

REFLECTIONS OF *RESILIENCE*

10

10 YEAR ANNIVERSARY

LOOKING BACK AT OUR PROGRESS.
LOOKING FORWARD TO A CURE.



ALD Connect

ANNUAL REPORT 2023

Dear ALD Community,

We are thrilled to extend heartfelt greetings as we commemorate a significant milestone — the 10th anniversary of ALD Connect. It is with immense pride and gratitude that we present to you our 2023 Annual Report, celebrating a decade of unwavering dedication, impactful initiatives, and transformative progress in the fight against adrenoleukodystrophy (ALD).

Over the past ten years, ALD Connect has stood as a beacon of hope for individuals and families affected by this rare genetic disorder. Our commitment to fostering collaboration, advancing research, and supporting patients and caregivers has been the cornerstone of our mission. This Annual Report serves as a testament to the remarkable achievements made possible by the collective efforts of our community this year, and since we were founded in 2013.

We have navigated challenges, celebrated breakthroughs, and fortified our resolve to drive positive change. Our journey has been marked by resilience, unity, and a shared vision of a world free from the burdens of ALD.

In this report, you will discover the impact of our initiatives, the stories of resilience from individuals touched by ALD, and the collaborative endeavors that have fueled our progress. As we reflect on the past and set our sights on the future, we invite you to join us in acknowledging the remarkable achievements of ALD Connect and the invaluable contributions of our partners, supporters, and stakeholders.

As we stand at the threshold of a new chapter, we remain steadfast in our commitment to driving innovation, promoting awareness, and advocating for those affected by ALD. Together, we can continue to make a profound difference in the lives of individuals and families impacted by this devastating condition.

We extend our deepest gratitude to you for your unwavering support and belief in our mission. Your partnership has been instrumental in our journey, and we look forward to continued collaboration in the years ahead.

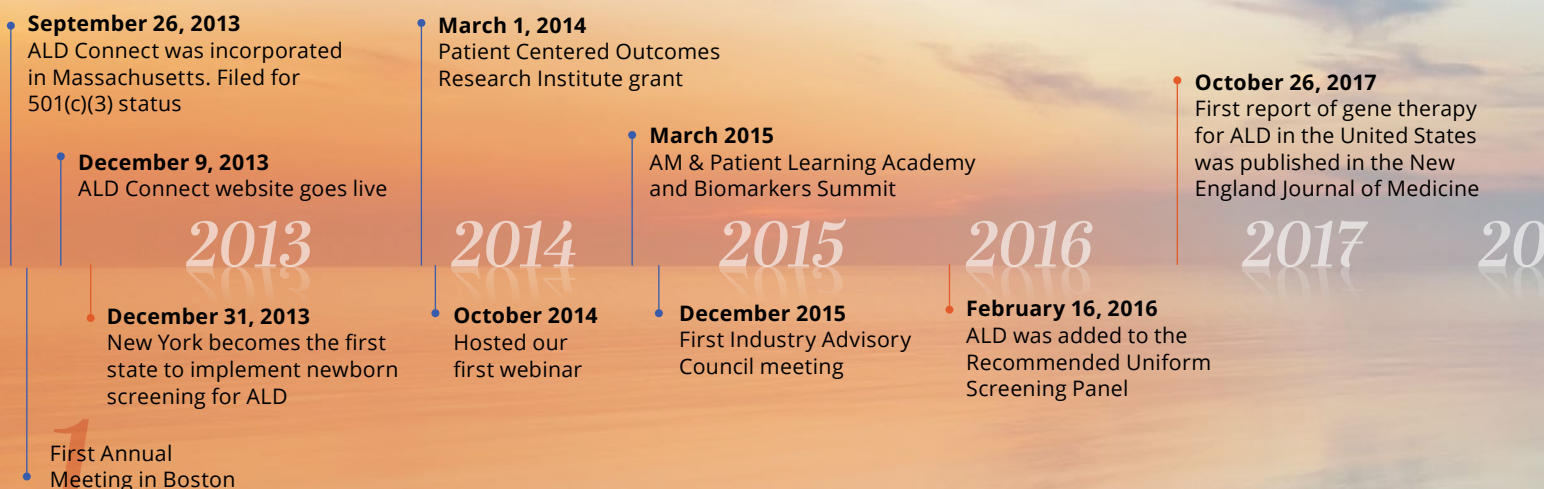
Thank you for joining us in celebrating a decade of progress and paving the way for a brighter future for those affected by ALD.



With Gratitude,

Florian Eichler, MD
Co-Founder, President

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



For 10 years, ALD Connect has been raising awareness, raising funds and raising spirits on behalf of the ALD community—and we're committed to doing it again and again, year after year.

Mission

Our mission is to improve health outcomes for patients with 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.



RESTORING HOPE

for Patients & Families

ERIC'S STORY

FINDING A BRIGHTER OUTLOOK

First of all, I feel like my case was unusually diagnosed. My feet and legs had been in pain and getting worse for several months. I attributed it to being on my feet all day on concrete. I was at work one day and fell off a small riser. I thought I had just lost my balance or stepped on something to cause the fall. After this, I was scheduled to see a neurosurgeon to rule out any back injuries. He could find nothing on the MRIs to cause the pain; then, he did a Babinski test. He said that I definitely had nerve damage, but not from anything surgical. He referred me to a neurologist who didn't have an opening for a couple of months.

