UNITED LEUKODYSTROPHY FOUNDATION | VOL. 41 • NO. 3 • SUMMER 2023

NEWSLETTER

united LEUKODYSTROPHY FOUNDATION

THE 2023 ULF FAMILY CONFERENCE IS ONE FOR THE RECORD BOOKS!

This year's meeting was truly an amazing experience for all involved. In 2019, we saw a record number of people attending our in-person meeting with approximately 330 people in attendance, and we did not offer any virtual option. After 2020 and 2021 being completely virtual and 2022 being our first attempt at a hybrid conference while the world still grew more comfortable with the relaxed health and safety guidelines, the response to our 2023 meeting was unexpected.

In-person and virtually, we estimate that nearly 600 people participated in the 2023 meetings, including families and professionals from all over the world. The feedback we have received so far has been so constructive and will help us continue to shape our event to accommodate the needs of families for years to come.

Please check out page 3 of this newsletter to see all of this year's sponsors and partners. We would not have been able to pull off this hugely successful meeting without them! We would also like to express our deepest gratitude to those who made item donations to this year's live and silent auctions, and raffle. Of particular note, one donor crocheted two blankets featuring the ULF's original logo and our current logo! These two items alone brought in $875 to the ULF's mission!

We are excited for September and Leukodystrophy Awareness Month, so keep an eye on our social media platforms to learn about more ways to get involved, spread awareness, and support the ULF's dedication to the leukodystrophy community!

2023 BY THE NUMBERS

- **400** In-Person Attendees
- Live and Silent Auctions raised over **$15,000**
- **10** Partner Organizations
- **17** Sponsor Organizations
- Over **100** Presenters
- **28** Leukodystrophies Represented
- **22** Countries Represented

2024

SAVE THE DATE FOR 2024! JUNE 27–29, 2024

Eaglewood Resort and Spa • Itasca, Illinois, USA (Chicago)
Virtual options will be available!

WHAT'S INSIDE

A LETTER FROM THE PRESIDENT OF THE BOARD

Ron, Marla, and their son Aiden

As the summer flies by, as it always seems to, I took a second to reflect on what was one of the absolute highlights of my summer. For those prone to guessing, no, I am not referring to breathing in the smoke-filled air sent straight from my neck of the woods (Canada, sorry eh…) nor the unseasonably hot weather that has bracketed most of our continent, if not the world.

I am instead referring to the ULF's 2023 Scientific Symposium and Family Conference held at the Eaglewood Resort in Itasca, Illinois in late June. It was amazing for me to see so many members of our community come together in one place and share and learn so much together in such a short period of time. Attendance was at an all-time high and was represented proportionally from the four pillars of our community; our families, our fellow advocacy groups, our generous and truly caring partners from the pharmaceutical industry and, of course, our beloved researchers and clinicians.

The feedback we have received has been overwhelmingly positive, but we do want to continually improve this event so please feel free to send the office an email and or complete the survey to let us know what you liked and what we can improve on.

Among the personal highlights for me was seeing so many young researchers interacting alongside their carefully crafted posters with our families as well as our more veteran scientists. The presentations during the Scientific portion were at an extremely high level and provide great hope that new treatments and trials will continue to make their way to our affected individuals. In my 20 plus years of coming to the conference, I have never seen such a diverse agenda for the Family Conference where the opportunity to learn about anything related to leukodystrophy was in one presentation or another.

All that said, my favourite moments at this year's conference occurred as I walked through the halls and witnessed all the great collaboration happening. I was incredibly grateful that so many of the wonderful patient advocacy groups that serve our families chose to make this a truly united conference hosting individual breakout sessions focussed on their specific subtype. My thanks go out to VWM Families Foundation, Cure LBSL, Cure MLD, CTX Alliance, Maxie's Hope, Global Dare, Sister's Hope, Cure Cadasil, AGSAA, Foundation To Fight H-ABC and Alliance MLC for their partnership in making the entire event such a smashing success.

I write this to you as I spend a few days in beautiful Ellicottville, New York enjoying all the activities and people attending the Hunter's Hope annual conference. Our missions are all essentially the same: to serve our families and individuals in any and every way possible and we can all better meet this goal when we work together. I look forward to continuing to work together with Jacque and her fantastic team at Hunter's Hope along with all the other organizations that inhabit the leukodystrophy space.

