Newsletter of the Lowe Syndrome Association • Summer 2025 • Vol. 44, No. 2

LSA ON THE BEAM

Circle of Care | Vision for a Cure



LSA Board Members and top local donors at the LSA 2025 Conference in Cincinnati, OH, June 27, 2025.

Igniting Action in Cincinnati: The 2025 LSA Conference

The 18th International LSA Conference is now in the books! Thank you to all the families and friends who traveled across the U.S. and internationally to join us at the Summit Hotel in Cincinnati, Ohio. We are proud to have served a record number of children in childcare this year, and financially accommodate a record number of families this year through stipend funds raised by the LSA Board of Directors.

Thank you to all the families and friends who traveled across the U.S. and internationally to join us in Ohio.

Additionally, we were overjoyed to recognize and honor a few of our incredible local donors in Cincinnati during the conference. This was a milestone moment for our organization—the first time top local donors joined us in person at the conference to be publicly recognized for their powerful impact on our community. Their generosity has made a lasting difference by helping fund travel stipends for families, cover critical conference expenses, and support our ongoing research initiatives. It was truly special to honor these champions face-to-face, celebrate their contributions, and show them the heart of the community they are helping to sustain and grow.

Finally, we would like to thank our wonderful conference sponsors for their generous support! Your partnership helped make this event possible, allowing us to provide meaningful programming, resources, and connection for families affected by Lowe syndrome.

Our 2025 conference brought renewed energy and passion to our mission. We appreciate the robust feedback via our post conference survey which included many highlights and ways we can continue to make future events even better – from accommodations and entertainment preferences to specific agenda recommendations – thank you LSA community!

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LOWE SYNDROME ASSOCIATION

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The Lowe Syndrome Association, Inc. is a non-profit corporation dedicated to the following purposes:

Improve the lives of persons with Lowe syndrome and their families through fostering communication, providing education and supporting research so that individuals can attain their highest potential.

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PRESIDENT'S MESSAGE



Dear LSA Community

What an incredible few days we shared at the 18th International Lowe Syndrome Association Conference in Cincinnati, Ohio! From June 27 - 29, families from across the globe came together in person – many for the first time in years – to learn, connect, and simply be together. **There is something powerful and healing about seeing our boys and men with Lowe syndrome, along with their siblings, laugh, play, and form lasting bonds.**

A heartfelt thank you to our Medical and Scientific Advisory Board members, our local donors and all the speakers who volunteered their time and expertise throughout the weekend. Your dedication continues to fuel our understanding, empower our families, and move us forward.

This year, our conference theme, Igniting Action, was reflected in every corner of our time together. We placed intentional focus on caregivers, acknowledging the immense stress and responsibility that comes with this role. We heard one another, supported one another, and strengthened our collective resolve to care – not just for our loved ones, but for ourselves and each other.

While we missed several families who had to withdraw due to health challenges, please know that our thoughts and prayers are with you. You remain an integral part of this community, always.

As we closed out the weekend, we shared actionable ideas for how we can each contribute to the mission – from grassroots fundraising to community outreach – and we're excited to share more on this soon. While no event is ever perfect, we are proud that the overwhelming majority of attendees felt satisfied with the experience, and more than 90% expressed interest in attending our next gathering in 2027. If you missed my opening comments that outline a few of our priorities - you can **rewatch the video here**.

Looking ahead, there is much to be hopeful about:

- **Giving Tuesday** is just around the corner – a perfect time to come together for a collective fundraiser. More to come on that!
- Every day we continue to expand our partnership with **Citizen Health** and explore how their platform can better support our families.
- We are working towards building **a simple, accessible parent directory** to foster deeper connections across our community.
- We're continuing our efforts to **build champions on Capitol Hill**, ensuring the LSA and our families have a voice where it matters most.
- And yes – we will explore **venues for our 2027 conference**!

Thank you for being part of this incredible journey. Your involvement, optimism, and unwavering support are what keep this organization moving forward. **Together, we are igniting action – and we are just getting started.**

With gratitude,

Jeri Kubicki
President, Lowe Syndrome Association

FAMILY STORIES



Charlotte and Elliot Tsai

Our Lowe Syndrome Journey

By Caroline Tsai

The sun blazes overhead without a cloud in sight. Only a high of 90 degrees today—typical for an October afternoon in Phoenix. My Fitbit reads Monday the 14th, as I push a stroller into Phoenix Children's Hospital with my son Elliot, who is almost 18-months-old. The air conditioning blasting as we enter through the automatic doors. Before long, I am listening to the geneticist explain the results of the test to me, but not comprehending much. My eyes fill up with tears as I hold my son close. What did this all mean? How did I not even suspect my son had a genetic condition? How did I not know that I was a carrier? And for goodness sakes, what is a nephrologist? The genetics appointment I thought was going to be insignificant has now changed our lives forever. My heart shattered.

Over the next several months, and many tears later, my husband and I learned about Lowe syndrome through various medical articles and organizations such as the Lowe Syndrome Association (LSA) and the National Organization for Rare Disorders (NORD). We felt slightly more grounded and informed, but we still were missing something: connection. Explaining to our friends about our son's newfound circumstances was both heart-wrenching and draining. We sought out a local special needs group at Foundation for Blind Children, which provided us with a support network of parents experiencing similar challenges with their young children, but not surprisingly, no children with Lowe syndrome were present.

Flash forward to eight months after receiving our son's LS diagnosis. My husband and I decided to fly our family across the country to Ohio to attend the 2025 Lowe Syndrome Association conference. We were both anxious and excited for this opportunity to learn more about the syndrome from experts in the field, and to meet other families impacted by LS. We were also searching to be a part of something greater than ourselves, while we continued to face the life-long and unpredictable journey of our son's health and development.

At the conference, my husband and I connected with so many families who were warm, friendly and open to sharing their stories, their struggles and their fears. We heard experiences of hardship that resonated with our own. Parents shared accounts of constantly advocating for their son's education, their therapy services and their medical treatment. We identified with others about the endless explaining to medical professionals about the complexities of LS. We discussed the challenges of navigating sibling relationships and how living with the uncertainty of our son's futures was both frightening and exhausting.

