

# REACHING OUT

FALL 2022

Spotlight:  
**Behavioral Health  
Concerns in Individuals  
with CdLS**

**ON THE COVER**

*Sam Luce*



**CdLS Foundation**

Cornelia de Lange Syndrome Foundation, Inc.

# Director's Message

Welcome to the fall issue of *Reaching Out*. The days are getting cooler, and the nights are getting longer. Soon we will be spending more time indoors, snuggled close to the ones we love. This time of the year is my favorite – the leaves are turning into vibrant hues of orange, yellow and red, and pumpkins line the walkways up and down the street. It reminds me that with every new season, the CdLS Foundation continues to grow and change with the needs of the great community we care for.

Like the seasons your needs are ever changing – but the mission of the Foundation remains the same – to support you wherever you are in your CdLS journey.

You will discover the strength of a grieving mother. The power of a mom who wants to better care for her son who has selective mutism. You'll read how to identify and address behavioral concerns and learn about psychiatric medications.

Read about all the exciting events that our amazing community members have and are planning to host. Learn ways you can be more involved in our fundraising activities.

There is power in our community. This season we can continue to look inward while we collectively enjoy the colors nature is creating.

Together we can be CdLS strong.

*Bonnie*



Bonnie Royster, CdLS Foundation  
Executive Director

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## SYMPOSIUM UPDATE



Antonie D. Kline, M.D., CdLS  
Foundation Medical Director

We had the tenth Scientific Symposium, virtually, on CdLS and cohesin.

The first group addressed various aspects of behavior in CdLS. Dr. Beatriz Allegri (Italy) discussed differences in behavior based on the type of gene mutation in *NIPBL*. She recommended augmentative and alternative communication intervention and cognitive behavioral therapy. Dr.

Katherine Ellis (UK) spoke about how individuals with CdLS were more social than individuals with other conditions. Jessica Mingins (UK) discussed triggers for individuals with CdLS and how these can lead to anxiety. Dr. Kayla Smith (UK) defined intolerance of uncertainty, a relatively new term in behavioral psychology, and how this is found frequently in individuals with CdLS who desire predictability.

My session discussed COVID-19 and its impact on the CdLS community. Of 41 known individuals with CdLS who had COVID-19, nearly 10% needed some ventilation assistance, and 5% died of a cause related to the infection. There was a 27% increase in Ask the Expert questions during the pandemic, behavior and GI topics were more frequent. Dr. Carol Li (USA) described a study related to sleep, funded by the CdLS Foundation, which found an increased risk for moderate to severe obstructive sleep apnea and 66% with sleep-disordered breathing. Also, 16% of teens with CdLS have daytime sleepiness which is less than the general population, if they are sleepy, it is in the morning. Dr. Gholson Lyon (USA) described new aspects of KBG syndrome due to mutations in the *ANKRD11* gene, which can cause CdLS spectrum from different mutations.

The second day started with talks about embryonic aspects of CdLS or cohesin effects. Dr. Natalie Blagowidow (USA) described several cases of individuals with CdLS who have developed embryonic or germ cell tumors, some from genetic literature, and how these could be related to the disruption of cohesin. Dr. Wei-Ting Yueh (USA) spoke on cohesin in early embryonic development, and how the *SMC3* gene helps maintain genomic stability and embryo formation. Stephenson Chea (USA) reviewed his ongoing work on the origin of birth defects in CdLS, particularly cardiac. Dr. Antonio Musio (Italy) discussed genome instability of CdLS cells.

The next sessions discussed CdLS and cohesin complex gene testing. Sara Raible (USA) summarized 10 years of the multidisciplinary CdLS clinic held at Children's Hospital of Philadelphia. Anna Platt (USA) gave an overview of molecular genomic testing results given to individuals with CdLS who had initially tested negative, often years after their sample was obtained. Justin Blair (USA) further described genome testing results and gene identification, of which 15% had variants in non-CdLS genes. Maninder Kaur (USA) described a related gene to the immediate cohesin genes, *NAALADL2*. Dr. Philip Boone (USA) spoke about the *WAPL* gene and its overlap with CdLS clinically.

Finally, a panel of experts addressed future treatments and how researchers and clinicians collaborate. The experts from the USA included: Dr. Ian Krantz; Sarah Raible; Dr. Arthur Lander; Dr. David Litwak; and Dr. Rich Haaland. The group plans on forming a coalition of working with scientists, families, and clinicians to discuss gene testing results, build a registry, and interact with the FDA. The symposium was exciting and informative - a success!

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# Identifying & Addressing Behavioral Health Concerns in Individuals with CdLS

*Julia O'Connor, Ph.D., Psychologist, Kennedy Krieger Institute and CdLS Foundation Professional Development Committee (PDC) Member*

Individuals with CdLS experience significant challenges including behavioral health concerns, such as anxiety, depression, and behavior issues. Anxiety has been estimated to occur in 10-64% of individuals with CdLS and includes separation anxiety, social avoidance as well as selective mutism. Depression has been reported in 11-50% and presents as low mood, decreased interests or excessive irritability, especially in adolescence and early adulthood (Groves, Oliver, & Moss, 2021).

These underlying mental health diagnoses may manifest as overt behaviors in CdLS. For example, **self-injurious behavior (SIB)** may be how they expresses anxiety or depression. Alternatively, these overt behaviors may be a means to communicate. The prevalence rate for significant SIB has been reported to be 55.6% in people with CdLS (Oliver, Sloneem, Hall, & Arron, 2009). Additionally, 32% of individuals exhibited physical aggression and 41% engaged in property destruction.