On the way home, I stopped to visit with my cousin because I knew that he had some issues with his back and nerves. He has never been open about any issues he was having, and I had never heard of any diagnosis he had received. He told me about his condition, and he suggested that I get a genetic test. He gave me the contact information for the geneticist that diagnosed him. After contacting her, I received my kit in the mail and sent off my sample. I don't think I'll ever forget the day I got a phone call from her. I was at a craft show a couple of weeks before Christmas, 2018. She told me who she was, and that my test had come back. I was told I had a mutation in my *ABCD1* gene which meant I had AMN. That was about the extent of the call. I didn't know what that meant. I had only just heard of this by name just a few weeks prior.

To Google I went. Trying to find any information I could about this disease and what it meant for me. All that did was create a fear and uncertainty for me. I quit doing searches and decided to wait until I spoke to the

I found a group online called ALD Connect... I was absolutely amazed. I felt like I belonged.

Eric Quinney



neurologist. That too was an unforgettable experience. I gave her the genetic report when she came in the room. She looked it over, looked at me, back at it, and then me. She said, "You just made me feel like I hit the lottery." I didn't know if I needed to stay or run after that comment. She quickly said that it was because of the rarity of the disease. She then went into detailing the symptoms and trying to find when I started with any and where I was in my progression. We concluded my symptoms had started in my late 20's to early 30's.

I progressed from using a cane to using a rollator because of the spasticity in my legs. My neurologist started recommending getting tested to see if a Baclofen pump would help. I kept refusing, mainly because I didn't want a surgical procedure, and an implant in me. I broke down after a couple of years of the spasticity progressing and agreed to get tested. I was scared of the spinal tap required for the test, but once it was done, the results were amazing. My legs were not jerking uncontrollably; I could walk more easily, as well as the tightened muscles in my abdomen loosening up. I was ready to schedule surgery. Since the implant, I have gone from using a rollator, to using hiking sticks just to help me keep my balance. I feel blessed to have Dr. Kelli be a part of this

journey with me. Her thinking outside of the box and her encouragement in everything has been something I hope everyone can find in their doctors.

A couple of years later, my wife and I got a divorce. That was devastating; dealing with this disease and the future of dealing with it alone. My thought was, "who would want to marry someone as broken as me?" Since then, I met Michelle who now will be the future Mrs. Quinney in October. Her encouragement has helped me so much and is proof there is someone out there for everyone. Her family has accepted me, and has been very encouraging and accommodating to me also.

During this time, I had found a group online called ALD Connect. I read quite a few articles online, and watched several videos. I joined a few community calls even. It was great to connect with men that were dealing with the same issues, and I wanted to go to an Annual Meeting and Patient Learning Academy. I finally made it to the one in Houston last year, with Michelle I would like to add. I was absolutely amazed. I felt like I belonged. I met many wonderful people from doctors and researchers, people with AMN, and families coping with children with ALD. I felt normal for a change, something I had not felt in a few years.

Thank you ALD Connect and everyone involved for all you do for this community. To feel like I belong and seeing all that is being researched and accomplished through all you do has given me a brighter outlook on the future both for me and my daughter. Keep up the great work, and I plan to see you all at future meetings.

Eric Quinney



KEEPING A SLOW AND STEADY PACE

Dylan was born full term. We had no complications during pregnancy or delivery. The pediatrician came to the hospital to check him out, handed him to me and said, “He’s perfect!”

Had we only known then.

Dylan hit all of his milestones and matured into a sweet, funny, loving little boy. Dylan loved to play soccer, basketball, swim, tell outrageous stories, he absolutely adores music and loved to have sleepovers with his friends and most of all, he loved to draw. He was an exceptional artist.

In first grade, Dylan’s teacher brought to our attention that Dylan had trouble staying focused. We went to his pediatrician as we suspected ADHD. He did some testing and agreed. We then started our journey on the path of trying to find the right ADHD medication that didn’t either turn our child into a zombie or a troll.

We found a good option after a lot of trial and error, but Dylan continued to struggle. He started to get really annoyed with sticky hands, sounds, people chewing, etc. Back to the pediatrician we went, to be diagnosed with Sensory Processing Disorder. We started to see a psychologist to help Dylan with his sensory issues and anger management around those triggers. This was Fall of 2019. Simultaneously, Dylan had some testing at school and was regressing. The school called a meeting and suspected a learning disability, and asked our permission for more testing. We agreed. We had a meeting and the findings were inconclusive. My husband was Dylan’s basketball coach, and we began to notice that Dylan acted confused, like he didn’t know what he was supposed to be doing, whereas before that, he was a great little ball player.