We did not just experience grief and sadness at the conference. We listened to stories of resilience and hope, about how milestones such as eating, walking, and communicating, were to be celebrated with every small success. Collectively, the various speakers at the conference touched our

Tsai Journey continued on page 4

FAMILY STORIES

Tsai Journey continued from page 3

hearts, helped us continue to process our own grief, and educated us about complex medical aspects of this rare syndrome. Hearing personal accounts of raising a son with Lowe syndrome during the parent panel was particularly moving, deepening our connection with those individuals who chose to share their most vulnerable feelings about their experiences.

While our 2-year-old son, Elliot, has a diagnosis of Lowe syndrome, his syndrome does not define him. He is a social boy who loves making hand motions to Wheels on the Bus. He will look at you and wave hello, all whilst having a big grin upon his face. His energetic and inquisitive 5-year-old sister, Charlotte, continues to learn that her brother is on his own timeline for reaching a variety of milestones. She tells others how her brother uses a gait trainer to walk and an iPad to communicate, obviously proud of his accomplishments. When asked about her favorite part of the conference, she replied, "the butterfly art," which hangs in her room above her dresser.



Elliot

We are grateful we traveled to Cincinnati to experience the supportive and caring nature of the Lowe Syndrome Association firsthand. The highlight of our visit was having the opportunity to meet many amazing families who made the conference extremely meaningful and beneficial to us, including the board members who coordinated this event. Nothing can change what our sons have gone through and will go through, and nothing can alter the weight we as parents carry everyday with the unknown ahead of us. Even so, getting to know other families has made the reality of our son's diagnosis more manageable, knowing that there are other people who understand what we are going through.

As I sip my morning coffee, a rocket blasts off on my LSA mug, the words "IGNITING HOPE" giving me a mantra to hold close to my heart. I watch as Elliot side-steps along our couch, a major milestone we celebrated a few weeks prior. Lists of appointments to schedule bombard me from every direction: therapies, blood draws, medical follow-ups and MRIs.

"I gave Elliot his meds," my husband informs me before I have a chance to ask. With a quick kiss, he leaves to take Charlotte to summer camp before going to work. Elliot waves, "bye bye," to daddy and sister, another major achievement by our special boy. I look down at my LS bracelet, the yellow letters reading "Lowe Syndrome Association" contrasting with the bright blue band. A tangible reminder that there are other families like us. Other families who have joined together to promote awareness of Lowe syndrome and ongoing research efforts. While the future of our sons may be out of our hands, the bonds we have with each other can help us to stay strong.

I recall a powerful quote by Dr. Martin Luther King Jr., "If you can't fly then run, if you can't run then walk, if you can't walk then crawl, but whatever you do, you have to keep moving forward." Progress matters. No matter how small. Little victories are worth celebrating both in our sons and in ourselves as caregivers.

I carefully place my empty mug in the dishwasher, glancing again at the blue circle of hope encircling my wrist. You are not alone, it seems to whisper to me. I hear a robotic and repetitive "Eggs. Eggs. Eggs," coming from my son's speech generating device. As I take the egg carton out of the fridge, I inhale deeply, remembering to take each moment one at a time.

FAMILY STORIES



Left to Right: Kelsey Daniels, Gina Daniels, Susan Brown, and Angela Torres.



Left to Right: Kelsey Daniels, Dr. Robert Nussbaum, and Gina Daniels.

Dear LSA Families,

My nephew, Grayson, is seven months old and was diagnosed with Lowe syndrome shortly after birth. Overwhelmed by the diagnosis and what it meant for our family, when my Mom and I learned about the LSA, we were relieved to see an organization set up for this community of families and other boys facing the same challenges. We were also glad to see a conference was coming up and decided to attend with Grayson's paternal Grandmother and Aunt.

Attending the LSA conference was such a meaningful experience for us because we connected with so many families and moms and for the first time were able to talk about Lowe syndrome with other people who understood. We hope that my sister, her husband, and Grayson will attend the next conference and also feel supported by the love and camaraderie we experienced during these few days.

We also had the chance to meet the doctors and researchers that have been deeply involved in Lowe syndrome treatment and learned so much information that we're bringing back to Grayson's doctors at CHOP. For example, we never would have known a pediatric dentist was important to find early on without hearing Dr. Tesini and Dr. Gennantonio's presentation. We were so impressed that these doctors, like Dr. Nussbaum, Dr. Brewer, and Dr. Gallop, spent time with the families outside of their presentations and clearly care about the cause. Learning about the research and clinical trials underway gave us hope and is something we are sharing with others to hopefully get more doctors involved.

Overall, we're so glad to have found this community as our family navigates this new world and to have learned what we did so it can help Grayson. We are looking forward to helping spread knowledge about this ultra-rare disease and will be dedicated to supporting other families and the organization in the future.

The Daniels Family

FAMILY STORIES



The Escobedo Family

by Sara Escobedo

My name is Sara Escobedo, my husband's name is Ismael Escobedo, and we are the proud parents of four beautiful children – a daughter and three sons. Each of them has their own personality,but today I want to share more about our youngest, Mikalo, and the journey our family has been walking since he was born.

Mikalo was born prematurely at 36 weeks on October 15, 2024. From the very beginning, he's shown us how much strength can be found in the smallest body. He spent two weeks in the NICU due to feeding difficulties and low muscle tone which we later learned was one of the earliest signs of his diagnosis.

Since birth, Mikalo has been closely followed by a team of specialists. His early months were filled with tests, appointments, and long nights of worry but also with hope.

One of the major challenges came when Mikalo was diagnosed with congenital bilateral cataracts. At just under four months old, on February 13, 2025, he underwent surgery on both eyes and spent the night in the hospital for observation. Then, just weeks later on February 26, he was admitted to the hospital again this time for failure to thrive. His weight gain had stalled, and he was aspirating when eating. He spent 10 difficult days hospitalized before being discharged on March 7.

Because feeding continued to be a struggle, doctors placed a G-tube for nutrition support on April 3, alongside a hernia repair. He's also had genetic testing, which confirmed his diagnosis of Lowe syndrome. We were heart broken – our world changed forever that day.

I will say one of the most powerful turning points in our journey was being able to attend the Lowe Syndrome Association National Conference. That experience gave us something we didn't even know we were missing – community. I met other parents who understood. I saw children/men who looked like Mikalo, who were thriving in their own ways. I felt a level of emotional support I hadn't felt since his diagnosis. This conference gave me the space to grieve, but also the strength and hope. It reminded me that it's okay to feel a mix of emotions to mourn what we imagined life to be. This is something I still struggle with on a daily basis with every battle I still question when things will get better?