Given these statistics, it is important to monitor mental health and problem behaviors. To address these concerns, first rule out medical/physiological problems. Additionally, address head directed SIB and seek medical evaluation as there could be internal injuries. Protective equipment such as helmets, arm guards, and protective clothing are options to prevent tissue damage.

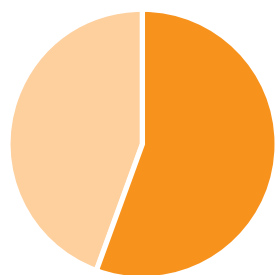
For ongoing behavioral issues, a referral to a behavior psychologist/analyst would be appropriate. Using the techniques of applied behavior analysis, the therapist conducts a functional behavioral assessment. Function is determined by evaluating the contexts in which the behaviors occur and gathering information about antecedents, behaviors, and consequences.

Once function is identified, a behavior plan is developed to reduce the problem behavior while increasing appropriate skills. Behavior plans may include treatment components such as: structured schedule, leisure activities, as well as teaching communication, adaptive skills, and coping strategies. All caregivers should be trained in the behavior plan, so the plan can be implemented consistently and to monitor effectiveness. The behavior plan may also include planned ignoring, redirection, and blocking. If indicated, it is recommended that you consult a psychiatrist/developmental pediatrician with experience in CdLS.

The COVID-19 pandemic greatly affected everyone's mental health and behavior, but further increased feelings of physical and mental exhaustion, isolation, stress, and disconnect from the community for families of individuals with CdLS. Access to community resources were greatly limited as evidenced by lack of in-person services and supports as well as increased staff turnover.

Finally, practicing self-care can improve caregiver physical, mental, social, and emotional well-being. Without their own mental well-being, caregivers will not be able to address the well-being of their child. Key areas are getting adequate sleep, eating healthy, getting exercise, relaxing, seeking social supports. It is important to identify which self-care strategies work for you and to make a point of doing them as often as possible. Start by building one new habit at a time and explore other activities to expand your well-being toolkit.

**Overall, it is important to not only address the behavioral and mental health concerns in individuals with CdLS, but also remember to focus on personal well-being for the caregivers.**



**55.6%** of people with CdLS have significant SIB

# Self-Care for Parents of Children with Disabilities

*Adapted from: Navigate Life Texas*

Parenting a child with a disability or special health care needs is rewarding, fills us with love, and can also take its toll. When we are constantly in the role of caregiver, it is easy to neglect ourselves. The best way we can help our children is to take care of ourselves.

We all face challenges with time, money, and how comfortable we feel being away from our children. But the bottom line is that parenting a child with a disability or special health care needs is challenging and taking care of ourselves is crucial.



Here are some ideas that we hope help you and your family.

## Why Taking Care of Ourselves Is So Important

- By taking care of ourselves first (hard to do but super important), we will be better able to take care of our family.
- It helps to recharge our batteries every day, even for just 5 minutes, so we don't feel as tired and run down.

## Daily Self-Care Tips

Be gentle with yourself. If self-care is new to you, take baby steps. Don't feel like you have to do everything suggested on this list. See if any of the suggestions below are helpful—if not, create your own.

- Try to get enough sleep each night, drink lots of water, and eat healthy foods that nourish your body.
- Work movement or exercise daily, even if you can only do 5-10 minutes of dancing in the kitchen with your kids.
- Schedule and keep your doctor's appointments to help you stay physically healthy.
- Find time each day for a few minutes of quiet time to write in a journal, read, or listen to music.
- Deep breathing can go a long way in helping us relax and feel more centered. Breathe deeply through your nose while feeling your diaphragm and belly expand, hold your breath for a few seconds, and exhale through your nose.
- Go outside. Nature has a way of calming and centering us. Go for a walk, run, or hike through a park or your neighborhood. Kids love being outside, too, so this is a good one to do even when your kids are with you.
- Spend time with your spouse or partner, even if it's challenging to go on regular dates. Set time each day to reconnect, even if you have time for coffee together in the morning for 5-10 minutes.
- Take people up on their offers of help or look into respite care, so you can have time to do other things like read a book or spend time with your other children.
- Join friends for coffee, a monthly book club, or a moms' / dads' night out. Build your support community, especially if you are a single parent. Start with a few activities a month, and then add more as you get comfortable and find more time.
- Nurture your emotional and spiritual needs. Some parents find talking with a therapist or clergy member helpful. Others practice yoga, attend a spiritual community, journal, paint, or read poetry. Do what works for you and helps you.

# Maladaptive Behaviors and Medication Management for CdLS

Marco Grados, M.D., MPH, Professor of Psychiatry & Behavioral Sciences, Johns Hopkins University School of Medicine, and Member of the CdLS Foundation Clinical Advisory Committee (CAB)

Pharmacological treatments of maladaptive behaviors and emotional dysregulation in CdLS requires careful consideration of symptoms, medication choice, and safety profiles. Medications for individuals with intellectual disability (ID) and autism spectrum disorder (ASD) are helpful to individuals with CdLS. However, additional considerations are warranted; empirical evidence strongly suggests that individuals with CdLS may also have unusual reactions to psychotropic medications. We review below current indications for psychotropics in ID, ASD, and individuals with CdLS.

