

# Reaching Out

The Newsletter of the Cornelia de Lange Syndrome (CdLS) USA Foundation, Inc.  Fourth Quarter 2014

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The National CdLS  
Registry is Here!

### Spotlights:

Selective Mutism and  
Social Anxiety in CdLS

Physical Therapy  
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### Super Siblings:

Easton





## Director's Message



### The National CdLS Registry is Here!

Patient registries are designed to collect, store and curate data on individuals and their medical conditions. They can be tools to establish natural history studies, establish prevalence of conditions, and connect patients with researchers.

A national CdLS registry has been on the Foundation's to-do list for many years. In 2010, we were selected to participate in the pilot program of the National Institute of Health's (NIH) Rare Diseases Registry. Unfortunately, the program's funding was cut by the NIH in 2013 before it came to realization, leaving the Foundation with a survey and no home for it.

Determined to keep the momentum going, we researched other options, deciding finally on the Coordination of Rare Diseases at Sanford (CoRDS). CoRDS was established in 2010 to serve as a central resource for data on rare conditions with the aim of accelerating research into those conditions. The CdLS Registry at CoRDS will house basic clinical information on individuals diagnosed with CdLS who choose to enroll. Once entered into the registry, participants 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 CoRDS to access anonymous data in order to determine if any individuals in the registry are qualified for their research studies. The information in the registry may even peak researchers' interests in CdLS or a certain aspect of the syndrome.

If you are a parent/caregiver or person with CdLS and would like to participate in the registry, go to the CoRDS web site at [www.sanfordresearch.org/CoRDS/enroll](http://www.sanfordresearch.org/CoRDS/enroll) to begin the process. Whether you enroll by mail or via the online portal (set to launch this fall), you will receive a username/password and be able to update the registry online annually, choose to participate in approved research studies, and eventually see anonymous data on others in the CdLS registry.

If you have questions about the registry, contact me or go to [www.sanfordresearch.org/cords/faqs](http://www.sanfordresearch.org/cords/faqs).

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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# What a Difference a Difference Can Make



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

We are all different. Our differences contribute to our growth and development, passage into adulthood, relationships, family dynamics, child-rearing and education. Having a syndrome, or having similar characteristics due to a single underlying cause, may cause individuals to resemble each other more than other family members, and often have specific medical

complications in common. It is important not to be too quick to judge others, based on superficial characteristics.

This is, of course, true for CdLS. Physically, in CdLS, there are typical facial features, an abundance of hair, including excessive hair on the extremities and back, and often smaller body size, including small hands and feet. There are, however, many potential differences among individuals with CdLS, and this was probably evident at the recent national conference in Costa Mesa in June. Some children will have more hair than others, or be bigger at a similar age than others, or have smaller hands or missing fingers.

We now know that CdLS is due to mutations in more than a single gene (actually, five known currently), and it is partly because of this that the differences exist. But family genes are also present and contribute as well. Medical involvement can vary greatly, and some children can be born with a life-threatening problem, develop severe reflux, or have difficulty managing seizures as they age. This is also true for developmental skills. Although almost everyone learns to walk, not everyone with CdLS does. The majority of children with CdLS have the most difficulty with speech and language, yet there is a huge range of verbal abilities. In addition, behavior varies greatly. It is often the children who have mastered developmental milestones earlier than others, who will develop more severe behaviors. Despite learning to walk relatively early and communicating or speaking well, these children may manifest behavioral issues. Furthermore, self-injury or aggression can worsen, particularly in adolescence, and lead to other behavioral complications.

Thus, when observing other children with CdLS at the national conference, online, or at regional gatherings, please do not compare your child to the others. It would be too difficult to assess that the other children are “more severe” or “less severe” than yours based on a few hours or days of observation. It would not be fair to any of them to determine that their issues and complications have more or less effects on their families’ lives. A severe heart defect can cause just as many hours of worry and anxiety as severe self-injury, but neither may be evident externally. Our national conference should be a time of joining together and acquisition of knowledge, and each child and family deserves their place alongside one another. Be open, be fair and be empathetic with all, and all of our differences will play a more positive role.

