (Heidi) Lempradl, PhD

April Rickle was born and raised in New Jersey. She attended Rutgers University for her undergraduate studies where she worked in the Vershon lab studying Sir2 protein cofactor interactions in baker's yeast. She graduated in 2019 with a BA in Molecular Biology and Biochemistry and joined Van Andel Institute Graduate School for her doctoral studies. She joined the Lempradl lab and began her work studying ALD via a fruit fly model. She hopes to continue researching rare diseases throughout her career. In her free time, she enjoys spending time with her cats, crafting and baking.

Rickle's Project: Testing The Impact Of The Drug Leriglitazone In A Fruit Fly Model Of ALD

Leriglitazone is a promising new drug candidate for the treatment of ALD. While research has been done in cell culture and in mice, we do not fully understand how leriglitazone treatment affects the brain as a whole. We will treat a fruit fly model of ALD with leriglitazone and test how it affects movement and behavior; brain tissue degeneration; metabolism; and gene expression. By testing the effects of leriglitazone on our ALD model flies and comparing to our healthy flies, we can determine which aspects of disease leriglitazone treats and how it affects them. In addition, examining these factors in healthy and untreated ALD model flies will give us insight to the molecular mechanisms behind ALD overall. This research will give us a better understanding of how leriglitazone treats ALD, and how ALD works in a broader sense.

PURPOSE THROUGH *Mentoring*

PEER MENTOR PROGRAM

This year we onboarded three new Peer Mentors: Ken Dieffenbach, Christie Higuera and Christopher Kaag! We are proud to know so many wonderful community members who are willing to support other ALD patients and families by listening, providing encouragement and sharing their own experience.

"When we first got the diagnosis, one of the hardest elements of navigating our new reality was how isolated we suddenly felt. ALD is difficult to explain, let alone to friends and family when you are scared, shocked and confused yourself. In the early days, I found myself grieving for my son's future and feeling very alone. But after reaching out to ALD Connect, I was matched with Sydne as my peer mentor and I immediately felt like I had a friend in my corner who knew exactly what we were going through, as she had already gone through so much of it herself. I don't know what I would have done without her after those first few doctor's visits when I was feeling so lost. She pulled me up in my lowest moments and supported me through the initial darkness. Each doctor's visit, test, blood draw and meeting with a specialist was less daunting because I had someone to reach out to who could put each test result in perspective. I was impressed with how thoughtful ALD Connect was with who I was matched with. I truly feel like Sydne is a lifelong friend and we are each invested in the health and wellbeing of each other's families." — Sophia Rowland

Sydne Pantaleon and Sophia Rowland

> Achieving goals has provided me a fulfilling and rewarding life that I otherwise would not have experienced if not for my life changing diagnosis. I want to redefine what being adaptive looks like and provide motivation and inspiration to others that may have similar obstacles to overcome."
>
> **— CHRISTOPHER KAAG**

MEET A MENTOR: CHRISTOPHER KAAG

Christopher Kaag is Founder and Executive Director of the IM ABLE Foundation and Founder and President of Corps Fitness Crossfit Berks. Through programs associated with the IM ABLE Foundation, as well as Corps Fitness, Kaag works to promote active lifestyles for all and to raise money to support active lifestyles for the adaptive community. Kaag runs daily workout classes that bring his experience as a US Marine into his no-frills fitness facility.

Shortly after graduating from high school, Kaag entered the United States Marine Corps, where he spent almost five years in avionics. In 1998, he left the Marines after being diagnosed with adrenomyeloneuropathy, a rare degenerative condition affecting the myelin sheath of his nerve cells. Upon his honorable discharge from the Marine Corps, Kaag returned to Pennsylvania and began pursuing a business degree at Penn State Berks while working full time.

