

Reaching Out

The Newsletter of the Cornelia de Lange Syndrome (CdLS) USA Foundation, Inc.  Second Quarter 2017

Highlights

Director's Message:

The Yellow Brick Road
to Awareness

Spotlights:

Sleep and CdLS Research

Alternative Communication
in the Education System

Super Siblings

David and Michael



Photo by Laura Kilgus



Director's Message



The Yellow Brick Road to Awareness

In the classic American tale, *The Wizard of Oz*, Dorothy is thrust from her home by a tornado with her puppy Toto into a strange new land. Feeling out of sorts, her only goal is to get back home. As she moves along her journey she encounters a lion with no courage, a tinman without a heart and a scarecrow who doesn't have a brain. Together, as they make their way to see the Wizard of OZ, they uncover courage they didn't realize they had, feelings and emotions that propelled them forward and untapped wisdom in the face of uncertain circumstances.

Like Dorothy and her companions, as you walk the path toward raising the CdLS banner of awareness for your loved ones, you are not alone. There are many wonderful parents who advocate on behalf of their child for school services and appropriate medical attention. As soldiers on the ground – you are a wonderful resource. Below are a few ideas on how you can expand your advocacy and bring awareness to CdLS:

1. Write a short letter to the editor of your local newspaper about the ways your school has been supportive of your child or ways you think the school district could change to be more accommodating.
2. Reach out to the Foundation for support and insight into how you may be able to tackle an obstacle or road block with your local school district or medical professional.
3. Take a refresher course on advocacy. Many local school districts and advocacy organizations offer free advocacy training. This will be beneficial as you navigate the educational system for your child, and perhaps you can share it to help other children with special needs. Perhaps this will even inspire you to become an Awareness Coordinator for the Foundation.
4. Encourage fellow parents to sign up for the CoRDS Registry to create a database for researchers to better understand the syndrome and further educate medical professionals. Go to <http://bit.ly/29OBqz3> to sign up.

The CdLS Foundation strongly believes that your willingness to be engaged will be ever-rewarding. Taking the journey with us on behalf of your loved one will uncover untold courage, engagement with your heart and untapped wisdom.

Sincerely,

Bonnie Royster
Executive Director

Our Mission

The Cornelia de Lange Syndrome Foundation is a family support organization that exists to ensure early and accurate diagnosis of CdLS, promote research into the causes and manifestations of the syndrome, and help people with a diagnosis of CdLS, and others with similar characteristics, make informed decisions throughout their lives.

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Aging Research in CdLS



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

Advances in medical care, nutrition and fitness, along with early intervention therapy and ongoing physical activity, have led to individuals with genetic syndromes living longer into adulthood. For those with Cornelia de Lange Syndrome (CdLS), this was a significant area of research when heading into the new millennium. Although much information had been published about children with CdLS, not much was known about the syndrome during adulthood. Due

to the lack of information my team and I decided to focus on assessments of adolescents and adults with CdLS through the generosity of the hospital where I work, Greater Baltimore Medical Center (GBMC). Since 2001, we have held multidisciplinary aging clinics twice a year, and since its inception we've seen over one hundred individuals with CdLS and their families.

It is probably most important to note what we have not seen. In the oldest group of patients, there have been no instances of cancer and only several benign tumors. We have seen only a few people with hypertension (high blood pressure), and some with high cholesterol or lipids, and often there was also a family history of these. There has been one individual with heart failure, which was treatable, and no one with a heart attack or sudden cardiac death. To put this into perspective, according to the Center for Disease Control in Atlanta, 29% of the population in the USA has hypertension and 2-3% has heart failure—most adults with CdLS are exceeding these odds. Furthermore, according to the National Cancer Institute, 4.5% of all men and women develop cancer every year. Thus, our numbers for these in CdLS are very low, which is quite reassuring. Overall, we have found that adults with CdLS are very strong, healthy and generally disease-free.

We have noted continued involvement of several specific body systems. The gastrointestinal (GI) system is always one of the most commonly involved. Gastroesophageal reflux is present in nearly every individual with CdLS. If not, it should be assessed for in the event that there is new onset abdominal pain or a change in or worsening of behaviors. Symptoms can come and go. There also can be constipation or diarrhea, with poor bowel motility as the cause. Some GI complications can occur at an earlier age than in the general population. One such GI complication is Barrett's esophagus.

The nervous system also has continued involvement. Seizures may occur in adults if they were present in childhood, and occasionally if they were not. Intellectual disability and autistic features will persist into adulthood. In terms of psychiatric diagnoses, both depression and anxiety can develop, but incidence is difficult to assess. Many adults have some obsessive-compulsive traits, but very few have obsessive-compulsive disorder. Behavioral problems persist as well, sometimes amenable to behavioral modification or other treatments; these include outbursts, aggression and self-injury, and often subside in the 20's and older. The urinary tract and genitalia remain stable, and we have not seen kidney complications.

