

# Reaching Out

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



## Highlights

**Director's Message:**  
Change is Good

**Spotlights:**  
Quality of Life Findings

The Individualized  
Education Plan

**Super Siblings:**  
Matthew & Robynne



## Director's Message



### Change is Good

The Cornelia de Lange Syndrome (CdLS) Foundation has experienced remarkable growth and transformation over the past 33 years, especially in the past decade. In addition to providing services for families caring for babies and young children with CdLS, it's now serving a more diverse group of people with the syndrome, as well as older individuals with CdLS. With the Foundation's programs and services shifting to meet the needs of its constituency, it was time to change our block logo—first introduced in 1997.

In the summer of 2013, the development process began. Multiple graphic artists submitted designs, our Public Affairs Committee whittled them down, and the Board of Directors approved a design in November. Focus groups with families and "outsiders" followed and alterations were made.

The new logo—created pro bono by Connecticut designer Svetlana Sudeykina—incorporates many of the elements that represent the values and mission of this organization now and into the future: caring, family, growth and protection.

On National CdLS Awareness Day, May 10, the block was officially retired and the new logo revealed (shown below).

I hope you will join me in embracing the Foundation's new logo. We're certainly excited about the new look, and the future of the organization.

Marie Concklin-Mallo  
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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# Why Research Matters



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

As the deadline for research proposals submitted to the CdLS Foundation for funding for 2014 concluded, the question that many of our families and friends might ask is: "How can money spent on research help my/my friend's child with CdLS?" The real question is, "How can research funding help the child with CdLS practically, clinically and successfully?"

As of 2013 the Board of Directors for the CdLS Foundation has committed \$20,000 annually to help support research related to CdLS and its biologic origins. The projects are submitted and the Research Committee leads a formal review process in order to decide which project(s) should get funding. There have been both clinical and basic science proposals submitted, and each year both types have been funded. Clinical research often looks at a symptom or secondary effect of CdLS and investigates how to treat or reverse it. This could obviously be relevant to your child.

It is often less obvious how basic science research can be directly helpful. By funding research, particularly into the developmental biology behind the condition, the goals are to find out further information about the causes of the symptoms, how to alleviate those symptoms and how to prevent the symptoms or find a cure for a specific condition. Often, this works. We know that CdLS is caused by mutations in genes related to the protein complex called cohesin, which carries out many tasks in our developing body as fetuses and after birth. Basic science research focuses on those tasks that are not working properly and tries to find substances that might reverse them or correct them in a cellular or animal model.

It is a very real possibility that substances will be identified that seem to work. Shifting from research to human use is very lengthy, highly regulated and also very expensive, and currently national and governmental funding is increasingly difficult to obtain. That is why we have previously funded these types of projects.

Certainly for other genetic syndromes, with the help of funding from their parent support group, research resulting in specific treatment has been successful, although often with much larger budgets than the CdLS Foundation can currently allot. Still, we feel that it is so important to support all kinds of research as well as we can, and hope that you will agree.

**You can help.** You can help the CdLS Foundation support research projects that may save lives. Please identify "Research" when specifying your gift type, either online or by mail.

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## Quality of Life Findings



*By Alena Egense, MGC, CGC,  
University of Maryland*

As children with CdLS age, family discussions may start to include questions about long-term care and whether a residential care center is the next best step. This can be a very difficult decision: What is best for our child? If

and when should we make this transition? How will this decision, either way, impact our family? The effect of residential care decisions on a family with a child with CdLS had not been researched; this study aimed to explore this topic and help address some of these questions for caretakers of individuals with CdLS.

Overall, the study found that there is no difference in family well-being, defined as satisfaction with overall family life, between families whose child with CdLS lives at home and families whose child lives in a residential care center. Parents of a child with CdLS can be reassured that overall family well-being does not significantly differ based simply on the residence of the individual with CdLS. The study is consistent with the current literature that describes how family well-being is multifaceted and defined differently for each family.

