REACHING OUT

WINTER 2022

Spotlights:

Updates on CdLS Symposium Research

ON THE COVER

Ezra Robinson

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

Director's Message

Bonnie Royster, CdLS Foundation
Executive Director

As we approach 2023, we have a moment to reflect on the many ways that the CdLS Foundation is evolving to support you wherever you may be in your journey. We held our Scientific Symposium a few months ago. It was heart-warming to see and hear the scientists and researchers worldwide working together and sharing their learning relative to CdLS. We asked a few scientists if they would be willing to share their findings with all of you. They answered YES. The resulting pages are summaries they wrote specifically for you.

We learned there that there is a significant correlation between Autism and CdLS. Both conditions have a high intolerance of uncertainty. Critical triggers of anxiety in individuals with CdLS are:

- *Changes to the routine*
- *Loud or unexpected noises*
- *Sensory sensitivities*

Parents, do these triggers sound familiar? The results of the UK study are that targeting the intolerance of uncertainty as an intervention could be beneficial for people with CdLS. Also, remedies that work for Autism could effectively treat anxiety in CdLS.

We also learned from another powerful presentation that there is a higher prevalence of sleep disorders in individuals with CdLS compared to the general population. Individuals with CdLS were more active during the morning.

We hope you can take some solace in the fact that these efforts are leading us to interventions that will make a difference in the quality of life for your loved ones in the future.

Bonnie

Reaching Out
Published Since 1977
(ISSN 1097-3052)
Cornelia de Lange Syndrome
Foundation, Inc.
Incorporated December 1981
email: outreach@CdLSusa.org
www.CdLSusa.org

GENE THERAPY WORKSHOP SUMMARY

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

Many genetic conditions, like CdLS, could be treated by gene therapy. Gene therapy is an emerging technology that can "fix" what the variant did to cause the condition. If the DNA sequence of a gene is like a recipe, the gene variant is a change in one of the ingredients of the recipe, and the gene product is the finished cooked food. Gene therapy either changes the gene's DNA sequence back to what it should have been (rewrites the recipe), replaces a missing gene (substitutes another ingredient), or alters a faulty gene product (changes the final food).

Potential gene therapy is developed and investigated in the laboratory to see if it can fix a specific gene variant. If the research is promising, it is tested on animal models. Then after many years, there are human clinical trials. These trials must demonstrate that the benefits of the therapy outweigh its risks. It must show that the therapy is safe and makes a difference in measures that matter to the subjects and/or their families. There must be enough subjects receiving the therapy to compare to untreated individuals meaningfully.

In CdLS, some unique obstacles make it less straightforward to figure out gene therapy. At least 7-8 genes, likely more, cause CdLS. Not one specific organ or cell type is affected in CdLS – almost all are! Some effects of being born with CdLS may not be reversible or changeable. There is so much variability in CdLS not explained by just the gene changes. And we are only starting to know what measures the families would want to prioritize with this type of research. For these reasons, it may take a long time before gene therapy for CdLS is a reality.

The involvement of individuals with CdLS and their families is critical to overcoming these obstacles. Over ten years ago, we formed the "Advanced Clinical Trials (ACT) in CdLS" committee (clinicians and researchers, scientists, educators, allied health professionals, ethicists, family members, and CdLS Foundation staff) to discuss future clinical trials related to CdLS. To understand the preferences of our community, we conducted a voluntary survey of families associated with the Foundation for direct feedback. There was good reception in general for future studies. Respondents indicated that gastrointestinal issues, communication, and anxiety were the top three quality-of-life complications of CdLS for trials to address. Most families would consider enrolling their child in a study that might improve quality of life-related to feeding or early intervention developmental services. Sixty-eight percent would consider an investigation involving an already FDA-approved drug. And both 40% and 40% would not consider such a study using a non-FDA-approved medicine.

Future next steps will include: establishing an accurate and complete registry of all individuals with CdLS through the Foundation to be ready for clinical trials, including knowing the gene variants; forming a family advisory panel to discuss and prioritize preferences and interests; asking families and individuals with CdLS to help educate the FDA; and continuing current research.

TABLE OF CONTENTS

Genomic Analyses in Cornelia de Lange Syndrome and Related Diagnoses

Justin Blair, Division of Human Genetics, Roberts Individualized Medical Genetics Center, The Children's Hospital of Philadelphia (CHOP)

Cornelia de Lange Syndrome (CdLS) can present in various ways with different effects and levels of severity, including different degrees of growth and developmental delays, structural differences, and other systemic involvement. Several genes can cause CdLS when changed. In many cases, variations can be explained by what gene has a change (called a "mutation" or "variant") and by the type and location of a specific mutation in a particular gene. Each gene is a blueprint for producing a protein (the items that does the work in our cells, organs, and tissues) that is 3-dimensional. A change in one area of a gene can have a different effect on the structure of the protein, which can have other effects on how that protein functions in our cells.

We looked at the genetic code of a large population of individuals with CdLS, or others with similar diagnoses, to determine what genetic changes were causing their condition. Significant contributors to causing classic/typical features of CdLS were variants in the genes encoding a group of proteins called the cohesin complex. The cohesin complex comprises many individual proteins and works as a "master switch" in our cells, regulating the number of other proteins made in our cells – a process essential during development. Some critical genes involved in the cohesin complex include *NIPBL, SMC1A, SMC3, HDAC8*, and *RAD21*.