One thing we also recently got set up with was vision services and one of the questions [asked was] "What did I see for his future?" – I simply told her, "I just want him to be here." I'm sure we all hope for the same things for our boys! To other families navigating rare diagnoses I see you. I know the weight you carry. And I hope you find connection and comfort in knowing you are never truly alone.

2025 LSA CONFERENCE

Conference Session Highlights

"I really liked the depth and breadth of different speakers at the conference. I also really liked the different breakout groups. Most of all I liked meeting all the other Lowe syndrome families."

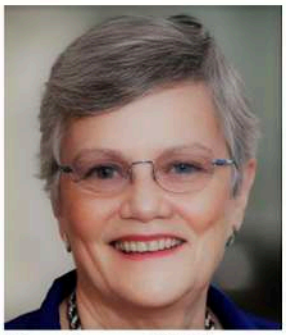


Left to Right: **Theresa Haugen, Caroline Otani, Clare Gromoll**, and **Patricia Bass**

"I especially liked the opportunities to connect with other women - Clare did a great job of facilitating that so that each person could share and also discuss in smaller groups.

I appreciated the research and kidney talks too ***– it is so rare to hear from professionals who understand LS, so I appreciated those talks and the time each doctor spent talking with me individually."***

"Dr. Brewer's presentation was excellent. I found Donna McCartney's presentation supportive and helpful."



Eileen Brewer, *LS & the Kidneys Panel Discussion*

Click here to see Dr. Brewer's presentation slides.



Donna McCartney, *Grief & Emotional Support for the LS Family / Family Panel*

"Direct access to professionals."

2025 LSA CONFERENCE

"Meeting other families, access to doctors and specialists. The childcare worked well and was super helpful to allow us to engage in the content and the conversations."



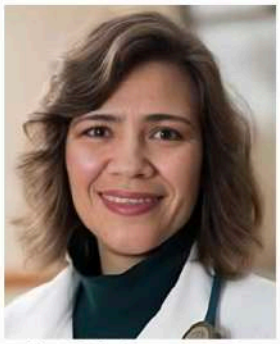
Skidaddles Team Member and **Jonah Brehm**

"I'm thankful that my kids had a place to be and that they were excited to go there each day. ***It was so nice to be able to focus on the presentations knowing my kids were safe and cared for."***



Skidaddles Team Members, **Tyrese Floyd**, and **Jonathan Ankrom**

"It focused a lot on the caregivers. *The ways for them to cope, help the boys and self-care."*



Naila Goldenberg,
Self-Care for the Caregiver



Bodhe and Krew Samaniego

2025 LSA CONFERENCE



Left to Right: ***Jenny Gallop**, *Transforming Possibility into Reality: From Research to Finding a Treatment*, and **Robert Nussbaum**

What aspects of the conference did you find most valuable?

"[LS] 101 with MSAB, grief panel, [Ruchi's] opening Keynote, Gallop talk and fundraising conversation."



Yang Sun, LS *101 – New Parent Session with MSAB*

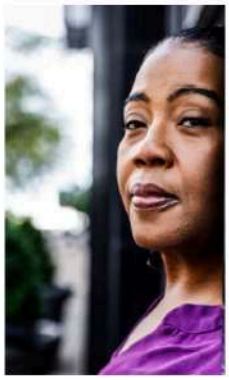


***Jenny Gallop**, *LS 101 – New Parent Session with MSAB*



Robert Nussbaum, LS *101 – New Parent Session with MSAB*

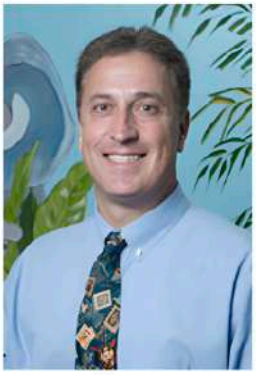
"Ruchi's talk was great."



Crystal Hall, Seizure Training & Certification / Family Panel

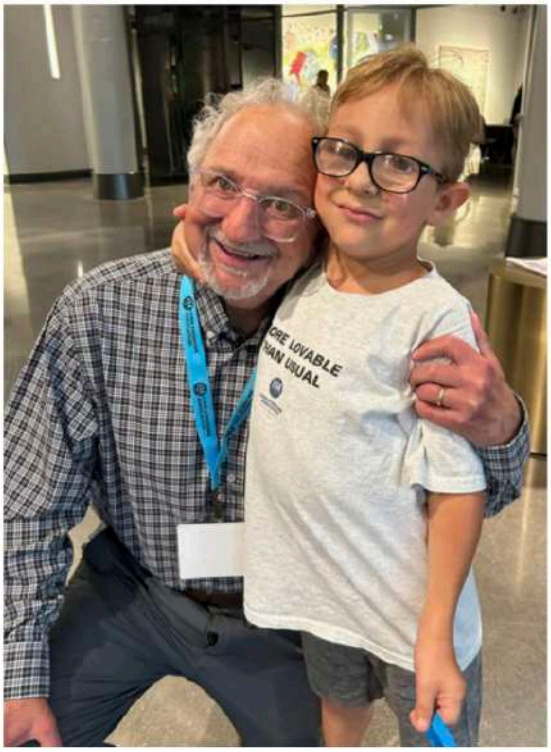


Ruchi Koval, *Living With and Loving Your Special Needs Child*



John Gennantonio, *Onboarding A New Dentist*

"I valued the interaction with the other parents/mingling, as well as the kidney review session, dental session, epilepsy session, and the grief session."



David Tesini, *Onboarding A New Dentist*, and **Blake Kubicki**

2025 LSA CONFERENCE



Back Row, Left to Right: **Dustin Rasmuson**, **Coral Karsky**, **Andy Karsky**, **Will Karsky**, **Jonah Gromoll**, **Clare Gromoll**, **Theresa Haugen**
Front Row, Left to Right: **Casey Rasmuson**, **Cole Rasmuson**, **Jacob Karsky**, **Joshua Karsky**, **Sam Gromoll**, **Aidan Gromoll**

"Meeting the families and sharing stories and bonding over LS. ***Not feeling as alone."***



Left to Right: **James Carbonneau** and **Brian Samaniego**



Left to Right: **Sara Escobedo**, **Theresa Haugen**, and **Lisa Waldbaum**

2025 LSA CONFERENCE



"My son gets to be around boys like him."