Certain psychiatric conditions target emotional and behavioral challenges and are treated with psychotropic medication. Before any medication approach, there is a thorough review of medical conditions (toothache, ear infections, reflux). A professional should also conduct a functional analysis of the target behaviors to understand if the maladaptive behaviors have a practical component amenable to behavioral therapy interventions. If these assessments happen and symptoms persist, consideration for psychotropic use is an option. Four general categories of behavioral and emotional challenges can guide medication choice in one scheme. The four broad categories of psychopathology are 1) hyperactive and hyperarousal states, including attention-deficit hyperactivity disorder (ADHD) and stress-related disorders (post-traumatic stress disorder (PTSD)); 2) depressive and anxiety states, including major depression, anxiety disorders, and obsessive-compulsive disorder (OCD); 3) major affective disorders such as bipolar disorder; 4) psychotic disorders or aggressive/disorganized behaviors. These four categories lend themselves to a 1-1 mapping of psychotropic classes and a similar analogy for side effects, from mild to severe.

**1 DISORDERS OF HYPERACTIVITY AND HYPERAROUSAL:** stimulant medications are used to treat attentional challenges and hypermobility (ADHD),  $\alpha_2$  and alpha-agonists can be used for either ADHD or hyperarousal states (PTSD). These include stimulants methylphenidate (MPH), dextro-amphetamines (d-AMP), and alpha agonists (clonidine, guanfacine).

*Side effects:* stimulants are simple to manage, with precise dose titration (0.3 mg/kg MPH per dose; twice daily; half the amount for d-AMP). When dosed appropriately, stimulants' side effect profile is overall benign, mainly decreased appetite; others are possible (anxiety, repetitive behaviors), but they subside with a decrease in dose or stopping the stimulant. Alpha agonists are blood pressure medications that decrease sympathetic system activity in the body, the system responsible for higher blood pressure and increased heart rate (stress reaction). The calming effect of alpha agonists decreases hyperactivity and overarousal-which can result in hypermobility. Side effects are benign, with lower blood pressure requiring monitoring and fully reversible on lowering the dose or discontinuation.



**2 DISORDERS OF ANXIETY AND DEPRESSIVE STATES:** selective serotonin reuptake inhibitors (SSRIs), as well as other types of anti-anxiety and anti-depressant formulations for common symptoms of generalized anxiety, selective mutism, and social anxiety.

*Side effects:* SSRIs can cause “behavioral activation” (agitation, insomnia, irritability) if started at too high a dose or increased too quickly, “start low and go slow” is a good rule of thumb for this and other categories of psychotropics. Other side effects, such as mild headaches and GI distress, are less common but should be watched for.

**3 DISORDERS OF SIGNIFICANT AFFECTIVE DYSREGULATION:** mood-stabilizers can be used for the management of severe “mood swings”, cyclical mood conditions (bipolar disorder), and chronic irritability that is severe and impairing. These include lithium, valproic acid, lamotrigine, carbamazepine, oxcarbazepine among others.



*Side effects:* most mood stabilizers require blood level monitoring and, in rare instances, can cause end-organ damage, especially in chronic use. Chronic use of lithium can cause kidney malfunction, valproic acid liver damage, carbamazepine low blood counts (oxcarbazepine, similar to carbamazepine, is an exception). When used judiciously, mood stabilizers can effectively treat very severe mood states, such as bipolar mania.

**4 DISORDERS OF PSYCHOTIC PHENOMENA AND SEVERE BEHAVIORAL AGGRESSION/ DYSREGULATION:** neuroleptics are used in instances where psychosis (hallucinations, delusions, disorganized and bizarre behaviors) is the predominant presentation. Controlled clinical trials also support the use of neuroleptics for aggressive behaviors in autism. Classes of neuroleptics include older “first-generation antipsychotics” haloperidol (high potency), chlorpromazine (low strength), and “second-generation antipsychotics” (SGAs), risperidone, olanzapine, quetiapine, aripiprazole, and ziprasidone.

*Side effects:* first-generation antipsychotics, especially the high potency class (haloperidol), can cause movement disorders known as extrapyramidal symptoms. The extrapyramidal system controls the ease of movements, including muscle tone, so that extrapyramidal symptom side effects due to first-generation neuroleptics include rigidity and shuffling gait (like in Parkinson’s disease), as well as dystonias (sudden, stiffening of muscle groups). On the other hand, while SGAs do not easily cause extrapyramidal symptoms (although they can occur), SGAs can cause metabolic syndrome, including weight gain, insulin resistance (pre-diabetic states), and increased appetite. Baseline and routine periodic monitoring of blood glucose, lipid profiles, hemoglobin A1c (a biomarker of diabetes), and weight are required when using SGAs.

In summary, a careful characterization of psychopathology in children, adolescents, and adults with CdLS is required, after a medical exam and behavioral assessment, if psychotropics are being considered. Classifying emotional and behavioral challenges into the four categories of psychopathology can streamline understanding of the disorders and allow for a tiered safety approach to psychotropic medication.

**Disclosure:** non-FDA-approved use of psychotropics is in the article.

# HIGHLIGHTED EVENTS

## PENNSYLVANIA BACKYARD FUNDRAISER BREAKS \$50,000

The Feehan family in Pennsylvania has done it again! Led by CdLS Foundation Board Vice President, Mike Feehan, the family raised more than \$50,000 for the CdLS Foundation with their third annual Feehan Backyard Obstacle Course. The event honored Mike's 12-year-old son Connor, who has CdLS.



With a starting goal of \$20,000, Mike began reaching out to family, friends, and colleagues in early May requesting that they support his event, which was held in July. The support he received was incredible.

We continue to hold up the Feehan Backyard Obstacle Course as the gold standard for running a simple, inexpensive, and successful fundraising event.

According to Mike it's all about creating an event with low overhead, and most importantly being consistent with outreach to your supporters, including thanking them and sharing event photos after the event is over.

Looking for more pointers to create your own fundraising event for the CdLS Foundation? Contact us at [events@cdlsusa.org](mailto:events@cdlsusa.org).