We also found several variants in genes that can cause features seen in CdLS: changes found in genes such as *ANKRD11* and *BRD4*, as well as others like *EP300, AFF4, TAF1*, and others. The overall population of individuals studied included 716 individuals with CdLS and others with similar diagnoses and with samples collected for 25 years. Variants identified 31 genes using screening techniques that have dramatically improved over the years. The most recent and thorough look into this population's genes was in a subgroup of 176 individuals with no genetic answer found using previous tests. For this population, we conducted Whole Genome Sequencing (WGS), which analyzes every single part of an individual's genetic material, including over 20,000 genes and the large expanses of DNA between these genes. Our genes only make up ~ 1-2 % of all our DNA, with billions of letters representing the base pairs that are the blueprint for all of our features. From this subgroup with WGS, causative gene changes were found in 31% of individuals, 16% in known cohesin complex genes, and 15% in different genes that result in a similar clinical picture to CdLS when mutated.

Additionally, several new CdLS candidate genes that were not previously associated with this diagnosis in the past were identified. Using complex bioinformatic analytic tools, we could show similarities in all these genes' functions. We found multiple individuals with the same variant, often having very similar features, and found specific "hot spot" places in the genes where variants were more commonly seen.

Even with this powerful genome sequencing technology, there are still a significant number of individuals with CdLS that we could not identify an underlying genetic cause. We have received additional funding from the National Institutes of Health (NIH) to take the following steps to find answers for these families. This includes looking at something called "RNA" instead of DNA. RNA is produced from DNA sequencing and processed to help shed light on whether there are "cryptic" mutations or variants missed when looking for them in the DNAS sequence alone.

This research will help us provide valuable information to families with a child or family member with CdLS and expand our knowledge of the causes of CdLS. It will also shed light on isolated birth differences (which can be seen in CdLS, such as diaphragmatic hernias) for which genetic causes are poorly understood and hopefully provide insights into strategies to develop potential therapeutic options for CdLS.

The Relationship between Anxiety and Intolerance of Uncertainty in Cornelia de Lange Syndrome

Victoria Perry, School of Psychological, Social and Behavioural Sciences, Coventry University, Priory Street, Coventry, UK, CV1 5FB; Kayla Smith and Hayley Crawford, Cerebra Network for Neurodevelopmental Disorders and Mental Health and Wellbeing Unit, Warwick Medical School, University of Warwick, UK, CV4 7AL; and Laura Groves, Cerebra Network for Neurodevelopmental Disorders and School of Psychology, University of Birmingham, Edgbaston, Birmingham, UK, B15 2TT

People with Cornelia de Lange syndrome (CdLS) are more likely to experience co-occurring anxiety and autism than the general population and people with an intellectual disability with no known genetic cause. However, the relationship between anxiety and autism is unclear, and it can also be challenging to identify these conditions in people with CdLS. Recent studies suggest that intolerance of uncertainty* may help explain the relationship between anxiety and autism. Additionally, intolerance of uncertainty has been identified as a significant factor in the development and maintenance of anxiety in autistic people. Many people with CdLS are likely to experience both anxiety and autism. Understanding the relationship between anxiety, autism, and intolerance of uncertainty is vital for informing interventions and future research on anxiety for people with CdLS.

In this study, we aimed to look at the relationship between the severity of anxiety symptoms, autism traits, and levels of intolerance of uncertainty and determine if intolerance of uncertainty could explain the relationship between the severity of anxiety symptoms and autism traits in people with CdLS.

This study included 33 people with CdLS, who were at least four years old, and had mild to moderate intellectual disability. The severity of anxiety symptoms, autism traits, and levels of intolerance of uncertainty were examined using a variety of questionnaires completed by a parent or caregiver.

Results from this study suggest that autism traits and levels of intolerance of uncertainty significantly predicted the severity of anxiety symptoms. Additionally, levels of intolerance of uncertainty thoroughly explained the relationship between the severity of anxiety symptoms and autism traits in people with CdLS, like the association seen in people with autism.

The results of this study suggest that intolerance of uncertainty plays an essential role in the presence of anxiety in CdLS. As a result, it is possible that targeting intolerance of uncertainty as an intervention for anxiety may be beneficial for people with CdLS. Recent studies suggest that cognitive behavioural therapy (CBT) targeting intolerance of uncertainty has been helpful in reducing anxiety in the public. Similarly, autism-specific interventions, such as the 'Coping with Uncertainty in Everyday Situations' (CUES), have shown promising results in reducing intolerance of uncertainty and anxiety in autistic people. Although these interventions have not been used with people with rare genetic syndromes, these programs are promising options for people with CdLS.

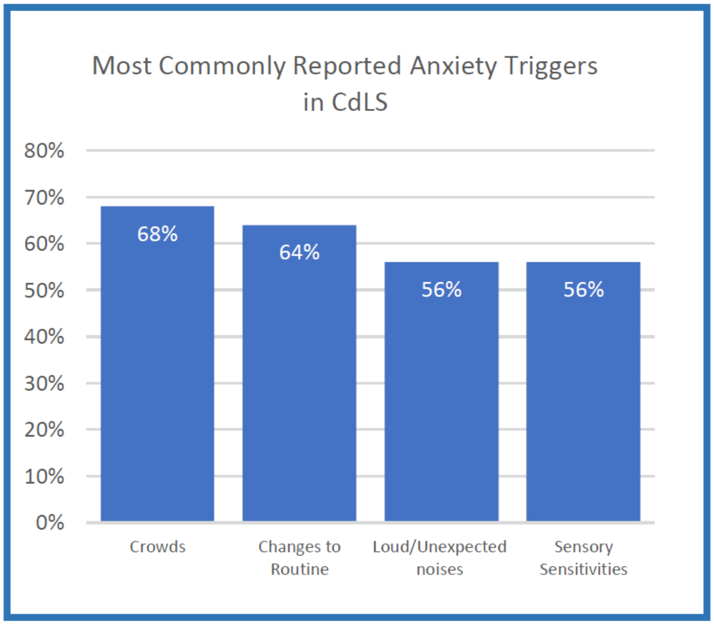
*Intolerance of uncertainty is commonly defined as the tendency to react negatively or experience distress when faced with uncertain or unpredictable situations.