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Sleep and CdLS Research



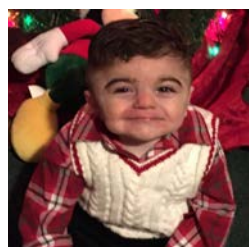
*By Stacey Ishman, M.D., M.P.H.,
Pediatric Otolaryngology, CdLS
Foundation Clinical Advisory
Board*

Studies show that 1-4 percent of children have moderate to severe sleep-disordered breathing (SDB); however, sleep disturbance is not well characterized in CdLS. In 2010, a team of researchers surveyed the caregivers of 22 patients with CdLS using three validated Sleep Questionnaires: Pediatric Sleep Questionnaire (PSQ), Pediatric Daytime Sleepiness Scale (PDSS), and Obstructive Sleep Apnea-18 (OSA-18). The questionnaires use a number of inquiries to measure symptoms of sleep-disordered breathing, daytime sleepiness, snoring, and inattentive/hyperactive behavior. Results of the study illustrated that 35-36 percent of those with CdLS had symptoms consistent with SDB; while, 23-36 percent of participants were characterized as sleepy.

Previous evaluations of individuals with Smith-Magenis Syndrome, who have similar levels of self-injurious behavior and intellectual disability as CdLS, suggest there is a correlation between sleep disturbance and insomnia or circadian rhythm disorders. In 2012, we collated the results of a sleep history questionnaire completed by the caregivers of 31 patients with CdLS to assess the prevalence of insomnia or circadian rhythm disorders. The results showed that the CdLS study population had significant difficulty falling asleep (75 percent pediatric, 33 percent adult) and difficulty staying asleep (52 percent pediatric, 33 percent adult). The data suggests that individuals with CdLS have a predisposition towards insomnia and circadian rhythm disorders; however, more study is necessary to better characterize the relationship between sleep disturbance and CdLS.

To further characterize this relationship, we are currently asking caregivers of patients with CdLS to complete a survey that more specifically looks at these symptoms. It is available in print or online at: <http://bit.ly/Mztf97>. We are also collecting sleep studies already carried out at home institutions. Please contact us with any questions or concerns at researchsleep@gmail.com.

A Closer Look at Sleep: Rocco



Thanks to Rocco's mom, Kim, for sharing his story

Rocco is a very active 18-month-old, with a contagious smile.

Since the beginning, we had concerns about Rocco's breathing. At 4 weeks old, which

was 2 weeks earlier than his original due date, Rocco became seriously ill with Rhinovirus. Shortly upon arriving at the ER he had several apnea episodes, in turn requiring intubation. Rocco spent the next 24 days in the hospital. During his stay, he had many desaturations.

After his extubation Rocco's oxygen desaturation level would occasionally drop into the 70s and 80s. A bronchoscopy and sleep study were inconclusive, and we were sent home with many doubts and concerns.

At this point, Rocco was not formally diagnosed with CdLS, although it was being considered. Following our scary start to life and his respiratory issues, I purchased a monitor that would measure his oxygen level and heart rate as he slept. Again, Rocco seemed to average in the low 90s and occasionally the high 80s. This made me feel very unconfident, and I wondered if the machine was accurate. Along with the fact that his breathing was noisy, his pulmonologist and I felt that a sleep study would be beneficial.

Getting Rocco to sleep with all those leads and wires was a challenge. He woke up throughout the night, but although they did not feel it was the best study, the specialists felt like they had enough data to make accurate conclusions. The doctor diagnosed him with a higher level of mild apnea. I was told that the apnea was categorized as obstruction, as opposed to central. The test would eventually have to be repeated to hopefully achieve a better study. However, his oxygen levels did remain above 95.

We were told to follow up with an ENT to discuss the causes of the obstruction. After a quick evaluation, the doctor determined it was not being caused by his tonsils or adenoids. He explained to me that it was most likely being caused by low muscle tone. They are not currently doing anything to treat his apnea and we will be repeating Rocco's sleep study in a few months to see how he is doing.



A Closer Look at Sleep: Zackery



Thanks to Zackery's mom, Tiffany, for sharing his story

At first, Zackery slept like a normal infant. Around 16 months, it all changed. He would sleep for 6 hours a day. It was in spells of 30 to 45 minutes or, if I was lucky, an hour. I would tell

his doctors at the appointments, but no one could give me any solution. I stayed up with Zackery and slept when I could so my husband could work. I slept on his days off, unless Zackery had a doctor's appointment. Finally, after 18 months of this sleep schedule, we saw a geneticist, Dr. Allen. He recommended that we use melatonin and see a pediatric neurologist. We used the melatonin and documented his sleep patterns; it worked most nights.