Before I leave you, I just want to once again extend my congratulations to this year's two ULF award winners. Both have given tirelessly of themselves in service to this community and the strength of the ULF is a testament to their personal sacrifices. Thank you, Bob Rauner, winner of the ULF's Legacy Award and Dr. Geneviève Bernard, the recipient of the Hugo Moser Service Award. Two more richly deserving names to add to a list that reads like a who's who when it comes to making life-altering contributions towards the care of those suffering from a leukodystrophy.

I can't wait to see you all next year, from June 28 to the 30th in Itasca again.

Best wishes to all,

RON CHAPLEAU

ULF Board President

THANK YOU

2023 SPONSORS

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VIKING THERAPEUTICS

Calico

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Orchard therapeutics

bluebirdbio

FORGE BIOLOGICS

affinia THERAPEUTICS

poxel

DENALI THERAPEUTICS

Aspa a bridgebio company

Maxie's Hope

2023 PARTNERS

AGSAA

Alliance MLC

CTX alliance

Envision the Cure — cure CADASIL — Embrace the Challenge

Cure LBSL

Cure MLD

Foundation to Fight H-ABC

Global DARE Foundation #CureRefsum

sisters' hope FOUNDATION

WM Families Foundation

2023 CONFERENCE YouTube PLAYLIST

Thanks to the generosity of our Conference sponsors, we were able to stream and record all the sessions at this year's meeting! Here is a highlight list of the recordings that are now available to view on the **ULF Office YouTube channel**, under the 2023 ULF Family Conference Playlist:

AGS

Presented by AGSAA, speakers include: **Patrick Winters (AGSAA)** on the work of AGSAA on advancing AGS advocacy in the leukodystrophy community. **Lauren Lowery (AGSAA)** on caregiver burnout and ways to overcome the challenges of leukodystrophy parents/families.

ALD & AMN

Speakers include: **Elisa Seeger (ALD Alliance)** on efforts for ALD newborn screening expansion. **Dr. Stephan Kemp (Amsterdam UMC)** on "variants of uncertain significance" and how they complicate the diagnostic process. **Dr. Marc Engelen (Amsterdam UMC)** on the outcome measures in research that leads to developing therapeutic interventions. **SwanBio** on their study of AAV gene therapy for AMN. **Dr. Wolfgang Koehler (Uni. of Leipzig)** on the status of the first international controlled clinical trial with an AMN drug for men. **Dr. Patricia Musolino (Mass. General Hospital)** on the efficacy of using Leriglitazone to treat childhood cALD. **Dr. Amena Fine (Kennedy Krieger Institute)** on Minoryx's study on cALD patients. **Sophie Bozec (Poxel)** on Poxel's work with ALD. **Lizbeth De La Rosa Abreu (Mass. General Hospital)** on the current work of the studies of ALD/AMN Women.

Alexander Disease

Dr. Florian Eichler (Mass. General Hospital) on current topics in Alexander disease.

ALSP

Speakers include: **Dr. Jennifer Orthmann-Murphy (Uni. of Penn.), Dr. Caroline Bergner (Uni. of Leipzig), Aaron Baldwin (Uni. of Penn.),** and **Erin Sullivan (Sister's Hope Foundation)** on a discussion panel regarding a patient's first visit. **Dr. Ivana Magovcevic-Liebisch (Vigil Neruo)** on an overview of Vigil Neuro, and their commitment to the ALSP community. **Dr. Andreas Meier (Vigil Neuro)** on ALSP research and development of clinical treatments. **Dr. Jennifer Orthmann-Murphy (Uni. of Penn.)** on the natural history study and how industry and academia work together

CADASIL

Speakers include: **Bertram Kasiske (Cure CADASIL)** on Foundation updates. **Dr. Jane Gunther (Cure CADASIL)** on patient engagement to drive therapy development. **Dr. Stephen Fitzsimmons (Mount Sinai)** on therapeutic targets for treatment options. **Dr. Jane Paulsen (Uni. of Wisconsin-Madison)** on the CADASIL natural history study. **Dr. Manfred Boehm (NIH)** on understanding mechanism and clinical phenotypes of CADASIL. **Dr. Joseph Arboleda-Velasquez (Harvard Medical School)** on targets for treatment.