Triggers and Correlates of Anxiety in Cornelia de Lange Syndrome

Jessica Mingins, Dr. Jane Waite, Dr. Hayley Crawford, and Dr. Joanne Tarver

People with CdLS often have high anxiety levels, which may be specific to CdLS. However, not much research has broadly focused on anxiety triggers in people with CdLS. Current research on anxiety has not been representative of many people with CdLS, because there is a broader and more severe range of intellectual disability (ID) levels and often speak few to no words are used in speech

The goals of this research project were to: identify the most reported anxiety triggers in people with CdLS who speak few or no words, compare the proportion of people with CdLS who reported these triggers to groups of autistic people and people with Fragile X Syndrome (FXS), another rare genetic syndrome associated with ID (both autism and FXS are associated with high levels of anxiety), and identify differences between the people with CdLS who reported each of the most reported triggers compared to those who did not.



We recruited 150 parents or caregivers of people with CdLS, FXS, or autism through NHS trusts, mailing lists, and syndrome support groups. They filled in an online questionnaire that asked about anxiety triggers, autism characteristics, sensory sensitivities, and intolerance of uncertainty in the person in their care. The participants filled in the questionnaire on behalf of 26 people with CdLS, 22 people with FXS, and 116 autistic people aged 4-63 years. Most were male (70%), many had an anxiety disorder (27%), and most never spoke a word or spoke odd words only (64%).

The three most reported anxiety triggers for people with CdLS were crowds (68%), changes to routine (64%), loud or unexpected noises (56%), and sensory sensitivities (56%). There were no significant differences between the proportion of people with FXS and autism. They reported these triggers, which shows how high anxiety is in CdLS. People with CdLS who had crowds, changes to routine, loud or unexpected noises, and sensory sensitivities as anxiety triggers often had higher autism characteristics, intolerance of uncertainty, and auditory sensory sensitivities than people with CdLS who did not have these anxiety triggers.

What does this mean? The most reported triggers of anxiety in CdLS are also common in autism. People with CdLS who had these triggers often had high levels of autism and increased intolerance of uncertainty and auditory sensory sensitivities, both associated with autism. These results help us to understand what factors lead to anxiety in people with CdLS. We may be able to adapt assessments and interventions for anxiety, which are used in autistic people for people with CdLS. Future research should continue investigating the link between autism characteristics and anxiety in people with CdLS.

Evaluation of Sleep Disturbance and Patterns in Patients with CdLS

Carol Li, M.D., Pediatric Otolaryngologist, Division of Pediatric Otolaryngology-Head and Neck Surgery, Cincinnati Children's Hospital Medical Center

Insufficient sleep can negatively affect cognitive function and mood and be associated with obesity, high blood pressure, and diabetes. Sleep disorders are more prevalent in patients with neurodevelopmental disorders. This study aimed to identify sleep disturbances in individuals with CdLS.

In the first part of this study, we used validated sleep surveys administered to caregivers of patients with CdLS. These surveys were the Obstructive Sleep Apnea-18 (OSA-18) questionnaire, pediatric sleep questionnaire (PSQ), pediatric daytime sleepiness scale (PDSS), and children's chronotype questionnaire (CCTQ). The results of these surveys have been shown in other studies to predict obstructive sleep apnea and sleep-disordered breathing. We found a more significant percentage of individuals with CdLS were at high risk of sleep-disordered breathing compared to the population norm (Figure 1).

Although a higher rate of individuals with CdLS were at risk for sleep-disordered breathing, the PDSS scores showed that these individuals were not overly sleepy during the day compared to the general population (Figure 2).

Finally, the fourth survey, the CCTQ, showed that the circadian (daily changes in) rhythms of those with CdLS tended to be morning-type individuals (Figure 3).

The second part of this study used actigraphy (small sensors worn on the body to measure motor activity) to assess sleep-wake cycles. We compared results from actigraphs worn by individuals with CdLS to those worn by their family members. Results showed that most sleep measures, like total sleep time and the number of arousals per hour, were similar between individuals with CdLS and their family members. However, the family members without CdLS did take a longer time to go to sleep.

In summary, this study showed a higher prevalence of sleep disorders in individuals with CdLS compared to the general population. However, individuals with CdLS did not report sleeping excessively during the day. Compared to the general population, individuals with CdLS were more active during the morning.

These data may be useful in addressing sleep-related issues for children with CdLS. **We appreciate the funding received from the CdLS Foundation.**