Fast forward to 2020. Dylan takes an eye exam at school and passes it. Then COVID hits. I notice, because I’m now his teacher and home with him all the time, that he is bouncing around the page when he’s reading, he’s falling a lot, he’s having trouble focusing....I call to get him in to an eye doctor, but it’s COVID. It’s a pandemic. We finally get him in May 29, 2020. The absolute worst day of our lives. Dylan is rushed to the ER because the eye doctor sees that he has bilateral papillary edema. My husband calls me and tells me they are going to give him steroids, and they should be out soon. They are going to do an MRI to check him out, but should be home soon. He calls me back and says, “Elissa, you have to get down here. They are talking about some crazy disease....I just can’t even believe this is happening.” So, my father-in-law drives me because I am too upset to drive myself. Because it’s COVID, they won’t let him come in with me. He has to leave. They take me and my husband into a room and tell us that they found something on the MRI. They tell us it’s called Adrenoleukodystrophy. They explain it is X linked—meaning I gave it to him as Mom, and that there is a 50/50 chance our other boys have it. At this point, my husband loses it. I’m still in denial and shock. Then they tell us there is no cure, they think his Loes score is high, too high for any salvation, and that we probably have 2 years. They tell us to go home, and enjoy our time. Oh, and by the way, he also has Addison’s disease, which explains why his skin is so tan and he craves salt all the time. We have to be admitted to get his Addison’s disease under control.

Our family motto has become, ‘slow and steady wins the race.’ We still do a lot of the same things we love, we just do them different. We have decided that we are NOT going to let ALD stop us, or him, from living.

Elissa Findling



Since it was COVID, only one of us could stay with him, which was obviously Mom (me). I didn't sleep that night. I got online and researched and found ALD Connect. I got on and joined and asked for guidance. I was immediately put in contact with the University of Minnesota and Dr. Loes, Lund and Gupta. Dr. Gupta called me the next day. We got the MRI sent over to him and he and Dr. Loes read it. Dylan's Loes score was an 11. Damn.

A Loes score of 10 or under is really where you need to be to qualify for transplant. That coupled with the symptoms Dylan was already having, it didn't sound good. However, Dr. Gupta offered us some hope in that he had an idea for a cocktail of meds to "put out the fire" and attempt to slow the progression of Dylan's disease. We decided to try it. And Dylan's disease has not progressed via MRI since diagnosis. However, his physical symptoms have manifested quickly. We went from still walking and talking in July of 2020 to unable to talk, eat, or walk in September. We ended up with a g-tube and a wheelchair in a matter of weeks.

Dylan has remained stable. He is cognitively aware. He can move to answer yes or no. We have an AAC device that we use to help him communicate. He knows what he wants and we have to ask yes or no questions to get there. He is mainstreamed in school and attends middle school. He pushes into Gen Ed classes and they do what they can to incorporate him into the classroom.

Aside from an occasional seizure, which he typically only has if he's about to come down with a virus, most days are pretty good days! We keep Dylan busy with music therapy, PT, OT, speech, school, play dates with friends and cousins, family trips, massages, hyperbaric oxygen therapy, reiki, and lots of prayers and hope in our Lord and Savior Jesus Christ.

Our family motto has become, "slow and steady wins the race." We still do a lot of the same things we love, we just do them different. We have decided that we are NOT going to let ALD stop us, or him, from living. None of us know how long we have, and we intend to make the most of whatever time we have. We have promised Dylan that we will always do what we can to make sure he looks good, feels good, smells good, and is included in as much as we can. We also promised him to always let him make choices where he can.

By the grace of God, I am not a carrier of ALD, so I do not have it, and my other two sons don't have it either. I know that is a blessing, because we have met so many families who have lost all of their sons to ALD. They are all impacted. Devastating. It's a *de novo* ALD that Dylan has.



If we had only known when he was born. Things would be so different. We need more funding, more research for these boys that have late stage ALD. The ones who get left behind. The ones who don't have the opportunity to get a BMT or interventional therapies. They get left behind. I hope we can change that.

We are grateful for the opportunity to share our story. God bless. Slow and steady wins the race!



KINGDOM'S STORY

GATHERING INFORMATION AND SUPPORT

Kingdom Andrews was born on June 1, 2022 in Charlotte, North Carolina. Our journey with ALD started on June 9, 2022. That was the day we found out Kingdom was picked up on newborn screening; he was 9 days old. As a first time mom, I had so many emotions, but the one that stuck to me most was denial. I just knew this had to be incorrect as the pediatrician had a difficult time explaining to us what it all meant. He handed us an educational printout from the internet and sent us on our way.

As I battled emotions that come with new motherhood, I was in a difficult place. My husband was still in denial but supportive in any way that he could be. We waited for what felt like an eternity, to have a visit to UNC Chapel Hill for confirmation testing. The staff at UNC Chapel Hill was much more knowledgeable and had a team of providers, ready to care for our son. It was reassuring. I left with more information and continued to educate myself on what our journey will look like. As I researched further, I stumbled upon many organizations that supported ALD. ALD Connect stood out for me.