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Incorporated December 1981

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## Selective Mutism and Social Anxiety in CdLS



*Blake D. Hansen, Ph.D; David C. Ball, Brigham Young University*

Selective mutism is an anxiety-related disorder that is characterized by the persistent failure to speak when speaking is expected (such as school or social situations). It is rare in the general population, some biological conditions, such as those associated with CdLS

make it more likely that children will demonstrate higher levels of selective mutism and social anxiety than others (Goodban, 1993; Richards et al., 2009). Anxiety and other challenges are sometimes ignored in individuals with intellectual and developmental disabilities, these problems have correlations to other problems, including repetitive behaviors, aggression, and self-injury.

In 2013, our research group conducted a parent questionnaire to identify selective mutism and social anxiety characteristics in individuals with CdLS. Parents completed the Spence Children's Anxiety Scale (SCAS; Nauta et al., 2004) and the Selective Mutism Questionnaire (Bergman et al., 2008), a normed questionnaire that identifies characteristics and severity of selective mutism symptoms. All participants completed the SCAS and parents of children who are verbal completed the SMQ. In addition we requested information about challenging behaviors to look at relationships between anxiety, self-injury, and aggressive behaviors. We found that 15% of individuals with CdLS who are verbal are within the clinical range for selective mutism. In addition, we found that 61% of individuals with CdLS fell within the clinical range for any anxiety disorder. There was no relationship between anxiety levels and self-injury, however, there was a significant relationship between anxiety and aggression. These findings indicate that a majority of individuals with CdLS have anxiety disorders, and these anxiety disorders place individuals at-risk for aggressive behaviors.

Due to the limited presence in research literature, those interested in looking for treatments for these problems in CdLS should look at the extensive literature on the treatment of selective mutism for children with typical

development. There are several handbooks and treatment manuals (see green box on page 5). The main treatments are based on research-based assessment and behavioral treatment approaches.

When assessing selective mutism it is important to identify situations where the child will speak and where the child struggles. A description of the situations should include information about who is present and where the difficulty speaking occurs. It is also important to be certain that the failure to speak is not due to speech and language impairments rather than anxiety. For example, if a child does not know how to engage in back-and-forth conversation or the demand is too challenging, then the failure to speak is not necessarily selective mutism.

Behavior principles such as positive reinforcement, shaping, and fading are used to help children with selective mutism speak in more situations. In addition, there has been a large amount of research done on an intervention called video self-modeling. A complete description of these interventions would be too extensive for this publication, but the resources on page 5 may be helpful for clinicians in identifying some approaches to intervention for children with CdLS.

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## A Closer Look at Selective Mutism: Andre's Story

*We want to thank Glenda, Andre's grandmother, for sharing Andre's story with us.*



Andre is 15 years old. He talks normally to family members, but stopped talking at school, and to others he had previously talked to, two years ago. He was diagnosed last year with selective

mutism, an anxiety disorder in which extreme anxiety prevents children from speaking in some social settings. Andre probably had the anxiety since grade school where he would sometimes talk in a quiet voice or whisper.

Over the course of several years, Andre spoke in school less and less, and during an IEP meeting we were informed he wasn't talking at all at school.

Last year, I spent an hour twice a week in one of his high school classes to help him try to communicate in his class. He would whisper to me, but only when no one was looking. He was scared to talk for some reason. By the end of the year, he would quietly whisper a few words to the teacher or the assistant when I would ask him questions in her office.

Genetics of CdLS may contribute to his selective mutism, and the two ADHD medications he takes have side effects of anxiety. Andre also has social anxiety, as he doesn't like to eat in front of people, dance, swim, or other activities where there are a lot of people. We tried generic Prozac for a month to help with anxiety, but it caused his hands to tremble, so that medication was stopped. I plan to take him to a psychologist for help with the social anxiety and selective mutism.

I have researched information and found selective mutism experts say the most effective treatment is through exposure therapy to the anxiety to gradually overcome it. Teachers and others need to be trained to ask students with selective mutism forced choice questions or open-

ended questions, and to wait five seconds for a response. If no response, repeat or rephrase the question. They should avoid asking yes/no questions or mind reading. Parents shouldn't rescue the child by answering for them when asked questions. Some may believe the child is shy or being oppositional when anxiety is preventing them from answering.

More information I found useful about selective mutism can be found at the Child Mind Institute website ([www.childmind.org/en/sm-workshops/](http://www.childmind.org/en/sm-workshops/)), which has past and future workshops with videos and PowerPoint presentations.

