



Circle of Care | Vision for a Cure™

IN THE BEAM

LSA SPOTLIGHT



John, Catherine, and Thomas

Grandparents

Growing up, I was blessed to have a wonderful grandfather who helped raise me in my younger years. Despite saying good-bye to him when I was only seven, I can still remember a lot of wonderful memories I made with him. My grandfather instilled good morals, showed me what genuine love and kindness was, and most importantly, always just encouraged me to be myself.

Their unconditional love, wisdom, and life experiences enrich the lives of their grandchildren and offer support to their own children as they navigate parenthood.

If we should be so blessed to have grandparents in our lives, they truly touch a place in our hearts like no other. Their unconditional love, wisdom, and life experiences enrich the lives of their grandchildren and offer support to their own children as they navigate parenthood. They create a safe place; building trust, confidence, and happiness within their grandchildren. Their entertaining stories teach life lessons.

Grandparents, continued on page 3

Celebrating Grandparents

It takes a full, supportive community to raise a child with Lowe syndrome. So this edition of our newsletter highlights the important role that Grandparents can play in our son's lives. We want to thank all the grandparents for your love and support.

In this issue

- 2 President's Message
- 3 Family Stories
- 4 Memorials
- 6 Research Update
- 10 Fundraising Corner
- 11 Get the Lowe Down
- 13 LSA Supporters



**LOWE SYNDROME
ASSOCIATION**

Circle of Care | Vision for a Cure

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.

On the Beam, ISSN #0740-218X, All contributions are tax-deductible under Section 501(c)(3) of the Internal Revenue Code. Reproduction of this newsletter or any parts thereof is not allowed without the prior written consent of the Lowe Syndrome Association

8190 Beechmont Avenue, Suite 111, Cincinnati,
Ohio 45255.

Tel: 216-630-7723

E-mail: info@lowesyndrome.org

www.lowesyndrome.org

LSA Board of Directors

Jeri Kubicki, (President)
Cincinnati, OH

<jgkubicki@gmail.com>

Cristen Alberts, (Treasurer)
Virginia Beach, VA

<alberts.cristen@gmail.com>

Cori DeGori
Kings Park, NY

<cdegori@aol.com>

Theresa Haugen, (Marketing)
Blaine, MN 55014

<haug0016@umn.edu>

Laura Kim (Social Media)
Los Angeles, CA

<laurakimster@gmail.com>

John Macdonald (Legal)
Jacksonville, FL

<jmacdonald@akerman.com>

Barry Ross Rinehart (Creative|Technology)
Boca Raton, FL

<barryrinehart@gmail.com>

Lisa Waldbaum
Solon, OH

<LisaWaldbaum@lowesyndrome.org>

Kaye McSpadden (Founder)

Medical and Scientific Advisory Board

Eileen D. Brewer, M.D.

Jenny Gallop, M.D., Ph.D.

Richard Lewis, M.D.

Robert L. Nussbaum, M.D.

Elahna Paul, M.D., Ph.D.

Yang Sun, M.D., Ph.D.

David A. Tesini, D.S.D., M.S.

Philip Zeitler, M.D., Ph.D.

PRESIDENT'S MESSAGE



Happy New Year LSA Friends!

In this third edition of the LSA's digital newsletter, I hope you learn something new and are inspired to become an even more engaged member of our community. 2023 was a busy year- full of many accomplishments: several funded research proposals, an international research meeting, a refresh of our website (check it out here- www.lowesyndrome.org), new educational and caregiver support zooms sessions, and the launch of an awareness campaign that educated over 70 offices in our U.S. Congress. We continue to seek financial support for our efforts- from grassroots fundraisers to grants commissioned by various organizations. All these things are in support of our mission to educate about Lowe syndrome, foster communication and drive key research that will lead to better treatments, and someday, we hope a cure for those living with Lowe syndrome.

2024 brings even more research efforts and collaboration. Don't miss the article from Dr. Aguilar that outlines the 2024 Research Symposium hosted by Purdue University in November. We are thrilled to have two international research meetings in two consecutive years! As we get closer to the event, stay tuned for ways our local families and their Lowe syndrome sons may get involved in this effort.

In closing, I am also pleased to share that we are working very hard to secure a venue for our 2025 Lowe Syndrome Conference. For planning purposes our target dates are June 27-29, 2025. We strive to provide something for everyone in this 2025 conference- expert talks, a track for new families and our veteran families, childcare, a space for our older boys and men to play a game or listen to music, evening social activities, and group meals. 2025 will be an epic year- we will have accomplished so much and will look forward to seeing our broad community celebrate in person!

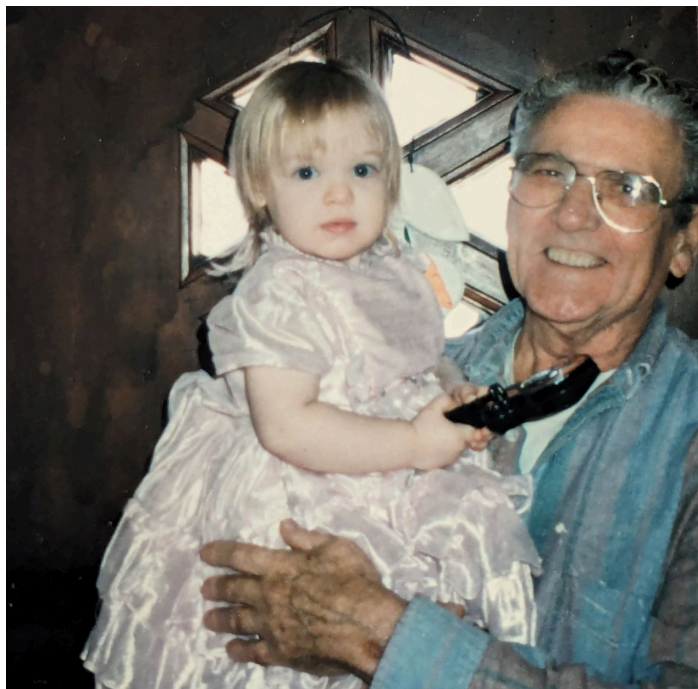
As we secure more conference details, we will be in touch via email and social media. If you need to update your contact information, including your email or mailing address please let Heidi know at lsa-admin@lowesyndrome.org.