I gathered as much information as I could from ALD Connect and various resources that were available on the website and YouTube page. When I found out that ALD Connect had monthly "support groups," I was SOLD! The entire journey thus far has been very isolating and I had no one to talk to. My husband is a truck driver and spends his week over the road, so this felt like the perfect way to not feel so alone. I joined my first Newborn Screening, Young Families, Parents of Asymptomatic Children call and immediately felt connected. When things turned difficult during the monitoring phase, I was quickly paired with a mentor who could provide one-on-one support during times of uncertainty. Kingy is still in the asymptomatic, monitoring stage of ALD.

I continue to attend the community calls to obtain support, insight, and strength to endure our journey with ALD. What lies ahead of us is unknown—which is often the most difficult part—but ALD Connect has helped me feel empowered and supported in a way that is unimaginable. I will continue to support the organization as it has significantly changed our lives in a very short amount of time.

Toyia Andrews



STAYING STRONG AND MOBILE

Howdy from Texas!

As a child, I watched my mom struggle with her balance and gait. She moved from a cane to crutches and then to her wheelchair. None of us kids realized that we would be facing the same fate. My mom was never provided with a correct diagnosis.

I started having difficulty walking in my early 30's when I was pregnant with my son. After his delivery, my symptoms seemed to have vanished except for an occasional loss of balance or stumble which resulted in a fall to the ground. My husband said that I was just clumsy.

In my mid 40's, I started to have frequent falls. After seeing several different neurologists and having quite a few tests completed, I still did not have a diagnosis.

In 2017, I was accepted into the Undiagnosed Diseases Network (UDN) at Baylor University in Houston. Blood samples were provided from all five members of our family. A whole exome genetic study was performed on all samples. We were told that it could take up to a year to possibly find an answer. Four months later, I was asked to come to UDN at Baylor. On day one of my visit to UDN, I was told that they had a diagnosis—Adrenoleukodystrophy.

All five members of my family have a mutation of the ABCD1 gene. It took me around seven years to finally get a diagnosis. I was so relieved to get a diagnosis, but no cure was in sight. The doctors suggested that I have my young adult son genetically tested. He refused but finally had his VLCFA levels tested. His results were normal. I am so thankful that my son will not have to deal with this disease, and it stops with me.

I have been exercising on a seated elliptical bike since 2017. This is helpful with my spasticity and my leg strength. I also go to Physical Therapy every other week and see a chiropractor that specializes in Neurology. My family is extremely supportive. We purchased a 4-wheel electric bike (The Rig) so I could enjoy being outdoors, but this didn't provide me with any physical activity. In the past year, we purchased a recumbent trike (e-Catrike) so I can increase my muscle strength and stay mobile as long as possible. The time I spend outdoors on The Rig, or the trike brings me the most joy in life.

I enjoy participating in the monthly ALD Connect Women's Community Calls. This year, I was able to attend the Annual Meeting in Texas and thought it was a great experience.



I'm hopeful for a cure soon that will either stop or slow the progression for all of us.

Cheryl Johnson

REWARDING GRANTS

that meet patient and family needs

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 2023, we funded nine grant applications for help with travel to medical appointments, mortgage payments and car payments.

● April 2023

We paid two months rent for a father in New Jersey who has a 13-year-old with ALD.

● May 2023

We paid for mileage for a patient to travel from Montana to Utah for an appointment.

● July 2023

We purchased a wheelchair for a father in the Philippines.

We purchased a wheelchair for a mother in the Philippines.

We paid for a family to travel from Tennessee to Minnesota for appointments.

We paid for a mother to travel to an MRI appointment in Florida.

● September 2023

We made a car payment and paid a phone bill for the mother of a cerebral ALD patient at CHOP.

We made a payment on an electric wheelchair for a man with AMN and cerebral ALD.

We made a mortgage payment for a family in Indiana with a son who has cerebral ALD.

A very special thank you to Michael Benton's family and friends for continuing to support the Michael Benton Travel Scholarship Fund and helping us provide financial assistance in his memory.



We continue to bring together all ALD stakeholders to create a sense of belonging and community while fostering an environment of better understanding of ALD via our annual conferences and monthly webinars. By supporting ALD Connect, you are helping to advance critical research and rare disease advocacy, while also providing much needed financial support for ALD patients and families in need.

Greg Benton

Meet our RESOURCEFUL STAFF



DR. AMENA SMITH FINE | New Board of Directors Member

Amena Smith Fine is an Assistant Professor of Neurology and Developmental Medicine, Kennedy Krieger Institute and Johns Hopkins School of Medicine. Dr. Fine is a physician scientist working in the Moser Center for Leukodystrophies. The focus of Dr. Fine's primary research study is to use advanced MR imaging sequences of the brain and spinal cord in conjunction with quantitative measures of gait and balance obtained with wearable accelerometers to 1) determine the neural substrates of clinical impairment in ALD and LBSL and 2) validate the longitudinal use of wearable technology to identify clinically meaningful quantitative performance measures.



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 13 years. She has a Master of Healthcare Administration degree from the University of Minnesota.