His contact teacher and I plan to try again this year to do more to help him relax and talk at school. Hopefully, we will succeed this year, but it is harder to overcome in an adolescent who has had selective mutism a long time. It helps to have parent, teacher, psychiatrist, psychologist, and speech therapist to work together as a team to achieve the goal.

### Treatment Resources

#### Books

R. Lindsey Bergman, *Treatment for Children with Selective Mutism: An Integrative Behavioral Approach*, 2012, Oxford University Press.

Christopher Kearney, *Helping Children with Selective Mutism and Their Parents: A Guide for School-Based Professionals*, 2010, Oxford University Press.

#### Websites

Selective mutism group – Childhood Anxiety Network: <http://www.selectivemutism.org>

#### Research Studies

Kehle, T. J., Madaus, M. R., Baratta, V. S., & Bray, M. A. (1998). Augmented self-modeling as a treatment for children with selective mutism. *Journal of School Psychology*, 36(3), 247-260.

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## Physical Therapy Concerns and Answers



*Katherine Farr, D.P.T., Texas Children's Hospital, Houston, TX, CdLS Foundation Clinical Advisory Board Member*

After attending the Cornelia de Lange Syndrome (CdLS) conference this past year in California, I noticed a common theme for questions and concerns regarding physical therapy and CdLS. I chose to focus on two main subjects: 1) *Is Physical Therapy (PT) hurting my child?* 2) *How can I help him/her progress towards walking?*

First: PT does not hurt your child. While physical therapists are trained to know the limits of the child, it is important to note that you should also develop a relationship and good rapport with the therapist. Knowing the plan for therapy will allow for better understanding of when the child might be crying to manipulate the situation or when the child is truly in pain. The physical therapist has already performed a thorough evaluation of your child and knows the limits to which he/she can safely be pushed. It has been my experience that a patient crying during PT sessions is due to the patient being pushed past his/her comfort level rather than actual physical pain. In order for any child to progress, though, he/she must be pushed to levels higher than where he/she is currently functioning.

Second: Issues and concerns with walking. No one can predict when your child will walk; however, there are some key areas to focus on to progress him or her towards upright standing and walking skills. No matter the child's impairments (e.g. limb abnormalities) the key to developing upright standing and walking skills all points back to core strength. The child must be able to control his or her core in various positions (tummy, back, sitting, tall kneeling and standing) to allow for upright standing posture, balance and mobility to progress towards walking. Individualized treatment programs are best for your child; however some commonly seen treatment techniques and equipment used to increase the core strength include various positions on a dynamic surface (Physioball, BOSU ball, peanuts) with multi-directional movements.

To utilize equipment for incorporating balance with core strengthening, use items such as a platform swing, bolster swing, and the U-gallop machine (mimics movements of riding a horse). Utilizing gravity-assisted equipment such as the Universal Exercise Unit allows the child to be in various positions without the effects of gravity inhibiting them from attaining a position.

Please consult with your child's physical therapist prior to initiating any core strengthening activity to ensure that it is safe and appropriate for your child's current level of functioning. Each child's progress toward the goal for walking varies based on family support, patient motivation and severity of the syndrome.

No matter what advice you receive, always know that your child will only be as successful as you expect him/her to be. Remember, even when it seems that the ultimate goal is not being focused on (e.g. walking), PTs are training and preparing the child for the ultimate goal, just as an adult training for a marathon. There are months of preparation, even years in some cases, before a person competes in a marathon; the same holds true for a child with CdLS attaining physical milestones.

This is only a brief summary of a few of the PT concerns that have been brought to my attention. Each child is unique and differs from the others, requiring an individualized treatment approach based on his or her abilities.

*If you have any further questions, please contact Deirdre at the CdLS Foundation at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org).*

Katherine shadowed Professional Development Committee member Amy Metrena, M.S.P.T., during family consultations at the 2014 CdLS National Family Conference. We are always looking for other professionals to gain exposure working with individuals with CdLS.

If you, or a professional you know is interested in shadowing, please contact Deirdre Summa at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org) or call 800.753.2357.





## A Closer Look at Physical Therapy: Jordyn's Story



*We want to thank Jordyn's mother, Jennifer, for sharing Jordyn's story with us.*

From the moment Jordyn was born we were told that her legs were hypertonic, meaning she had very high muscle tension. This caused her to be extremely stiff from the waist down almost constantly and hindered her from sitting, crawling, or later, walking correctly. We started her sessions with a physical therapist shortly after the age of one to help her with these issues. At that time, she was getting visits from her therapist twice a week. I know this was a great help to her motor skills because not long after starting PT, she could finally sit up alone at 13 months old. Shortly after, around 18 months, she crawled, and then walked just before turning two. She has continued on with her therapy through the years to help her learn to do things like run, jump, and maneuver her body correctly. Currently, Jordyn is receiving PT once a week to keep her on track and moving efficiently.