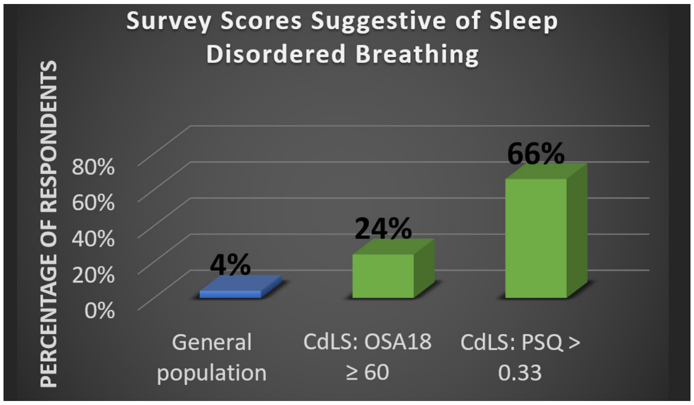


Figure 1

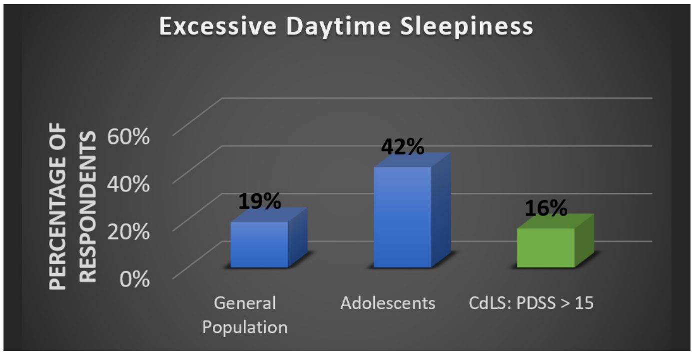


Figure 2

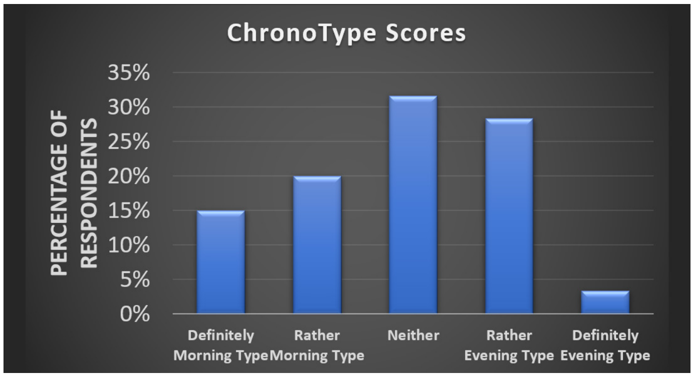


Figure 3

HIGHLIGHTED **EVENTS**

TEAM CdLS HITS THE PAVEMENT IN CHICAGO AND BALTIMORE

What a run! Members of Team CdLS hit the streets in Chicago and Baltimore this fall, raising more than $86,000 to support individuals with CdLS and their families!

Thirteen runners from four states and two countries raised more than $70,000 by participating in the October 2022 Bank of America Chicago Marathon. The CdLS Foundation has had a team running in this event for twenty years. One of this year's runners, Edgar Solis Chavez, whose 4-year-old son Daniel has CdLS, traveled with his family from Mexico to run in Chicago with the team. To spur him on, Edgar ran every mile of the 26.2-mile race in honor of a different child with CdLS from another nation. That is just one of many amazing inspirational stories from Team CdLS in Chicago.

Meanwhile, not to be outdone, Team CdLS in Baltimore had a record number of participants in the Baltimore Running Festival this year, totaling 34! This is the most participants Team CdLS has ever had at the Baltimore Run. The team's co-captains are CdLS Foundation Medical Director Dr. Tonie Kline and Tasha Howland, who runs on behalf of her precious daughter Peyton, who has CdLS. This group - participating in the full marathon, half-marathon, 5k, or walk - exceeded their fundraising goal by raising more than $16,000. The group also raised awareness about CdLS and the Foundation as they stormed the streets of Baltimore.

Thanks to our runners and curb crew volunteers for making this year's Team CdLS events the best! Your dedication has made a difference.

CHICAGO MARATHON 2023 REGISTRATION NOW OPEN

That's right! No grass grows under the Bank of America Chicago Marathon organizers' feet. (Pun intended!)

Registration for the big race is already up and running!

The CdLS Foundation has nineteen regular registration allotments and two 2020 deferred registration allotments for Team CdLS in 2023. These allotments are hot commodities! If you are interested and want to learn more about joining Team CdLS in Chicago for an incredible event in October 2023, please connect with Sherry Waitsman at swaitsman@cdlsusa.org.

WALGREENS RAISES FUNDS AND CdLS AWARENESS

Make a donation, make a difference
DONATE $1, $5 OR $10
AT THE REGISTER
JUNE 13–29

FOLLOW THIS LINK TO LEARN MORE ABOUT ALL OF OUR CdLS FOUNDATION AMBASSADORS

MEET TARYN!

Taryn is a 14-year-old from Sutton. She loves music and movies. Taryn has CdLS, a rare genetic disorder that causes a broad range of physical, cognitive and medical challenges. It is found in both genders and all ethnicities. While CdLS is a part of Taryn's life, it does not define her. **Taryn is resilient and captures the hearts of everyone she meets.**

Thank you to Walgreens and their customers for digging deep to raise $14,331 in funds and raising awareness for the CdLS Foundation! Thank you also to Mike Christie for making it all happen.

Mike, who works for Walgreens, also happens to be the dad to 14-year-old Taryn and is a CdLS

Foundation Board Member. For the past several years, Mike has submitted the CdLS Foundation as one of Walgreen's charities of choice for their Scannable Fundraising Event held at Walgreens stores across southeastern Massachusetts.


HELP SUPPORT THOSE WITH CdLS

Walgreens Scannable Event

Make a donation, make a difference

DONATE $1, $5 OR $10

AT THE REGISTER

JUNE 13–29

Walgreens | CdLS Foundation

FOLLOW THIS LINK TO LEARN MORE ABOUT ALL OF OUR CdLS FOUNDATION AMBASSADORS

MEET THOMAS!

Thomas is an 12-year-old Foxboro resident who enjoys listening to music, playing with his sister and being outside. Thomas has CdLS, a rare genetic disorder that causes a broad range of physical, cognitive and medical challenges. It is found in both genders and all ethnic backgrounds. While CdLS is a part of Thomas' life, it does not define him. **Thomas is resilient and captures the hearts of everyone he meets.**

For two weeks in June, posters featuring the stories and photos of local CdLS Foundation ambassadors were displayed at Walgreens stores in Massachusetts from Milford to Boston, south to New Bedford, and Cape Cod. With these informational posters throughout the stores and on the counters, customers were made aware of CdLS and asked to make donations at checkout.

"Walgreens serves over 8 million customers and patients per day. We believe that we are part of every community we serve and can help make our team members and customers' lives a little more joyful and healthier through our personalized interactions with them daily," Mike commented. "By supporting local and national charitable organizations at the local level, Walgreens attempts to use our brand recognition and footprint to help those who need it, like the CdLS community. We sponsor events like the scannable periodically throughout the year."

