

# Reaching Out

The Newsletter of the Cornelia de Lange Syndrome (CdLS) USA Foundation, Inc.  First Quarter 2015

## Highlights

### Director's Message:

Looking Back

### Spotlights:

Travel Training for  
Increased Independence

Twins and CdLS

### Super Relatives

Katie and Jenna





## Director's Message



### Looking back...

The CdLS Foundation ended the year on a high note, both in regards to programs/services and fundraising. Here are some of the highlights, many made possible due to our steadfast donors and volunteers:

- We've welcomed 121 new families to the Foundation, bringing the total number of families we serve nearly to 2,700. Many of these family members were among the nearly 600 people who attended a record eight Foundation-sponsored family gatherings and three non-sponsored throughout the country.
- Santa Clara Valley Medical Center in San Jose, CA, became the home of the third CdLS clinic in the country and the first one west of the Mississippi.
- The Foundation hosted 133 families at its 27th National Family Conference in Costa Mesa, CA, in June.
- The CdLS Foundation retired the block logo and launched its new heart logo on National CdLS Awareness Day.
- Three research projects shared \$33,750 in funding. The scientists will look at the cellular functions of the genes involved in CdLS, which may lead to further understanding of how the syndrome happens and the therapies that could improve the quality of life for those affected.
- The Foundation partnered with the Coordination of Rare Diseases at Sanford (CoRDS) to launch a CdLS Registry. This patient-reported database will help the Foundation staff and researchers identify clinical trends, among others.

I'm excited to see what 2015 has in store for the organization, and for you all. The Foundation staff is ready to tackle new projects and continue to grow our various efforts.

Marie Concklin-Malloy  
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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## West Coast Clinic has Arrived



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

By the time you read this newsletter, I will have returned from the CdLS Foundation's first West Coast multidisciplinary clinic. Organizing this took over a year, but was well worth the wait.

The first multidisciplinary settings were held at the national conferences, organized initially by Dr. Laird Jackson at Jefferson Medical College in Philadelphia, with whom I trained. Professionals who could join the conference were able to meet individually with families. The initial specialists included genetics, ophthalmology, speech pathology, dentistry and psychiatry.

This model has continued at the national conferences, although specialties have expanded into 17 different fields. Assessments of aging and adulthood in CdLS and gathering families with older children and adults to meet together were the goals when I established biannual free multidisciplinary clinics at my hospital, Greater Baltimore Medical Center (GBMC), in 2001, now comprised of 12 specialties. In 2009, The Children's Hospital of Philadelphia (CHOP) set up its multidisciplinary Center for CdLS and Related Diagnoses, where various specialists throughout the hospital see attendees. Thus, these opportunities were localized to the mid-Atlantic East Coast.

We explored hospitals in California, and identified the Santa Clara Valley Medical Center as the perfect site. The pediatric geneticist there, Dr. Robert Wallerstein, did his fellowship under Dr. Jackson and had personal experience in evaluating individuals with CdLS. He has put together a team of five to six specialists, some of whom attended the national conference in Costa Mesa this past June. Clinics will be held twice a year, for all ages. Trained parent volunteers will be present and represent the Foundation, which is done at CHOP clinic as well.

We anticipate wide interest and further development of expertise as the clinic grows. We recognize the many benefits of attending a clinic, including receiving personalized recommendations that can be shared with the individuals' local specialists. We are extremely grateful to everyone who has helped bring this about, and we look forward to future opportunities to set up similar clinics in other areas of the United States! A recent survey conducted by the CdLS Foundation showed that more than half of the participants cited adding regional clinics as their number one desire.

For more information on CdLS clinics, call 800.753.2357.

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## Travel Training for Increased Independence



*Linda G. Shepard Salzer, Ways2Go Travel Trainer*

Identifying and accessing appropriate services becomes a priority when a family member receives a diagnosis, as you or your family member approaches transition age, as caregivers age, or if you plan to relocate to another city. Many of you probably already know about and take advantage of early intervention, speech therapy, physical therapy, occupational therapy, and more.

In the search for resources and services, you may or may not have heard of travel training. Travel Trainers teach seniors and persons with disabilities how to safely use public transportation. Schools, social service agencies, and transportation authorities, as well as a variety of other organizations may provide travel instruction.

Travel training generally refers to individualized one-to-one instruction, based on the individual's abilities, needs, and preferred destinations. In addition to information about the transit vehicles, stations, and rules, travel trainers also cover subjects like street crossing, trip planning, safety, and more.

Travel instruction refers to orientation, familiarization, and travel training. Some organizations provide all three, others provide one or more of the above. Some programs provide small group or travel buddy options.

Travel trainers realize that individuals have different learning styles and learn at different paces. Generally, trainers practice routes with the individual multiple times, if needed. Skilled practitioners create or use a variety of approaches and teaching tools based on the individual's needs. These may include route cards or booklets with words, images, or a combination of the two; color coded destination cards; number matching cards, and more.

Some individuals may focus on specific routes, to and from school or work, for example. Others work on generalizing those skills and teaching trip planning skills.

Safe travel on public transportation also involves time management, safety, contingency planning, self-advocacy, and personal responsibility—all helpful skills as one becomes more independent.

Learning to safely use public transportation in cities and towns where it's available increases an individual's independence and options for getting to work, school, and social activities. The ability to use public transportation increases one's ability to get and keep a job. Other benefits of using public transportation include flexibility, independence, increased self-esteem, and saving money.