Three months later we went to the University of North Carolina Children's Hospital and met with Dr. Greenwood for pediatric neurology, who reviewed the sleep documents. He noticed that Zackery's sleep pattern was disrupted around the time of a full moon and diagnosed him with circadian rhythm disorder, but before we could treat it, he needed a sleep study. The study showed obstructive sleep apnea. We went back to the ENT to have his tonsils and adenoids removed, but they were only able to remove the adenoids due to Zackery's airway being so small.

Six months later we repeated the sleep study. This time it showed central sleep apnea, and Zackery was put on 1 liter of oxygen at night along with a pulse oximeter machine, which measured his oxygen saturation. It was a struggle, as Zackery would remove the nasal cannula setting off the alarm and I would have to replace it. I asked the pulmonologist, ENT, pediatrician, and neurologist PA if there was anything to help. We were finally referred to a pediatric sleep specialist at UNC Hospitals who recommend Diamox, and it has made a great difference. After using Diamox for a month, a sleep study showed that he only had half the episodes as before, and we were able to eliminate using oxygen at night.

My suggestions for parents is to listen to your instincts. Keep telling the doctors what is going on until one listens.

CdLS Registry: Enroll Today!

There are so many questions that accompany a diagnosis of CdLS. Questions you have are probably the same as many other families across the country, and world. To get answers, medical professionals rely on information from families to guide their hypotheses and research. This is why the CdLS Foundation has established a CdLS Registry at the Coordination of Rare Diseases at Sanford (CoRDS).

CoRDS Registry

Coordination of
Rare Diseases at Sanford



You can help researchers progress forward into determining more about the causes and manifestations of CdLS. For those of you interested in participating in this registry, visit <http://bit.ly/CdLSReg> to begin the process.

Aging Research in CdLS Cont.

Menopause appears to be at a typical age. Men can have prostate enlargement earlier than the general population. For some people, the skin and face appear older than would be expected based on age. Hair tends to become grey slightly earlier than other family members. The bones can lose their density earlier than expected as well.

We have published recommendations for evaluations and assessments in adults, available through the Foundation at www.CdLSusa.org, by searching "adulthood." We also welcome you and your adolescent or adult with CdLS at our aging clinic in Baltimore!



Alternative Communication in the Education System



*Maurice Belote, Project Coordinator,
California Deafblind Services*

From the time I first became a teacher of children with combined deafness and visual impairment, there has been a focus on educators

and family members working together, not in isolation of each other or, worse yet, in opposition to each other. But what does parent/professional collaboration look like at the classroom level, and how can everyone work together to support a child's communication and language development? There is probably no other educational domain in which a strong partnership is more important than it is with communication.

I often tell families that one of the greatest gifts a school program can give a student is an effective, formal communication system that meets the child's receptive and expressive needs. What I mean by the word "formal" is not that the communication system necessarily includes symbolic modes such as speech or American Sign Language, but that the system is documented in writing and/or video. This documentation can be in the form of a personal communication dictionary, a personal passport, or as simple as a written list or a few short video clips.

The communication systems for students I serve typically include multiple modes (e.g., photos, line drawings, tactile name cues, objects, signs, spoken words, etc.). This may seem unusual because most other students are often characterized by one mode, such as saying a child is a "signer" or a "talker." In addition, for many children I serve, expressive and receptive communication modes might not be the same. For example, a child might respond to signed communication receptively but use pictures and a voice output device to communicate expressively. This is actually much closer to the way we all communicate. I may use speech as my primary mode but will revert to gestures, facial expressions, behaviors, etc. when I am fatigued, stressed, or feel like I am not being understood.

What can families expect from educational programs? First, they should expect a home visit by their child's teacher if the family is agreeable to this. Seeing a child's home

environment will help the teacher to better understand the whole child and will help the teacher understand the context of much of the child's communication. Second, a family should expect that members of their child's educational team will contact them to learn how the child communicates. This will be the time to find out if the child uses "home signs" or has other idiosyncratic ways of communicating. (Family members tell stories of making up a sign on the spot because they didn't have access to a sign language dictionary or app at the moment they needed to know a particular sign, thinking that the child wouldn't remember the sign. And then, much to their dismay, this is a sign the child never forgets and they have to spend years explaining this made up sign!) Families should expect that the child's communication system will be used consistently throughout the child's school program.

Families should also expect that the communication system will meet everyone's needs, so that words and phrases are included that the family, the child and the child's peers all want to communicate about. And lastly, families should expect that program staff will strive to expand the child's communication system and not be limited only to the words and signs the child knows and regularly uses.

For more information, check out the communication page from the National Center on Deaf-Blindness website: <https://nationaldb.org/library/list/14/> or contact me for ideas and strategies that have worked for others. I can be reached at mbelote@sfsu.edu.