More recently, we've noticed a stunt in growth in Jordyn's left foot. It being significantly smaller than her right foot causes her to lean in on one side and drag the other to walk evenly. This led us to request a consultation with the physical therapist, Amy Metrena, at the CdLS National Family Conference. Jordyn's feet were examined and Amy had tons of information for us regarding our concerns. We were shown exercises to do daily to help stretch both feet and given a recommendation for an orthotic; either a Dynamic Ankle Foot Orthosis (DAFO) from online or we could have one made specifically for Jordyn. She also suggested that we purchase more supportive shoe brands and watch for worsening symptoms so, if need be, we could see a specialist. This made us feel like we had options to work on the problem, rather than jump into something more invasive.

The advice and instructions were invaluable to us and I am so grateful to the Foundation for arranging the meeting and the therapist for donating her time.

## Get your FREE Education Toolkit

Thanks to a grant from the American Legion Child Welfare Foundation, the CdLS Foundation is able to provide a CdLS Education Toolkit for Parents and Professionals later this fall.

Elements of the toolkit include the Foundation's Educational Handbook for parents and teachers, CdLS Management Guideline cards, and the new CdLS informational video on USB. The toolkit helps ensure children with CdLS have a positive experience in their school setting, and educates teachers and other school personnel about best practices for teaching children with CdLS.

Contact Deirdre Summa at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org) to request your FREE Education Toolkit today.

Or, for families of teens and adults, ask for our newly redesigned Guide to Transition.

## Don't throw away those lost teeth!

Dr. Lawrence T. Reiter at the University of Tennessee Health Science Center in Memphis is conducting a study to find out if nerve cells can be grown from the dental pulp of people with various neurogenetic conditions, including Cornelia de Lange Syndrome (CdLS).

Nerve cells are the key cells that function in the brain. If you take part in this program, you might help researchers develop new treatments for patients with autism and autism related syndromes, like CdLS.

To help with this research, you will be asked to answer several questions about your child's genetic status. Dr. Reiter will provide a tube of cell growth solution and a prepaid return envelope to you at no cost. If you agree to take part, you will only need to provide a fresh tooth, either extracted or one that fell out on its own. The tooth should be placed in the cell growth solution and mailed right away. **The tooth must arrive at Dr. Reiter's laboratory no more than 48 hours after the time it came out of the mouth.** To request a kit or learn more about taking part, please email [lreiter@uthsc.edu](mailto:lreiter@uthsc.edu).



## Mailbag

### – Lessons in LifeSkills –

Kerrie Kuntz, Special Education Teacher at John Jay High School in Katonah, NY, has worked with students in the LifeSkills program there for three years. One of her students is Luca (pictured right side), who has CdLS.



“Everyone deserves an education,” said Kuntz. “Our goal is to get the students in this program to be as independent as possible, giving them as fulfilling a life as possible. That may mean that the students need to understand that they learn differently, learning about their challenges and understanding their abilities and how that can be applied in the future.”

The LifeSkills program works with students ages 14-21 who have multiple disabilities (cognitive and/or physical), and works on vocational and everyday skills, such as cooking, laundry and speech.

“We try to break the habit of having someone else do things for them,” said Kuntz. “The students also work on vocational skills, such as counting money, making a purchase, telling time and reading a schedule.”

Kuntz explained that many businesses in the community have opened their doors to these students.

“At Kelloggs & Lawrence Hardware, students stocked shelves,” said Kuntz. “That involved looking at a paint can, understanding that it was orange paint, and finding the other cans like it. At other businesses, they might sweep, bag groceries, and much more.”

Kuntz also shared that there is a social and community aspect to the class that is very important.

“It’s also about socializing these students into the

community too,” said Kuntz. “They need to feel integrated into the community and community members need to meet them as well.”

Thank you to family friend, Alice, for sharing Luca’s LifeSkills class’ experiences with us.