While many individuals, even those with significant challenges, can learn to safely use public transportation, some individuals may need to work on safety issues or social skills before beginning travel training. Organizations which provide travel instruction can help you determine if and when travel training would be appropriate for you, a friend, or family member.

For more information about Travel Training please visit the Association of Travel Instruction website: <http://www.travelinstruction.org/travel-training>. The Consortium for the Educational Advancement of Travel Instruction (CEATI) also has information on their website: <http://www.ceati-travelinstruction.org>.



## A Closer Look at Travel: Valerie's Story



*We want to thank Dianne, Valerie's mother, for sharing Valerie's story with us.*

Valerie has traveled most of her life, starting with a road trip to Florida with us when she was six months old. She made nine trips cross-country and back by car and/or motorhome, and has been in all 50 states, Canada, Mexico and many places in the Caribbean.

She went to visit her cousin in Virginia when she was 12, and flew by herself. I was at the gate in Boston, and my sister was at the gate in Virginia. Everything went well and she was very proud of herself. She made her second flight by herself to Florida to visit her grandmother. I was at the gate in Boston and my mother was at the gate in Tampa, but Valerie was on a plane that was touching down in Baltimore. This was a little worrisome for Valerie even though she didn't have to get off of the plane and she ended up getting sick on the plane, but did fine on the trip back.

For many years, Valerie, has done a lot of traveling with a group called "Trips R Us." They provide recreation for special needs people. She went on many road trips with this group. One particular group of friends used to go on a trip each summer called "Roller Coaster Dudes." They would ride to different states to visit amusement parks that had great roller coasters. These trips were made by van. They visited places like Busch Gardens in Virginia (along with visiting Williamsburg), Six Flags in New Jersey, and Cedar Point in Ohio.

Valerie then started going on trips where the group would fly to different places. A few times she went to Florida, and also to Hawaii. Cruises were then added to the mix, the first one from Seattle to Alaska on the Inside Passage (she's made it to Alaska before we did).

These trips above are in addition to all of the traveling she has done with us including a trip through the Panama Canal when we also visited Mexico, Costa Rica, and Columbia.

She usually tells me about the plans for her next trip when we are picking her up from the airport from her current trip. Ask Valerie if she wants to travel somewhere and she responds with "when should I pack my bags?"

Once Valerie was in her apartment and on her own, DDS (DMR) provided her with training to go on commuter rail and the regular "T"(train) in Boston. I forget how many sessions she had. She uses the T, and also has access to "The Ride" where she calls up and sets up a pick up. They pick her up at her apartment and she goes places like the mall, or to visit a friend. She has also gone a number of times to the Boston Museum of Science with her friend. One time the driver tried to drop her off at the Museum of Fine Arts because he didn't know the difference. She wouldn't get out of the car and told him he was at the wrong place.



Photo credit <http://www.tripsrus.org>

One funny story was when Valerie asked me if I thought she could get a job in Boston. I said, no I didn't think she could take the train and get around Boston to wherever her job might be. One Saturday I received a call from Valerie on her cell. I asked her what she was doing, and she said "you probably don't want to hear it." She said, "a friend and I are in Boston at Faneuil Hall." I said, "how did you get there?" She said, "We took commuter rail. Now we're going over to where all of the vegetables are at (Haymarket Square)." I asked how she was going to find her way back to North Station where the commuter rail is. She said, "We are just going to follow the (Zakin) bridge." She made it back fine, and sure showed me didn't she?



## Twins and CdLS

*Sarah Noon, M.S. and Mary Pipan, M.D., Children's Hospital of Philadelphia*



Twin pregnancies account for a small percentage of all births; however, over the past few decades the rate of multiple pregnancies has been rising. According to the National Center for Health Statistics, in 2012 the overall twin birth rate was 33.1 per 1,000 total births in the United States which has risen by over 70% since 1980, due to the use of assisted reproductive technologies.

Identical twins (monozygotic twins) develop from a single embryo, which is created from the union of a single sperm from a man and single egg from a woman, then divides into two embryos. Monozygotic twins are mostly genetically identical, thus being very similar in looks, physiology and development. Fraternal twins (dizygotic twins) result from two separate eggs that are fertilized by two separate sperm. Fraternal twins on average share half of their genetic material, which is the same amount shared between any two siblings with the same biologic parents.

The occurrence of Cornelia de Lange Syndrome in twins is rare but possible. Though CdLS is a genetic diagnosis, it is rarely inherited from a parent to a child, but rather is the results of a sporadic mutation of a gene related to the cohesin complex. These mutations could occur during the formation of the sperm, the egg or after the embryo is formed.

Coping with twins can be challenging, and raising twins of whom one has a significant disability can be even more stressful, depending on the level of resources available to the family. The differences in development and behavior from an early age will be much more obvious to a parent in a twin pair, similar to the differences seen when siblings are close in age. One study showed that most parents in these situations consider their twins to be siblings rather than twins. Such differentiation likely helps parents accept

each child for who they are, rather than having unrealistic expectations of either.