Thanks for being a part of our growing community. Let's accomplish many more milestones in 2024!



FAMILY STORIES

Grandparents, continued from page 1



Chantel with Grandfather Ed

Now that I am a parent myself, I've come to experience the importance of grandparents in new light. When I was young, my grandfather was always ready and willing to teach me something new. At the time, I loved building toy planes, helping in the kitchen, and was genuinely curious about everything. He embraced my every interest, unconditionally, and one of my favorite memories is him teaching my five year old self how to brew the best cup of

coffee. I still remember how proud I was serving my parents and grandparents coffee all on my own after he taught me how.

Of course I didn't know it then, but looking back, I often reflect on how important that time truly was with my grandfather at such a young age. He is the reason I am so resilient and adaptive today. Isn't it interesting the things we realize later on in life? Now I get to watch my son experience the same unconditional love and support from his own grandparents.

The greatest gift from grandparents is their unconditional love, support, and the joy they bring to their grandchildren. Our family's grandparents show our son every day the sky is the limit, he can do anything he puts his mind to, embracing his every interest – they show him he is so much more than a diagnosis. They have embraced our family and the challenges we face every day.

They are our biggest supporters and have helped bring out the advocate in me, which is exactly what our child needed, especially with a rare diagnosis. Grandparents truly play an important role in our children's lives – no matter how big or how small, those memories will live on in their hearts as they grow. They are helping us as parents perform the most important job in the world, raising and caring for our wonderful young men.

Chantel Jablonski

More Grandparents



Above: Hunter with his Grandpa, Gene Haugen on a fishing trip in northern Minnesota. Center: Blake Kubicki with his Grandpa Ben Gillespie. Right: Blake Kubicki with his Grandpa Ron Kubicki..



MEMORIALS

Kang-Suk Andrew Lee

September 8, 1983 - December 10, 2023



Andrew Lee

Kang-Suk Andrew Lee was born in Boston, Massachusetts on September 8, 1983 to Young Hee Lee and Jay Kyoon Lee. He was baptized at St. Aidan's Church in Brookline, MA and received his first communion at Holy Cross Church in DeWitt, NY. Andrew was a very happy baby and there are many memories of his beautiful smile and chubby cheeks. He loved being in the water and especially playing around in the bathtub. Being in a city with experienced doctors and specialists allowed Andy to receive the care he needed from birth. Andy had glaucoma surgery followed by removal of his cataracts at two months. He was diagnosed with Lowe syndrome around age 1. Andy had overall good physical health and a strong immune system. However, his biggest challenges were his developmental delays. He needed to be fed, bathed and changed, and needed assistance using the restroom. He learned to walk at age 6. He was non-verbal and had his own way of communicating his needs and wants.

The family moved to Syracuse, New York in 1985, where Andy and his big sister, Jean Ah, were soon joined by a new baby brother, Michael. Jean Ah took on the role of junior mom, taking care of her two little brothers growing

up—playing with them, changing their diapers, and being Mom and Dad's little helper. Andy attended a BOCES program at Stonehedge Elementary School until 1990 when he began attending Tecumseh Elementary School in his neighborhood. Michael joined Andy at Tecumseh and they were two peas in a pod, attending middle and high school, and even sharing a bedroom together until they graduated together from Jamesville-DeWitt High School in 2005. Upon graduation, Andy attended the ARC of Onondaga day habilitation program.

Despite his disabilities, Andy was blessed to live a rich life full of activities that he loved. When he was just seven years old, Dad started carrying Andy down the ski slopes. In 1997, he joined the ARISE and Ski program for people with disabilities and skied with Dad. Since 2004, Chuck Bertuch has been volunteering and skiing with Andy and Dad, and had plans to continue this ski season. Andy started therapeutic horseback riding while in high school and continued throughout the remainder of his life with From The Ground Up



Andrew' riding in a rowboat



Andrew's 40th Birthday

and ARISE at the Farm. In middle school, Andy learned to swim at the school pool in PE with Coach Dodge. In later years, Andy continued to swim regularly with volunteers from the JD High School and LeMoyne College swim teams.

There were also so many simple things that brought Andy joy. He watched Disney movies, listened and danced along to Dad singing Korean karaoke songs, played peek-a-boo, and laughed at fart jokes. Andy loved outings to Shoppingtown Mall in DeWitt and spent many hours there with family or with various volunteers and caregivers. He knew the mall and the library like the back of his hand and had his route and routine. He enjoyed riding the elevator and escalator and would often giggle in response to being asked if he wanted to go to the mall. He also enjoyed walks with Mom and Dad at Green Lakes State Park and Clark Reservation State Park. Andy enjoyed row boat and canoe rides which were connected to the overall theme of him loving being in the water.

In February 2014, Andy moved to a New York State operated group home, which was a difficult decision for the family, but one that allowed Andy to be an adult and live with housemates with special needs. Fortunately, he was only 1.9 miles from home. This allowed Andy to have frequent interaction and visits

Lee, continued on page 5

MEMORIALS

Lee, continued from page 4

with Mom and Dad. He lived with five other housemates and was cared for by numerous staff members. It took him a while to adjust to a new environment but he eventually learned to call it home. The staff at his house gave him loving care.

Throughout his life Andy was surrounded by incredible people supporting him, including teachers, therapists, direct service professionals, social workers, home health aides and nurses, bus drivers, volunteers for outings, horseback riding, swimming and skiing, and of course, many physicians. Andy's biggest champion was Mom. She was his personal chef, nutritionist, nurse, tailor, stylist, and executive assistant. As a registered nurse, she took charge of his medical needs and made sure to understand and question all decisions made around his health. She prepared healthy meals and snacks for him, even after he moved to his new home. She is a pro seamstress, adjusting all of his clothes, sewing his bedding, and reupholstering his favorite chair four times. She made sure that Andy wanted for nothing.