This is the third time the store has featured the CdLS Foundation, thanks to Mike's efforts.

GOLF CLASSIC RAISES MONEY IN ST. LOUIS IN MEMORY OF AMBER AND IN HONOR OF ALL WITH CdLS

Golfers and sponsors came together again to see that the MCWP and PLM 34th Annual Amber Gaines Memorial Golf Classic was another huge success.

With more than fifty sponsors and 120 golfers, this fundraiser grossed close to $75,000 for the CdLS Foundation. Held at the beautiful Tapawingo National Golf Club, the event is traditionally scheduled for Columbus Day each year.

In its 34-year history, it is estimated that this golf classic has grossed approximately a million dollars for the CdLS Foundation.

A special thank you goes to title sponsors Madison County Wood Products, PLM Companies, the Gaines, Kesting, and Snodsmith families, and major sponsors: Daniel & Henry, Lakenan, LPS Equipment, Magnum – Mid Continent Steel, TAG Truck Center, and UHY LLP.

CALENDAR 2023

February 12, 2023
A Day of Remembrance

February 28, 2023
Rare Disease Day

March 2023
Family Gathering
California

March 8, 2023
Multidisciplinary Clinic
Salt Lake City, Utah

April 1, 2023
Multidisciplinary Clinic for
Adolescents and Adults
Baltimore, MD

May 2023
New England Golf Classic
Registrations Open in January
Ipswich, MA

May 13, 2023
CdLS Awareness Day

June 2023
Family Gathering
Illinois

October 8, 2023
Bank of America Chicago
Registration to Join Team CdLS
NOW OPEN
Chicago, IL

October 9, 2023
35th Annual Amber Gaines Memorial
Golf Classic

Using Your Donations Wisely

If you have a loved one with CdLS and have used the CdLS Foundation services, then you already know that the services we provide are at NO COST to families. We prioritize utilizing our resources, much of which comes from our families, in ways most beneficial to those we serve.

The CdLS Foundation continues to boast of being an award-winning nonprofit organization because of its dedication to serving adults and children living with CdLS and using your donations wisely.

When you give to the CdLS Foundation, you are giving to an organization that bears America's Best Charities Seal of Excellence. This is awarded to charitable organizations that meet the highest public accountability standards, efficiency, and cost-effectiveness.

The CdLS Foundation is among the few organizations that earn the GuideStar Platinum Award. GuideStar is the world's largest source of information on nonprofit organizations.

In addition, the CdLS Foundation is also considered a Four-Star Charity - the top honor - by Charity Navigator. It rates charities by evaluating the organization's financial health, accountability, and transparency.

As the only nonprofit organization in the nation dedicated to CdLS, the CdLS Foundation sees to it that your donations make the most significant impact possible for those who need it most.

When you donate to the CdLS Foundation, you can rest assured that your gifts are being used well. Thank you to everyone who continues to support the CdLS Foundation and its critical mission.

The Sustainers Circle: A Special Nod of Thanks to Our Recurring Donors

Whether for a personal household or a business, anyone planning a budget and paying bills knows how important it is to be able to rely on a regular income. It provides a sense of security and relief. That is what our recurring donors provide to the CdLS Foundation.

These donors make a significant impact by contributing a set amount on a weekly or monthly basis. They help sustain the CdLS Foundation. Some donate online by selecting the "recurring" option. In contrast, others contribute through direct deposit from their paychecks or by sending in a check. We call these donors members of our Sustainers Circle, and we are very grateful to them:

Veronica Airey-Wilson
Joyce Bell
Brooke Bender
Vincent Berardini
Chris Bernhardt
Praveen Bhagavatula
Cheri Carrico
Kamisha Coleman
Darlene Davis
Beth Dixon
Nancy Drach
Janice Duff
Jill Dupuis
Katherine Frank
Gerome Technologies
Fernando Gonzalez
Ellen Graper
David Harvey
Weber Hoen
Karen Huang
Donald Johnsen
Ann Johnson
Penelope Keating
Farah Kennedy
Dorothy Laumann
Dianne Lessa
Jeff Linsk
Tim Luce
Tracey Maguire
Janis Mamayek
Debi Marks
Lynnette Miller
Hien Nguyen
Katherina Terhune
Karl Rissland
Bonnie Royster
Donna Shuford
Peter and Julia Swanson
Nicole Tiongson
Carmen Valentin Swerling
Darl VanderMeulen
Sibyl Wyatt
Angie Young

If you want to become a part of our very special Sustainers Circle, use this link to make an online donation and select the "Recurring Donation" option: **bit.ly/CdLS_Sustainers** or contact Annette Scheidecker at ascheidecker@cdlsusa.org or 860.899.1391.

Finance Report

Workplace Giving

Did you know that your employer may be offering a workplace giving campaign?

Many companies offer employees the opportunity to make a one-time charitable gift or give through payroll deduction during an annual workplace giving campaign. Workplace giving is an easy and efficient way to work with your company to benefit the CdLS community.

Don't hesitate to contact your Human Resources department to determine whether your company offers a workplace giving campaign. Giving policies vary among companies.

Donating through your workplace giving program offers the convenience of automatic payroll deductions without losing the tax benefits of charitable giving. Employers often match their employees' contributions, so the impact of your gift can go even further!

You shop. Amazon Gives.