CTX

Speakers include: **Dr. Robert Steiner (Uni. of Wisconsin)** on case examples in CTX. **Dr. Bart Duell (Oregon Health & Science Uni.)** on clinical overview of CTX. **Dr. Phillip Swanson (UW Medicine)** on the discovery of CTX disease biomarkers. **Dr. Hidde Huidekoper (Erasmus MC)** on autism with CTX and the benefit of early diagnosis. **Dr. Brian Wishart (Mass. General Hospital)** on psychiatric and behavioral issues in children with CTX.

H-ABC

Speakers include: **Michele Sloan (Foundation to Fight H-ABC)** on foundation updates. **Dr. Dominic Gessler (UMass)** on gene therapy for H-ABC. Dr. Adeline Vanderver (Children's Hospital of Philadelphia) on a disease overview and research updates.

LBSL

Speakers include: **Dr. Christina Nemeth Mertz (Kennedy Krieger Institute)** on scientific updates for LBSL. **Melody Kisor (Cure LBSL)** and **Michael McGinn (Cure LBSL)** on foundation updates.

MLD

Speakers include: **Angela Tom (Orchard)** on the status of Orchard's therapeutic development options. **Dr. Laura Adang (Children's Hosp. of Phil.)** on the impact of development treatments on MLD families. **Lesa Brackbill (Leukodystrophy Newborn Screening Action Network)** on the newborn screening efforts for MLD in the USA. **Maria Kefalas (Cure MLD, The Calliope Joy Foundation)** thanks the community for all the hard work and accomplishments so far and anticipated in the future.

MLC

Speakers include: **Dr. Marjo van der Knaap (Amsterdam UMC)** on the MLC Consortium and registry. **Christina Marouda (Alliance MLC)** on foundation updates. **Dr. Rogier Min (Amsterdam UMC)** on a disease overview. **Dr. Jigiyasha Sinha (Narayana Health)** on the status of MLC in India. **Marla Chapleau (Alliance MLC)** on adaptive activities for people with MLC.

Refsum

Speakers include: **Kristie DeMarco (Global DARE)** on Foundation updates for the Refsum community. **Dr. Joseph Hacia (USC)** on the status of research for adult Refsum. **Dr. Florian Eichler (Mass. General Hospital), Dr. Paul Watkins (Kennedy Krieger Institute), Ann Moser (Kennedy Krieger Institute)**, and **Dr. Wedad Fallatah (Kennedy Krieger Institute)** on historical perspectives on Refsum.

VWM

Speakers include: **Dr. Marjo van der Knaap (Amsterdam UMC)** on therapy targets, the VWM Consortium, registry, and trial development. **Dr. Lezanne Ooi (Uni. of Wollongong)** on VWM patient astrocytes and research. **Dr. Allison Bradbury (Nationwide Children's Hospital)** on gene therapy for VWM. **Romy van Voorst (Amsterdam UMC)** on disease impact on VWM families. **Allyson Buck (VWM Families Foundation)** on the merging of the VWM Families Foundation, VWM Foundation, and Saving Chloe Saxby Foundation. **Dr. William Cho (Calico)** on Calico's clinical trial development. **Dr. Joshua Bonkowsky (Uni. of Utah)** on the phases of clinical trial research.

2023 PLAYLIST ALSO INCLUDES:

- Adaptive Toys
- Brain & Tissue Banking
- Communication Aids
- Forming Your Medical Team
- Forward Movement in the UK
- Gene Therapy 101
- Navigating Hospital & ER Visits
- Modalities of Leukodystrophy Treatments
- Palliative & Hospice Care
- Parents as Teammates for Hospital Social Workers
- Transition from Pediatric to Adult Care
- Physical Therapy Topics
- Disease Research – A Complex Journey
- Occupational Therapy
- Service Animals
- Sharing Your Story
- Speech Therapy
- Special Needs Students – Transition FAQs
- Traveling with Leukodystrophies
- The Unaffected Child(ren)
- Vision Therapy
- Restorative Yoga
- Caregiving Caregivers
- Drug Development and Clinical Trials
- Genetic Counseling & Family Planning
- Leukodystrophy 101
- Making Memories with Your Loved One
- Newborn Screening Updates
- Scientific Symposium Session Summaries

HONORING THE ULF'S BEST

Honoring Dr. Geneviève Bernard and Bob Rauner on their work to better the lives of the leukodystrophy community.