Overall, Andy was in good health until his unexpected kidney failure, which resulted in his passing on December 30, 2023. He was at Upstate



Andrew' skiing with his Dad

Douglas Mitchell

January 9, 1987 - December 12, 2023

It was an honor and privilege to be Doug's mother and 24/7 caregiver for the last 36 years. It was never a burden and I accepted my calling.

Doug loved the simple things in life. He enjoyed the great outdoors, the sunshine in his face, music, riding in the paratransit bus to all of his activities, people (especially the ladies) and holding hands. I will always remember Doug's laughter and love.

I loved Doug with all my heart, BUT God loved him more. Doug spent

Christmas with Jesus last year and now for all eternity. One day we will be together again.

With comfort and peace,
Diane Mitchell

No eye has seen,
no ear has heard,
and no mind has imagined
what God has prepared
for those who love Him.
1 Corinthians 2:9 NLT



The JK Lee family, 2015

University Hospital with Mom and Dad by his side. Andy loved his family and had a special relationship with each of us. He changed our lives in so many ways. He made us better people and taught us every day about patience, love and grace. Dad used to call him "little Jesus" in our family. We are so grateful for the love and peace he brought into our family. He truly was a blessing. Our family is so loved and by so many because of Andy. Andy is survived by his mother and father, Young and Jay

Lee, his older sister Jean Ah, younger brother Michael, brother-in-law Tim and niece Olivia. He was loved by so many family and friends and will be missed.

Jay and Young Lee (Syracuse, NY)
Jean Ah Lee, Tim Walsh, Olivia Walsh
(San Diego, CA)
Michael Lee (Berkeley, CA)

Lenny

LSA Funds Key Research for 2024 and Beyond

In the fall of 2023, several research proposals were submitted to the LSA for consideration of funding. I'm pleased to share that upon considerable review and discussion, in alignment with our Medical and Science Advisory Board, the LSA Board of Directors selected the following initiatives to financially support. A brief summary of each is included below:

Research Team & Location

Dr Jonathan Gadsby and Dr Jenny Gallop, University of Cambridge, Cambridge UK

Research Title & Summary

Mechanism & cell type specificity of alpelisib action on the cellular phenotypes of Lowe syndrome

The mutations in OCRL1 that cause Lowe syndrome increase the levels of an important lipid inside cells, phosphatidylinositol (4,5) biphosphate (PI(4,5)P2), that has multiple effects on cell functions that in turn cause brain, kidney and eye problems that are disabling and life-limiting. My previous research on how proteins and lipids control cellular activity suggested that a drug, alpelisib, which blocks a protein that acts on PI(4,5)P2, called phosphoinositide 3-kinase (PI3K), could be used to treat Lowe syndrome and Dent 2 disease. We found an improvement in the protein lost in urine when the mouse model of Lowe syndrome and Dent 2 disease was treated with alpelisib. Alpelisib has shown good results

in treating patients with metastatic breast cancer and some rare developmental disorders, where it is now approved and in long-term use. In this grant we will explore new opportunities for Lowe syndrome treatment through mechanistic study of the actions of alpelisib on kidney and brain cells. Study of disease mechanisms using new ways to measure changes in Lowe syndrome neuronal cells offers hope that future treatments can be tested for their effect on neurological symptoms before clinical trials.

Timeline:

2024- Growing human kidney 2 and induced pluripotent stem (iPS) cells, optimizing conditions, initial cell treatments

2025- Cell measurements, data collection and analysis

What We Hope to Learn:

The molecular details of how alpelisib is working in kidney cells and how it affects brain cells derived from Lowe syndrome iPS cells

Research Team & Location

Dr Francesco Emma (Ospedale Pediatrico Bambino Gesù, Rome, Italy), Dr Barbara Ruggerio (Ospedale Pediatrico Bambino Gesù, Rome, Italy), Dr Jenny Gallop (University of Cambridge, UK) and Dr Olivier Devuyst (University of Zurich, Switzerland)

Research Title & Summary:

Alpelisib for the treatment of Dent 2 disease or mild Lowe syndrome

Mutations in OCRL1 cause Lowe syndrome and another ultra-rare genetic kidney disease, Dent 2 disease. In Dent 2 disease the kidney symptoms are similar to Lowe syndrome, there can be some mild learning disabilities and no cataracts. How OCRL1 mutations cause kidney disease is most understood of all the body tissues and it is similar between Lowe syndrome and Dent 2 disease. Previously, we found an improvement in the protein lost in urine when the mouse model (which mimics both Lowe syndrome and Dent 2 disease) was treated with alpelisib, which blocks a protein called phosphoinositide 3-kinase (PI3K). Alpelisib is an existing drug for a rare developmental disorder and breast cancer, has shown good results, and is now approved and in long-term use. This means that the drug development for alpelisib has already been completed and it could be prescribed "off-label" by clinicians with a relatively small amount of additional work. We are performing an initial pilot study treating 4 young adults with Dent 2 disease or mild Lowe syndrome, where there is high proteinuria with unaffected glomerular filtration rate. We will treat patients with alpelisib for one month and determine whether kidney uptake of protein in the urine is improved and how alpelisib is tolerated by patients that show kidney symptoms and do not have substantial care needs or intellectual disability, as the least harmful way to test whether alpelisib will be helpful to people with OCRL1 mutations. This pilot study will be co-sponsored in equal amounts by

LS Research continued on page 7

RESEARCH LSA UPDATE



LS Research, continued from page 6

the Lowe Syndrome Association, the Lowe Syndrome Trust, and the Dent Disease Foundation.

Timeline:

Jan-June 2024: Ethical review:

June-Dec 2024 : Study

2025 : Analysis and write up

What We Hope to Learn:

Whether alpelisib is safe in short term treatment, whether alpelisib is likely to help kidney function in Dent 2 disease patients and by analogy in Lowe syndrome. Whether there is clinical justification for considering a larger trial in future that also includes Lowe syndrome children.