FELICITY EMERSON | Project Manager

Felicity joined ALD Connect as a Volunteer in 2020 and was hired as a Project Manager in 2021. In May of 2023, she completed a Ph.D. in Biomedical and Biological Sciences at Cornell University, and in the fall of 2023, she started a Genetic Counseling Masters program at Rutgers University. Felicity values every opportunity she has to learn from the ALD community about the lived experiences of families with ALD.



KATIE DOW | Project Manager

Katie joined ALD Connect in 2022. Katie helps with a variety of projects. She enjoys learning more about the ALD community.



EMMA HAYES | Project Manager

Emma joined ALD Connect in 2022 as a Project Manager. 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 as a rare disease patient liaison at a pharmaceutical company. She is a dedicated advocate for ALD which stems from a family history with the disease.

Since joining ALD Connect, I have learned so much about the power of community, the strength in enduring and advocating, and the expertise in the patient population.

Felicity Emerson

RECONVENING

to Share Insights



2023 Annual Meeting and Patient Learning Academy

2023 ANNUAL MEETING AND PATIENT LEARNING ACADEMY

Our 2023 Annual Meeting and Patient Learning Academy was held on November 3–4, 2023 in Houston, Texas. At our meeting, we heard exciting updates from experts on ALD research and important recommendations for ALD monitoring, care, and mental health. This year, we included patient-friendly summaries of each talk in our agenda. With our patient panels, we ensured that doctors, researchers, and industry members heard the patient perspective. To celebrate 10 years of ALD Connect, we hosted a birthday party at the end of the first meeting day to reflect on the progress we have made and the work yet to be done. You can check out the recordings from the Annual Meeting on our website or YouTube channel. We are already looking forward to our 2024 Annual Meeting and Patient Learning Academy.

Thank you to our sponsors:



ALD Connect has given me the opportunity to turn our lived ALD experience into meaningful change and support and education for our ALD community.

Kathleen O'Sullivan-Fortin

WORK GROUP 4

ALD Connect hosts a working group meeting called “Work Group 4,” to bring together all ALD community members to receive feedback on our programming and brainstorm community-wide initiatives. Our 2023 fundraiser, ALD Makes Me Blue, arose from fruitful discussions during our Work Group 4 meetings. Any patients, family members, or advocacy organizations working in the ALD space are welcome to join.

In 2024, this group will transition to a new name: Patient-Led Planning Committee.



Sign up for the Work Group 4 newsletter by scanning the QR code or email Felicity at felicity.emerson@aldconnect.org.

ALD MAKES ME BLUE

For Rare Disease Day 2023, we launched the “ALD Makes Me Blue” campaign. This was a collaborative fundraising initiative with other ALD patient organizations, including ALD Alliance, Alex TLC, Arrivederci ALD, the Leukodystrophy Resource Research Organisation Incorporated (LRRO), Knockout ALD, and the Stop ALD Foundation.

ALD Makes Me Blue was the “brain child” of Fight ALD founder, Janis Sherwood (pictured on right). As a way to pass the torch to the community before retiring, she realized there was a lot of potential to still spread awareness, raise money for ALD organizations, and perhaps have a little fun while doing so. Thus, the idea for “ALD Makes Me Blue” was born.

The color blue has historically been associated with leukodystrophy awareness, so it is only fitting that the fundraiser is centered around this color. During this fundraiser, we asked the community to show how ALD made them “blue” by dyeing their hair, painting their nails, wearing blue, or becoming blue in any way they could and sharing the challenge on social media.



Fight ALD Founder, Janis Sherwood



Making Meaningful CONNECTIONS

AARON GARDNER

While I've been involved with ALD Connect for a few years now, and had previously interviewed a provider during a virtual conference and shared at the drug development meeting last year, I had never previously attended an ALD Connect conference in person. When I was presented with the opportunity to not only attend the most recent conference in Houston, but also to speak on the patient experience panel, I jumped at the chance.

It is no exaggeration to say that this was one of the best and most memorable weekends of my life. ALD Connect is an amazing organization, led by some incredible individuals, both on staff and on the Board of Directors. The broader ALD/AMN community is also filled with the most wonderful people. I can't recall how many times over the weekend I heard people comment, "This is a crappy (or more colorful colloquialisms) disease, but the most amazing people." I couldn't agree more.

Whether it was reconnecting with old friends who I've previously met in person, or finally meeting face-to-face those I've only known through online interactions, or feeling an immediate kinship with other members of our community (one sweet lady without affected siblings instantly identified me as her "ALD brother"), the connections I made this weekend were incredibly moving

and meaningful. In this community, I feel loved, appreciated, seen and heard, and most importantly, valued.

The Saturday breakout session for men with AMN was itself worth the entire event for me. To sit in a roundtable not only with other men living with AMN, but also a handful of the leading experts on our disease, was an experience I will never forget. I was able to ask some very specific questions that I've never had answered previously, and the camaraderie amongst the male attendees in this breakout was strong. My AMN brothers are great guys, and I admire how each of them is managing living with this illness in his own way.

I've very rarely been presented with the opportunity to speak publicly about my family's and my experiences with ALD/AMN, so I was deeply grateful for this platform.