## Super Siblings: Easton

One sibling stood out at the CdLS Foundation National Family Conference as he went around asking for donations on the night of banquet:

“I heard a little voice,” said Deirdre, staff member at the Foundation. “It was Easton, holding up a pale and asking if I wanted to make a donation. I asked, ‘what are you collecting money for?’ and he said, ‘for kids with CdLS, so they can get medicine to feel better.’”



“I put in my donation,” said Deirdre, “and only a few moments later, he circled around, and was back in front of me, saying, ‘I’m trying to reach \$100...’ I laughed, and put more money into his bucket.”

Easton presented \$75.17 to Executive Director Marie Malloy at breakfast the next morning. He made sure she included, “and 17 cents” as she announced his wonderful fundraising total.

### Share your Mailbag or Super Sibling Story!

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

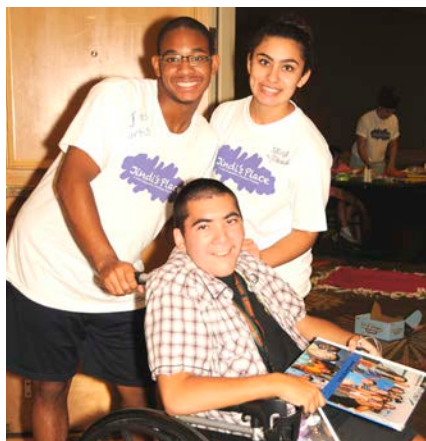
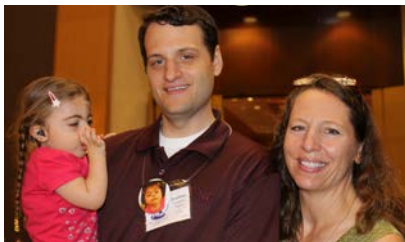




# 2014 CdLS Foundation National Family Conference

*We dreamed. We explored. We discovered.*

For those who attended, and those professionals who donated their time and efforts to support the Foundation, we say thank you!



# 21<sup>st</sup> century

CONFERENCE FUND

**CdLS Foundation**

Cornelia de Lange Syndrome Foundation, Inc.

Our eight-year-old daughter, Hannah Greyson “Gracie” lives a wonderful life. When she was diagnosed, we could never imagine uttering those words, but today we are amazed how well Gracie has succeeded despite her challenges.

The 2010 National Family Conference in Dallas, TX, was a life-changing experience for us and was critical to Gracie’s future. Today, we ask you to help us support the 21st

Century Conference Fund, a fund established to ensure that conference is here for families in the future. Our family will match all donations, up to \$10,000.

We know first-hand, how important it is to support the Foundation’s efforts and feel responsible to help them continue. This initiative provides a means to secure the financial stability of this event and it will continue to grow because of your care.

*David and Christine Fry, Gracie’s proud parents, Charlotte, NC*

**This year the Fry family will match your gift, dollar for dollar, up to \$10,000, making your gift have twice the impact.** Your gift not only is greatly appreciated, but it also connects a community of people that share a common bond: CdLS.

To have your donation matched, donate online and choose *21<sup>st</sup> Century Conference Fund*, write *Match it!* on your check or call 800.753.2357.



## de Lange Society Class of 2014 Inducted

The de Lange Society induction was held June 29 at the Crowne Plaza Costa Mesa Orange County. We also honored those in our 2013 class who were unable to attend the event last April. Additionally, co-founder of the *Reaching Out* publication, Carol Power (formerly Carol Welsh), spoke to an audience of 50 people.



Congratulations to the de Lange Society members, class of 2013 & 2014 (listed left to right): Joanne Gersuk, Angela Young, Dick Mungo, D.D.S., Karen Prada, Hunter Jackson, Ph.D., Jan Abbott, Lynne Kerr, M.D., Jim Abbott, Julie Gonella, Roy Gonella, Jim Kenney, and Susan Drexler.



Garth and Molly Black receiving the Sue Anthony Award at the CdLS Foundation National Family Conference.

## Award Recipients Honored at 2014 Conference

The CdLS Foundation relies on our volunteers around the county to successfully implement many services and ideas. The following individuals were recognized for their years of dedication and efforts to create new programs, enhance scientific endeavors and provide a significant impact on the Foundation and the families we serve.

### **Sue Anthony Award: Garth and Molly Black**

The Sue Anthony Award is the Foundation's highest award and is named for our primary Founder and the co-ordinator of *Reaching Out*. Sue Anthony represented innovation, commitment, integrity, and enduring contributions to families living with CdLS.