CanFite Pharmaceuticals Plans to Target Lowe Syndrome with drug Piclidenoson

Building off the research findings from Dr. Antonella De Matteis, Professor of Biology, University of Naples, Italy, CanFite Biopharma plans to move towards clinical studies to further test the potential use of Piclidenoson for treating those with Lowe syndrome.

“Having tested thousands of compounds in search of a treatment for Lowe syndrome, Piclidenoson is the only compound we’ve found to date that has shown to be effective in preclinical studies. Importantly, we observed that Piclidenoson treatment in mouse models of Lowe syndrome leads to a significant decrease of the urinary loss of proteins in diseased animals,” Dr. De Matteis stated. “We chose to investigate Piclidenoson based on the availability of extensive scientific data showing its excellent safety, coupled with efficacy in this disease which involves renal, cerebral, and ocular manifestations.”

Can-Fite Chairman Dr. Pnina Fishman commented, “We are hopeful that Piclidenoson can offer a much needed treatment for infants, children, and young people living with

Lowe syndrome. Based on Piclidenoson’s proven safety profile in clinical trials to date, and because Lowe is a rare disease in dire need of treatment, we plan to move into an advanced stage clinical study which may open a path to approval. Dr. De Matteis and her team have made an impactful discovery with Piclidenoson and we look forward to working with her and Fondazione Telethon.”

The LSA will stay close to this development in hopes of supporting CanFite’s endeavors through community engagement and fundraising.

A press release announcement can be found here

<https://ir.canfite.com/news-events/press-releases/detail/1027/can-fite-enters-rare-genetic-disease-field-based-on>



CORDS Registry

Support Research by Helping the LSA Build Our Patient Registry!

All families should have received an email from the LSA requesting your support of our research efforts by growing our registry. In addition to updating your registry, please also include a copy of your son’s de-identified genetic testing results. This may include blood and/or enzyme testing results.

If you have any questions about this effort, feel free to contact Cori Degori at cdegori@aol.com

Purdue University to Host 2024 Lowe Syndrome Research Symposium

The International Lowe Syndrome Symposium is being planned to take place at Purdue University in October 2024. The primary objective of this symposium is to convene an interdisciplinary and international community, including scientists, medical professionals, clinicians, patients, families, and other relevant organizations. This collaborative platform aims to advance research, increase awareness, and improve clinical outcomes related to Lowe syndrome. The symposium is organized by R. Claudio Aguilar from Purdue University's Department of Biological Sciences, in collaboration with Thomas G Sors, the Director of Scientific Strategy and Relations of the Purdue Institute of Inflammation, Immunology and Infectious Disease.

Purdue is partnering with the LSA, the Lowe Syndrome Trust, Purdue Institute for Integrative Neuroscience, Institute for Drug Discovery and Institute of Inflammation, Immunology and Infectious Disease, which reflects our collective determination to improve the lives of those affected by Lowe syndrome and to bring about broad awareness for sustained improvements in clinical outcomes.

Key Symposium Objectives include:

1 Scientific Collaboration and Exploration: This symposium will serve as a hub for the exchange of technical knowledge and insights among international researchers, clinicians, and medical professionals. It will provide a platform for in-depth discussions on Lowe syndrome's etiology, clinical manifestations, and potential therapeutic interventions.



2 Diverse Representation: We are committed to promoting diversity, equity, and inclusion (DEI) throughout the symposium. We aim to engage individuals with diverse backgrounds, underrepresented groups, and clinicians from various specialties to enrich discussions and perspectives on Lowe syndrome.

3 Awareness and Education: By actively involving patients, families, and patient advocacy groups such as the LSA, we intend to raise awareness of the disease. Educational sessions will address research findings in layperson terms, management strategies, and avenues for patient support.

4 Clinician-Researcher Collaboration: The symposium will create a unique opportunity for clinicians and researchers to establish and strengthen collaborations. We aspire to bridge the gap between scientific discoveries and clinical applications, ultimately enhancing patient care.

5 Support for Early Career Investigators: To nurture the next generation of researchers and clinicians, we will primarily use the R13 funding to support "Bring a Junior Colleague" awards. This initiative will foster knowledge transfer and community building between experienced professionals and early career investigators.

With our accomplished speakers, workshops, and networking opportunities, we are confident that the Lowe Syndrome International Symposium will be a significant step toward advancing our understanding of Lowe syndrome and fostering collaboration within the research community.

Gene Editing- How is Lowe Syndrome Impacted?

As a community of caregivers, relatives and health professionals we often share news articles on various topics that may be of interest for those impacted by Lowe syndrome. One such article regarding the promise of gene editing for individuals with high cholesterol was posted last fall in the New York Times and illuminated by community member Katherine Barrow (thank you again Katherine). The article underscores the point that if gene editing works, it could be a tool to prevent heart disease in those with genetically high cholesterol. The article can be found here:

<https://www.nytimes.com/2023/11/12/health/cholesterol-gene-editing-study.html>

The LSA membership has expressed curiosity about whether and how this technology might impact Lowe syndrome. Dr. Nussbaum and Dr. Jenny Gallop are helping us parse out the potential and challenges of this technology.

Gene editing can be used in different ways. One is to go in and correct the specific alteration (mutation) in a particular disease gene that is causing the disease, thereby restoring its function. Although precise correction of each patient's specific mutation in a disease gene IS possible with CRISPR base and prime editing technology, it is more challenging than an alternative strategy, which is to counteract and compensate for the loss of function of the disease gene by deleting a piece of another gene, NOT the disease gene itself, thereby destroying its function. This strategy only works if destroying this other gene's function has been shown to

compensate for loss of function of the disease gene and its function can be eliminated without harming the patient.