Every year, the ULF honors a worthy physician, clinician, or researcher with the Moser Service Award, and a person in the leukodystrophy community who has furthered advocacy efforts in an extraordinary way with the Legacy Award. The 2023 recipients are Dr. Geneviève Bernard and Bob Rauner.

MOSER SERVICE AWARD WINNER: Dr. Geneviève Bernard

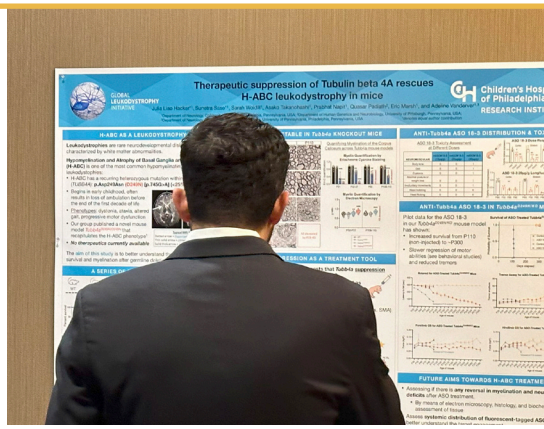
Dr. Bernard has been a pillar of the leukodystrophy community since at least 2014, leading research on disorders including POLR3 and EPRS1 gene variants and other leukodystrophies. Dr. Bernard joined the ULF's Medical and Scientific Advisory Board in 2017, and in 2020 became the Chairperson. She guided us through the Covid-19 pandemic with grace, sage advice, patience, and tact.

LEGACY AWARD WINNER: Bob Rauner

Bob is a familiar presence to many families, medical professionals, and pharma representatives associated with the leukodystrophy community. Bob has been involved in the ULF for 30 years in various roles, but notably served as Board President from 2014 until the end of 2022. Bob steered the ULF through some tumultuous transitions during his tenure, advocated for newborn screening efforts across the country, supported international leukodystrophy initiatives, and worked tirelessly to strengthen the ties between the dozens of leukodystrophy organizations that all strive to bring about a better quality of life for affected families.

NURTURING THE FUTURE OF RESEARCH

The ULF presents the first annual Dr. Paul A. Watkins Excellence in Research Poster Presentation



At this year's Conference, we put a call out for abstracts from researchers to present their work at a poster presentation that took place during the Scientific Symposium on June 22. We were thrilled to have 29 posters on display in the hallways of the Eaglewood! Trainees who submitted a poster were also eligible to participate in the competition portion of the presentation and we were thrilled to honor three exemplary researchers with awards this year.

During the award presentation, Dr. Joe Hacia, friend and colleague, awarded Dr. Watkins with the honor of having the poster presentation named him. Dr. Watkins is a familiar face at the ULF's Annual Meetings and we here at the ULF are eternally grateful for his unwavering dedication to leukodystrophy research. Dr. Watkins was presented with a plaque and a gumball machine as a nod to his work with the "bubblegum gene" in mice. The first place winner of the poster competition was also presented with a gumball machine and trophy, which will continue to be the tradition for years to come as the ULF continues to nurture the future of leukodystrophy researchers all around the globe.

2023 WINNERS

FIRST PLACE: Jessica Herstine, Nationwide Children's Hospital in Columbus, Ohio, USA
"Gene replacement therapy displays early efficacy in two models of Vanishing White Matter disease"

SECOND PLACE: Quinty Bisseling, Amsterdam UMC in Amsterdam, The Netherlands
"Unraveling astrocyte dysfunction in the white matter disease MLC: linking the cytoskeleton to volume-regulated ion channels"

THIRD PLACE: Alexandra Chapleau, McGill University in Montreal, Canada
"Expanding the Phenotypic Spectrum of EPRS1-Related Disorder"

Left to right:
Jessica Herstine, Dr. Joe Hacia, Alexandra Chapleau, Quinty Bisseling, and Dr. Paul Watkins

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DID YOU KNOW?

Did you know you can double your support for the United Leukodystrophy Foundation, without even giving a dollar more? You can if your employer participates in a **Matching Gifts Program**.

You can see if your employer will match employee donations to the ULF on our website! If your company does offer matches, there are easy instructions for how to activate that employer gift.

Thank you for your support! Learn more here: **https://ulf.org/get-involved/**