## NEW ENGLAND KIDS FOR KIDS DANCE TO RAISE FUNDS FOR INDIVIDUALS LIVING WITH CdLS



Young dancers competed in the Kids for Kids, Dancing for Life Performing Arts Contest in North Haven, CT, last spring. The event brought in \$5,000 to help support services offered by the CdLS Foundation.

Kids for Kids, Dancing for Life, Inc. is a nonprofit 501(c)3 corporation dedicated to raising funds for children in medical or financial need. Proceeds from contest registration fees are given to local charities.

The CdLS Foundation is very grateful to have been chosen among the charities to benefit from the kindness of Kids for Kids.

## NEW ENGLAND GOLF FUNDRAISER SELLS OUT

For the first time ever in its 29-year history, the New England Golf Classic to benefit the CdLS Foundation was a sold-out event! Golfers from across the region converged on the Ipswich Country Club in MA, in May, raising more than \$60,000 to help individuals with CdLS and their families.



During dinner, golfers heard heartfelt messages from fellow golfers, Frank Mairano, the Foundation's co-founder, and Board member Eric Johnson, both of whom had daughters born with CdLS. Golf Committee Chair Pat Lyons also shared the difficulties of raising a son with CdLS.

Showing that they understood that the event was about more than golf, the golfers gave generously by spending money on silent auction items and raffle prizes to help ensure that the Foundation can continue its mission of serving those with CdLS and their families.

A very special thanks goes out to major sponsors and supporters: **Code 42, The Teixeira Family, GreenPages, Matter Communications, Envision Technology Advisors and Workers Credit Union.**

## GEORGIA HIGH SCHOOL SENIORS HOST 5K FUNDRAISER



The One Love One Heart 5k hosted by high school seniors Hannah Pomfret and Kira Czech of Decatur, GA, was a fantastic endeavor that brought both awareness and funding to help individuals with CdLS.

The teens, runners and supporters raised over \$10,000 for their August event. Two other families with children with CdLS traveled to join in the race as well!

This 5k, which had been hosted by Hannah's parents years ago in honor of their older daughter Maya, who has CdLS, was taken over by Hannah and Kira last year for their school community service project. This year they decided once again to continue to carry on the One Love One Heart 5K legacy, and we are very grateful that they did!

## NEW YORK ANTIQUE CAR ENTHUSIASTS HELP INDIVIDUALS WITH CdLS

They have a love of antique cars and a love for those who live with CdLS! The Western New York Oldsmobile/GM Club presented the CdLS Foundation with a check for \$1,200 in July.



This is all thanks to club member Sue Bolger, who has a special relationship with someone with CdLS - her darling niece Aubrey. It is because of that sweet relationship that Sue brought the information about CdLS and the CdLS Foundation before the club members, who agreed to make the donation.

As part of the enjoyment of their antique car hobby, the club members host car shows. With the support of the Western New York car hobby community and local businesses they raise funds to donate to organizations like ours. Thank you, WNY Oldsmobile/GM Club, Sue, and Aubrey!

# CALENDAR 2022

### October 9

Bank of America  
Chicago Marathon  
Chicago, IL

### October 10

Amber Gaines Memorial  
Golf Classic  
Sunset Hills, MO

### October 15

Baltimore Running Festival  
Baltimore, MD

### October 23

Walk for Will & Hope  
Shelby Township, MI

### November 5

Multidisciplinary Clinic for  
Adolescents and Adults  
Baltimore, MD

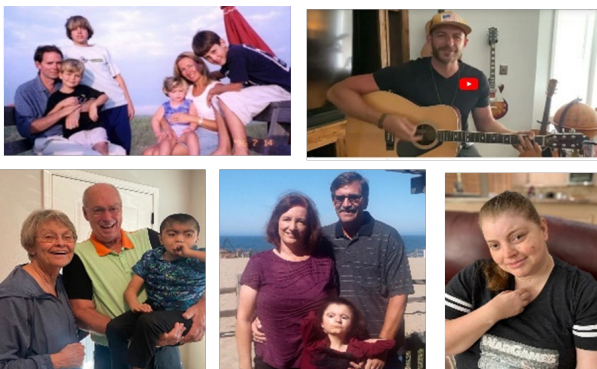
### November 29

Giving Tuesday  
Virtual

## We Couldn't Do It Without You

In June, families, clinicians, and medical professionals came together for a powerful week of information and sharing during the 2022 National Virtual Family Conference. The cost to create and host this event for you, our amazing CdLS community, exceeded \$44,000.

We are incredibly grateful to our corporate and family sponsors, who helped to cover much of the costs. Because of their wonderful generosity we were able to offer this very special conference, which helped our attendees in so many ways.