The key challenges with this technology as it pertains specifically to Lowe syndrome are clear:

1 There is no single gene mutation in the Lowe syndrome gene that could be edited to correct the misspelling responsible for Lowe syndrome in all individuals. Each individual patient's variant causing the disease would have to be edited to correct the misspelling. This would require a specialized gene targeting therapy for each patient unless a patient has exactly the same disease-causing variant as another patient (which does happen, but in general, variants are very heterogeneous). This type of precise gene correction is not what is being done for conditions that are showing gene editing promise and received much publicity such as hypercholesterolemia or even in sickle cell anemia (where there is only one particular DNA misspelling responsible for the disease). Instead with these diseases, researchers are taking the strategy of targeting other genes in order to compensate for the initial disease-causing variant(s).

2 There is no other gene that we know we could delete in order to counteract the effect of the loss of the Lowe syndrome gene without harming the patient. While today we do not have another such specific gene identified, future research could bring more insight to the promise of this technology. Many questions remain, such as might the PI3-kinase that Alpelisib inhibits be a possibility? Currently the drug use is a partial inhibition and we do not know

what potential adverse side-effects could occur following permanent complete loss of function of that gene. Also, we may gain more insight into other genes that, when inhibited, improve Lowe syndrome such as Piclidenoson developed by CanFite Pharmaceutical).

3 There is not one tissue to focus on in which to correct the effect of loss of the Lowe syndrome gene function in Lowe syndrome. Multiple tissues are impacted. In the case of gene editing for high cholesterol or sickle cell anemia, for example, gene editing need only be used to destroy the function of a gene in only one tissue, the liver or bone marrow respectively, to be effective. Gene editing for Lowe syndrome would need to occur with high efficiency in multiple tissues, including the central nervous system, which is particularly difficult to target with gene replacement or gene editing therapies. Some gene therapies, such as Zolgensma for Spinal Muscle Atrophy can access the central nervous system by injection into the space around the spinal cord. The kidney is not easy to target. Thus, there are multiple cell types and not all can be targeted equally.

We will continue to share any updates or changes in this thinking as technology and our Lowe syndrome research continues to advance.



FUNDRAISING CORNER

LSA Facebook & Instagram

Over the past several months, we have been experiencing issues with both of these platforms due to changes with Meta and Paypal Giving. We learned of this particularly during Giving Tuesday when meta inadvertently linked personal facebook accounts with the LSA facebook account resulting in some errors in giving receipts. This Facebook issue has been resolved. Additionally, we found it necessary to completely delete and re-open a new LSA Instagram account. If you are an instagram user please follow us at @lowesyndromeassoc

Thank you to all of our Facebook and Instagram supporters! We know many of you host fundraisers on these platforms to benefit the LSA, and we appreciate this so very much! Let's plan to make 2024 the most successful Giving Tuesday yet by engaging all our contacts on social media to share in our community giving efforts.

Mark your calendars for Tuesday December 3, 2024- which is later than previous years!

We Want YOU!

Fundraising is the backbone of a rare disease nonprofit like ours! Do you have any fundraising experience and may want to lend some time to our community efforts? Would you be interested in participating in a fundraising committee with other like-minded folks? Do you have any friends, family or colleagues who have this type of experience and may be looking for ways to support an organization part time? The LSA is looking for your feedback, ideas and support in all things fundraising.

Please reach out to Jeri (president@lowesyndrome.org) or Theresa (haug0016@umn.edu) if you have any insight to share!



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.



GET THE LOWE DOWN

Did you Move?

Let's stay in touch!

If you moved within the last year or so, please help us keep our database updated by sharing your new address.

We had more than 20 annual holiday cards returned- so please contact Heidi Payot with your new information at Isa-admin@lowesyn-drome.org

LSA Caregiver Support – Monthly Zoom Meetings

In July 2023, we launched an experimental offering for all caregivers of boys and men with Lowe syndrome to share compassion, questions, and wisdom across age and geography. Building supportive community is the aim of our monthly Zoom meetings. We currently meet monthly on a weeknight at 8pm (CST). LSA volunteers Clare Gromoll and Cori Langert-Degori co-facilitate our gatherings.

Topics explored thus far:

- Making the best of the summer months
- Managing behaviors
- Supporting nutrition
- Advocacy in school, medical, and other settings
- Transitioning into adulthood with Lowe syndrome
- Lowe-related grief & stress during winter holidays



- Fresh starts: trying something new as a caregiver
- The early years: diagnosis and next steps