Coping then becomes similar to how families cope with any sibling group in which one has significant challenges. Coping strategies include:

- Consider each child as unique, recognizing their strengths, and needs. This means having different expectations, based on their individual capabilities, not the exact same expectations.
- Encourage siblings to see the strengths in each other and find activities that all family members can enjoy together and make this a regular part of the week.
- Arrange for each child to have their own activities independent of the other. The difficulty with twins or siblings who are close in age is the expectation that they will do everything together and not allowing for individual differences.
- Give each child time alone with a parent. Arrange to divide and conquer if they are in a two parent household. In a single parent household, use relatives, family friends, play dates, or babysitters to do this.
- Allow each child his/her own space in the house, which is off limits to other siblings. This can be a bedroom, but if you don't have the space, it could be a desk, or a cordoned off area in a common living area.
- Avoid making the typical child the 'grown up' too early. They can and should take part in helping their sibling, but not to the point of giving up their own childhood.

### **Coping with Twins Discordant for Intellectual Disabilities: The Mothers' View.**

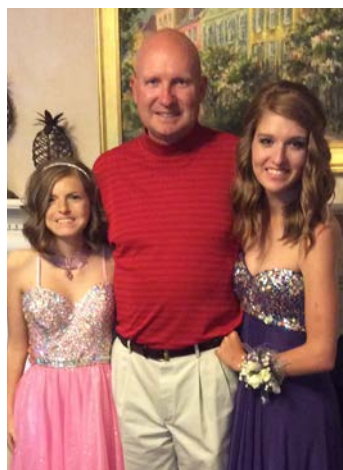
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*Sarah Noon serves on the Foundation's Clinical Advisory Board; Mary Pipan serves on the Professional Development Committee.*





## A Closer Look at Fraternal Twins: Grace & Susannah



*We want to thank Beth, Susannah and Grace's mother, for sharing their story with us.*

Having twin girls, Susannah who is typically developing, and Grace who has CdLS, had its positives and negatives as the girls grew up. One benefit we found was that Grace always had a typical peer to model after, which helped her meet a lot of milestones that she might not have met

as quickly. It gave her so much more determination. If Susannah stood up or climbed, Grace wanted to do it also. We clearly saw her trying to do what her sister was doing. However, when Grace couldn't do what her sister was doing, it was heartbreaking.

Things were tough at the very beginning, like when Grace was in NICU, but Susannah couldn't go in – we just tried to manage both twins. Those times when all babies need lots of attention would be tough because Grace needed a little extra attention. Some days could be really hard. My husband, Glenn, and I call it the “foggy years”—when the kids were young and it was a little crazy (Grace and Susannah have two brothers too). We don't have a lot of baby pictures or pretty baby books, or vacation albums. We were just trying to manage.

We'd be out in public and people would ask their ages. When I told them they were twins, they'd say, “That can't be.” We'd get a lot of funny looks because there were size and facial differences, so I started carrying CdLS Facts brochures in the diaper bag because people would ask.

The girls are definitely good friends. They like to listen to music and watch movies together. Susannah has that watchful spirit for her sister. When she introduces Grace as her older sister (she was the first twin to emerge), Grace really likes it and always lights up.

## A Closer Look at Identical Twins: Jack & Ethan

*We want to thank Cindy, Jack and Ethan's mother, for sharing their story with us.*



Jack and Ethan were born in April 2008 and are identical twins. They were clinically diagnosed with CdLS at three months. Their blood tests are negative for any of the known genetic mutations. We went on to have another baby boy in 2010 (Max), and he is typically developing.

After the twins were diagnosed with CdLS and I had some time to process my feelings of “how does this happen... times two?” I was grateful that the boys would always have each other. As they get older and their differences are more apparent, or if they have difficulty making friends, they will always have a blood buddy who will love them unconditionally.

The twins met most of their early milestones within weeks or a couple of months of each other. It was so great to see that. One would start crawling, then the other, then you would see the two of them crawling into trouble together! Then it was cruising, followed by walking. We could get lost in those happy moments and forget all about CdLS. Sending them off to school together, and watching them integrate into the school system together was so exciting!

Twins are difficult, special needs or not. Having twins with special needs might not be twice as difficult, but it certainly presents its own set of challenges.



## Research Grants Program Updates

The CdLS Foundation commits funds annually to research in order to find treatments to help individuals with CdLS overcome many of the challenges they face. The following are results from research that was funded by the CdLS Foundation in 2013:

*Treatment of Zebrafish Models for CdLS with L-leucine, Jennifer Gerton, PhD, Stowers Institute for Medical Research (Missouri)*

My lab at the Stowers Institute for Medical Research in Kansas City studies the basic processes within living cells that, when functioning improperly, can lead to human diseases like Cornelia de Lange syndrome. In our recent studies, we have used zebrafish with characteristics similar to human CdLS patients to study the disease and search for potential treatments. The work funded by the CdLS Small Grants Program has focused on treating these affected zebrafish with a common and nontoxic amino acid called L-leucine. Our study showed that this treatment rescued many of the physical defects in zebrafish and improved proper cell function.

In some ways, this study was a natural follow-up to our earlier work on Roberts syndrome – a condition related in many ways to CdLS. We had previously shown that treating zebrafish that model Roberts syndrome with L-leucine rescued many of the cellular and developmental defects of that disease. Based on the success of our zebrafish study in CdLS, we will soon extend our work to a mouse model for CdLS. A postdoc in the lab, Baoshan Xu, received a CdLS foundation grant for 2014-2015 to continue this line of investigation in human CdLS cells, so we will provide you with an update next year.