### **John King Award: Eileen Ahearn, M.D., Ph.D.**

The John King Award is presented to an individual whose long-term efforts on behalf of the Foundation have made a notable impact.

### **Special Recognition Award: Elizabeth "Jo" Bruen, Julie Gonella, and Maureen Feighan-Kurth**

Special Recognition Awards are presented to individuals or organizations whose efforts on behalf of the Foundation have made a specific impact.

### **Dr. Laird Jackson Award: Matthew Deardorff, M.D., Ph.D.**

The Dr. Laird Jackson Award, inspired by the Foundation's first medical director, recognizes an individual's significant medical or scientific contributions toward understanding CdLS and improving the lives of people affected by the syndrome.

### **Outstanding Philanthropy Award: Jim and Jen Pomfret, and Frank and Denise Teixeira**

The Outstanding Philanthropy Award recognizes outstanding individuals or organizations whose long-term contributions of financial support, time and talent have made a significant impact for individuals with CdLS.





## Event Highlights



### Battle of the Bands

IPC Systems Inc. hosted its 7th annual Battle of the Bands on July 10 in New York City and tapped the CdLS Foundation as its charity of choice, raising \$15,000. Lisa and Mike Lewin (pictured above), parents of Lauren who has CdLS, connected the Foundation to IPC.



### Dirty in Philly

A little mud didn't scare off the Team CdLS mud run crews near Philadelphia, PA, this summer. Runners participated in the Merrell Down & Dirty race in July and the Warrior Dash in August. Combined, the teams raised over \$30,000 in honor of Andrew, Connor and Howie, and in memory of Layla Ann, Layla Ann's father, Shawn, and other family members are at the finish line pictured above.

## 2014 CALENDAR

### October 4

Maddy's Run  
Salt Lake City, UT

### October 25

Texas Family Gathering  
Dallas, TX

### October 12

Bank of America  
Chicago Marathon  
Chicago, IL

### November 1

Multidisciplinary Clinic  
for Adolescents and Adults  
with CdLS  
Baltimore, MD

Walk for Will and Hope  
Shelby Township, MI

### November 2

TCS New York City Marathon  
New York, NY

### October 18

Southeast Foundation  
Sponsored Family Gathering  
Sweetwater, TN

### November 9

Sarah's Bowl  
Batesville, IN

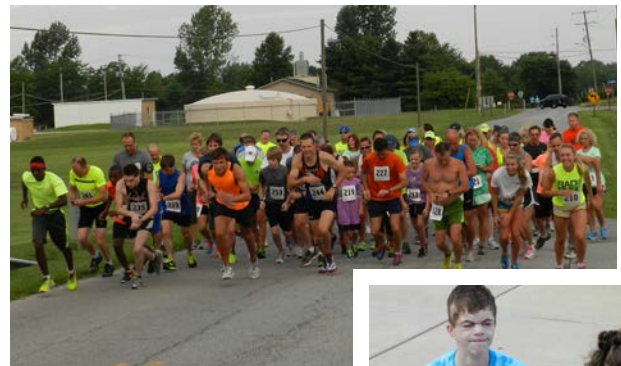
Baltimore Running Festival  
Baltimore, MD

### December 6

One Love, One Heart  
5k walk/run  
Decatur, GA

### October 24-25

Board of Directors meeting  
Dallas, TX



### Lapel Village Fair

For the 14th year, the Hersberger family and their community have come together during their town fair for a race supporting the CdLS Foundation and in turn Conrad and Sarah (pictured above), who both have CdLS.





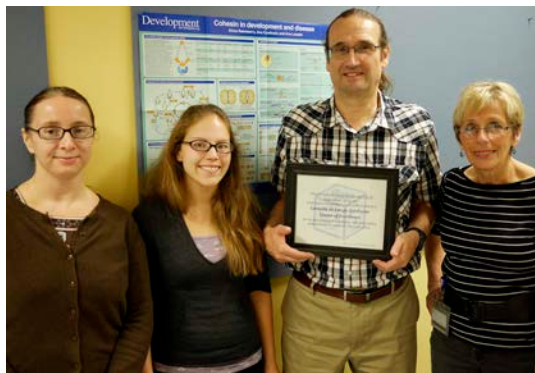


## Center for Excellence Profile: St. Louis University

*Dale Dorsett, Ph.D., Biochemistry & Molecular Biology,  
Saint Louis University School of Medicine, CdLS Foundation  
Clinical Advisory Board member*

Saint Louis University (SLU) School of Medicine's involvement with the CdLS Foundation has been primarily through me and my laboratory: We conduct research into the causes and possible treatments for CdLS, I have previously been a member of the CdLS Foundation Board of Directors, and I've chaired the Research Committee in the past. I helped obtain NIH funding to support the CdLS Foundation's Scientific Symposia held in conjunction with the national family conference.