A Closer Look at Alternative Communication: Lindsey



Our daughter Lindsey, now 5, was diagnosed with CdLS at 18 months through genetic testing. We live in Connecticut and were put in contact with our local Birth to Three office, which is run by the Connecticut Office of Early Childhood and assists families to meet the developmental and

health-related needs of infants and toddlers who have delays or disabilities. We began to learn ASL with a speech pathologist using basic signs to communicate hunger, feelings and needs.

When Lindsey turned 3 she qualified for our local elementary school's preschool special education program. While in the program, they gave her a school issued iPad and she began to use Proloquo2Go. It is an Augmentative or Alternative Communication (AAC) program that uses symbols and pictures for those who cannot speak.

I was very against her relying solely on a device for communication for the simple fact that if the battery dies, or she breaks it or we forgot it somewhere, what were we to do? What would the option be, to not talk to her? So I pushed for sign language and received resistance. Professionals told me that signing was not the best method because if she signed incorrectly, it would be working against her.

However, Proloquo2Go has been life changing, and the support and assistance we received has not gone unnoticed. Lindsey's special education teacher both speaks and signs simultaneously to her, which allows Lindsey to use a multitude of communication skills. Her Proloquo2Go fills in the gaps where she does not yet have the ability to communicate. Lindsey is now integrated in a regular education classroom with 1:1 support, plus her iPad.

We have seen a decrease in frustration both at school and at home with Lindsey being able to select pictures, letters and numbers to communicate her exact needs

and desires. She is able to participate in her classroom's morning meeting, telling the class the month and day as well as the weather outside. We are so thankful to have a program that is easy to use and school faculty that not only has the resources, but is educated and willing to help.

On behalf of all of our non/limited verbal sons, daughters, brothers, and sisters... We love you!

The Fernandez Family

Michael, Ashley, Lindsey & Lilliana

Equipment Exchange

The CdLS Foundation has researched existing programs that offer equipment exchange. The top sites that were user-friendly and inclusive of many types of medical and adaptive equipment are highlighted. We encourage you to visit the sites to see what is available or consider using them to offer equipment and/or medical supplies you no longer need.

You can find the sites posted at the bottom of the main page of our website www.CdLSusa.org under "Outside Links."



Raising Awareness Through Your Passions

As a parent or caregiver of someone with CdLS, it may feel frustrating at times to explain your child's syndrome over and over again. This is an unfortunate reality of the lack of information about CdLS known to the public. But don't get discouraged—do something about raising awareness! A good first step is to start at the source; present information to the child's peers.



While Riley Rissland participates in Special Olympics with his middle school, interacting with dozens of peers, his mother, Former Board Member Fran Rissland, also serves as a Girl Scout Leader in Alpharetta, GA. She recently used the opportunity to raise awareness about CdLS in a unique way:

“Being a Girl Scout Leader is such an honor! I have been my daughter's troop co-leader for the past 4 years and I absolutely love helping the girls build courage, confidence and character to help make the world a better place.

As I'm a certified group fitness instructor, one of our sister troop's recently reached out to me to assist in the troop's fitness badge. Of course I jumped at the chance, since fitness is my passion. I led this awesome group of 5th graders in a basic 'boot camp' style fitness training routine at a local park. The girls learned to use body weight awareness as they did push ups on park benches, sit ups and push ups on the grass, lunges down a trail, sprinting from one tree to the next, and running and walking through out the park.

When asked what I would like as a thank you for teaching them, I asked them to make a donation to the CdLS Foundation in honor of my son, Riley, who is 13 years old and has CdLS and they did just that!

Thank you Troop 12741 for allowing me to help your girls earn their fitness badge, spread awareness of CdLS in the community and for your donation to the CdLS Foundation.”

This is a great example of combining passion and awareness—performing regular day-to-day duties but adding a CdLS angle; and nonetheless to a group of children who may meet someone with the syndrome. We thank Fran for her efforts!

WELCOME NEW FAMILIES

Florida

Rachel and Michael and son Kaden, born August 5, 2016

Florida

Betsy and son Hunter, born March 3, 2009

Illinois

Dana and Matthew and daughter Evelyn, born July 6, 2015

Indiana

Leslie and Garrison and daughter Emma, born July 7, 2002

Maryland

Sharon and Justin and daughter Janayla, born February 28, 2006

Missouri

Wendi and Greg and son Jayse, born July 3, 2013

New York

Patrica and Andres and son Jose, born March 31, 2015

Ohio

Janelle and Johnny and daughter Aubrynn, born September 2016

Nicole and daughter, born May 15, 2015

North Carolina

Tianna and Ian and daughter Lauren born July 6, 2016



Awareness Day Preparation: It's Not too Late!

National CdLS Awareness Day is quickly approaching, and we need all hands on deck across the country for celebrations. There are numerous ways to get involved ranging from hosting a picnic, to giving a presentation, to having your local government officials sign a CdLS Awareness Day proclamation.