On the other hand, there are similarities between families, and the study identified a few common themes introduced by many study participants. For example, aggressive behavior was a major concern; results suggest that families whose child with CdLS has more severe behavior problems have a lower overall satisfaction with life. Additionally, from these common themes emerged suggestions for caretakers of individuals with CdLS on ways to improve and protect family well-being:

- Establish a relationship with a behavioral psychologist early on in the child's development. For example, a behavioral psychologist can help caretakers of a child with CdLS determine the best timing for transitioning to a residential living situation; possibly before behavior problems for the child with CdLS become too severe.

- Be proactive in seeking intervention at the first sign behavioral problems are developing to help address the problem-behaviors before they escalate.
- Children with CdLS can have a difficult time adjusting to transitions, both daily and long-term. For transitions to living situations outside the home, families may want to consider slowly introducing their child to the new location. For example, one family found it helpful to start by driving by the new residence, then walking in to look around, followed by the individual with CdLS being able to show it to his/her friends; this method provides time for the child to internalize and fully adjust to the change.
- Family support, not just support by friends and neighbors, but by relatives, is important to prevent isolation and stress-fatigue. Engage other family members early on to walk with you through the joys and difficulties of raising a child with CdLS. Whether it be emotional, physical, or financial, allow your family to help support you in this journey.

Family well-being is not determined by a single decision of living arrangements, but involves many aspects of family life. There is still much to learn about what specific factors have the strongest impact on family satisfaction of life; however, through the invaluable participation and thoughts of the participants, this study has been able to provide a few considerations for caretakers of individuals with CdLS as they navigate the ups and downs of life.

As a final thought, thank you to all the families who filled out the survey; this study would not have been possible without you. In addition, thank you to the CdLS Foundation for your support, resources and guidance of my research.



## A Closer Look at Living Arrangements: Molly

*We want to thank Molly's mother, Kathy, for sharing Molly's story with us.*

When thinking about future living situations for your son or daughter with CdLS, there is no "right answer." There are excellent arguments on both sides of the issue of residential placement versus living at home. This is why this decision is so difficult for some families, including ours.

My perspective and advice is as a parent of a daughter who is severely affected with CdLS. Molly is 26 years old and is about the size of a first grader. She is completely non-verbal and requires total care. She walks short distances when holding someone's hand. She functions on the level of an infant/toddler in most areas. Functional level is an important factor in the choices available to your family. We had only one agency in our area that could handle someone with Molly's level of need.



There were the practical concerns, which were mired with emotional responses to this decision. We were a bit stuck. Fear, guilt, grief, sadness and anxiety are always very present when we discuss the future for Molly. Each parent may have very different ideas and experience different emotions as well. This can raise the stress bar even higher.

We started thinking about residential care when Molly was around 21, and were not enthusiastic. While we were trying to make up our minds, I was nearly killed in a car accident. This was a major wake up call. Our own mortality became frighteningly real and we began to look at options with a different mindset.

We were exhausted from this traumatic experience, trying to rehabilitate my two broken legs, finding extra help with Molly and trying to run our business all at the same time. In retrospect, this was a very bad time to make a big decision like this, and I wish we had waited another year or so after the accident. I think we would have made a better informed choice. We were overwhelmed and didn't know the right questions to ask.

We let the agency do most of the talking, and were heavily influenced by the case manager. We toured the home a couple of times, and asked all the questions we could think of. The house seemed very well maintained and clean and the residents seemed well cared for. The staff was very welcoming. Unexpectedly, a bed became available, and our fear was that if we didn't take the placement, it could be years before another opening became available. We spent an intensive few weeks getting staff properly trained and getting Molly used to the home.

We became aware of the limitations of this particular agency: the house manager, the staff and particularly the nursing case. After eight months of trying to make the situation work, we made the very difficult decision to pull Molly out and bring her home again. We have not regretted this choice, but it is not without its own set of challenges.

My advice: start early and get help. This should be a large part of your transition planning that starts in school at age 14. Information is power, so don't stick your head in the sand. Use your state case manager, and talk to other parents. Group homes are not the only option for out of home care, so identify all of the housing options in your area. If you start early enough, you may even come up with a solution unique to your child, using your own connections and resources.