The work funded by the CdLS grant has been accepted for publication in *Human Molecular Genetics*. Funding for additional experiments has been provided by the March of Dimes and the Stowers Institute for Medical Research.

*Airway and Anesthetic Telephone Survey for Children and Adults with CdLS, Yvon Bryan, M.D., Wake Forest Baptist Medical Center (North Carolina)*

The preliminary findings from the survey of families of children and adults with CdLS were related to problems

with airway and anesthesia. The challenges in younger children were related to problems with oxygenation and intubation, while in the older children and adults were with anesthetic challenges related to effects of different medications and agents used for general anesthesia. The unpredictable response to medications in patients were that certain sedatives made children more excited while in others it made them very sleepy.

In terms of airway management, there does not seem to be one anatomic feature that correlates to either having problems placing a breathing tube (intubation) and/or preventing problems with oxygen levels. The variability in the anatomic features of the head and neck in patients may also lead to the inability to be able to predict which patients or techniques prove to be safest or best.

There needs to be continuing research related to which airway and anesthetic techniques prove safest and provide the best outcomes in patients. Until then, families need to report both good and bad outcomes in order to create protocols and educational material for physicians and families.

*Mitochondrial Dysfunction in CdLS, Clair Francomano, M.D., and Antonie Kline, M.D., Greater Baltimore Medical Center (Maryland)*

We have been searching for causes for premature aging in CdLS that we have noted in our multidisciplinary aging clinics. Some research studies in animal models and human cell lines with mutations causing CdLS have shown a poor reaction to “oxidative stress,” with a higher build-up of unwanted byproducts from chemical pathways than in non-CdLS cells. This suggested the possibility that the mitochondria in the cells in CdLS are not functioning properly; mitochondria are involved in energy production, but also aging and cell death. If we found evidence for mitochondrial dysfunction in CdLS, then specific treatments could be proposed which might help some of the symptoms.

Our project looked at the function of mitochondria in 10 individuals with CdLS by chemically measuring, from blood and urine, breakdown products and some vitamins and cofactors for the reactions. We also wanted to assess for the underlying mutation causing CdLS in some of these individuals. We found all vitamins and cofactors to be normal and no lactic acid build-up (sometimes seen in mitochondrial disorders). Looking at the byproducts of





the oxidative pathways in the mitochondria did, however, show evidence for mitochondrial dysfunction, particularly in one enzyme complex, in about half of those tested. Future work is needed to confirm these findings.

*Heterozygous Drosophila Models for CdLS. Yaning Wu, PhD, Children's Hospital of Philadelphia (Pennsylvania)*

CdLS is caused by mutations in one of the five genes, *NIPBL*, *SMC1A*, *SMC3*, *RAD21* and *HDAC8*. Mutations in *NIPBL* are the most common and generally seen in moderate and severe CdLS patients. Mutations in the other four genes are usually seen in mild cases.

Our study has demonstrated that *Drosophila* Nipped-B (fly version of *NIPBL*) manifest deficits in learning, memory and sleep. In addition, nearly half of Nipped-B flies have abnormal brain structures, which may explain the learning/memory deficiencies seen in these flies. These findings confirmed that the human *NIPBL* and fly Nipped-B have similar functions in the brain and fly Nipped-B flies are a valid model for CdLS.

This fly Nipped-B CdLS model can now be used to investigate why mutations in *NIPBL*/Nipped-B are detrimental to brain development and provide means for searching potential therapeutic compounds to ameliorate such damage.

## CdLS Foundation Now Accepting Research Grant Requests

The 2015 Small Grants Program is now open. Funding is available to investigators at academic and research institutions in the United States. Fellows and graduate students are encouraged to apply. Overhead or indirect costs are not supported by these grants. The maximum funding request per application in 2015 is \$15,000. A Letter of Intent is required by April 15. Final applications are due May 15, with funding distributed the first week of September.

For more information, email [director@CdLSusa.org](mailto:director@CdLSusa.org) or go to [www.CdLSusa.org/research/grants.htm](http://www.CdLSusa.org/research/grants.htm) to review the guidelines and download the application.

## Stowers Institute now a CdLS Center for Excellence

The Stowers Institute for Medical Research received a five-year designation from the Cornelia de Lange Syndrome (CdLS) Foundation as a CdLS Center for Excellence. Stowers joins the ranks of other Centers for Excellence: Greater Baltimore Medical Center, Saint Louis University School of Medicine, Children's Hospital of Philadelphia, and the University of California, Irvine. The work at these institutions has changed the landscape of what is known about CdLS.



"My lab members and I are thrilled by this recognition of our research efforts," said Stowers investigator Jennifer Gerton, PhD. "I believe that understanding the basic biological function of the genes mutated in CdLS will reveal insights into potential therapeutic approaches."

In order to receive this designation, an institution must pursue research that provides a significant contribution towards understanding the science behind CdLS, demonstrate a strong knowledge base of CdLS, and commit to furthering scientific knowledge of CdLS through collaboration with others, among other criteria.



## Mailbag – Troy –

My nephew, Troy, has CdLS. He is 17 years old, and attends Cedar Falls High School with his sister. He recently got a job at a car dealership in the detail department as part of a school class that teaches life skills. He is preparing for life after high school, and we've watched him develop in all areas.