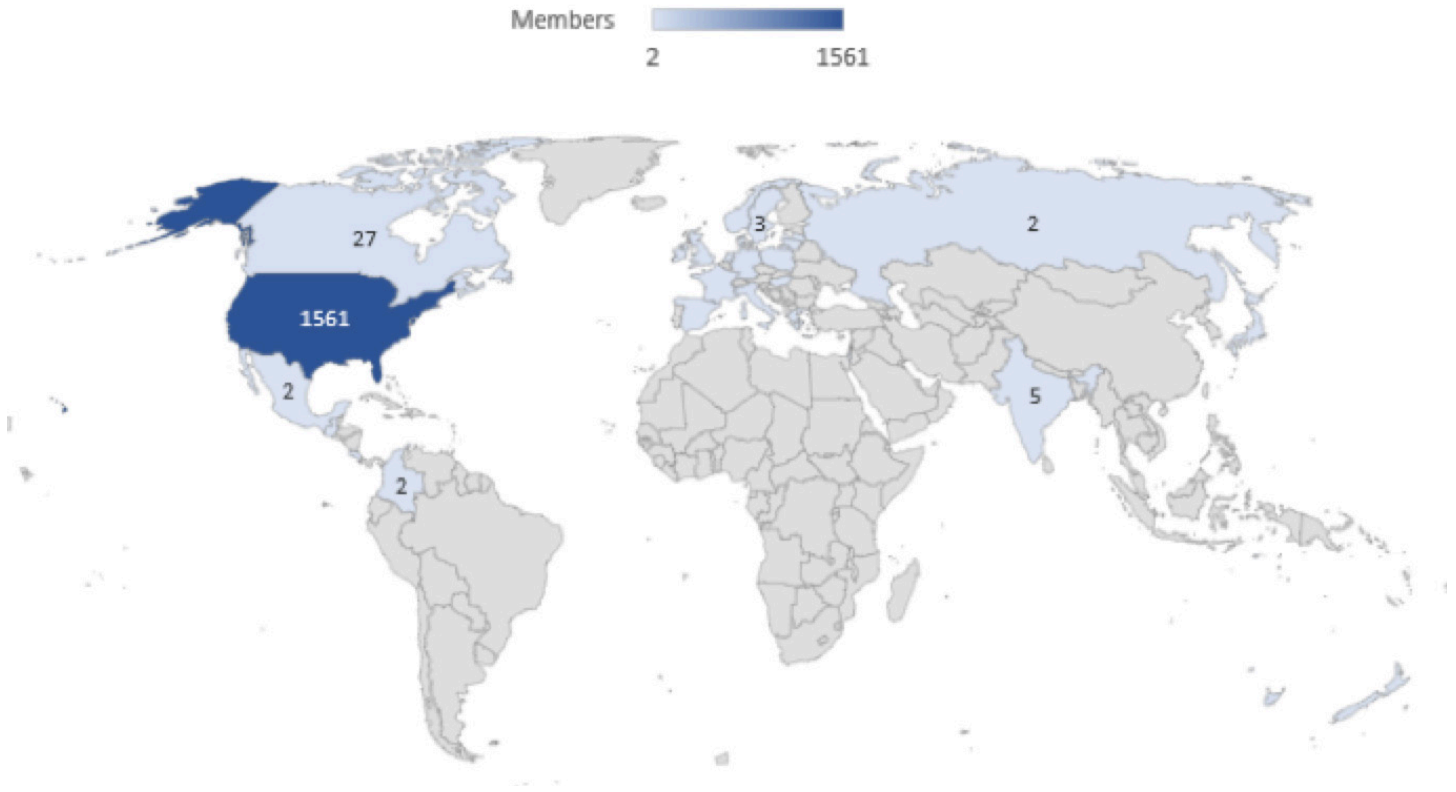
Meeting reminders (including the topic and recurring Zoom link) are posted on our “LSA Community” Facebook group and sent by email through our LSA distribution list. Due to the sensitive nature of our discussions, we do not record our meetings. Given that our community exists across all time zones, we are open to expanding our caregiver support network by creating a second monthly meeting at another time of day. Please contact Cori (cdegori@aol.com) or Clare (claregromoll@yahoo.com) with questions or topic ideas!

LSA Refreshes our Website

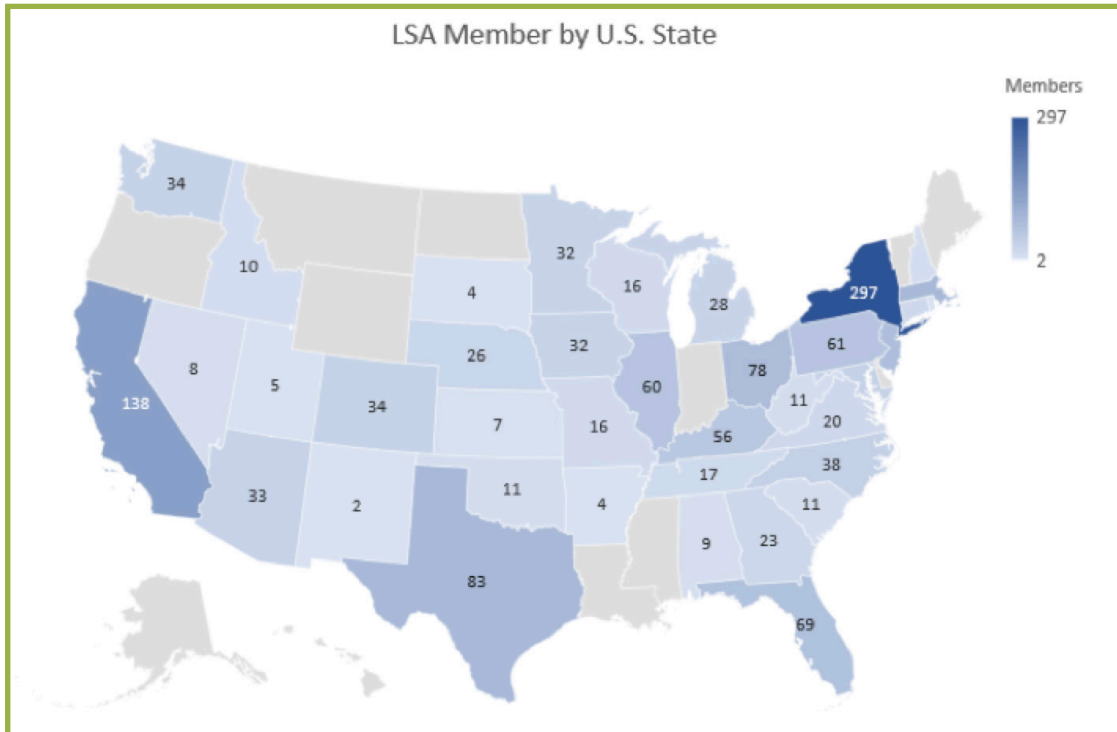
Please check out a few key changes we recently made to the LSA website, including:

- Reorganizing content and updating key links
- Adding access to the LSA digital newsletters, new videos and space for past and upcoming conference information
- Updating Wellcare and our RFP process

LSA Members Around the World



LSA Member by U.S. State



THANK YOU



The LSA extends our deepest thanks to our generous donors. Your contributions uplift Lowe families and fuel critical research that gives us hope. We are sincerely grateful for your compassion and support.

\$5000

Hrncir Household, Dallas, TX.
Padebettu Household, Jersey City, NJ.