Shop at smile.amazon.com and 0.5% of your eligible purchases will be donated to the CdLS Foundation – no fees, no extra costs. Just follow the steps below to get started:

1. In your internet browser go to **smile.amazon.com** and log in using your Amazon credentials.
2. Under **"pick your own charitable organization"**, type in Cornelia de Lange Syndrome Foundation, Inc.
3. Click **"Search"** and from the list, select **Cornelia de Lange Syndrome Foundation.**
4. Check the **YES** box to confirm. You must visit smile.amazon.com each time you shop to support the CdLS Foundation.
5. When redirected, bookmark your AmazonSmile page so it comes up automatically when ordering items.
6. **That's It!**

Self-Care Tips for Parents and Families During the Holidays

Michele Kane, Chair, Parent, Family & Community Network; Professor of Special Education, Northeastern Illinois University

Almost every culture has a festival of light during the winter months. Some are religious and some are secular, but all traditions typically include an increase in activities related to music, celebrations, specialized food preparation, and exchanging of gifts. For families with gifted people who are already dealing with the stress that comes with intensity, sensitivity, and perfectionism, this whirlwind of events can easily drift towards overload. Often, busy caregivers put themselves last and are short-changed without time to recharge and renew. The following tips are some ways to enhance self-care either solo or in tandem with other family members:

1. Take time out each day for a few minutes of alone time/me time. When adults model that this is an essential part of life then they send the message that respite is important, just like work. Microbursts of me-time can help to reset when daily challenges threaten to overwhelm. Five minutes of quiet reading, practicing an instrument, or walking the dog can be energizing. Brainstorm with your family how each person can find some individual favorites for finding their quiet center and then make and post the lists.

2. The research on mindfulness is becoming more and more clear—mindfulness or noticing the present with kindness and curiosity— is effective in creating a sense of peace and calm. Whether you have a formal mindfulness practice like meditation or an informal practice like intentional walking, there remains a focus on what is happening in the here-and-now. Delighting in the seasonal sounds and tastes, savoring the scents, and giving attention to natural surrounds heightens awareness and slows the pace of life. Guiding gifted youngsters in these practices creates a shared bond of experience.

3. Breathe—just breathe. It sounds so silly to be reminded of something obvious but in times of stress the breathing often becomes erratic and escalates a sense of loss of control. Controlling one's breath with a simple activity like breath in, hold for four counts, breath out, and hold for four counts can generate a sense of control and tranquility. For those who prefer more direction there are increasing numbers of free apps for kids and adults alike that provide guided breathing or relaxation exercises. Have family members locate the ones they favor for tone, cadence, and overall soothing ability. It is often revealing to note the wide range of preferences within the family.

4. Find time to demonstrate moving empathy to compassion. Compassion is the willingness to shift towards action in relationship to kindness or thoughtfulness towards others. It isn't difficult to find others in need but sometimes it can be tough to find other families who share similar concerns. Websites like The Honeycomb Project can provide resources for families who share a vision towards helping a particular cause or group. Researching and reading more about issues can provide gifted youngsters with a deeper understanding of the complexity of social justice issues.

Enjoyment of the holidays begins with maintaining a sense of equilibrium despite the many challenges brought on by the increase in activities and events. Joining together and sharing helpful strategies within and among families can lead to festivities that create positive memories for all.

A Letter of Encouragement Inspires Special Needs Families

Ewa Omahen, Ph.D., Autism Parenting Magazine

A Letter to a Fellow Parent Walking in My Shoes:

I know how hard it is to let go of dreams and expectations. The reminders of possibilities of what could have been and will not be around wherever I look. My friends' and family's kids are growing up, graduating from high school then college, getting jobs, starting families. With every graduation celebration and every wedding, we attend, I reminisce some more about what could have been and is not in the cards for us as a family. It is hard to let go of one's plans and dreams. There are more tears, more sadness, more grief, more worries about our children's future.

In the midst of all this, I often neglect to celebrate my child for who he is. I do not notice the joy on his face of simply being, living in the moment, enjoying simple things. I grieve, yet he continues to look up to me with so much love, innocence, trust, and forgiveness.

Each day, I make a promise to myself that I will try harder to celebrate the gifts he brings daily to me and to this world. I will get better at understanding his way of teaching me and others about patience, compassion, and unconditional love.

Today I will stop and look more carefully into his eyes and once more ask for his forgiveness for my disappointment, sadness, and loss. I will take the time to notice the beauty, innocence, and light he brings into this world. Once again, I will thank him for the gifts he brings into my life. Today I will worry less and celebrate him more for the special gift that he is.

- Mom of a "Differently-abled" Child

Ezra Robinson

By Cassandra Brown

Ezra is 2.5 years old, born on the first day of "Heart Week" on February 7, 2020. He is such a happy-go-lucky kid, always laughing and smiling. He lights up any environment and is always friendly and outgoing. Ezra was diagnosed when he was 11 days old with the *NIPBL* gene mutation. While Ezra is happy, he does suffer from chronic illness due to CdLS. He has complex congenital heart disease, mild kidney failure, severe reflux, a small airway, and low muscle tone. Due to these issues, he gets breathing treatments 3x a day. He's g-tube dependent and takes ten medications daily, a few every 8 hours. He also is dependent on oxygen while sleeping. He has developmental delays, so he doesn't do things other almost 3-year-olds would do independently, like feeding himself, walking, or talking.

He loves to snuggle and chill out some days, but most days, he loves to get into trouble with his Kitty, Nova. He also loves going on walks and car rides and enjoys playing with his sensory toys. Ezra has faced many challenges. The most severe being a lengthy NICU stay, open heart surgery at six months old, and complications from heart surgery. We almost lost him, but I am so grateful he pulled through!

Having a child with CdLS is so nerve-wracking at first. Google does not help with the anxiety that comes with the shock. However, despite all his struggles, Ezra is one of the sweetest kids I've ever known. If Ezra didn't have CdLS, he wouldn't be who he is, and while that is bittersweet due to his health problems, I couldn't have asked for a better kid.

The CdLS Foundation has helped connect us with other families through Facebook groups and the newsletter.