# The Individualized Education Plan



**Featured in the CdLS Foundation's free Education Handbook**

*By Shelly Champion, M.Ed.,  
Co-chair of the Foundation's Professional Development Committee*

Each public school child (age three to 22) who receives special education must have an Individualized Education Program (IEP) as required by IDEA (Individuals with Disabilities Act). This is a document written specifically for an individual child. The IEP creates an opportunity for teachers, parents, school administrators, related services personnel, and students (when appropriate) to work together to improve educational results for children with disabilities. The IEP team comes together to look closely at the student's unique needs. The IEP guides the delivery of special education supports and services for the student with a disability.

The IDEA requires that certain information must be included in each child's IEP, however, states and school systems often include additional information to document that they have met certain aspects of federal or state law. IEPs may look different from state to state, or even school system to school system because school systems design their own IEP forms; however, they must include the same information regarding a plan for the child's education.

## **The IEP Process**

The formation of an individualized program involves six steps, beginning with pre-referral and ending with evaluation of the child's program. These steps are:

1. Pre-referral
2. Referral
3. Identification and Eligibility
4. Development of the IEP
5. Implementation of the IEP
6. Reviews and reevaluation

Many children with CdLS who were diagnosed at birth or before age three may have been receiving services necessary to facilitate their development through an Individual Family Service Plan (IFSP). When these children turn three, an IEP is developed for their education in the school system. These children bypass the referral, identification and eligibility steps and proceed directly to development of an IEP. Children with CdLS who are diagnosed after age three need to participate in the traditional referral steps of the IEP process.

## **Pre-referral**

Prior to developing an IEP, the school will implement various interventions depending on the kind of problem a child is exhibiting. The student's difficulties and challenges would be documented. Effectiveness of classroom accommodations and modifications would be tested and the student's progress monitored. Students whose learning remains challenged are referred to special education through the next step: Referral.

## **Referral**

Parents or school professional(s) may request an evaluation to determine if a child has a disability. Parents must consent to an evaluation. This evaluation must be completed within a reasonable time after consent is provided.

## **Identification and Eligibility**

A group of qualified professionals and the parents look at the child's evaluation results. Together they decide if the child has a disability as defined by IDEA. For many children with CdLS this is evident, however, for some children with a mild form of CdLS, the disability might not be as evident. If parents disagree with the evaluation, they have the right to take their child for an Independent Educational Evaluation (IEE) and ask the school to pay for it.

## **Development of an IEP**

The school system schedules and conducts the IEP meeting. School staff must:

- contact the participants, including the parents;
- notify parents early enough to make sure they have an opportunity to attend;



- schedule the meeting at a time and place agreeable to parents and the school;
- tell the parents the purpose, time and location of the meeting;
- tell the parents who are attending; and,
- tell the parents that they may invite people to the meeting who have knowledge or special expertise about the child.

The IEP team gathers to talk about the child's needs and write the IEP. Parents and the student (when appropriate) are part of the team. If a different group decides the child's placement, the parents must be part of that group as well.

Before the school system provides special education and related services to the child for the first time, the parents must give consent. The child begins to receive services as soon as possible after the meeting.

If the parents do not agree with the IEP and placement, they may discuss their concerns with other members of the IEP team and try to work out an agreement. If they still disagree, parents can ask for mediation, or the school may offer mediation. Parents may file a complaint with the state education agency and may request a due process hearing, at which time mediation must be available.

### **Implementation of the IEP**

The school makes sure that the child's IEP is being carried out as written. Parents are given a copy of the IEP. Each of the child's teachers and service providers has access to the IEP and knows his/her specific responsibilities for carrying it out. This includes the accommodations, modifications, and supports that must be provided to the child.

The child's progress toward the annual goals is measured, as stated in the IEP. Parents are regularly informed of their child's progress and whether it is enough for the child to achieve the goals by the end of the year. These progress reports must be given to parents at least as often as parents are informed of their typical children's progress.