Can the Foundation count on you to raise awareness for CdLS on Saturday, May 13th?

Events

A simple way to celebrate Awareness Day is by hosting a picnic at your local park. Invite friends, family and professionals to celebrate; have everyone wear purple or CdLS Foundation swag as this helps make your group stand out. Make sure you're armed with awareness materials, too—there is no doubt people will become curious about your brightly dressed group!



Awareness materials are always available from the Foundation free of charge. Our extensive list includes fact brochures, publications, temporary tattoos, stickers and

more. We are even in the process of ordering new, fun promotional materials like sunglasses! Contact Samantha (smanns@CdLSusa.org) well in advance to make sure you have everything you need for May 13th.

Presentations

We have recently received many requests for talking points and ideas for presentations at events like business seminars and college courses. The possibilities are endless, but the best presentations are ones that mean something to you. Parents, does your child with CdLS often experience questions about his or her syndrome? Professionals, how did you determine a treatment plan for your patient with CdLS? A meaningful experience creates a gripping tale, and will captivate your audience into learning more about CdLS. The Foundation can help provide PowerPoint templates and ideas on how you can make it more about your personal experiences.

Proclamation Requests



In the past, families have reached out to government officials in their towns and states to proclaim the second Saturday of May as the official CdLS Awareness Day. This has led to meetings with governors, newspaper features and new awareness initiatives.

What could your request create?

All you have to do is identify your local officials—the Foundation can help with the rest. We have templates for request letters and official proclamations available for you to personalize.

Board Member and National Family Conference Chair Angie Young kick started her Awareness Day efforts early—and it's paying off.

Angie is currently in the process of getting her proclamation signed by Florida Governor Rick Scott. After a phone call from an aide in Tallahassee, all the materials were forwarded and now she is waiting for confirmation! If it gets approved, she hopes to receive the proclamation in a ceremony with Governor Scott, inviting all local media outlets.

Her second request was to the County Commissioners. Angie recently received an email asking if she could be present on May 2 at the Board of County Commissioners meeting. Of course, she said yes and will receive the proclamation at the official gathering.

All we can say is WOW—what a great job of taking initiative early and reaping the rewards. Angie's says, "It's really a matter of finding the right person to connect with."

Join Angie and take on a leadership role by hosting an Awareness Day activity for the Foundation on May 13.



Super Siblings - David and Michael



Thank you to David, Michael's brother, for sharing his story with us.

Sitting down to write this article might have seemed easy, but in a true sense it was difficult. There isn't anything I wouldn't do for my

brother but for some reason what to write was hard. Do I write about our relationship? Do I offer advice to other siblings? Do I retell the stories of long ago that most people already know? Do I write about new experiences or challenges? Talking about Mike is easy and, of course, always fun, so I decided to give you a little bit of everything.

Michael and I have always had a strong bond. Even though I no longer live close to Mike, each and every time I go back home to Geneva, N.Y., I always find time to spend with him. It doesn't take a lot to make Mike happy. He used to enjoy going out to Wal-Mart for stickers, but since losing his vision, he likes to stay home where it's familiar. He used to love to go to dances and boogie the night away with every girl he could meet, but a lot of noise just doesn't do it for him anymore. What hasn't changed is his love of ice cream sandwiches, stickers, and Barry Manilow—yes Frank Sinatra has taken a back seat!

Mike can only see shadows and movement now, so he is becoming more anti-social and having new behaviors. As a family we adapt to change, and Mike's needs have become more of a top priority. I have learned to have even more patience with him as he adapts to his new "situation." I know how frustrating it is for us to adapt to his "behaviors," but I can't even begin to understand what it's like for him to adjust and not be able to articulate what he's going through. Being Mike's stand by guardian has allowed me to understand more of the complexities of what there will be to deal with in the future. Having to process his needs and what would be best for him is easier now because of our bond.

Prior to Thanksgiving, I did not see Mike since the National Family Conference that took place in June. I

spoke to him on the phone often, but it's not the same. When I went to his residence to pick him up for dinner, my 45-year-old brother literally jumped into my arms and gave me the biggest hug anyone could imagine. This is something that he has never done before and now I know that he needs that physical contact even more now than ever. I put Mike in the car and played his Barry Manilow songs and sang to him—trust me I am no American Idol—but he was as happy as a child on Christmas morning.

Every time I am home or on vacation with the family, I am always learning something new about Mike. I share these experiences with my students at school so that they are able to see that there are differences out there and how to accept them. Mike has been to some of my schools so that my students can meet him and ask questions about him or CdLS. It's never just an ordinary day, as anyone with a child with CdLS can tell you.

I have been given the opportunity to work with this amazing Foundation because Mike is my brother. Volunteering at the Conferences has allowed me to meet and interact with amazing families and to mentor other siblings. I look forward to this opportunity every time it arises. As a sibling, we will never know "normal," because what we live with is our normal. There will be times when we can be frustrated, angry, upset, and even embarrassed, but I know that I wouldn't change anything or want to live everyone else's "normal." There will always be tough days but the greater days outweigh everything else.