On the patient panel, I was able to share about my extensive family history with this disorder (spanning over at least five generations), my own history with the disorder since I was diagnosed with AMN 8.5 years ago, and some of the things that I've found to be most helpful for me. These include receiving professional counseling to help process learning to live with chronic illness, discovering adaptive sports and adaptive recreation opportunities in my area (I've played my primary sport of wheelchair basketball for over 4.5 years now), and making connections with others who share this disorder and with experts from around the country (which is one of the greatest resources that ALD

Connect provides). I am deeply appreciative of the many kind words and warm embraces that were shared after my presentation, and these connections will not be forgotten.

I didn't want the event to end. I've rarely felt so at home in my whole life. I am so thankful to Kathleen O'Sullivan-Fortin for encouraging me to speak at this conference and to ALD Connect for all of the hard work that goes into making it happen. I would strongly encourage anyone who has considered attending one of these conferences in person to commit to making it happen. There is no better way to build community, and I promise you won't regret it!



ALD Connect is an amazing organization, led by some incredible individuals, both on staff and on the Board of Directors.

Aaron Gardner



Mentoring RELATIONSHIPS

PEER MENTOR PROGRAM

The purpose of the ALD Connect Peer Mentor Program is to connect with individuals with ALD/AMN, family members, or caregivers with an ALD Connect mentor so they can form a relationship that is maintained with encouragement, respect, and confidentiality. This year, the program has continued to grow and we matched 9 mentees with their respective mentors. We also onboarded a new mentor, Suzanne Flynn, for monitoring stage and newborn screening families.




SUZANNE FLYNN

Suzanne Flynn is a graduate of the Connell School of Nursing at Boston College. A pediatric nurse for 23 years, she lives in Crestwood, New York with her husband, Ken and three children—Charlotte (16 years), Gavin (14 years), and Patrick (9 years). Her youngest child, Patrick, was diagnosed with ALD at birth through newborn screening thanks to Aidan’s Law in New York State. Her older son, Gavin, was then tested and was also found to have ALD. Both boys are under the care of Dr. Florian Eichler at Mass General Hospital in Boston. They are asymptomatic and doing very well.

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. This year, we added a structured Community Call with a focus on mental health, run by ALD community member and counselor, Jesse Torrey, LAC. These calls are not recorded.

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



ALD Connect is the bridge connecting scientists, doctors, and families in a shared pursuit of answers. Positioned at the intersection of research and compassion, it actively addresses crucial questions and amplifies the voices of those affected, driving progress within the community.

Stephan Kemp, PhD

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.

FEBRUARY 12, 2023

The Grey Zone Introductory Webinar

Dr. Stephan Kemp
Dr. Troy Lund

MAY 24, 2023

Travel Considerations for ALD and AMN Patients

Katy Ehnstrom

AUGUST 2, 2023

Siblings: What Works, What Hurts

Jesse Torrey, Counselor
Anna Torrey, Panelist
Kathleen O'Sullivan-Fortin, Panelist
Maggie Fortin, Panelist

NOVEMBER 20, 2023

From Natural History to Clinical Trials: A Family Guide to Engaging in Research

Dr. Camille Corre
Jennifer Steichen, ALD Family Panelist
Emily Reimer, ALD Family Panelist
Troy Reimer, ALD Family Panelist

Revolutionizing 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.



Virginia B. Toulmin Foundation

In 2023, we received a \$150,000 grant to support community-wide ALD initiatives, including The Grey Zone project.

THE GREY ZONE

ALD Connect is proud to have continued our funding and support of the "Grey Zone" project in 2023. This community-wide initiative is a great fit for ALD Connect as it includes many of our board members!

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.

- **Was your baby diagnosed with adrenoleukodystrophy via newborn screening?**
- **Is the *ABCD1* gene mutation a Variant of Uncertain Significance?**
- **Is everyone in your family who has this *ABCD1* mutation asymptomatic?**

If you can answer YES to all three questions, you may be eligible to participate in The Grey Zone project. Please reach out to our team at aldconnect.org/contact-us.



DONATE TODAY!

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

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. Each IAC meeting also features a patient perspective to ensure that industry members and ALD Connect Board members center the experience of ALD families in their work.

FEBRUARY 2023

Global Outreach Strategies

Abhaya Sundhar

Fundación Lautaro te Necesita—

Leukodystrophy

Foundation: Assessment of needs in Argentina

Verónica de Pablo

JUNE 2023

Empowering Patients through Knowledge

Camille Corre

SEPTEMBER 2023

A blood test to track brain damage: Predicting the early onset of neuroinflammation in X-linked

adrenoleukodystrophy

Dr. Isabelle Weinhofer

DECEMBER 2023

Overview of clinically meaningful sensorimotor outcome measures to assess myeloneuropathy in ALD

Dr. Amena Smith Fine

PUBLISHED VOICE OF THE PATIENT REPORT

The information in the Voice of the Patient summarizes our July 2022 Externally-Led Patient-Focused Drug Development Meeting, which highlighted the experience of patients who are affected with adult manifestations of ALD. This report may be used to guide therapeutic development and inform the FDA's benefit-risk evaluations when assessing therapies to address ALD. The hope is that this information will catalyze better treatments and ultimately a cure for those affected by ALD.