Left to Right: **Jordan Chattaway**, **Tyler Jackson**, and **Tyler Haubert**



Left to Right: **Shane Call**, **Alex Waldbaum**, and **Ross Hernaez**



Left to Right: **Jordan Chattaway** and **Sam Gromoll**

2025 LSA CONFERENCE



Left to Right: **Tyrese Floyd** and **Cricket Alberts**



Esterlina and **Ian MacInnes**



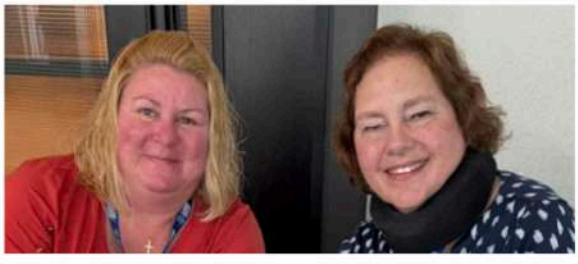
Jan and **Jordan Chattaway**



Left to Right: **Terrence Burke**, *Wump Mucket Puppets*, **Blake Kubicki**, and **Alex Waldbaum**



Tamra and **Tyrese Floyd**



Left to Right: **Tammy Jackson** and **Jan Chattaway**

2025 LSA CONFERENCE



Jonathan Ankrom



Rickey and **Ross Herneaz**



Above and Below: **Erin** and **Oliver Reynolds**



Left to Right: **Shane Call**, **Tyler Jackson**, **Tyrese Floyd**, and Skidaddles Team Member





Andrew Otani

2025 LSA CONFERENCE



Rod and **Michelle Ankrom**



Terry, Matthew, and **Andy Johnson**



Sam and **Clare Gromoll**



Shane Call

2025 LSA CONFERENCE



Left to Right: **Scott Richards** and Skidaddles Team Member



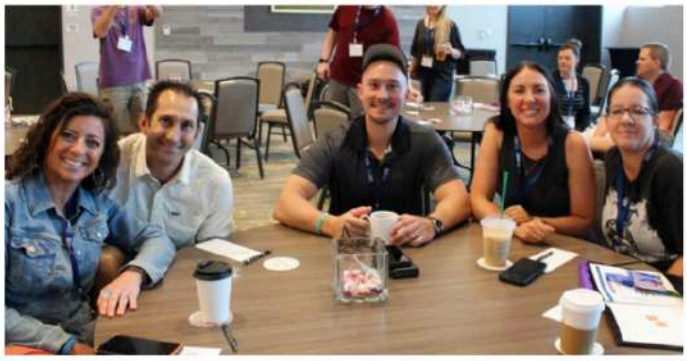
Allen, Nathan, and **Jennifer Kwitkowski**



Left to Right: **Clayton Arnold** and **Terrence Burke**, *Wump Mucket Puppets*



Left to Right: **John Macdonald**, **Clare Gromoll**, **Barry Rinehart**, **Lisa Waldbaum**, **Jeri Kubicki**, **Theresa Haugen**, and **Anna Farris**



Left to Right: **Julie and Brian Samaniego**, **Stephanie and James Carbonneau**, and **Patricia Bass**



Left to Right: **Ismael and Sara Escobedo**, **Lisa Waldbaum**, and **Theresa Haugen**

"This is the first trial in patients with Dent disease due to OCRL1 mutations, based on repurposing a drug that targets the underlying mechanism causing the disease. It aims to find out if the drug improves abnormal kidney function."

Dr. Francesco Emma

Press Release: First Therapeutic Trial in Dent Disease caused by OCRL 1 gene mutations

Joint Press release from Bambino Gesù Children's Hospital, Rome; Department of Biochemistry and Gurdon Institute, University of Cambridge, UK; Institute of Physiology, University of Zurich, Switzerland; Dent Disease Foundation USA; Lowe Syndrome Association, USA; and Lowe Syndrome Trust, UK.

The Lowe Syndrome Trust UK, Dent Disease Foundation USA, and the Lowe Syndrome Association USA have each awarded grants of €20,000 towards a pilot clinical study led by Professor Francesco Emma, Head of the Nephrology unit at the Bambino Gesù Children's hospital in Rome, Italy. The first patients were enrolled into this Phase 2 clinical trial of alpelisib in Dent disease 2 in June, 2025. Dr. Emma said, "This is the first trial in patients with Dent disease due to OCRL1 mutations, based on repurposing a drug that targets the underlying mechanism causing the disease. It aims to find out if the drug improves abnormal kidney function."

Dent disease (type 2) is a rare condition due to inactivating mutations in the OCRL1 gene that affects males. The most common manifestation of the disease is dysfunction of the proximal tubule of the kidney, which manifests as low-molecular-weight (LMW) proteinuria. The disease may be difficult to recognize and can lead to life-threatening complications and kidney failure at a young age. Dent disease can be cured with kidney transplant but there is a shortage of available organs, and a transplant requires lifelong immunosuppression – so a drug treatment would be a significant step forward. Jill Goodrich, Co-Executive Director of the Dent Disease Foundation said, "If a treatment is available, we hope that testing for LMW proteinuria will become more widespread to the benefit of not only Dent disease 2 patients but also the other genetic causes of Dent disease. The progress to chronic kidney disease is a burden that affects whole families, and we look forward to hearing about the progress of the research."

Lowe syndrome is caused by other mutations in the same OCRL1 gene and is a more severe multi-organ condition causing congenital cataracts, intellectual disability, autism-like symptoms, and seizures amongst other symptoms. These disorders have a profound effect on patients and their families. Longstanding supporters of the Lowe Syndrome Trust include patrons Jonathan Ross OBE, Penny Lancaster Stewart, and Tony Hadley MBE. Penny Lancaster, Patron of the Lowe Syndrome Trust, commented, "We are delighted that the Lowe Syndrome Charity is supporting this clinical trial and hopeful that this will be a major breakthrough in the treatment of Dent 2 and Lowe Syndrome and transform the lives of the affected families."

The trial has been made possible by an international consortium of

The Trial, continued on page 17

The Trial, continued from page 16

scientists, clinicians, and the patient groups that are funding the study. The drug compound being used – alpelisib – was identified by a collaboration between Dr. Jennifer Gallop's laboratory at the Gurdon Institute and Department of Biochemistry, University of Cambridge UK, and Professor Olivier Devuyst's laboratory at the Institute of Physiology, University of Zurich, Switzerland.