Being recognized as a CdLS Center for Excellence has affected us primarily by keeping us more focused on the research questions most immediately relevant to CdLS. We do basic research on genetic mechanisms, and we can take many different directions. We now include potential relevance to CdLS as one of the criteria for setting research priorities.



We've hosted CdLS Foundation events at SLU, which included members of the CdLS Foundation Board, and donors from Saint Louis. I have given tours of our laboratory to board members and local interested families. My lab has contributed to all of the CdLS Scientific Symposia events.

Our status as a CdLS Center for Excellence has helped keep us focused on the ultimate goal of helping CdLS families. Although we have been recognized for our research into the basic molecular mechanisms underlying CdLS, this is work that we actually began before we knew the connection to CdLS. However, we have undertaken

projects aimed at searching for potential treatments, which is not something we would have done if not for our connection to the CdLS Foundation. For example, we obtained supplemental grant money from the NIH to conduct a screen for FDA-approved drugs that might have beneficial effects using yeast and *Drosophila*. We have had promising results, and hopefully this work will be able to continue.

I think the future of CdLS research will be in multiple directions. There is still much to learn about the basic molecular mechanisms, which is what my lab does, and understanding these will likely be very helpful in developing therapies. There may be more genes involved, and so the human genetics of CdLS will continue. There are many things we still don't know about the physical and mental aspects of CdLS, including behavior, and there is a strong need for more research into these areas, which could be very beneficial for helping families.

## New West Coast Clinic Opens

The CdLS Clinic at Santa Clara Valley Medical Center in San Jose, CA, plans to open its doors this fall. The core members of this team will include a geneticist, genetic counselor, neurologist, gastroenterologist, psychiatrist, nurse, and a social worker. A gynecologist will also be available. The clinic will be held twice annually and is open to all ages.

Can't make it to California? Two existing CdLS clinics are available:

- The CdLS Multidisciplinary Clinic for Adolescents and Adults at Greater Baltimore Medical Center (GBMC) is for individuals ages 12 and over. The clinic is held in the spring and fall. The next one is Saturday, November 1.
- The Center for Cornelia de Lange Syndrome and Related Diagnoses at the Children's Hospital of Philadelphia (CHOP) meets the third Thursday at each month and is open to all ages.

For information on any of the clinics, contact Deirdre Summa at 800.753.2357 or [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org).



## Board Corner

*Bob Boneberg, President, Board of Directors*



It is sometimes convenient to think about the CdLS Foundation's plans as falling into three broad categories. First is the annual business or operational plan, which sets forth in detail exactly what is to be achieved during the coming year. Second is the strategic plan, which sets forth in more general terms what the Foundation

wishes to achieve over the three years or so that are the subject of the plan. The strategic plan will often reflect new programs or new emphasis. Third is the strategic vision, which attempts to develop an understanding as to where the Foundation should be going during the next 3-10 years.

The current strategic plan, which is intended to provide direction for 2013-2015, was developed after much thinking, many conversations, and input from many people. Those who attended the Conference in Lincolnshire, IL, may remember attending one of the strategic planning focus groups. Our current plan placed a new emphasis in some areas of the Foundation's activities. For example, we are working to expand the number of regional and other family gatherings. We are also directing more resources to support research.

Now, the Board is working on the strategic plan for 2016-2018. Once again, we need and welcome your thoughts and suggestions. Should we expand our support for research? How can we best support families? Is there a better way to educate and inform professionals about CdLS? Perhaps most exciting are ideas that might change our vision of what the Foundation will be 10 years from now.

Whatever your thoughts may be, please share them with us so that together we can build a Foundation that will best serve all the CdLS families now and in the future.