In 2004, he graduated with his bachelor's degree in Business. Upon graduation, Kaag organized the first annual **Got the Nerve? Triathlon** and also established Corps Fitness and began running fitness classes modeled after his Marine Corps training. In 2007, he launched the IM ABLE Foundation, building on his philosophy of the mental and physical benefits of activity regardless of one's physical circumstances. He continues to uphold IM ABLE's mission by providing adaptive recreational grants to the adaptive community, thereby providing opportunities for everyone to **Get Up and Move!**

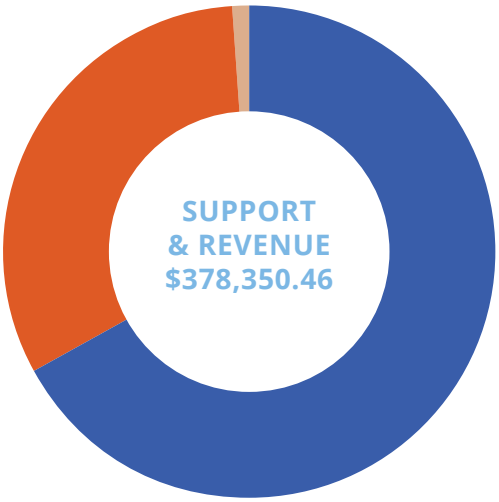
Chris speaks in the community to audiences of all ages to help change perceptions of what a person with different challenges can accomplish and to encourage everyone to view those challenges as stepping stones not crutches.

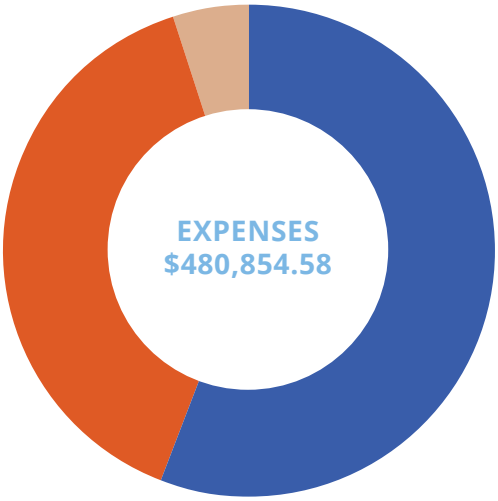
"The Marine Corps saved my life and gave me the ability to look at any obstacle as a challenge to overcome. Achieving goals has provided me a fulfilling and rewarding life that I otherwise would not have experienced if not for my life changing diagnosis. I want to redefine what being adaptive looks like and provide motivation and inspiration to others that may have similar obstacles to overcome." Chris has received many accolades during his career including: PSU Berks Commencement Speaker (2010), Induction into the Central Chapter of the Pennsylvania Sports Hall of Fame as a Courageous Athlete (2008), Penn State Alumni Achievement Award honoring outstanding young alumni (2009), a Pennsylvania State Resolution by the House of Representatives (2012), the American Red Cross Community Impact Hero Award (2015) and Greater Reading Rising Star Award (2015).

Kaag lives in Wyomissing, PA with his wife Gretchen and son Carter.

FINANCIALS & *the Future*

January 1, 2022–December 31, 2022





SUPPORT & REVENUE	**$378,350.46**
Contributions	$255,593.35 (67%)
Securities	$121,446.00 (32%)
Sponsorships	$1,311.11 (>1%)
EXPENSES	**$480,854.58**
Management & General	$267,580.68 (56%)
Events & Conferences	$188,984.22 (39%)
Programs	$24,289.86 (5%)
NET INCOME	**$-102,504.12***

*Please note that this was an expected and planned budget deficit.

2023 Initiatives

LOOKING FORWARD

We look forward to seeing you at an upcoming Community Call, webinar or our 2023 Annual Meeting and Patient Learning Academy on November 3–4, 2023 in Houston, Texas.

> Thank you ALD Connect for providing financial assistance for Michael's custom wheelchair. We are most appreciative for the support and generosity."
>
> — LOLITA & MICHAEL JONES

Thank You

to all our partners and members of the ALD community for continuing to support us.

DONATE TODAY!

Our programs and projects would not be possible without your continued support.
To make a donation please visit aldconnect.org/donate or scan the QR code.

ALDConnect

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