If I could tell the world one thing, I'd say, "Don't apologize! His life is not a sorry one". When they ask about his diagnosis, many people always say, "aw, I'm so sorry; he'll be okay, though," and it doesn't hit the way they want it. Change your approach to disabled kids and their parents.

Logan Fowler

Susan's Story, Logan's Mom

Logan is a hero.

He was born 33 years ago and diagnosed at the age of three months with CdLS. We didn't know at that time what was really going to happen to him. We have been extremely blessed. He is a wonderful, smart, sensitive person. He has a wonderful life able to ride bikes, ski and live a normal life adapting to his special needs.

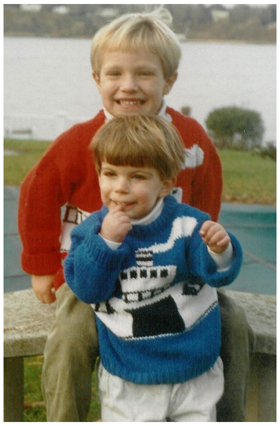
He is now a part of a facility in Westbrook Connecticut, Vista Life Innovations. There he is in his own house and rents two of the rooms out to other members of Vista. He does everything -laundry, orders food from Instacart, meets repair people, writes checks, pays bills, gets himself to a personal trainer, etc.

Doctors mean well and try to do what they can. However, we found they - we - did too much. He had issues with hearing, renal reflux, hernia, testosterone, and anxiety panic attacks. Now Logan won't go to doctors. At one point when he was about 18, and having anxiety issues, he had tried 32 different meds. He was in terrible shape - hallucinating, bolting from the house, knocking over things. We worked hard to take him off his medications. He hasn't been on meds for about nine years. Thank God he has been healthy. But it is a worry that he now refuses to go to the doctor. How do you know what to do?

Logan's Story

Living with CdLS can be hard at times, but other times it is not so tough. I feel like CdLS causes some issues for very high anxiety, depression, and OCD. Sometimes, I really must push myself to do things, for example going out to run errands. It also causes to me crave sugar and weight problems.

I don't really trust doctors. It is very tough for me to go see a doctor. Medication and I don't mix and when I was 18, I was overmedicated and loopy - in and out of the hospital all the time. I enjoy and am in tune with astrology, tarot cards and spiritual tv shows, like Joel Osteen. I also keep physically healthy.



When I was in school, I loved it. I had days that were difficult, but overall it wasn't too bad. I hang out with my friends. I like to go to Dunkin Donuts and Starbucks. I like to go on walks and watch TV shows and movies. For the last 10 years I have owned a home in Westbrook, CT. Through a program called Vista, I rent out a couple rooms to individuals.

In the future, I want to be an astrologer or a tarot card reader.

If you have medication questions please speak with your prescribing provider. If you have additional questions or concerns, contact Family Service at 800.753.2357, press 1, or email **familyservicesteam@CdLSusa.org**.

Helping Your Family Thrive During the Holiday Season

The holiday season can be joyous! But a whirlwind of activity can quickly turn things stressful!

Here are some suggestions to help your family during the holiday season.

1. Remember that the small things are almost always the big things.

The Sibling Support Project asked fifty adult siblings of people with disabilities: "When you were younger, what did your parents, family members, and service providers do to make you feel special and let you know they cared?" None of their responses mentioned the need for expensive gifts or grand gestures.

Reflect on some of their responses. See if you can include similar things even during the holiday season.

- "My mom and I had "dates" and did something that I wanted to do. I know planning this was sometimes difficult for her, especially if my brother was having a bad day. But she always made it happen one way or another".
- "My mom would lay in bed with me after putting my brother to bed. We would talk about anything and everything. Those fifteen to thirty minutes of uninterrupted time with her daily put our relationship on the right track."
- "Even the smallest reassurance can make someone's day."

2. Review your schedule. Prioritize what your children like.

It may not be possible to cater to everyone's wants and needs. Including activities most important to your children in your holiday celebrations can help reinforce how much you support them.

3. Try and prepare everyone.

Create and share a calendar of your plans for the holiday season. When possible, make your children aware of what to expect, how long an event lasts, who will be there, and what they should do if they need a break.

When getting together with those who aren't very familiar with your child with CdLS, consider providing a little tutorial in advance. Sharing information may help avoid confusion or hurt feelings if your child does not behave as others expect. Sharing how your child may react during the visit can be helpful. For example, consider letting people know if your child prefers high-fives over hugs or if they are hesitant to try new foods.

4. Maintain at least some of your typical routines

Schedules are interrupted during the holiday season but choose a few routines to maintain. Know that a change in sleep schedules can be incredibly disruptive. Build in time for rest or let children sleep later than usual. Preserving pre-bedtime rituals like bathing or storytelling can be helpful.

5. Utilize Helpful Tools

Reducing frustration and anxiety helps everyone! Physical or Occupational Therapists may recommend ways to help your child with CdLS be more mobile or comfortable. Products that improve posture or stability may help your child expend less energy. Some children benefit from weighted blankets or fidget toys to help reduce anxiety. Adjust gifts by loosening ribbons, unsealing envelopes, or minimizing tape for those that struggle with fine motor skills.

6. Identify how your village can help.

Identify things others can help with or do on your behalf. If there is an event your child with CdLS may not manage well, but their sibling(s) want to attend, can a friend or family member provide transportation? When others ask what they should get you for the holidays, ask for help that allows you to spend time with your child's siblings with CdLS or for a few hours of "me time" to recharge. A little goes a long way!

Additional planning may be necessary for your family to thrive during this time. But with preparation and communication, the holiday season can be a magical time of year.

We are wishing you and yours a healthy, happy holiday season!