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

## WELCOME NEW FAMILIES

### California

Flor and Julio and son Adam,  
born December 2, 2010

Christina and son Greyson,  
born January 24, 2012

Lauren and daughter  
Emberbeth, born  
February 10, 2014

### Georgia

Maresa and Quantazius and  
son Hendrix, born  
July 13, 2014

### Illinois

Tara and John and daughter  
Colbie, born May 8, 2014

### Kansas

Erin and Matt and daughter  
Madison, born  
February 18, 2014

### Minnesota

Valentene, born  
January 6, 1992

### New Hampshire

Crystal and daughter Sarah,  
born April 22, 2009

### New Mexico

Laura and daughter Melanie,  
born January 13, 2011

### New York

Terry and Joe and son Sammy,  
born May 27, 2011

### Oklahoma

Heather and Jeremy and son  
William, born  
December 17, 2012

Kate and Scott and daughter  
Alena, born October 18, 2011

### South Carolina

Sherry and Tyrone and son  
Tyrone, born June 7, 1986

### Virginia

Paula and Karl and son Will,  
born December 29, 2013

### Washington

Kiaira and Dimari and son  
Dimari, born March 24, 2014



Donations from 4/30/2014-7/02/2014

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Keith and Magdalena Cooper

Susan and Lyle Tryba

Veronica and Ernest Tryba

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## On the Cover

Mason is one of the happiest six year olds you can ever meet. His smile is infectious and he brings joy to everyone around him! Mason loves all music and any toy that lights up, and makes noise. Although, one of his favorite toys is his stuffed bird that tweets and chirps when he squeezes it. He loves to climb and hang off of almost anything he can get his hands on and is very persistent! Although he is non-verbal, Mason speaks volumes to everyone he interacts with, especially his big brother Reece! He also loves playing on his iPad. One of our favorite memories with Mason was in 2010, when we traveled to Dallas to meet other CdLS families for the biennial Conference. This fall, Mason will start Kindergarten in a special needs program at our local public school. We are positive he will continue to amaze us with his abilities and are excited for what is to come.

Megan G., mom of Mason

## Grieving Booklet Now Available

A booklet covering the topic of grieving the death of a child with CdLS is now available. Thanks to the generous time and efforts by many family members, this booklet has finally come to fruition, and is available to families who have lost a child. While this is a publication that we never want to give a family, we hope it provides comfort when needed. To request a booklet, contact Lynn Audette at [families@CdLSusa.org](mailto:families@CdLSusa.org) or call 800.753.2357.



### Our Deepest Sympathy



**Dré Hemphill**  
May 1, 1997 – December 12, 2013  
Son of La Gunda Brown  
1510 E. Beverly Rd  
Shorewood, WI 53211

**Andrew Schwalbe**  
June 28, 1990 – June 22, 2014  
Son of Karen and Jon Schwalbe  
2660 1st St N  
North St. Paul, MN 55109-1812

**Lacey Gray**  
January 30, 1988 – July 12, 2014  
Daughter of Belinda and Glenn Conyers  
and Bill Gray  
17 Bob White Ct  
Oakley CA 94561

**Skyler J.A. Hauck**  
April 16, 2004 – April 25, 2013  
Son of Boni Hauck and Justin Peterson  
P.O. Box 632  
East Helena, MT 59635

**Jason Kyle**  
June 10, 2000 – May 26, 2014  
Son of Sylvia and Jesse Kyle  
11408 Muller St  
Santa Fe Springs, CA 90670-4328

**Kelsi Emma Ahlfield**  
August 23, 1993 – June 7, 2014  
Daughter of Jennifer Burnett  
and Reggie Ahlfield  
166 Stagecoach Rd  
New Franklin, MO 65274

**Brandy Kay Neels**  
June 4, 1978- March 27, 2014  
Daughter of Ed and Sharon Neels  
368 S 46th Rd  
Dunbar, NE 68346-8802

**Robin Wakeman**  
April 19, 1976 – June 9, 2014  
Daughter of Glenda  
and Richard Wakeman  
6638 Aintree Cir  
Dallas, TX 75214



## CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.  
302 West Main Street #100  
Avon, CT 06001-3681  
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**Yes, I want to help people with CdLS.**

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The Circle of Caring is the legacy society of the CdLS Foundation that honors individuals who make bequests or other planned gifts to secure the Foundation's future.

For more information on leaving your legacy, call Marie Malloy at 800.753.2357 or request more information by emailing [director@CdLSusa.org](mailto:director@CdLSusa.org).



CIRCLE of CARING  
CdLS Foundation



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