**We extend our thanks to these wonderful Major Sponsors:**

### **The Kerr-Jackson Family**

Hero Level – Maddy's Virtual Ice Cream Social Sponsor

### **Dianne & Rich Haaland**

Champion Level – Sibling Support Sponsor

### **The Prada Family**

Friend Level – Scholarship Sponsor

### **The Family of Katie Luyt**

Friend Sponsor

### **Angie & Tim Young**

Friend Sponsor

### **Pat Pollifrone**

Entertainment Sponsor

### **Corporate Sponsors**



## Missouri Truck Stop Presented with Fundraising Plaque

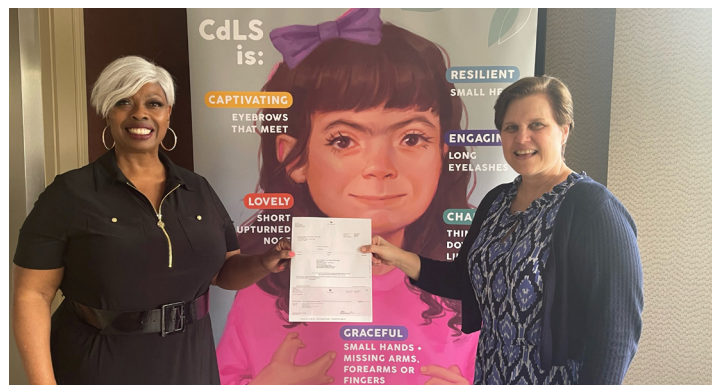
James Robbins, owner of the Cherokee Pass Mobil truck stop in Missouri was presented with a plaque by Doug Gaines, father to the late Amber Gaines and VP of Madison County Wood Products (MCWP), in appreciation of five years of fundraising for individuals with CdLS.



James and his customers have raised about \$4,300 for the CdLS Foundation by dropping coins in a special Hope for Change jar at the cash register counter since 2017.

Cherokee Pass Mobil is also a sponsor of the Amber Gaines Memorial Golf Classic each year in October.

## Connecticut Organization Makes Donation



CdLS Foundation Board Member, Tricia Wise (right), presented a check for \$4,000 to CdLS Foundation Executive Director Bonnie Royster (left) from the Crane Fund for Widows and Children in August.

Tricia, Vice President of Talent Development for Crane Co., recommended the CdLS Foundation as one of the recipients of the company's annual Charitable Giving Campaign.

Having joined the CdLS Foundation Board of Directors in 2021, Tricia was quick to embrace her company's -spirit of giving, and eagerly suggested that the CdLS Foundation be one of their grant beneficiaries.

"One of the attractions to me for working at Crane is their philanthropic nature," Tricia explained. "I also get a lot of satisfaction out of volunteering for a worthwhile organization like the CdLS Foundation. So, bringing the two together with this grant is very rewarding to me."

This is the second year that Crane Fund for Widows and Children has donated to the CdLS Foundation thanks to Tricia.

## East Coast Company Gives Back



Not only do they sell tires, but Town Fair Tire also gives back to their community. The Town Fair Tire Foundation has chosen to award the CdLS Foundation a \$2,000 grant as part of their Charitable Giving Campaign.

Town Fair Tire Foundation is a private, not-for-profit philanthropic organization established by Town Fair Tire owner Neil Mellen in 2000. Their mission is to financially support organizations that help people in need of social, health, welfare, educational and other human services throughout New England and beyond.

The organization reached out to the CdLS Foundation based on our mission to serve individuals with CdLS and their families, and on our excellent Platinum Guidestar rating.

## New Mexico Dad Raises Money from Southwest Airlines Pilots

Eric Roberts of New Mexico and father to sweet Penelope, was instrumental in convincing his classmates in pilot training for Southwest Airlines to donate a total of \$737 to the CdLS Foundation in honor of his precious little one, who has CdLS. The total amount '737' is in reference to the

Boeing 737 aircraft flown by Southwest.

"Southwest Airlines has an amazing and rich tradition of its new hire class donating \$737 to a charity of their choice," Eric explained. "My family and I reached out to the CdLS Foundation in 2021 for help when we were struggling to understand

CdLS and looking for answers regarding our amazing daughter, Penelope."

It was only fitting that Eric suggest the CdLS Foundation as the charity of choice to his classmates. "My class had such big hearts that we chose to donate \$737.11 to three different charities! One of them being the CdLS Foundation! (Eleven cents because we are the 11th new hire pilot class of 2022), he added."

## Gearing Up for Giving Tuesday – November 29th

November 29th will mark the 10th anniversary of Giving Tuesday – a movement that was started in 2012 to "unleash the power of radical generosity around the world".

Described as a Global Day of Giving, it is held each year on the Tuesday following Thanksgiving, with a call to action for people to give to their favorite charities.

The CdLS Foundation is so grateful to all of you who have chosen us as your charity of choice on this annual day of giving. Thanks to your generosity, last year the Foundation received approximately \$15,000 in donations. Those who gave not only helped to ensure that we ended the year on budget, but they also donated in time to receive a tax credit for 2021.

# GIVING TUESDAY

Our goal this year is to hit last year's total of \$15,000. Help us hit that goal and put Giving Tuesday – November 29th, on your calendar. Spread the word to encourage your family and friends to donate to the CdLS Foundation Giving Tuesday page using this link: [bit.ly/GTCdLS2022](https://bit.ly/GTCdLS2022).

Posting photos and sharing stories about your loved one with CdLS on your social media sites will help to raise funds for the CdLS Foundation. It will also help to spread awareness about this rare syndrome – both are equally important for the future of the organization.

Thank you for your support, and for choosing to make a difference in the lives of all the precious ones served by the CdLS Foundation.



# Grieving the Loss of My Child

*Madeline Pond*

Grief is not something anyone “wants” to do, but it hits us all no matter what. It is sharp, quick, and unexpected. It blindsides you and takes your breath away. My son Odin was diagnosed prenatally with Cornelia de Lange Syndrome, he was my little firework. He came into the earth bright enough to light up a sky and loud enough to make everyone listen, he taught me so much, he taught so many people so much.

The day Odie was born a part of me was born; I was no longer a mom who was terrified of his diagnosis. I was a fierce advocate for each and every one of his needs, I was his voice. I was prepared to do this forever. The day Odin died a piece of me died as well, I was not only grieving him in that moment, but I was also grieving his first words, his first steps, him trying food for the first time and so much more.