The Family Services Team

WELCOME
New Families

California
Douglas Bodger and
daughter Penelope,
born 12/17/2018

Connecticut
Francesca Torres and son
Noah, born 7/22/2022

Illinois
Samantha Brown and
Edward Patterson and
daughter Naya,
born 2/22/2022

Minnesota
Tiffani and Taylor Hoffbeck
and son Riggins,
born 12/11/2020

Tennessee
Carrie and Zach Willmore
daughter Letty,
born 8/12/2018

Tribute Gifts 07.01.22 - 10.01.22

In Honor/ Celebration

Lizzie Bernhardt
Rita Linert

Donald Dellorco
Don and Mary Dellorco

Freya Dunning
William Taggart

Caitlin Igoe
Michael Igoe

Mason King
Kathy Brownell

Lauren Lewin
Lori Comber

David Melisa
Gloria Allen

Jonathan Swanson
Suzanne Soltess

Lillian Vangelder
Natasha Parise

Russell Zach
Deb Hood

In Memory Of

Diane Bockelman
Paul and Debbie Biggs
J. Buchanan
Jacqueline Conomikes
Dinny Elliot
Janette Elliot
William Elliot

Michael Cataline
Lynn Audette
Rosemary Balistreri
Elizabeth Brooke
George & Carol Bunce
Peter Butler
Mary Anne Calabrese
Carol and John Cataline
Paul & Jeanne Cataline
Casey Cherry
Lillian Collins
Deborah Combs
John D'Agostino
John and Catherine D'Agostino
Rahn Dagostino
Carol DeRuyter
Necia Dinan
Josh & Kelly Dinan
John Elliott
Stephany Felice
Finger Lakes Football Association
Erin Frahm
Charmaine Gallagher
Joanne Gersuk
Donald & Julie Guerrieri
Joanne Haers
Mary Hanna
Joann Harrison
Wanda High
Donna Holtby
Lisa Jimenez
Alex Levin
Teresa Malone
Carol McGuigan
Lynnette Miller
Gabrielle and Tim Nadeau
Patricia Notebaert
Tracie Pence
Donna Petrella
Elizabeth Poplawski
Nancy Powers
James Rinaldo
Misty and Michae lShuff
Jarrod Smith
Loretta Snyder-Frank
Florence Stajura
Dominick & Kathryn Vedora
Lindsay Willson
Norm and Barbara Winnerman
Elaine & Edwin Wittwer
Angela and Tim Young

Linda Marie Colombo
John Brancato
Michael Constantelo
Rose Cosenz
Joann Garofalo
Patricia Kopp
Nico Lechuga
Choloe Lechuga
Roberta & Angelo Libero
Meliss Mello
Stephanie Phang
Sophie Potvin
Tiffany Sunell
Lorraine Trautwein
Keith Whitmore
Steve Yurkunas

Joe Churchman
William Adkins
Rose Disanto

Nicholas Decillis
Barbara Peppas

Kamren Dennis
Kamisha Coleman

Casspin Hall
The Board and Staff at the CdLS Foundation

Waylon Kirchoff
The Board and Staff at the CdLS Foundation

Denny Koss
Rebecca McDonald

Jessi Anne McRobbie
Kathy and Gene van Buren
Benjamin Miller
Jeffrey and Melinda Miller

Ella Grace Musial
Joann Fordy

Molly Wagner
James and Ann Chleapas

Laurie Warren
Brooke Helton

Wyatt Welton
Matthew Steiner

Dennis P. Wrigley
Lori Comber
Giacomo Deannuntis
David J. Lafond
Mary Mammarella
Jason Mento

Our Deepest Sympathy

Michael Cataline
June 18, 1971 - July 28, 2022
Son to Carol and
John Cataline
18 Universal Avenue
Geneva, NY 14456

Casspian Hall
February 18, 2022 -
June 10, 2022
Son to Haley and Taaron Hall
104 West Cook Street
Mento, TX 75124

Waylon Kirchoff
May 27, 2022 - June 28, 2022
Son of Kaitlyn Engle
and Lane Kirchoff
1514 Cherry Street
Gainesville, TX 76240

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.

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.

YES! I want to continue to support the CdLS Foundation.

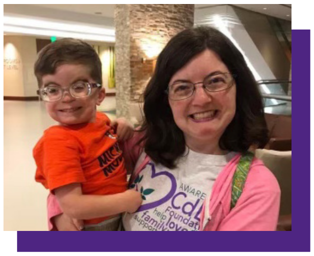
☐ **$1,000** ☐ **$500** ☐ **$250** ☐ **$100** ☐ **$50** ☐ **$**__________

☐ Check (payable to the CdLS Foundation)

☐ Credit Card

☐ Visa ☐ MasterCard ☐ American Express ☐ Discover

CC#:______________________________ CV:_____ Exp. (MM/YY):______
Signature:____
Email:____



Scan this QR code to **Donate online**.

CdLS Foundation | 30 Tower Lane, Suite 400, Avon, CT 06001

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.
30 Tower Lane, #400
Avon, Connecticut, USA 06001
800.753.2357
www.CdLSusa.org

NON-PROFIT
U.S. POSTAGE
PAID
HARTFORD, CT
PERMIT NO. 751

From Their Hearts to Yours

Your year-end gift helps ensure that the CdLS Foundation is fully funded in 2023 and able to continue to serve the incredible CdLS community.

Having a caring team to lean on means the world to so many living with CdLS. "The CdLS Foundation gives me the resources and support that I need, especially when I'm going through hard situations," explained thirty-five-year old Jesse.

Thirty-year-old Emily adds, "I've even had new doctors consult with the Foundation when they had questions."

Emily and Jesse echo the sentiments of many individuals who rely on the CdLS Foundation….just as the CdLS Foundation relies on you. Please consider donating by December 31st.

> "The donations you give mean I will always have access to the experts I need."
>
> -Emily, Adult with CdLS

Printing and distribution supported by:
The American Legion Child Welfare Foundation, Inc.