Troy loves cars and carries a large ring of keys everywhere he goes. He researches cars online and has a notebook that he keeps of all our relatives and friend's car registrations so he can look at them and remember them. Although he is very quiet around groups of people, he will call me and ask the year of one of my cars. He also will call his friends at the local car dealership to ask if his family can borrow a car while they are getting an oil change. He has a special relationship with his Grandma Carol. He enjoys visits at her house, and they have long conversations about cars, school and all his daily activities.

At the end of June this past summer, my brother Jamie, Troy's father, decided to run the Cedar Falls Iowa Sturgis Falls race in Troy's honor, and to assist the Foundation. In addition, a good friend of the family, Kevin, and I designed a T-shirt to wear at the race to raise awareness. We sold 70 shirts to friends and family and had a great turnout of people who ran and walked the race. The announcer at the finish line pointed out Troy over the loud speaker as he walked fast across the finish line with his family. She had noticed our shirts and wanted everyone to know who Troy was. We were all thrilled at the attention as everyone cheered his finish. It was a great day.

Jeff, Troy's uncle, IA

## Super Relatives: Katie and Jenna



I come from a family of people who look for ways to give their best towards everything that they hold dear. It's easy being a "Super Relative" in a family of such extraordinary people.

I wanted to do something for my family to assist us in understanding the needs of my cousin's daughter, Jenna, who has CdLS. Because of that, I am being awarded by an experience that was already rewarding and a Foundation that has already given so much for my family and me. On September 12, 2014, I published a children's book that donates half of its profits to the CdLS Foundation.

*Signing Together: A Guide to American Sign Language for Everyone* is available on Amazon, Createspace.com, and in store at Lift Bridge Book Store located in Saratoga Springs, NY. The book teaches a dozen vocabulary words in American Sign Language through illustrations and rhymes that encourage others to focus on assisting one another without judgment, resistance or self-regard. The CdLS Foundation is proof that when we work together we can overcome all obstacles.

During college I studied American Sign Language Through those lessons and a job where I assisted adults with cognitive and physical disabilities, I learned that American Sign Language could bridge the gap that Jenna and those in her life feel when we try to communicate. My goal is to find a way to communicate my gratitude to Jenna for the way that she has taught me and everyone that meets her how to put a small spark of kindness into the world and watch it glow.

Katie and Jenna, NY

**Share your Mailbag or  
Super Sibling Story!**

Send your story and photo to  
[bshepard@CdLSusa.org](mailto:bshepard@CdLSusa.org).



## Entrepreneurs Among Us

Whitney and Shelby didn't let CdLS get in the way of their entrepreneurial spirits. Both women have started their own mini businesses, selling items made by hand.

### So Whitney

Whitney started making and selling candles as a way to earn money to attend the CdLS Foundation Conference last summer. We didn't quite reach our goal but had great support from members of the CdLS Facebook page. Starting a candle business was all Whitney's idea. The name of her business is "So Whitney." They are 100 percent organic soy candles.



### Shelby's Dog Snacks

What started out as a simple craft idea one day has now turned into a thriving business for 21-year-old Shelby, who has CdLS. Shelby loves animals and adores her own dogs, Romeo and Ruby. She came across a dog snack recipe one day, and though it would be nice

to make some treats. Several neighborhood dogs got in on the snacks as well.

After making a few batches, Shelby realized she could make a profit and we supported her by supplying her with ingredients and special ribbons to make the packaging appealing. She has her own business cards and her own business name: "Shelby's Dog Snacks." She could make dog snacks every day of the week to keep up with demand, but also has other activities such as a job at a local pizza place as hostess, horseback riding and art projects.

## WELCOME NEW FAMILIES

### Arizona

Roxanna and Julio and daughter Anna, born September 21, 2014

### New Jersey

Padmini and son Kartik, born January 31, 2012

### Ohio

Melinda and Peter and daughter Abigail, born October 14, 2003

### California

Tina and daughter born March 3, 1996

Lynnette and Mark and son Connor, born June 10, 2014

### South Carolina

Tabatha and Cathy and daughter Skylann, born December 26, 2010

Martha and Matthew and daughter Kai'ana, born September 23, 2012

### Tennessee

Candy and daughter Neveah, born May 21, 2010

### Florida

Facundo and Diana and daughter Mia, born January 20, 2012

### Texas

Ashley and Jerry and son Toby, born November 11, 2013

### Georgia

Tulani and son Antonio, born October 8, 2014

Melissa and son Jayden, born March 3, 2005

### Louisiana

Kendra and daughter Nylah, born August 26, 2014

### Virginia

Amber and son Ryan, born February 24, 2014

### Minnesota

Lindsay and Matt and daughter Makayla, born January 29, 2009

## 2015 CALENDAR

### February 22

Hyannis Half and Full Marathon, 10k and relay  
Hyannis, MA

### April 11

Southeast Region Family Gathering  
Mobile, AL

### March 15

United Airlines NYC Half Marathon  
New York, NY

### May 18

23rd Annual CdLS Charity Golf Tournament  
Ipswich, MA

### March 28

West Coast Region Family Gathering  
Scottsdale, AZ

### August 24

27th Annual Golf Tournament for CdLS  
Sunset Hills, MO

CdLS Multidisciplinary Clinic for Adolescents and Adults  
Baltimore, MD





## 5th Annual Brew & BBQ Benefit

Since 2009, the Brew & BBQ event has raised nearly \$20,000 to help people with CdLS live better, fuller lives. This year, the event raised \$7,400.