I love my brother and there is nothing I won't do for him.

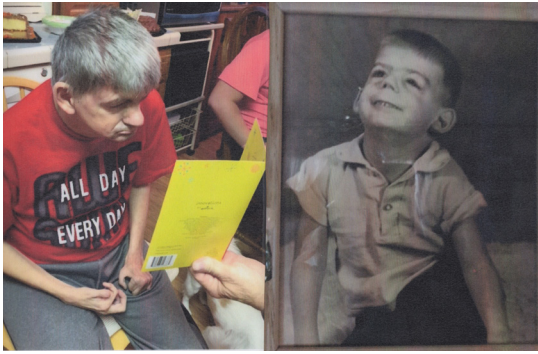
Share your Super Sibling Story!

Send your story and photo to
outreach@CdLSusa.org.



Mailbag – Stephen

Our family is one of a kind. Johnny Crawford Burchett was the second child of our four children; three boys and one girl. Johnny was born October 8, 1962. At that time, there were no schools or daycares for him to attend. Johnny didn't walk until he was five years old. He was around eight or nine years old before a nurse said she had seen another child that looked like Johnny.



We started looking through books and there was a whole page on CdLS. As time went on, Johnny had a few health problems. The biggest problem was finding a doctor to look after him. Johnny attended Thomasville Pediatrics until he was fifty years old. When Dr. Williams retired, I had to find another doctor. Johnny is nonverbal and still wears diapers, but he lets you know what his needs are or what he wants.

Johnny was raised in our daycare, so I feel like he picked a lot up from the children there. With four small children, I knew I had to help my husband financially. I started the daycare in my home, which let me spend more time working with Johnny.

In 1984, I moved the daycare out of our home and into a building. It took some time adjusting for Johnny but he came around. He has bad reflux, which we try to keep under control with medicine. He loves to take a bath, but hates our swimming pool. Johnny also loves to stay in his room and do nothing. What he loves the best is lying on the front porch; the hotter it is, the better he likes it. When the weather changes and Johnny tries to go outside, he turns around and stomps through the house—he's mad!

The best advice I can give to parents is to love their children with CdLS and remember all children are different, and including those with CdLS.

Faye Burchett, Thomasville, N.C.

Share your Mailbag Story!

Send your story and photo to
outreach@CdLSusa.org.

Add a Spring to your Step.

May 7

2017 TD Five Boro
Bike Tour
New York, NY

October 8

2017 Bank of America
Chicago Marathon
Chicago, IL

October 21

2017 Baltimore
Running Festival
Baltimore, MD

November 5

2017 TCS
New York City Marathon
New York, NY



Join Team CdLS!



Getting Involved with Fundraising

Everyone who volunteers does so for a different reason. Some live for the sense of personal achievement, others may feel a sense of satisfaction, but all know that they are making a difference in an area that they are passionate about.

The volunteers at the CdLS Foundation have found creative ways to incorporate awareness and fundraising into their social networks, community and workplace. There are many ways you can join them to let the world know about CdLS and how they can support the Foundation. Here are just a few ways you can get involved:

Support Local Events

There are many events hosted by families and friends across the country; from golf tournaments in Missouri and Massachusetts to Brew and BBQ in Atlanta, from Bowl-a-thons in Kentucky and Indiana to 5Ks and walks in Georgia and Michigan. Our volunteers are always looking to have other families attend events or to join them in coordinating.

Create Your Own

Ready to start a fundraiser on your own? Here are a few ideas that volunteers have done in the past: ask a restaurant to donate a portion of sales for the day, host a wine or beer tasting, host a paint night, organize a kickball tournament, or ask guests to make a gift to the Foundation in lieu of birthday or holiday gifts. The sky is your limit.



Cornelia's Garden

Imagine a garden of wildflowers representing the hope that the CdLS Foundation provides children with CdLS. You can help spread that hope by requesting a Cornelia's Garden Kit from the Foundation. The kit includes seed packets and fundraising materials to be distributed to friends and family so that they can plant a wildflower garden and make a gift in honor of your loved one. All those who participate will be rewarded by the beauty of the flowers and knowing they helped a child with CdLS grow and bloom.



Dress Down Day

Get comfy for a good cause. It's simple to raise funds and awareness for the CdLS Foundation by organizing a Dress Down Day in your workplace. It's fun, easy and gets everyone in the group involved. We will provide everything you need to make this day a success.



Yard Sale Across America

If you're ready to eliminate clutter, do so while supporting a great cause by participating in the annual Yard Sale Across America. Get your neighbors, friends and relatives to donate items to your sale, or volunteer to organize a neighborhood sale, with proceeds benefiting the Foundation.