There were some days I would lay and look at the ceiling completely convinced that the pain I was feeling would kill me, and sometimes I hoped it would, wherever Odie was, I wanted to be. The pain was swallowing me whole, it was so much that it would numb me at times. I felt so angry at the world, (I still am sometimes), that it would give me the biggest blessing and steal it from me, that it let his body fail him.

I knew I needed to seek help. I reached out to multiple local mental health resources and spent many nights on the phone with crisis hotlines, I had to find my reason again. There is a huge shortage of resources to help with people in a mental health crisis, and I swore if I heard one more person tell me “I understand why you’re feeling that way.” I was going to lose it! How could they possibly understand what it felt like to lose him? You must let it hurt to heal. You must be angry to heal. There is no therapy, medicine or coping skill that will lessen the blow of losing a part of you, of losing a life that you grew for 9 months.



**Grief is not something anyone *wants* to do, but it hits us all no matter what.**

Over time however, therapy can give you tools to put these feelings into little folders in your brain and find your purpose again. Over time with therapy (It’s still only been 6 months) I’ve found Odie still is my purpose. I wake up every day, and feel the sun hit my skin, for him. I enjoy new foods, for him. I go for walks, for him. I’m trying to live the way he wanted to, he fought so hard to be here. I’ve joined many Facebook groups for grieving parents, I’ve found other parents who’ve lost children understand it better than anyone else. I share my journey on social media. I talk about everything I feel, unapologetically.

***Talk about your child, keep them alive in memory, but most of all let it hurt.***

We are here to help guide you on the complicated journey after losing your loved one. Contact our Family Service Coordinators at 800.753.2357 or [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org).

# Asmita and Kavin Mistry

Kavin loves music. He sings in multiple languages and multiple genres. Music motivates, energizes, calms, and soothes him. He is my role model.

Kavin was evaluated multiple times without clear answers. At age 8, a geneticist felt Kavin had features of CdLS. We took him to Children's Hospital of Philadelphia (CHOP). Doctors examined him. His blood work did not match any of the gene markers known for CdLS at that time.

The doctors promised they would save his blood samples in case more markers were discovered in the future, Kavin was diagnosed at age 15 at the CdLS Clinic in Baltimore based on physical characteristics. Years after that, we received a call from CHOP. Kavin's saved blood matched the *SMC1A* marker.

Working full-time while raising a family, the years flew by. But, in my mind I always knew if I learned more, I could help more. The pandemic made that happen.

The transition from in-person to virtual education taught me at-school Kavin was **completely** different than at-home Kavin. At home Kavin was funny and chatty and never stopped asking questions. At school he was quiet, frozen, a deer-in-the-headlights. When encouraged to speak, he opened his mouth, and no voice came out. His body was visibly tense.

This paralyzed child was at-school Kavin. School staff knew nothing of at-home Kavin. We were shocked! We learned Kavin suffers from selective mutism and social anxiety. Selective mutism is an anxiety-based disorder debilitating disorder. We missed it for years. I was appalled. I am a speech-language pathologist and had not noticed it. Where did the communication breakdown between home and school occur?

In 2020 I began my Clinical doctorate in Speech Language Pathology. I conducted a study on selective mutism in people with CdLS, what it looks like and how to diagnose and treat this devastating disorder. Other communication challenges often present in individuals with CdLS can make it harder to diagnose selective mutism. My research led me to other parents who had not realized their child was suffering from selective mutism until participating in my study.



CdLS is like a box of 1,000 puzzle pieces. You can put the pieces together, but the picture is still incomplete. Despite all the research and knowledge, we are still missing pieces of the puzzle. With puzzle pieces missing, we have a life of trial and error. We need more research. We need more puzzle pieces.

If it takes a village to raise a child, our children need a city. That city can include siblings, family, friends, co-workers, and community members. As Kavin's needs continue to change, our city helps us take one step at a time.

Our family has received unconditional support, understanding and guidance over the years from the CdLS Foundation. As a parent and as a professional, I am grateful to have met professionals and families through the Foundation. You are now an integral part of our city.

I want to collaborate with the CdLS Foundation to spread awareness about selective mutism. I want to help other CdLS families get their children assessed and diagnosed earlier and receive timely help. We are truly blessed to be part of a world full of angels living beside us.



***Asmita's full story doesn't end here. To read its entirety, scan the QR code.***



*Tim Luce and Miguel Avila*

### **Tim's Story ...**

Sam, my son, was born on October 19, 1987. When Sam was young, I called and followed all the children I could find that were mildly affected. I wanted hope and was trying to see what our life would be like with Sam. We were Regional Coordinators for Washington and held gatherings at our home. We have attended many conventions in the past.

I would like to offer that hope to some parents going forward. Sam lives in a group home and works at Microsoft at the main campus as a supported employee. He can take the bus by himself and has flown to California alone. He can't make change for a dollar but can use his debit card.



He swims Special Olympics and regularly makes State, He has no medical issues but does wear hearing aids. He loves to go or do anything. We bowl every week, and I must try with all my might to have a win, He likes cornhole and Yahtzee (if we help him count)!

Sam is very quiet and doesn't talk much. When he does, he can carry a conversation. He comes home every Sunday. Sam is number two of four siblings and is loved by them.

### **Miguel's Story ...**

I am Sam's Manager at Café 34; we are delighted to have him working with us. He is so kind and well mannered. All our guests and team members love Sam!

Sam is a busser for our café at Microsoft and his duties include wiping down tables and chairs, stocking napkins and folding pizza boxes. He is a great team player, happy to help with any task asked of him. Sam is very dependable and reliable. He has impeccable timeclock skills - clocking in and out at precisely the same time every day. We monitor all time clock errors and Sam has the best record of all our associates!