\$1000

Merit Energy Associates,
in honor of Noelle Otero.
Ankrom household, Masonville, IA.
in honor of Carol Ankrom.
Brunelle household, Colgate, WI.
*in honor of Joseph and Aurora Valdez,
and Eva Felkl.*
Clausen household,
Silver Spring, MD.
in honor of Ethan Clausen.
Erlandson household, Oak View, CA.
*in honor of Robert Erlandson, and
the boys/men we lost in 2023.*
Macdonald household,
Jacksonville, FL.
Myatt household, Moraga, CA.
in honor of Dylan Myatt.
Nichols household, Santa Paula, CA.
in honor of Scott Richards
Trustmark Foundation,
Lake Forest, IL.
in honor of Carol Nahrwold
Wright household, Springboro, OH.
in honor of Blake Kubicki.

\$500

Abundis household, Plymouth, IA.
Carbonneau household,
Stafford Springs, CT.
in honor of Maryann Watson
Gillespie household, Mason, OH.
Gromoll household, Shoreview, MN.
Guthrie household, Dallas, TX.
Nieberding household,
Cincinnati, OH.
Otani household, Aiea, HI.
in honor of Andrew Otani.
Otero household, Dallas, TX.
Risman household, San Diego, CA.
recurring donations.

Robinson household, Hope, AR.
in honor of Craig Robinson.
Smith household, Cincinnati, OH.
in honor of Blake Kubicki
Taverna household, Chelsea, MA.
*in honor of "Jimmy Hollywood"
James Montenero.*
Waldbaum household, Solon, OH.
in honor of the LSA Board.

\$200

Armes household, Dallas, TX.
in honor of Craig Robinson
Berry household,
Havre de Grace, MD.
in honor of Ryan Weiss.
Buchs-Hammonds household,
Moundsview, MN.
in honor of Sam Gromoll
Byles household, Cincinnati, OH.
in honor of Blake Kubicki
Daringer household, Ames, IA.
Dearth household, Kansas City, MO.
in honor of Austin Dearth
Fleming household, Bensalem, PA.
*In honor of John Fleming, for his
great-grandson Thomas.*
Germon household, Cape May, NJ.
in honor of John Michael Fleming
Getty household, Salem, NY.
Gill household,
West Sacramento, CA.
in honor of Robert Erlandson
Haugen household, Blaine, MN
in honor of Hunter Haugen.
Jerman household, Dix Hills, NY.
in honor of James Jerman.
Laurier household, Brooklyn, NY.
in honor of James Jerman.
Lee household, San Diego, CA.
in honor of Sam O'Brien.
Meynier household, Wayland, MA.
in honor of Amy Nichols.
Samoilenko household, Moscow,
Russia.

\$1000

This donation is made in loving memory of John Fleming of Cape May, New Jersey, by the Cape May County MLS, to the Lowe Syndrome Association.

John Fleming passed away suddenly on October 24, 2023. He is the late great-grandfather of Thomas Jablonski (2, PA). John was a successful real estate broker in Cape May, New Jersey, as well as a beloved husband, father, grandfather, and great-grandfather who will be deeply missed.

John held a special place in his heart for his great-grandson, Thomas, who was diagnosed with Lowe syndrome at 11 months old. Like Thomas, John also wore glasses at a young age, was incredibly bright, enjoyed reading, and became a man of many interests over the years. John often talked about the strength and resilience he saw in his great-grandson despite the many challenges he'd already faced.

Like his great-grandfather, Thomas has come to love Cape May, enjoying seafood by the water, and spending time with his family. His great-grandfather's playful and quick witted humor often entertained Thomas and the two shared genuine joy in each other's silliness. We are blessed that Thomas knew his great-grandfather, and we hope to demonstrate the love, integrity, morals, and strength to Thomas as John did for us.

THANK YOU LSA SUPPORTERS

Schroerlucke household,
Madison, WI.
Snow household, Gilbert, AZ
in honor of Mason Snow.
Stein household,
Port Washington, NY.
Suchy and Lavedan household,
Pinehurst, NC.
Walls household, Louisville, KY.
in honor of Matthew Forrest.
Westover Companies of King,
Prussia, PA.
Wickham household, Austin, TX.
Woodring household, Honolulu, HI.
in honor of Andrew Otani.

DONORS

Abbott household, Buffalo, NY.
in honor of Lincoln Atkins, for his late grandma.
Antoniades household,
Cape May, NJ.
in honor of John Fleming.
Avila Household, Belle Glade, FL.
in honor of Nicholas Avila.
Barrow household,
College Station, TX.
Barry household, Jersey City, NJ.
in honor of Nancy Bramwell Siconolfi.
Bass household, Greensboro, NC,
Kerrville, TX, and Plano, TX.
Benos household, Athens, Greece.
Black Household, Bedford, MA.
in honor of Kang-Suk Andrew Lee.
Broadnax household, Houston, TX.
in honor of Kaye Kirkpatrick.
Burke household, Bayville, NY.
in honor of Matthew Degori.
Colwell household, Raleigh, NC.
in honor of Tyler Richardson.
Dahlof household, Harlan, IA.
in honor of Myron Daringer.
Daugherty household,
San Francisco, CA.
in honor of Kang-Suk Andrew Lee.
De Jauregui household, Napa, CA.
Dentino household, Poway, CA.
in memory of Matthew Forrest.
Dickerson household, Dallas, TX.
in honor of Brooks Kirkpatrick.