## Sarah's Bowl



In it's sixth year, Sarah's Bowl for CdLS was filled with bowling, a silent auction and raffle, face painting, cake and lots of fun! Held at East Bowl Family Fun Center in Batesville, IN, Sarah's Bowl hosted around 160 people and raised nearly \$14,000.



Special thanks to MHMRTarrant and Daybreak Community Services for funding the Texas family gathering in October.

## Bees in our Collective Bonnets

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

In 2010, hearing about the decline of the bee population nationally decreasing by a third each year, my husband, Doug Clemens, was spurred into taking action. The implications of this national phenomenon are far-reaching since bees are important pollinators of most food crops. Helping maintain beehives became Doug's mission: he and our youngest child, Conrad, became urban beekeepers. They took classes, learned practical aspects,



bought (big and little) bee suits, and became certified.

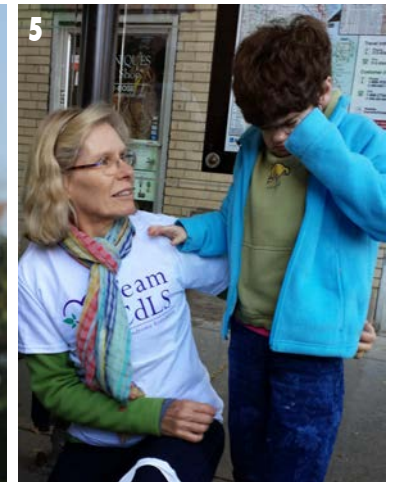
Baltimore City has an ordinance that allows only two hives and one nucleus colony per property, and is considered to be "at the forefront" of urban beekeeping. Doug now helps maintain 17 beehives in and around Baltimore City. He does everything he can to maintain them each winter; last winter in Maryland nearly 60% of all beehives were lost.

For his bar mitzvah project in 7th grade, Conrad chose to sell honey from the hives to benefit the Cornelia de Lange Syndrome Foundation. Conrad and Doug have continued to harvest and sell honey every year to benefit the Foundation. Doug has branched out into making beeswax candles and sells these as well. In addition to selling to neighbors and friends, the honey and candles have been sold at our temple, at regional CdLS gatherings including in a park, and at the pasta dinner before Team CdLS Baltimore ran in the Baltimore Running Festival this year!





## Thank you Team CdLS 2014!



 **Team  
CdLS**  
Cornelia de Lange Syndrome Foundation

1. TCS New York City Marathon Team
2. Warrior Dash Pennsylvania
3. Saratoga Palio
4. Baltimore Running Festival
5. Bank of America Chicago Marathon

6. Baltimore Running Festival
7. Bank of America Chicago Marathon
8. Merrell Down & Dirty National Mud Run Series
9. Baltimore Running Festival



## Board Corner

*Bob Boneberg, Esq., President, Board of Directors*



As usual, there is a lot happening with the Board of Directors that I would like to bring you up to date on:

A number of us were able to attend the National Family Conference in California in June. While there, many family members shared their thoughts, ideas, and hopes for the Foundation. A number of you were thinking about long-range research projects whereby we could learn more about CdLS and the best medical, educational, behavioral, and other interventions. We are very grateful for all comments and are considering them carefully as we go about our work.

Meanwhile, the Board's Strategic Planning Committee is continuing its work. Our current strategic plan is through 2015. We are evaluating our goals and plans in order to build upon and improve a strategic plan for the next three years. More about our current strategic plan can be found on the Foundation website. As always, please feel free to contact us with your thoughts and comments as to our goals, plans, and activities during 2016, 2017 and 2018.

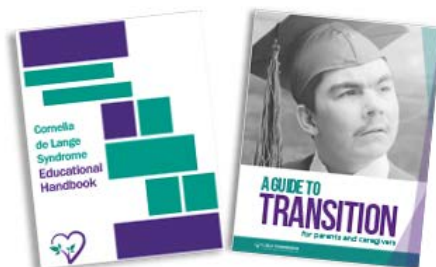
Also, we have been thinking about a longer-range vision for the Foundation. Put simply, if the Foundation could be anything we wanted it to be in 10 or 20 years, or perhaps even longer, what would it look like? What would it have achieved? What would be our priorities? Do you have any ideas in this regard? Perhaps a new direction for research? Or new or expanded services for families? Again, any thoughts would be appreciated.

As you may know, Board members are limited to serving two consecutive three years terms. As a result, we usually add and lose members every year. At the end of 2014, Eileen Ahearn, who is one of our most insightful and hard-working members, completed her term. Eileen will be greatly missed by the Board, but we are grateful that she will continue to be involved with the Foundation. Yet, we are very pleased that three new members will be joining us in 2015: Eric Johnson, Lynne Kerr and Pat Lyons (read more on page 15). We look forward to working with them and receiving their insights and guidance.

I would like to thank each of you for your continuing support for the Foundation. And, of course, I continue to welcome your thoughts and suggestions.

Best wishes always,  
Bob Boneberg  
[president@CdLSusa.org](mailto:president@CdLSusa.org)

## Get your **FREE** School Toolkit!



Includes the *Educational Handbook*, *A Guide to Transition* and a USB with our awareness video

To request a kit, contact Deirdre Summa at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org) or call 800.753.2357





## New Board Members

The new board members begin their three-year terms January 1, 2015.