## **A Closer Look at the CdLS Foundation's Education Handbook: Jonathan**

*We want to thank Jonathan's mother, Julia, for sharing Jonathan's story with us.*



I was not having particular difficulties when I requested the Education Handbook. I just like to always look to see if we have opportunities

to enhance what we are

doing for Jonathan. I used it as a reference to make sure

that we were on the right track with what we were doing

for Jonathan.

We are very fortunate with Jonathan's school. The school has always worked really well with us to do what we have requested. It is a center based program and they are very focused on meeting the needs of the population. There's the information written in the IEP and many other areas of focus. I think the processes outlined in the Foundations handbook match how things are happening in his school.

I think that entering the IEP process with the attitude that you are all on the same team, and want what is best for the child, is the best approach. Expecting an adversarial relationship often leads to communication and behavior that puts the staff on the defensive. I personally believe that the staff at Jonathan's school have very difficult jobs. They work very hard and often see minimal progress but remain dedicated to their calling and to the students. I like the quote that "you get more flies with honey" which I think is very true here. With that being said, I also know that we are very fortunate with Jonathan's educational setting and many are not as fortunate.

*See page 14 for information on how to order a copy of the Education Handbook.*



## Super Siblings: Matthew & Robynne



“Up until I was about four years old, I had an average life— I lived in East Brunswick, NJ with my mother and father and made friends with the children in my neighborhood. On January 20, 2003, everything changed; a new addition had been made to the family. When my brother was born, the

doctors didn't know what to say. They recognized that something was wrong but couldn't pinpoint exactly what. They later discovered he had Cornelia de Lange Syndrome, a rare genetic disorder that causes a variety of physical, cognitive, and medical problems.

At four years old, I didn't understand that my brother was disabled. Not until I saw him for the first time did I realize what my parents had tried explaining to me. He was not like the other babies I saw in the hospital. A nasal cannula was attached to his face with surgical tape and a feeding tube protruded from a hole in his stomach. His left hand consisted of a thumb, pointer finger, and a large flange of skin and tissue that would have been a pinky finger if given more time to develop in the womb. But these minor physical deformities did not make me love him any less; if anything, they increased my love and affection toward him.

I have never at any point in my life been ashamed of my brother. In public, people stare at him as though he is a monster of some kind. This blatant ignorance disgusts me and although I try to ignore it, it still pains me, since he does not understand the meaning behind the looks he gets. But if he doesn't seem to care, why should I? By being the happy-go-lucky child he is, my brother has inadvertently taught me countless life lessons that I would never have learned had it not been for his disability. My brother occupies the largest space in my heart and that will never change.”

## Mailbag – Kendall's Story –



My daughter, Kendall, and I have been raising awareness at a special needs fair for the past seven years. It is held at the Peoria Civic Center during “March Madness” high school basketball tournaments. When

this special needs fair got started it only had about 20-25 booths, but this year they had over 90 booths! Every booth offered something enriching and informative about special needs organizations, services or general information.

I don't get a lot of visitors, but I do get some that are interested to know about the syndrome. About six years ago I had a mother stop at my booth and she had a baby stroller with her in which the baby was sleeping so she had him covered up. She asked me a lot about CdLS and was very inquisitive, so I simply asked her if she had ever heard of CdLS. She replied, “Well...yes I have, my son was diagnosed with it.” I was so surprised, that when I asked her if I could see her son, she uncovered him and sure enough I could tell by looking at him that he had CdLS. Come to find out, she and her son live only about 15-20 miles from me.

About three years ago I had one couple stop by my booth because of my sign board with photos of all our kids and I was showing the CdLS awareness DVD video. They were very curious as to whether or not their daughter might have this syndrome. As soon as I saw their daughter and heard of her GI problems, feeding and swallowing issues, I instantly said, “Oh yes, let me give you some information on the CdLS Foundation and here is a contact card with a phone number for you to call.”

Kim and Kendall, Dunlap, IL

Share your Mailbag or  
Super Sibling Story!