Thank you to our sponsors:



View the full report by scanning the QR code or visiting aldconnect.org/pfdd.



ALD Connect brings all talents together to find new ways to improve care and strive for a cure for ALD. With shared purpose and diverse interests, we move forward.

Florian Eichler, MD
ALD Connect Co-Founder, President

RECOGNIZING

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.

We were proud to host our three 2022 emerging investigators and hear their research updates during our 2023 Annual Meeting and Patient Learning Academy, as well as welcome three new investigators who received grants in 2023. We look forward to hearing their updates next year! Check below to meet our 2023 grant recipients and find out what our 2022 emerging investigators achieved with our grants.

2023 EMERGING INVESTIGATORS



MANOUCHEHR AMANAT, MD

Dr. Manouchehr Amanat is currently working as post-doctoral research fellow at Kennedy Krieger Institute and Johns Hopkins Medical Institute. He achieved his Medical Doctorate degree from Tehran University of Medical Sciences in Tehran, Iran. He is developing novel RNA therapeutic agents such as antisense oligonucleotides in neurodevelopmental disorders. He is skilled at cell culture and growing induced pluripotent stem cells, advanced statistical methods and machine learning models using Python, and designing clinical trials.

Project: Antisense Oligonucleotide Therapy in X-linked Adrenoleukodystrophy

This project focuses on the application of antisense oligonucleotide therapy in the context of X-linked adrenoleukodystrophy (X-ALD). The primary objective of this research is to elevate the expression of the ABCD2-related protein as a compensatory mechanism for the deficiency of functional ABCD1-related protein in X-ALD patients. To achieve this, we have designed specific ASOs at Kennedy Krieger Institute to directly target ABCD2 mRNA to enhance protein expression. The experimental approach involves culturing fibroblasts derived from X-ALD patients, introducing ASOs, and subsequently quantifying the levels of ABCD2 mRNA and very long-chain fatty acids. A similar methodology will be applied to microglia cells derived from induced pluripotent stem cells. Ultimately, this project represents a significant step towards addressing the challenges posed by X-ALD.



YORRICK R.J. JASPERS

Yorrick R.J. Jaspers earned his Bachelor of Science in Chemistry from Leiden University and a Master of Science in Analytical Chemistry from the University of Amsterdam. Currently, Yorrick is in the final stages of completing his PhD at the Amsterdam University Medical Centra, where his research primarily revolves around advancing the field of lipidomics. His work focuses on developing and applying innovative lipidomics techniques to better understand and characterize metabolic disorders.

Project: Lipidomic Fingerprinting of X-linked Adrenoleukodystrophy Bloodspots

Newborn screening has significantly improved ALD diagnosis, but it has also led to the identification of genetic variants of unknown significance (VUS), which pose diagnostic challenges as we don't know if they cause the disease. Our project seeks to address this issue by using a specialized technique called lipidomics to create unique biochemical profiles (fingerprints) of ALD in bloodspot samples. Using bloodspot samples offers several advantages, including non-invasive collection through a simple finger prick, making it suitable for newborns and infants. It can also be done at home or at local healthcare facilities and is easily transportable via regular mail, thereby mitigating logistical hurdles and reducing associated costs compared to other sample types. We will analyze bloodspot samples from ALD patients and healthy individuals, using a state-of-the-art mass spectrometer. This instrument allows us to rapidly analyze a wide range of lipids in the bloodspot samples. By comparing the lipid profiles of known ALD cases with those of VUS, we hope to gain insights into the potential disease-causing effects of specific genetic variants.



COURTNEY DEMMITT-RICE

Courtney graduated with a Bachelor of Science in Biotechnology and did environmental toxicology research with honeybees prior to joining Dr. Josh Bonkowsky's lab as a Neurobiology PhD student. Courtney enjoys being outdoors with her family, arranging flowers, gardening, and carpentry. Her youngest child has a rare leukodystrophy—this has fueled Courtney's interest in clinical translational research and she looks forward to contributing to the rare disease community as both a caregiver and researcher.

Project: Transcriptomic and Lipidomic Signatures in ALD

Our project studies how ABCD1 mutations affect gene expression (transcriptomics) and metabolism of lipids and fats (lipidomics). We have two goals: first, to understand how these changes cause ALD disease pathology, especially of cerebral ALD, where the molecular change is poorly understood. Second, with the information from the "omics," we will test a potential new therapy identified from a large zebrafish drug screen.

Supporting ALD Connect means fostering this invaluable connection, where every contribution strengthens the foundation for a future filled with answers, support, and hope.