### **Eric Johnson, Danbury, CT**

Eric has served on the Foundation's Finance Committee for several years taking part in budget and financial oversight. He and his wife, Nancy, began their involvement with the Foundation in 1989 when their daughter, Emily, was diagnosed with CdLS. Emily died at age three from medical complications, and the family—which includes 22-year-old son, Anders—has maintained a relationship with the organization throughout the years.

Eric recently retired from the IBM Corporation after more than 35 years. His last position was chief financial officer of IBM Global Business Services. Prior to that, he served as a chief financial officer in IBM divisions in Japan and China and held a number of financial and operations roles in IBM's hardware, software and services brands. Eric has extensive experience in management, budgets and forecasts, business controls and planning.



### **Lynne Kerr, M.D., Salt Lake City, UT**

Lynne is a pediatric neurologist and member of the Foundation's Clinical Advisory Board (CAB). She has provided consults at conferences, contributes to Reaching Out and Ask the Expert, and co-authored the CdLS module for the Medical Home Portal, hosted by the University of Utah. She is currently working on a study on autonomic nervous system dysfunction in CdLS.

Lynne and her husband, Hunter Jackson, are the organizers of Maddy's Run 5K in honor of their daughter, Madelyn, who had CdLS and passed away at age 18 in 2010. The event is a family affair, with Maddy's three brothers participating.



### **Pat Lyons, Portsmouth, NH**

Pat works for Dell, Inc. as a channel account manager and is very active in his community, including his local PTA, Rotary, and the Portsmouth High School Business Education Collaborative. He also serves on the Workforce Investment Board and the Youth Council of the state of New Hampshire.

He and his wife, Karen, have three children, including Luke, age 6, with CdLS. Pat is a member of the CdLS New England Golf Tournament Committee and he has cheered on his wife during her two marathons with Team CdLS.

## Did you know?

The CdLS Foundation is here to listen and help. If you have a comment or suggestion, let us know using our new online suggestion box, located on the homepage of the web site.



## Jack & Ethan cont.

First, there are all of the doctor/specialist/therapy appointments times two. Most offices understand the need to co-schedule, but some do not and make it very difficult for us to try and schedule appointments efficiently.

Second, while the co-appointments are critical to our sanity, many times we find the boys get bundled together as one unit when they have very different CdLS characteristics and needs. It's constantly a challenge to maximize our appointments while being sensitive to each boys' uniqueness.



Third, sometimes our services also get bundled, instead of considering each boy as their own unit. We are grateful for every benefit we receive, but sometimes I wish they would provide separate benefits for each boy – they certainly make us complete all of the paperwork times two!

Fourth, with as difficult as it is trying to maintain their individuality while still leveraging overlapping appointment/therapy schedules, it gets even more complicated when their needs start to diverge. Jack has an eye condition (Duanne Retraction Syndrome) that requires appointments not needed by Ethan. Ethan was recently diagnosed with ADHD so he sees a specialist for that while Jack does not. The logistics of the separate appointments can be a nightmare as our care options are limited.

Finally, there's the constant worry – times two. All parents of children with special needs worry about their children's futures. We worry about who will care for them, what will they be capable of doing, where will they live, will there be enough money so they are well cared for? We worry

about that – times two. When we calculate how much money we want to set aside so our child will have a decent quality of life when we die, we have to multiply that times two. When we wonder who will care for them should we die unexpectedly we have to consider who could handle two children with special needs. The pool is very, very small. When we consider where they will live and what work, if any, they might be able to do, we have to factor in each child's uniqueness and abilities.

We identified and applied for all services for which we were eligible as soon as we could. Things like respite care and LNA care have been critical for our self-care. We also identified a trusted caregiver with special needs experience early on (we used a national online caregiver resource with great success). This allows us to get out of the house a couple of nights a month. I would like to say we take the opportunity to take a long weekend once or twice a year, but unfortunately we've only had the opportunity to do that twice in six years.

However, twins are just fun! The cute outfits, purchasing multiples of everything, the constant references to “double trouble!” Twins definitely generate plenty of attention. Their personalities and abilities complement each other more often than not. Jack is left-handed, and Ethan is right-handed. Ethan is a fine-motor expert, while Jack excels at gross-motor activities. Jack is an extrovert and very social, Ethan is an introvert and prefers solo activities. The school has equated it to “Yin and Yang.” They manifest their personalities – and CdLS – in very different ways.

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## Grace & Susannah cont.

Susannah is now a freshman in college studying communication sciences and disorders with a goal of becoming a speech therapist. She was inspired by how much speech therapy helped her sister. One of the challenges of being twins was that when Susannah went to college, Grace thought, “What's next for me?” Now, Grace has applied to a continued learning program at a university and is awaiting the decision. We're hoping she's ready for it, and that we're ready for it too.



## Autonomic Nervous System Dysfunction in CdLS Study

Researchers including Lynne Kerr, MD, a CdLS Foundation Board Member, Antonie Kline, MD, the CdLS Foundation Medical Director, and Amie Jones, MD, and Philip Fischer, MD, autonomic nervous system specialists from the Mayo Clinic, Rochester, MN, are conducting a survey. The autonomic nervous system is that part of the peripheral nervous system that controls functions usually below the level of our consciousness, such as digestion, heart rate, perspiration, and swallowing.