Sam continues to grow socially. I love to hear his stories about his swim meets and going to Dick's Drive-In on the weekends. I could go on and talking about Sam and how much we enjoy having him on our team.

## Helpful Resource to New and Veteran Families

*We are the CdLS Foundation Family Service Coordinators*

*Linda Pierce, M.S.W., Deirdre Summa, L.M.S.W., Whitney Rinaldi, B.S.W.*

As the Family Service Coordinators, we work you through all life's milestones.

With a compassionate ear, we can help you:

- Better understand CdLS
- Help your doctors better understand CdLS
- Get answers to questions about CdLS we cannot answer! We can contact experts on your behalf.
- Find providers in your area
- And so much more!

To reach the CdLS Foundation's Family Service Coordinators, please call 1.800.753.2357 (press one for Family Services) Monday – Friday from 8:30 am to 5:00 pm ET. You can also email at [familyserVICESTeam@cdlsusa.org](mailto:familyserVICESTeam@cdlsusa.org).

## WELCOME New Families



### THE MARKLE FAMILY

Noelle is an energetic 2.5-year-old who loves giraffes, dancing, books, building with Mega Blocks, and rocking her favorite sunglasses. She received her CdLS clinical diagnosis in January 2022 and we look forward to meeting more CdLS families! We thank God for blessing us with Noelle and are grateful for the support of our loving family and friends.

#### California

Nicole and Dalton Ambrose and daughter Blair born March 13, 2021

#### California

Theresa Ray and daughter Rylie born December 23, 2021

#### Florida

Catherine and Oscar Ortiz and daughter Isabella born July 2, 2016

#### Illinois

LaTonya and Steve Parham and daughter Zuri born June 18, 2021

#### Iowa

Liz Gansen and daughter Violet born October 6, 2021

#### Louisiana

Shelby Medines and Tyler Mellenthin and daughter Sophia, born November 17, 2021

#### Michigan

Leah and Brian Markle and daughter Noelle born January 21, 2020

#### Minnesota

Collen and Brian Heimann and daughter Audrey born October 14, 2017

#### Mississippi

Brie and Brandyn Smith and daughter Zoe born November 15, 2018

#### Missouri

Erin and Ryan Gifford and son Tate born October 4, 2021

#### North Carolina

Jayne and Vladimir Radionov and son Kaleb born July 11, 2008

#### Texas

Rachel and Patrick McCullough and Son Colton born June 14, 2021

#### Washington

Cherlyn and Tyler Emry and daughter Aubrielle born November 4, 2021

# The Sibling Support Project

*Emily Holl, Director Sibling Support Project Connect with Emily: [emilyholl@siblingsupport.org](mailto:emilyholl@siblingsupport.org)*

The Sibling Support Project is the first national program in the United States dedicated to supporting siblings of people with developmental, health, and mental health concerns. Founded by Don Meyer in Seattle in 1990, the Sibling Support Project is a program of Kinderling, the largest and most comprehensive neurodevelopmental center in Washington State. In recognition of the important role siblings play in their families and communities, we support their well-being through books and publications, Sibshops, online sibling communities, and workshops and trainings.

advise and problem-solve with one another, and then step aside to let the magic happen. Sibshops celebrate the many contributions of siblings, and above all acknowledge that siblings are special, too.

Sibshops led to the creation of the Sibling Support Project, and today we work to expand sibling supports around the globe by training and certifying others to run Sibshops in their local communities. Our goal is to make Sibshops accessible to as many siblings as possible.

As a sibling, I agree with other adult sibs who often say, "I wish I had Sibshops when I was a kid." I feel incredibly lucky to be able to experience Sibshops now, by teaching others how to facilitate them. I am deeply moved when young siblings attend their first Sibshop, quiet and uncertain at the start, and positively transformed by the end, asking when they can return for the next one. It is an astounding transformation that takes place in Sibshops everywhere.

**Want to start a Sibshop in your community?**  
**Learn more here: [bit.ly/cdlssibshop](http://bit.ly/cdlssibshop).**



Did you know that siblings share many of the same concerns as parents of individuals with disabilities, along with issues which are uniquely theirs? Siblings also often share the longest-lasting relationships with the family member with disabilities, impact the social development of their brothers and sisters, and serve as caregivers later in life. However, siblings have historically had few resources to help navigate the challenges and opportunities they commonly experience. Don Meyer first changed this by launching Sibshops in 1982 at the University of Washington.

Sibshops are pedal-to-the medal events designed for kids, which means that fun is an integral component. Sibshops enable school-age siblings of kids with disabilities to receive information and support in a highly recreational setting. We play fun games, discuss the ups and downs of having a sibling with a disability, and learn something about disabilities and support services.

At Sibshops, school-age siblings receive the priceless gift of validation from peers who understand and can relate to their experiences. Facilitators create space for the kids to comfort,

## Review of the 2021 Budget

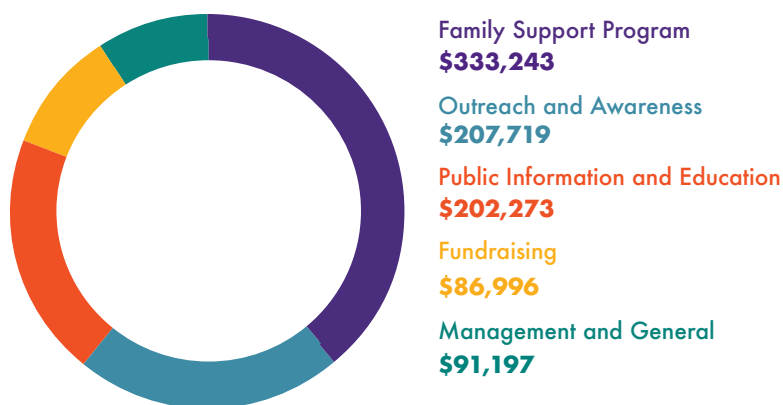
Your generous contributions allow us to sustain and enrich our efforts in supporting your loved one affected with CdLS, increase awareness, as well as connect you with other families in your communities.