Dr. Gallop gave a keynote lecture at the Lowe Syndrome Association conference on 29 June in Cincinnati, Ohio, USA. She talked about the route to treatments and the significance of the trial in Dent 2 patients for their goal of a meaningful treatment by 2030. Rod Ankrom, whose son lives with Lowe syndrome commented, "We have been coming to LSA conferences for 34 years, and for the research at first there were just guesses, and after decades of work, there now really might be a treatment coming down the line." Jeri Kubicki, President of the Lowe Syndrome Association said, "Our boys experience Lowe syndrome in different ways, at different stages. We are exploring multiple avenues and are excited to determine what this clinical trial tells us."

Dr. Gallop and her team at the University of Cambridge have been working on the fundamental science behind the trial for over 15 years. Their research, funded by the Wellcome Trust and Lowe Syndrome Association is on the actin cytoskeleton that is in all cells in our body and important for all functions, like pumping our heart and supporting our immune system. When Gallop worked out that specific lipid molecules in cell membranes are important in controlling the actin cytoskeleton, she saw that there was a link with Lowe syndrome and Dent disease 2, which causes alterations to these lipids. Gallop said, "And excitingly, we realized there's an existing drug that might be able to help."

At that time, Dr. Gallop reached to the group of Professor Olivier Devuyst (University of Zurich), who has done pioneering work on genetic disorders of the kidney proximal tubule, including the cellular mechanisms and models of Dent disease. Devuyst is co-directing the ITIN-ERARE Research Priority Program in Zurich, which works at translational programs to accelerate drug discovery for rare diseases. Devuyst, Berquez and colleagues were able to test the drug in model kidney cells, and then in a humanized mouse model of Dent disease 2 – that they had just characterized.

Working together, the team found that alpelisib did work to rebalance the actin cytoskeleton and improve kidney function in lab models of the diseases. Professor Devuyst commented, "This pioneering trial builds on decades of research establishing the crucial role of endolysosomal trafficking in kidney proximal tubule cell function. It also provides a rare opportunity to repurpose a drug that has been decisively useful in another set of rare disorders sharing problems with the small lipids." These fundamental insights were detailed in a joint publication (Berquez M et al. Kidney Int 2020, with Editorial Commentary), paving the way for the preparation of the Phase 2 clinical trial.

The trial is due to report in October 2026 and if it shows that alpelisib is effective in the Dent disease 2 participants, the hope is that it will also be beneficial to Lowe syndrome patients suffering with kidney disease.

Citizen Health Testing New AI Tool

In January, the LSA formally launched our partnership with Citizen Health – a platform that enables patients with rare conditions to get access to all their medical data in one place and accelerate critical research efforts with minimal effort.
Did you know that Citizen will feature an AI Advocate that acts like a personal assistant – helping you stay organized and easing the daily demands of caregiving? It can handle a wide range of tasks, from retrieving notes from past doctor visits to tracking symptoms over time and compiling medication lists. By typing in a few words, you can quickly access the information you need – making it easier to prepare for upcoming appointments, insurance appeals, or IEP meetings without having to dig through endless records. Please join Citizen now so you can have your child's records on the platform by the time AI Advocate is made more fully available to communities later this summer/early fall!

What is the value for the LSA community?
Citizen is the easiest way for communities to build natural history studies and help accelerate research for treatment options. Completely digital – no site visits required.

Citizen offers real value to patients by empowering them with all their medical data in one secure, digital place.

Patients are able to share their medical records electronically, for continuance of care, instantly, and directly from the platform

Citizen has already accelerated research into many rare conditions and is committed to the rare patient community. We can give researchers the answers they need to help design better clinical trials, faster with Citizen.

How does it work?
Create an account: In a simple, five minute onboarding process, patients or their caregiver will create a profile. You provide a few pieces of demographic information and a list of providers where they have received care. Guardianship papers (or other legal documentation) will also be needed in the case of a caregiver signing up on behalf of a patient 18 years or older.

Citizen will do all the work to collect the medical data from all of the patients' providers and provide patients access to their data in their own secure, digital profile.

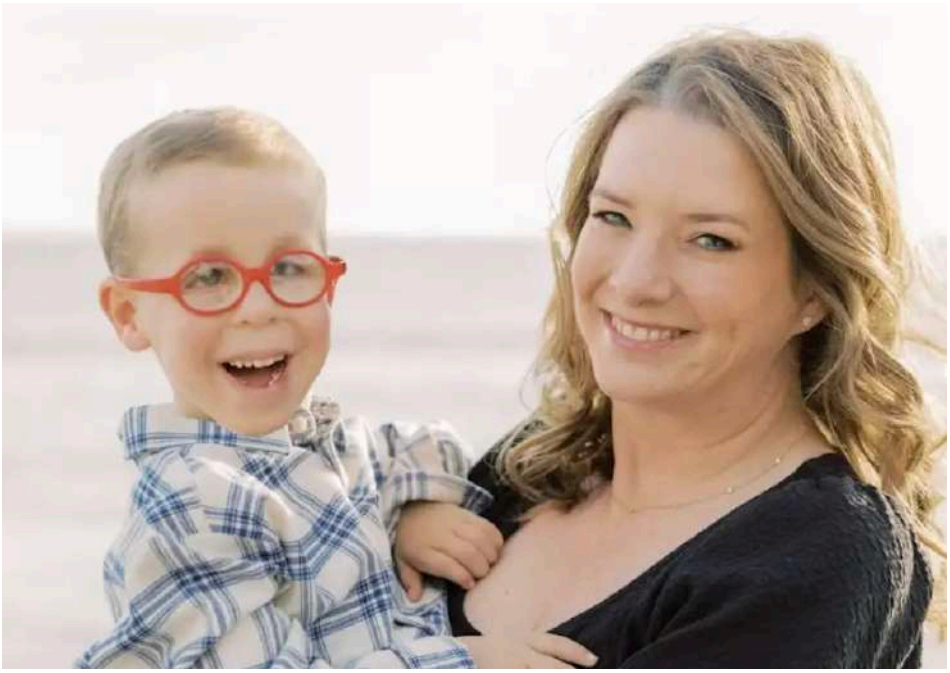
With patient data finally all in one place, the patient is in control of what happens next. Share the Citizen profile with your care team to help inform healthcare decisions, and/or choose to share your de-identified data stripped of any personal identifiers to help power Research.