The Foundation has many templates for letters, auction and sponsor forms to help get you started. Contact Kristi Larson at events@CdLSusa.org or 800.753.2357 to begin raising awareness and funds—we will be with you every step of the way.

In Cornelia's Garden,
wildflowers bloom
and there is hope.

Plant hope today! Email Kristi Larson at events@CdLSusa.org or call 800.753.2357 to get started.





Board Corner



*By Richard Haaland, Ph.D., President,
Board of Directors; CdLS Foundation
Research Committee Chair*

The CdLS Foundation Board of Directors will gather in April for the first of its two in-person meetings of the year. As we move into spring, this meeting is always a great opportunity to spend two days discussing where the Foundation is and where it should go.

This year, we'll spend the first day reviewing 2016 and the first few months of 2017 to look back at the great successes of the Foundation such as: the National Family Conference, CdLS clinics, Team CdLS, family gatherings, updated and additional resources for families, and new families who reached out to the Foundation. This also gives us the chance to identify how we can take some of these accomplishments and improve upon them even more to better serve the ever-evolving needs of each person affected by CdLS.

The second day will allow us to take these ideas and generate concrete ways to make those improvements a reality. This discussion really focuses on building a plan of action for the future of the Foundation so we're able to continue expanding upon our successes, but also to ensure that we're laying the groundwork for great things in 2018 and beyond.

One of the great things about the meetings is that we get to spend time getting to know new staff and Board

members who we might only communicate with over the phone or through email. This year is particularly exciting with a number of new faces among the staff and Board to join in the discussions.

We're thrilled to welcome Bonnie Royster as the new Executive Director of the Foundation. She has a wealth of experience in both for-profit and nonprofit organizations that will help the Foundation grow to improve its operations, and find new ways to meet the needs of families affected by CdLS. This Board meeting comes after she has taken a few months to get to know the staff, Board members, volunteers, parents, children and adults that make up the CdLS community. After an early start in January, Bonnie now has her feet firmly planted in the Foundation, and it's a perfect time for us to gather and carve out new opportunities to better serve everyone affected by the diagnosis.

With new faces come new ideas and ways of approaching the needs of the CdLS community. Each new person, whether Board Member, staff, volunteer, parent, or adult with CdLS, brings their own experiences to the discussion. Sharing these experiences is how we identify different ways to connect with each family affected, and meet their needs and those around them. These guiding principles help improve current services and ensures that we can develop innovative programs to better serve the CdLS community.

Please take a few moments to reach out to Bonnie, staff or Board Members to share your thoughts and ideas so we can start the process of creating an exciting future for the CdLS Foundation.

2017 CALENDAR

April 29
Kansas Family Gathering
Wichita, KS

April 30
Georgia Family Gathering
Johns Creek, GA

May 13
CdLS Awareness Day
Nationwide

May 21
Midwest Region Family
Gathering
Mount Prospect, IL

May 22
25th Annual CdLS Charity
Golf Tournament
Ipswich Country Club
Ipswich, MA

June 25
West Region Family
Gathering
Portland, OR

September 16
Northeast Region
Family Gathering
Wakefield, RI

October 9
29th Annual CdLS Charity
Golf Tournament
Sunset Hills, MO

November 4
Multidisciplinary Clinic for
Adolescents and Adults
Baltimore, MD



Donations from 1/5/17 - 3/6/17

Gifts that Count - In Honor/Celebration

Adam Jackson
Sara Lair

Annie Beaumont
Rita Deck

Barry Caudill
Melissa Kim

Brianne N. Prada
Denise Gelsimino

Brynnlee Beekman
Wiladene E. Heaston
Mary Dehaven
Bonita Boxell

Carolina Amor
Priscilla Amor

Conrad Clemens
Liselotte Davis

David S. Leonard
Lisa & Stephen Leonard

Elle Ponder
Melissa Ponder

Eric M. Loftus
Annie Fetzer

Ethel Silverman
Lori Chevette
David Hoffman
Ida Rosenberg
Karen Waxman
Preston Harcup