This is a preliminary study to gather information about autonomic nervous system dysfunction in individuals with CdLS. Symptoms may include reflux, dizziness, low blood pressure, skin mottling, temperature instability, and others.

Aggregated information will be shared with the families who have responded and with the CdLS Foundation in the hope that it will help individuals with CdLS and their families and their primary care providers in understanding autonomic nervous system dysfunction.

If there is reason to believe that there is substantial nervous system dysfunction in these individuals based on the information received, additional research is planned.

To participate in the survey, go to the CdLS Foundation web site, [www.CdLSusa.org](http://www.CdLSusa.org), click on Research, then Participate in Research.

## Upcycling for CdLS



Causes International, which upcycles electronics and donates proceeds to charities – including the CdLS Foundation – is accepting many electronic items you may have in your home or office. If your company or business has old computers, servers, phone systems, etc. to get rid of, the business gets a tax deduction and the CdLS Foundation receives funds. For smaller lots of electronics please refer to their Mail-In Product list for qualified product and shipping details. Causes International works with you to streamline shipping and packaging logistics. Packaging, pick-up, and shipping are underwritten by the Upcycle program. To learn more or request a pick up, go to <http://www.causesinternational.com/signup/pickup>.

## Join the CdLS Registry

The CdLS Foundation has established a CdLS Registry at the Coordination of Rare Diseases at Sanford (CoRDS). Patient registries are designed to collect, store and curate data on individuals to be used for a specified purpose. They can be tools to establish natural history studies, establish prevalence, and connect patients with researchers who study their conditions

The CdLS Registry at CoRDS houses basic contact and clinical information on any individual who chooses to enroll. Once enrolled, users receive a username/password and are able to update the registry online annually and choose to participate in approved research studies. Users have control over who can and cannot see the information.

What's the benefit of participating? Simply put, advancing the understanding of the syndrome and encouraging further research into its causes. Researchers with appropriate approval may apply to access an anonymous data set in order to determine if any individuals in the registry are eligible for their research studies. The information in the registry may even peak the researchers interest in CdLS or a certain aspect of the syndrome.

To learn more or enroll, go to [www.sanfordresearch.org/cords/patientsfamilies](http://www.sanfordresearch.org/cords/patientsfamilies).





Donations from 7/02/2014-12/01/2014

## Gifts that Count - In Honor/Celebration

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Dorothy and Daniel Steimke

### Ally Wineman

William Hart

### Amy F. Loftus

Karen Szukalski

### Andrea Needman

Diane and Michael Friedman

### Andrew Patitucci

Cindy and Robert Zamichieli

### Anna Shook and in memory of Chuck Shook

Genia Blackwell

### Aubrey Garigen

Jolene Burgess

### Barry Pugh

Cindy and Tom Trimble

Joe Churchman

### Benjamin Fisher

Martha and Robert Fisher

### Brandt Anderson

Fran and Wayne Shull

### Brenden Keating

Karin Csoltz

Susan and John Keating

### Camden Robbins

Teri and JD Robbins

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Diane and Michael Friedman

### Claire Ingold

Sally and Stanley Bell

### Diane Friedman

Lois Braverman

Ellen Braverman and  
Robert Fishel

Sandra and Stuart Kaufman

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Diane and Michael Friedman

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Anne and Mike Suttman

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## In memory

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Stephanie Smith  
Crystal and Jerry Smith

## On the Cover




Sydnee turned 10 on November 26. She is my “Little Diva” with a lot of personality. Sydnee is truly a social butterfly. There is not a lap that doesn’t have her name on it. For the past 10 years, she has given my life meaning and adventure. Sydnee is a handful, and there is never a dull

moment as her mother. She enjoys music, light up toys, long walks and sun bathing. Sydnee is thriving with the wonderful support from her school and therapy. Sydnee will put her ear to your mouth, as a request for you to sing with her, and lucky for me she isn’t picky if you can’t sing. Although Sydnee is non-verbal, she has mastered her version of the word “no.” Her level of communication has greatly increased with the use of “The Picture Exchange Communication System.” Sydnee also enjoys her time during her therapeutic horseback riding session. I am very excited to see where the next 10 years will take us.



### Our Deepest Sympathy



**Heather Perkins**  
October 14, 1981-August 4, 2014  
Daughter of Bonnie Ripani  
33 Bayview Ave  
Groton, CT 06340

**Megan Ramsey**  
October 27, 1978 – July 16, 2014  
Daughter of Diana Ramsey and Phil Gartlan  
7 Windmill Drive  
Morristown, NJ 07960  
and  
Allen and BettyLou Ramsey  
39 Meadow Drive  
Ephrata, PA 17522

## CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.  
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## 15 races for the 15th Anniversary

Team CdLS is celebrating its 15th anniversary in 2015. To honor the many years and many miles put in by runners, walkers and athletes across the country, we're aiming to host 15 races in 2015.

Whether you run short distances for pleasure or are an experienced marathoner, Team CdLS offers the opportunity to set your own pace. Join us and you will walk away with new friends, a sense of accomplishment and the satisfaction of making a difference. To join Team CdLS, to start your own event, or for more information, please contact us at [TeamCdLS@cdlsusa.org](mailto:TeamCdLS@cdlsusa.org), or call 800-753-2357.