Dowden household, Benton, LA.
in honor of Eric Dowden.
Driggers household,
Spring Lake, NC.
Dubrovsky household, Caldwell, NJ.
In memory of Lenny Dubrovsky.
Durant household, Fresno, CA.
Fritz household, Louisville, KY.
in honor of Charles Forrest.
Gann household, San Francisco, CA.
Germon household, Cape May, NJ.
in honor of John Michael Fleming.
Gjelsvik household, Forde, Norway.
Goggans Household, Belle Glade, FL.
Goldstone Household,
Cold Spring, KY.
in honor of Blake Kubicki.
Harris household, Bridgeport, WV.
Hartmann household,
Thousand Palms, CA.
Hays household, Salem, OH.
in honor of Thomas Allen Hays.
Hesse household,
Emmendingen, Germany.
Hrncir household, Dallas, TX.
in honor of Brooks Kirkpatrick.
Humble household, Willowick, OH.
Johnson household, Louisville, KY.
In honor of Charles Forrest.
Johnson household,
Liberty TWP, OH.
in honor of Ryan Weiss.
Johnson household, Sanford, FL.
Jones household, Nicholasville, KY.
Jordan household, Louisville, KY.
in honor of Charles F. Forrest.
Kan household,
Richmond Hill, Canada.
Kan household, Weston, MA.
Kearney household, Palmer, MA.
in honor of Dorothy Miller Gore.
Keough household, Weymouth, MA.
in honor of Roger J. Mondello.
Kieras household, New York, NY.
Kim and Carlile household,
Los Angeles, CA.
in honor of Ewan Carlile.
Kinkade household, Cincinnati, OH.
Kirkpatrick household
Corsicana, TX.
in honor of Brooks Kirkpatrick.

KP Graphic Solutions, Corsicana, TX.
in honor of Kaye Kirkpatrick.
Kubicki household, Cincinnati, OH.
in honor of Blake Kubicki.
Kubicki household,
Winston-Salem, NC.
LaRue Bennett household,
Elizabethtown, KY.
in honor of Charles Forrest.
Lava Household,
West Orange, NJ.
in honor of Ray Siconolfi's 70th Birthday.
Lebherz household, Frederick, MD.
in honor of Corbin Berry.
Lewis household, Louisville, KY.
in honor of Charles Forrest.
Lord household, Groveland, MA.
in honor of Steven Lord.
Lukens household, Cartersville, GA.
Mathews household, Cincinnati, OH.
in honor of Blake Kubicki.
McLemore household,
Fredericksburg, VA.
Mercier and Johnston household,
Levis Quebec, Canada.
Miller household, Iowa City, IA.
Mondello household, Braintree, MA.
in honor of Roger J. Mondello.
Mondella Household, Wayland, MA.
Morris household, Forest, VA.
in honor of Timmy Morris.
Morton household, Adrian, MI.
in honor of Michael Echelbarger.
Moskal household, Groveland, MA.
in honor of Steven Lord.
MyDoGood, LLC, New York, NY.
Nelson household, Binghamton, NY.
in honor of Daniel Basel.
Network For Good
O'Boyle household, New York, NY.
in honor of James Jerman.
O'Brien, III household,
Palm Beach Gardens, FL.
O'Donnell Household, Cape May, NJ.
in honor of John Fleming.
Orlando household, Dracut, MA.
Otani household of Waipahu, HI.
in memory of Matthew Forrest.
Padebettu household, Jersey City, NJ.

THANK YOU LSA SUPPORTERS



Pedicini household, Louisville, KY.

*in honor of Larry and
Mary Ann Baird.*

Peterson household.

Pharr household, Gretna, LA.

in honor of Will Bass.

Poore household, Adrian, MI.

in honor of Michael Echelbarger.

Priest household, Belle Glade, FL.

in honor of Nicholas Avila.

Redfield-Sears household,

Walnut Creek, CA.

in honor of Daniel Davis.

Reinwasser household,

Fountain Hills, AZ.

in honor of Colin Tietz.

Richards household, Rio Vista, CA.

in honor of Scott Richards.

Rimshaw household, Freeland, PA.

*in honor of Patrick Dinofrio and
Matthew Calvey.*

Rinehart household,

Boca Raton, FL.

*in honor of all the grandparents
supporting their families.*

Rockwell household, Adrian, MI.

in honor of Michael Echelbarger.

Ruffini household, East Windsor, NJ.

in honor of Jonathan Carman.

Rush Moore LLP, Waipahu, HI.

in honor of Andrew Otani.

Samaniego household, Orange, CA.

Seitz household, Cincinnati, OH.

in honor of Blake Kubicki.

Siconolfi household, Clark, NJ.

in honor of Casey Risola.

Sieg household, Louisville, KY.

In honor of Matthew Forrest.

Snow household, St. Petersburg, FL.

Sprenger household, Peoria, IL.

in honor of Terri Broms.

Springer household, Waukegan, IA.

Stein household,

Port Washington, NY.

in honor of Alec Everitt.

Stemp household,

Houghton le Spring, UK.

Stewart household, Louisville, KY.

in honor of Joshua Stewart.

Stroshine household,

Wilbraham, MA.

in honor of in honor of Dorothy L.

Gore's family- Kimberly & Jean

Paul Carbonneau.

The Trustmark Foundation,

Lake Forest, IL.

in honor of Carol Nahrwold.

Westover Companies,

King of Prussia, PA.

in honor of Thomas Jablonski.

Thompson household,

Smiths Grove, KY.

in honor of Charles Forrest.

Tietz household, Kewanee, IL,

in honor of Colin Tietz.

Tietz household, Chaska, MN.

Townsend household,

Max Meadows, VA.

in honor of Blake Henry Kubicki.

Toymil household, Patterson, NY.

in honor of James Paul Toymil.

Unmack household, Junortown, VIC.

in honor of Dylan Myatt.

VanderGoot household, Sussex, NJ.

Velooso household, Portsmouth, RI.

in honor of KitKat.

Ward household, Louisville, KY.

in honor of Matthew Forrest and

Charles Forrest.

Watson household, Westfield, MA.

in honor of Dorothy L. Gore's

family- Kimberly & Jean Paul

Carbonneau.

Whiley household, England.

Wientge household, Lebanon, OH.

in honor of Matthew Forrest.

Wilkins household, Hampton, VA.

in honor of Matthew Forrest.

Wollmar household,

Georgetown, ME.

in honor of Ethan and Mark

Mondello.

Wondoloski household,

Claremont, CA.

in honor of Carson Newton.

BUSINESS DONORS

Amazon Smile

KP Graphic Solutions

MyDoGood, LLC.

Network For Good, Matching

Rush Moore LLP.

Note: Donations listed in this issue were those received as of December 31, 2023. Donations received after that date will be included in the next On the Beam.