With your donations we are also able to provide a vast majority of printed materials and fund research.

We appreciate your kindness and continued support.

Your donations made a difference!

In 2021, the organization had operating expenditures of \$927,248:



## GIFTS THAT COUNT

01.01.22 - 06.30.22

### In Honor/ Celebration

Gregg Abbott  
Jim Abbott

Jillian Billings  
Sandra Beickert

Nicholas D'Angelo  
Layra D'Angelo

Jose Diaz  
Nancy Martin

Halston Dorow  
Nicole Johnson

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Adrienne Perry Fund

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Linda Duff  
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Natalie Fulgieri

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Sarah Griffith

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Rosalind Ferrara

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Sara Lair

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Mary Jetson

Myrna Kaplan  
Ann Swartz

Daniel Kliewer  
Brian Ewert

Charlotte Lawrence  
Diane Smith

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Paul Rockwell  
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Lynnette and Dean Miller

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Barbara Herochik

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Danielle Lowe

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Fred Vasquez  
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## In Memory Of

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Priscilla Amor

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TARC Inc.

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Helen Miller

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Molly & Garth Black

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Leslie Taito

Ira Thor

Stacey Weinstein

Ruthanne White

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New England Wire  
Technologies

Sandra Rossi

Stephanie Westover

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Kyndall Marshburn

Nicole Murray

### Paul Upham

Barbara Winnerman

### Molly Wagner

Karen Morgan

### Julia Wickness

Barbara Wickness

### Devonne Wilson

Georgia Riley

### Aaron Young

Judith Stilwell

Liz Hickman

# Our Deepest Sympathy

## Carolina Amor

September 11, 1996 - May 31, 2022  
Daughter of Priscilla and Fernando Amor  
407 Spring Valley Lane  
Altamonte Springs, FL 32714

## Marci Fisher

September 14, 1979 - March 17, 2022  
Daughter of John Fisher  
963 Tick Hill Road  
Lowell, OH 45744

## Rebecca Hahn

January 18, 1980 - April 23, 2022  
Daughter of Steven and Trim Hahn  
87 Main Street  
Petersborough, NH 03458

## Odin Pond

July 21, 2021 - February 23, 2022  
Son of Madeline and Andrew Pond  
68 Joe Lahout Lane Apt. 20  
Littleton, NH 75070

## Lisa Prom

November 29, 1971 - March 30, 2022  
Sister of Linda Mitchel and daughter of Steve Mitchel  
1021 Glenlyon Lane  
Port Washington, WI 53074

## Jeffrey Rowe

February 20, 1970 - April 8, 2022  
Daughter of Janis and James Rowe  
10329 Franklin Drive  
McKinney, TX 75070

## THE GRIEF SUPPORT ADVISORY GROUP IS HERE TO HELP

When a parent loses a child, the Foundation is here for them in many ways. Our Family Service Professionals are standing by to listen, guide and provide support. We also can connect a parent with another parent who has lost a child. Our unique team of 10 Grief Support volunteers understand, perhaps better than anyone else, what this kind of loss feels like. We also provide helpful and inspirational resources on our website's Bereavement page, as well as an In-Memoriam page where families can share loving memories and stories about their loved one. Visit: [bit.ly/CdLSbereavement](https://bit.ly/CdLSbereavement).

*If you have experienced the loss of a loved one with CdLS and are in need of support, be sure to reach out to [FamilyServicesTeam@CdLSusa.org](mailto:FamilyServicesTeam@CdLSusa.org).*



Yes, I want to **SUPPORT** the CdLS Foundation!

Enclosed is my tax-deductible gift of:

☐ \$1,000 ☐ \$500 ☐ \$250 ☐ \$100 ☐ \$40 ☐ Other \$\_\_\_\_

**Payments may be made by check or credit card.**

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☐ Charge my credit card  
☐ VISA ☐ MC ☐ AMEX  
Card Number: \_\_\_\_\_  
Expiration: \_\_\_\_\_ Security Code: \_\_\_\_\_  
Print name on card: \_\_\_\_\_  
Signature: \_\_\_\_\_  
Email address(es): \_\_\_\_\_

**Donate online at [www.CdLSusa.org](http://www.CdLSusa.org)**



**CdLS Foundation**

Cornelia de Lange Syndrome Foundation, Inc.

30 Tower Lane, #400  
Avon, Connecticut, USA 06001  
800.753.2357  
[www.CdLSusa.org](http://www.CdLSusa.org)

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# CURB CREW MEMBERS NEEDED

*Consider joining the 2022 Team CdLS Curb Crew in Chicago or Baltimore. As a member of this fun group, you will cheer on our runners during the race and attend pre- and post-race events. The dates of the races are 10/9 and 10/15.*

*These are some of the CdLS Foundation's most electric events and an incredible time to bond with other parents and volunteers. It will lift your heart, encourage you and change you in wonderful ways.*

*To learn more or volunteer, contact Sherry at 860.899.1227 or [swaitsman@CdLSusa.org](mailto:swaitsman@CdLSusa.org).*



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