Security and Privacy with Citizen
Privacy and security are at the foundation of everything that Citizen does. Their Chief Regulatory Officer, Deven Mcgraw, served as the Deputy Director for Health Information Privacy at the Office for Civil Rights and helped craft the HIPAA right of access laws protecting US patients and their health data today. In addition to their privacy policy online Citizen has a dedicated video series on this: Citizen Privacy and Security overview video with Chief Regulatory Officer, Deven McGraw.
A few highlights about Citizen's approach is listed below:

- Citizen Health is committed to protecting your privacy. The data governance followed by Citizen ensures secure storage of patient medical records. To learn more about the security measures Citizen takes, please visit https://www.ciitizen.com/privacy
- Citizen Health uses HIPAA-compliant cloud services Amazon Web Services (AWS) to maintain patient information.
    - AWS is an encrypted platform in widespread use by companies world wide, and their product reputation depends on keeping data secure.
    - Citizen encrypts data locally and have stringent policies for staff regarding access to data aimed at ensuring your data is protected at all times.
    - Citizen trains staff and contractors regularly on our company security policies and requirements.
    - We never share patient data with any third parties or access it for any other commercial purpose without the patient's explicit consent.
    - Citizen follows the HIPAA Privacy Rule's Expert Determination to confirm minimal risk of patient re-identification in data shared with research partners

How do I get support from the Citizen Health team?
Email support@citizenhealth.com.
Feel free to reach out to LSA Admin, Anna Farris, if you have any questions.

LSA NEWS



Scott andAmy Richards

Amy Richards Elected to LSA Board

The Lowe Syndrome Association is delighted to officially welcome Amy Richards to our Board of Directors.

For the past two years, Amy has been a behind-the-scenes powerhouse, generously offering her time, skills, and energy to support our mission. From event planning to strategic communication and tech know-how, Amy brings a thoughtful, can-do spirit that helps keep our work moving forward – especially when juggling the many demands of a small but mighty nonprofit.

What sets Amy apart isn't just her professional expertise – it's her deep commitment to our community. She shows up with positivity, optimism, and a collaborative mindset, always ready to support her fellow board members and the families we serve. Her presence is felt in countless ways, and we're thrilled to now welcome her as an official part of our leadership team.

Amy lives in Ventura, California, with her husband, Mark, and 6 year old son, Scott. Scott also has two older brothers, Matt and Jack, who are in college. In her professional life, Amy is an accomplished event professional with years of experience streamlining operations, leading teams, and managing high-stakes projects with grace. She is the head of events for a media company, The Information, where she focuses on producing events in the tech industry.

Please join us in extending a heartfelt welcome to Amy. We're grateful for her dedication and excited for what lies ahead!

Our Community Continues to Expand

We were thrilled to welcome several new families – both in-person at the LSA 2025 Conference and online to the LSA community! Of the families we welcomed at this year's conference, 47 were attending a LSA conference for the first time. We've also welcomed 133 new friends and supporters of the LSA through our website.

47 attended a LSA conference for the first time and we welcomed **133** new friends and supporters of the LSA through our website.

Families, relatives, and other supporters of boys and men living with LS can easily join the LSA community through our website, by following the Lowe Syndrome Association, Inc. Facebook group page, or by answering a few questions on our LSA Community Facebook page

LSA NEWS

The Normandy Group Partners with the LSA

by Alex Perez, Director of Federal Affairs, Normandy Group

We're proud to partner with the Lowe Syndrome Association (LSA) to support its mission and help elevate its presence in Washington, DC. The Normandy Group is a bipartisan federal advocacy firm with deep experience in patient advocacy, research funding, and health policy. Our team includes former senior congressional staff and Congressman Henry Bonilla, a longtime appropriator from Texas, who brings valuable insight into the federal funding process. We're excited to work alongside LSA to ensure the organization's priorities are heard and advanced at the highest levels of government.

"Right now, Congress is operating under a full year continuing resolution that holds funding at last year's levels through the end of September. That leaves about 60 days in the fiscal year and fewer than 25 working legislative days in the House to finalize key priorities.

Funding for health agencies like NIH remains frozen even as demand for research support increases. In this environment, we are focused on ensuring that Lowe syndrome priorities are included in the conversations shaping final spending decisions and future research investments. Looking ahead, we are exploring the potential of organizing a DC Fly In day, where families, advocates, and clinicians could come to Capitol Hill to share their stories and ask lawmakers for increased support.

These meetings are often the most impactful way to humanize federal policy and leave a lasting impression with Members of Congress and their staff. Whether it's calling for more research funding or simply raising awareness, these direct conversations can build long term champions for LSA's mission.

Together, we'll work to position LSA as a recognized voice in the health policy space. By strengthening relationships on the Hill, partnering with like-minded organizations, and engaging strategically throughout the federal budget process, we can help ensure Lowe syndrome gets the attention and support it needs in Washington.

About the Normandy Group
Our team works with local governments, nonprofits, and businesses to navigate federal government processes. We help clients tell their story, create new opportunities, and identify federal programs that can be of benefit.



Conference Merchandise

It's not too late! Conference and general LSA merchandise is still available for purchase here.

Please note that all conference gear will no longer be available after August 31, 2025, so don't miss out!



LSA NEWS



What is the LSA Community Facebook Group? Why is it Important?

The Lowe Syndrome Association (LSA) is a 501(c)3, established in 1983. The LSA Community facebook page, is an inclusive space open to anyone impacted by or seeking to learn about the condition. This can include parents, extended family members (like grandparents, siblings, or aunts/uncles), medical professionals, educators, therapists, researchers, and others in the broader community. It is our hope that we can continue to spread broader awareness about Lowe syndrome and we welcome those who would like to learn more about LS to join this group. The goal is to share reputable information, connect people across roles, and build wider awareness and support. We expect all group members to be kind and courteous, respecting our diversity of experience.

Beyond a Facebook Group, the LSA connects families with resources, webinars, clinical research developments, fundraisers, and in-person conferences. Dialogue within this group directly impacts the LSA's work.