Stephan Kemp, PhD

CHENXU LI

Developing Nervonic Acid, a Dietary Fatty Acid, as Therapy for Adrenoleukodystrophy by Targeting Mitochondrial Dysfunction and Cellular Oxidative Stress

X-linked Adrenoleukodystrophy (ALD) is caused by mutations in the ABCD1 gene, which encodes a peroxisomal ATP-binding cassette transporter protein (ALDP). Deficiency in ALDP results in the accumulation of saturated very long-chain fatty acids (VLCFA), mainly hexacosanoic acid (C26:0), in plasma, brain, spinal cord, and adrenal cortex. Oxidative stress and mitochondrial dysfunction induced by C26:0 are considered leading factors in disease progression and, therefore, promising therapeutic targets for ALD. Monounsaturated fatty acids have been demonstrated to reduce VLCFA accumulation in ALD patient-derived fibroblasts, suggesting their potential to lower the toxic C26:0 levels and delay disease onset or progression. We investigated nervonic acid (NA, C24:1), a naturally occurring monounsaturated fatty acid, as a potential therapy for ALD given its biochemical benefits in reducing C26:0 levels. Our study demonstrates that NA can significantly attenuate cellular oxidative stress and improve mitochondrial function in AMN-derived fibroblasts.

HEMMO YSKA, MD

Attitudes of Patients with Adrenoleukodystrophy towards Sex-specific Newborn Screening

Newborn screening (NBS) for X-linked adrenoleukodystrophy (ALD) can identify individuals before the onset of life-threatening manifestations. This study investigates the attitudes of persons with ALD towards sex-specific NBS for ALD. A questionnaire was sent to all patients in the Dutch ALD cohort. Invitees were asked who they thought should be screened for ALD: only boys, both boys and girls or neither. Motives and background characteristics of respondents were compared between screening preferences. Sixty-six of 108 invitees (61%) participated in this study. The majority of respondents (n=53, 80%) were in favor of screening both newborn boys and girls for ALD, while 20% preferred boys only. There were no differences in the background characteristics of the respondents between screening preferences. A variety of motives why respondents preferred a type of screening were identified. The results of this study can inform parties involved in the implementation of NBS programs.

APRIL RICKLE

Testing the impact of the drug leriglitazone in a fruit fly model of ALD

There is a dire need for a pharmaceutical treatment for ALD. Leriglitazone, an experimental drug that is currently in clinical trials, has been shown to protect neurons from damage in cell culture models of ALD and improve motor function in ABCD1 mutant mice. Thus far, metabolomic studies of leriglitazone in brain tissue have been limited to cell culture. While promising, these studies have not included brain tissue from an ALD model organism. Using a *Drosophila* model of ALD, we study how lifelong leriglitazone treatment affects brain metabolism on a molecular scale and neurodegeneration on a behavioral and histological scale. These results aid in understanding the effects of leriglitazone treatment on the brain as well as understanding ALD as a whole.



Over the last 10 years we have had breakthrough treatments, strengthened our voice with the FDA, and assisted many patients and caregivers with needed support. I'm proud to be a part of the ALD Connect community.

Timothy Maguire

2023 Financial REVIEW

January 1, 2023–December 31, 2023

SUPPORT & REVENUE **\$460,002.30**

CONTRIBUTIONS AND GRANTS \$400,002.30 (87%)

SPONSORSHIPS \$60,000 (13%)

EXPENSES **\$400,316.58**

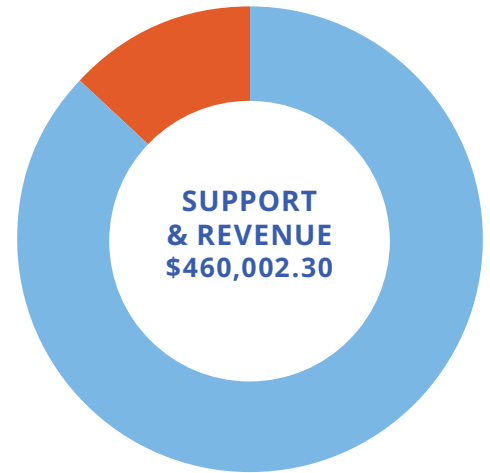
PERSONNEL AND PROGRAMS \$119,345.20 (36%)

RESEARCH \$113,200.00 (28%)

EVENTS AND CONFERENCES \$94,092.31 (24%)

OPERATING \$46,244.51 (12%)

NET INCOME **\$59,685.72**



RECOMMITTED to the Future

LOOKING FORWARD

We look forward to seeing you at an upcoming Community Call, webinar, or our 2024 Annual Meeting and Patient Learning Academy on November 8-9, 2024. Our 2024 initiatives include expanding our international reach, facilitating participation in the Grey Zone project, and expanding our mental health programming.

*Thank You to all our partners and members of the
ALD Connect 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.





2023 BOARD MEMBERS

Greg Benton

Josh Bonkowsky, MD, PhD*

Patti Chapman

Florian Eichler, MD*

Ali Fatemi, MD*

Stephan Kemp, PhD

Ben Lenail*

Troy Lund, MSMS, PhD, MD

Timothy Maguire

Patricia Musolino, MD, PhD

Kathleen O'Sullivan-Fortin*

Amber Salzman, PhD*

Alex Sherman*

Keith Van Haren, MD*

*Co-Founders

*We're proud of our board members' passion and commitment,
demonstrated by the high attendance rate in our monthly meetings.*

ALDCONNECT.ORG