Frances Wein
Diane Friedman

Grace Duke
Alison C. Duke

Jacob Weber
Keiko Nishiyama

Jake A. Marcus
Carrie Radomsky

Julie Champion
Winifred Connor
Gael Hoysgaard

Kelly S. Brown
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of Directors

Kennedi Ballard
Adcolor Inc

Parlay Social
Anita Fisher

Laura N. Dupuis
Jill Dupuis

Lillian M. Choong
Marjorie George

Linda Berger
Ronald Schanz

Lindsay Franco
Susan Coleridge

Logan Fowler
Christl Schambach

Luca C. Barone
Monica Ahrens

Maggie Johnson
Jennifer Johnson

Marny Borchardt
Lenora Borchardt

Meaghan O'Keefe
Colleen O'Keefe

Megan J. Stockwell
Brights Funds Foundation

Myrna Kaplan
Diane Miller

Nathan Larsen
Reta Larsen

Nicole M. Cardona
Juan Cardona

Nicole Roberts
Alisha Capeless

Olivia Roberts
Lois Braden

Ryan S. Elphingstone
Susan Elphingstone

Sarah Suttman
Michael Voegle

Sofia Teixeira
Andrew C. Roth

Trinity Malone
Nancy J. Malone

Tyler Macy
Heidi Macy

Vincent Pulice
Lewis Pulice

Wilder Thompson
Anna Wilson

In Memory

Allison Tryba
Veronica Tryba

Annabelle Schwindler
Mary Sue Elliott

Camille Rodriguez
Clara Rodriguez

Devon J. Downs
Bonnie LeMaster
Jennifer Watson

Emily K. Johnson
Elizabeth H. Johnson
A. M. Johnson

Francis Richardson
The Candor Faculty Association

Ida S. Friedman
Ellen Braverman

Isabel E. Lander
Arthur Lander & Anne Calof

Jameson L. Fischer
Scot & Stephanie Fischer

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Sara Peracchio
Carol K. Phillips

Trevor Michael Hertzfeld
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William G. Hanselmann
Helen A. Miller



Our Deepest Sympathy



Jared Miller
May 23, 1978 – January 3, 2017
Son of Jan and Glenn Miller
1935 Wehr Ave.
Allentown, PA 18104

Trevor Michael Hertzfeld
February 15, 1999 – December 26, 2016
Son of Beth and Brad Hertzfeld
8765 Heller Road
Whitehouse, OH 43571



On the Cover: Jaxon



Tiny little fingers and tiny little feet...Jaxon was born on December 31, 2015 in Providence, Rhode Island. He was diagnosed with CdLS clinically at birth, where he spent 28 days in the NICU. He was later diagnosed through genetic testing and has the *NIPBL*

gene mutation. Jaxon lives at home in North Providence with his mommy, daddy, grammy, grandpy, and his two siblings. Jaxon has a big brother Cameron, who is 9 years old, and a new baby sister, Avery, who is 2 months old.

Jaxon is known for his amazing smile that lights up the room. He loves to cuddle and play with all his toys, especially his giraffe guitar and Henry the helicopter. He loves to have "dance parties" with his daddy and play peek-a-boo with his mommy. He is not sitting up on his own yet, but he is rolling around all over the place and beginning to babble. He loves to laugh and be tickled, but dislikes being put down on his back and having his hair brushed—but you wouldn't know with his signature spiked hairdo!

His smile is truly magnetic. He's fantastic at using his two little thumbs to maneuver his toy cars and is soothed when he sucks his thumb. Every day is an adventure with our Jaxon.

Logo Usage

With a large influx of fundraisers originating from parents and family members, we thought it would be a good idea to remind everyone of our branding and logo usage policies.

Please be advised that the logo of the CdLS Foundation is trademarked. Before starting a fundraiser using the logo, you **MUST** get the design approved by the Public Affairs Committee to avoid infringement. We are so thankful for everyone's dedication to raising funds and awareness, but please remember to take the proper steps to ensure your efforts follow protocol.

Don't be afraid to bring it up to your Facebook "friends" as well—they may not know about the policy and you could help by encouraging them to contact the Foundation. Thank you to our CdLS community for your continuous fundraising efforts, and for remaining vigilant on this matter.

Find answers to frequently asked questions on our branding page or contact the Foundation at any time for additional assistance.



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.
302 West Main Street #100
Avon, CT 06001-3681
800.753.2357 • www.CdLSusa.org

Yes, I would like to contribute towards the reimagining of the iconic *Reaching Out* newsletter!

Enclosed is my tax-deductible gift of:

- ☐ Other \$ _____ ☐ \$500 ☐ \$250
☐ \$100 ☐ \$50 ☐ \$40

☐ I have included the CdLS Foundation in my will or trust.

One hundred percent of your contribution is tax deductible.



☐ Please charge \$ _____ to my credit card

☐ Once ☐ Monthly for _____ months

Charge my gift to: ☐ VISA ☐ MC ☐ AMEX

Card #: _____

Expiration: _____ Security Code: _____

Print name on card: _____

Signature: _____

Mailing address: _____

Email address: _____

Donate online at www.CdLSusa.org.

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National CdLS Awareness Day

May 13, 2017



Each year, parents, volunteers and friends request official Awareness Day proclamations from governors and local leaders; hang awareness fliers in public places; make presentations to civic groups or health workers; write their local newspapers; and much more.

For information, social media graphics and ideas for CdLS Awareness Day, visit
<http://www.cdlsusa.org/what-we-do/awareness-day.htm>

National Awareness Day
 **CdLS**
Cornelia de Lange Syndrome Foundation

May 13th, 2017