Joining this group ensures individuals will receive emailed newsletters and announcements throughout the year. Questions about this group? Please contact a group administrator- Amy Richards, Anna Farris or Clare Gromoll.

Separate from the LSA, there is also a parent-run Facebook Group, Lowe Syndrome Parents (not associated with the 501(c)3,) that we highly encourage parents impacted by LS to join. This group was created by parents, for parents. It's a private space where parents and primary caregivers of children with the condition can speak openly with others who share similar day-to-day experiences. Including things such as personal struggles, wins, emotional support, advice on navigating daily care and managing family life.

LSA NEWS



Emile's Voice in Action

We're thrilled to share a recent article featuring our very own Emile Eno-Daynes, a young man with Lowe syndrome in the UK who is using his unique voice and experiences to provide critical training.

Emile and Owen Torence-Smith have partnered with Ace Anglia, a disability advocacy service, to deliver the Oliver McGowan training, as people with lived experience of a learning disability and autism. Emile's work is helping educate staff on the lived experiences of individuals with complex medical needs.

"If this training wouldn't have happened there would have been more people with autism or learning disabilities in hospital having avoidable deaths. This training is crucial."

Emile's family shared this article with us in hopes it would be of interest to other families navigating the LS journey. We are honored to pass it along.

Read the full article here.









LSA Merchandise Available

LSA merchandise is now available at www.zazzle.com/lowe_syndrome_assoc. Options include apparel, mugs and phone cases. A portion of all sales are given to the Lowe Syndrome Association. Help us spread awareness of the good work of the Lowe Syndrome Association and support our boys.



THANK YOU LSA SUPPORTERS

Peer-to-Peer Fundraising – Become an LSA Champion

Thanks to all the families who contributed to our fundraising conversation at the conference- your insight helps us focus our efforts on what matters to you!

We are deeply moved by the dedication of families who've stepped up to fundraise for our shared mission. A heartfelt thank you to the Arnold, Gromoll, Ankrom, and Alcantara families for launching their own campaigns to raise awareness and support for Lowe syndrome.

- **The Arnold Family** hosted a garage sale to help spread awareness and raise funds to support research.
- **Clare and Sam Gromoll** created a Facebook birthday fundraiser, raising over $700!
- **The Ankrom Family** The Ankrom Family planned a fundraiser at their local Pizza Ranch (Rhinelander, WI) for August 18, where Jonathan Ankrom works part time 8-10 hours per week. Rhinelander's Pizza Ranch has agreed to donate 20% of all sales from 4-8 pm, as well as tips! Friends will bus and wipe down tables while the Ankrom Family greets guests. The Ankroms promoted the event on their local TV station. Check out Jonathon in action.
- **Katherine and Freddy Alcantara** are running a half marathon in September to raise money and work towards a goal amount.

Your efforts are already making a meaningful impact and we are honored to have your energy, creativity, and heart powering this work forward.

Start Your Own Fundraiser Today!

Your voice is powerful. Help us spread the word about Lowe syndrome and raise awareness by starting your own fundraising page on Give Lively. By doing so, you're helping the Lowe Syndrome Association reach more people, raise critical funds, and make a real impact.

"Peer-to-peer fundraising" means you invite your friends, family, and networks to support a cause you care about. When you fundraise on behalf of the LSA, you're not just raising money – you are raising awareness, building community, and making an impact in our LS community. You aren't limited to using Give Lively too! Whether you run a race, host a birthday fundraiser, or simply share your story, every effort counts.

It's Easy to Get Started!

1 Create an Account. Go to our Give Lively fundraising page. You'll be prompted to create a free Give Lively account (if you don't already have one).

2 Customize Your Fundraiser. After logging in, you can customize your fundraiser with a personal message, fundraising goal, and a photo.

3 Share! Share your unique fundraising page link with family, friends, and supporters

Important Notes for Donors Based Outside the U.S.

Because LSA is a U.S.-based nonprofit, all donations through Give Lively are processed in U.S. dollars (USD), even though the "$" symbol is used without specifying the currency. This is important for you and your donors to understand:

- A donation listed as "$20" on your page means "$20 USD."
- The actual amount charged will vary depending on the exchange rate at the time of donation.
- The donor's bank or credit card statement will reflect the converted amount.

Available Payment Methods for Donors Based Outside the U.S.
Donors can contribute via:

- Credit or debit cards (Visa, Mastercard, etc.)
- Digital wallets (Apple Pay or Google Pay, if available)
- PayPal (available in Australia)

Please note, Venmo is not available for non-U.S. donors.

Launch your campaign today and become part of something bigger. Together, we can improve the lives of persons with Lowe syndrome and their families.

THANK YOU LSA SUPPORTERS

Thank you to everyone who financially supported the LSA between January 1, 2025 and July 31, 2025. We are sincerely grateful for your compassion and continuous support. Lowe Syndrome Association (LSA) extends our deepest gratitude to our many generous donors. Your investment allows LSA to foster communication, provide education, and support research to improve the lives of persons with Lowe syndrome and their families, empowering individuals to attain their highest potential.

$1,000+

Bettencourt Household
in honor of Blake Kubicki
Erlandson Household
in honor of Robert Erlandson
Gillespie, Ben Household
in honor of Blake Kubicki
Nichols Household
in honor of Scott Richards
Padebettu Household
in honor of Angad Padebettu
Wientge Household
in honor of Oliver Reynolds
Wright Household
in honor of Blake Kubicki

$500 - $999

Arnold, Sue Household
in honor of Clayton and Levi Arnold
Eklof Household
in memory of Lindsey Edwards
Gray Household
in memory of Tim Gray
Gromoll, Norda and Carl Household
in honor of Sam Gromoll
McLeod Household
in memory of Lindsey Edwards
Nussbaum Household
Smith Household
in honor of Blake Kubicki
Teplitz Household
in memory of Lindsey Edwards
Waldbaum Household

$250 - $499

Abramowitz Household
Anonymous Donor
in honor of Blake Kubicki
Anonymous Donors
in memory of Lindsey Edwards
Byles Household
in honor of Blake Kubicki
Duffe Household
in honor of James Jerman and family
Gill Household
Gillespie, Brad Household
in honor of Blake Kubicki
Heatwole Household
in memory of Lindsey Edwards
Otani Household
in honor of Andrew Otani, Daniel Martin, and Hunter Haugen
Richards Household
in honor of Scott Richards
Suchy Household
Townsend Household
in honor of Blake Kubicki
Webb Household
in honor of Robert Erlandson
Wharam Household
Woodring Household

$100 - $249

Abundis Household
Baldwin Household
in memory of Lindsey Edwards
Broadbent Household
Buchs-Hammonds Household
in honor of Sam Gromoll
Chattaway Household
in memory of John Chattaway
DeGori Household
Gilbo Household
in honor of Ethan and Mark Mondello
Haugen Household
in honor of Hunter Haugen
Kidd Household
in memory of Lindsey Edwards
Kowalak Household
in honor of Grayson Alexander Brown
Kubicki Household
Leslie Household
in memory of Lindsey Edwards
Macdonald Household
O'Brien Household
in memory of Susan Sherman
Pollard Household
in memory of Lindsey Edwards

Special thanks to The UPS Store Anderson Township for printing the LSA 2025 Conference programs free of charge. Your generous in-kind donation allowed us to share essential conference information while keeping event costs low.

THANK YOU LSA SUPPORTERS

$100 - $249 continued

Risman Household
in honor of Raymond Hiett
Savage Household
in memory of Lindsey Edwards
Siconolfi Household
Tomilson Household
Wilbur Household
in memory of Lindsey Edwards; in honor of Matisse Tresca

$50 - $99

Alderson Household
in honor of Matisse Tresca
Arunachalam Household
Beane Household
in honor of great, great grandson, Kaison
Carbonneau Household
in memory of Susan Sherman
Daniels Household
in honor of nephew, Grayson
Gehring Household
in memory of Lindsey Edwards
Gromoll, Clare Household
Hoernschemeyer Household
Holcomb Household
in memory of Lindsey Edwards
Lambert Household
in honor of Clayton and Levi Arnold
LeGrand Household
in memory of Susan Sherman
Owen, Emma Household
in honor of Noah Owen
Paff Household
in memory of Mark Wolert
Rheem Household
in memory of Lindsey Edwards
Torres Household
in honor of Grayson
Veilleux Household
in memory of Susan Sherman
Vokbus Household
in memory of Susan Sherman
Young Household
in memory of Lindsey Edwards

Up to $50

Alberts Household
in honor of Kyle Alberts
Anonymous Donors
in memory of Susan Sherman
Burke Household
in honor of Matthew DeGori
Coppolo Household
in memory of Susan Sherman
Dinoia Household
in honor of Matthew DeGori
Farris Household
Hooker Household
in memory of Tim Gray
Kamerman Household
Kellenberger Household
in memory of Lindsey Edwards
Kordell Household
in honor of Hunter Haugen
Larsen Household
in memory of Tim Gray
O'Neill Household
in memory of Michael Rochvarg
Rich Household
Slikkers Household
in memory of Tim Gray
Sorrentino Household
in honor of Matthew DeGori
Tantillo Household
in memory of Susan Sherman
Watson Household
in memory of Susan Sherman

Note: Donations listed in this issue were those **received as of January 1, 2025 to July 31, 2025**. Donations received after that date will be included in the next *On the Beam* issue.

We would also like to thank the following corporations and foundations for their generous donations:

Up to $99
Kroger Community Rewards

$100 - $499
GoodCoin Giving Fund
In honor of Scott Richards

$500 - $999
Mother Murphy's Flavors
In honor of employee, Danielle Burns

Mueller Furniture Co.
In honor of employee, Bobby Morgan

$1,000 - $4,999
Service Roofing & Sheet Metal
In honor of employee, Mike Richardson

Theodore Eckert Foundation
In honor of Scott Richards

$5,000 - $15,000
S & P
In honor of employee, Rohit Padebettu

Goodwin Law
In support of our rare disease community

THANK YOU LSA SUPPORTERS

Donors by Fundraiser

Adam Snow's Fundraiser
Facebook
Lynne Household
Marks Household
Marson Household
Swartout Household
Thai Household

Amy Richard's Fundraiser
Facebook & Instagram
GoodCoin Giving Fund
Richards Household

Bethany Reynold's Fundraiser
Facebook
Bee Household
Borton Household
Dentino Household
Nelson Household
Schubert Household

Brenda Abundis' Fundraiser
Pampered Chef
Abundis Household
Wharam Household

Clare Gromoll's Fundraiser
Give Lively & Facebook
Hanson Household
Lemenager Household
Neuenfeldt Household
Thorson Household

Clayton & Levi Arnold's Fundraiser
Give Lively
Arnold, Sue Household
Lambert Household

Emma Owen's Fundraiser
Facebook & Instagram
Aspinall Household
Davies Household
Griffith Household
Leigh Household
Moran, Kevin Household
Moran, Mandy Household
Owen, Emma Household
Owen, Joanne Household
Owen, Yvonne Household
Parry Household
Roberts Household
Ross Household
Tejada Davis Household
Thomas Household

Hilary Rich's Fundraiser
Facebook
Chase Household
Crosson Household
Pale Household
Rich Household

Jennifer Caroline's Fundraiser
Facebook
Lombardi Household

Jeri Kubicki's Fundraiser
Give Lively
Bettencourt Household

Keri Loveday's Fundraiser
Facebook
Lehner Household

Matthew DeGori's Fundraiser
Give Lively
Burke Household
Dinoia Household
Sorrentino Household

We would like to thank the following individuals who created fundraisers on behalf of the LSA:

Special thanks to those that have raised:
$250 – $499, $500 – $999, or $1,000+ **in 2025**!

- **Adam Snow** (Facebook)
- **Amy Richards** (Facebook & Instagram)
- **Bethany Reynolds** (Facebook)
- **Brenda Abundis** (Pampered Chef Fundraiser)
- **Britt Marie** (Facebook)
- **Clare Gromoll** (Facebook & Give Lively)
- **Clayton & Levi Arnold** (Give Lively)
- **Crystal Rice** (Facebook)
- **Emma Owen** (Facebook & Instagram)
- **Hilary Rich** (Facebook)
- **Jennifer Caroline** (Facebook)
- **Jeri Kubicki** (Give Lively)
- **Kayla Guss Martin** (Facebook)
- **Keri Loveday** (Facebook)
- **Kimberly Haubert** (Facebook)
- **Lyndse Pepper** (Facebook)
- **Matthew DeGori** (Give Lively)
- **Rachel Siconolfi** (Facebook)