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



## CdLS Awareness Day Unites and Inspires Families Nationwide

Coast to coast, in communities large and small, activities were held in honor of the 2014 National CdLS Awareness Day on May 10. Events ranged from a Foundation sponsored family gathering, to informal barbeques in backyards. May 10 was made even more special with the announcement of a new CdLS Foundation logo.



Houston, TX

While the events varied, the message was the same, and it was told louder than ever: These very special individuals with a very rare disorder are very important.

We're confident that each flier, bookmark, and moment you spend sharing this information makes a difference in your communities, and our country.



Mt. Kisco, NY

## Opportunities for Research Participation at the Yale Child Study Autism Program

Researchers at the Yale Child Study Center Autism Program are launching a pilot research project on social and emotional development of children with Cornelia de Lange Syndrome 5 years or younger. Similar studies are being carried out with children with Autism Spectrum Disorders and other developmental disorders. The aim of the study is to evaluate mechanisms that give rise to social and emotional difficulties in children with CdLS. As part of their participation in the project, the children will undergo an evaluation of their cognitive, language, and social functioning using standard assessments and eye-tracking measures. Participating families receive a written report summarizing the clinical assessment results and recommendations for treatment.

For more information please contact Amy Margolis at 203.785.6237 or visit [www.childstudycenter.yale.edu/yescog](http://www.childstudycenter.yale.edu/yescog).

### WELCOME NEW FAMILIES

#### Alabama

Jennifer and Brian and son Cade, born August 8, 2002

#### Georgia

Heather and daughter Kierstyn, born August 29, 2008

#### Massachusetts

Lynnette and Ryan son Jacob, born August 21, 2003

#### Ohio

Lindsey and daughter Raelyn, born May 25, 2009

#### California

Melissa and Tim and son Austin, born March 14, 2014

#### Indiana

Sara and Michael and daughter Hannah, born August 30, 2013

#### Michigan

Becky and son Benjamin, born May 23, 1985

#### South Carolina

Rachel and Shawn and son Tobias, born December 14, 2002

Madison and Cory and son Austin, born March 4, 2014

#### Louisiana

Becky and daughter Nevaeh, born December 6, 2010

Misty and Tom and sons Andrew and Alex born October 12, 2009

#### West Virginia

Latisha and daughter Taryn, born March 20, 2006

#### Florida

Lisa and Jason and daughter Nellie, born February 15, 2011

#### Maryland

Sara and Amos and son Jake born July 24, 2008

#### New Hampshire

Scott and daughter Sarah, born April 22, 2009



## 22<sup>nd</sup> Annual CdLS Charity Golf Tourney a Success Once Again

The CdLS Foundation hosted a sold out 22<sup>nd</sup> Annual New England Golf Tournament for CdLS at Ipswich Country Club in Ipswich, MA, on May 19.

“The tournament was started many years ago when some dads of children with CdLS got together at a family gathering and wanted to do something for the Foundation and play golf,” said Shelly Champion, mom to Julie, who has CdLS.

Since 1993, the tournament has raised \$554,255 to help people with CdLS live better, fuller lives. Inspired by their children affected by CdLS, New England parents, and their friends have made the event an annual success.



Sequoya, Tanaya and Valerie were all smiles as they waited for the golfers to return.

“After a long cold winter, golfers in New England are looking forward to hitting the links and the golf committee thought it would be a good time to attract golfers, which has proven to be true,” said Shelly. “The

tournament has grown from just golf, dinner and raffle with a few prizes to a full day event starting with brunch, golf, prizes, live and silent auctions.”

In addition to Shelly, tournament organizers are: Ava Frank and David Molzan, Stephen Igoe, Dianne Lessa, Patrick Lyons, Brian O’Keefe, and Frank and Denise Teixeira.



Carol Mahoney, Denise Teixeira, Beth O’Kelly and Donna Harvey took home the Champions trophy for the team with the lowest score overall for the second year in a row.

Tournament sponsors included RBC Capital Markets, Renaissance Macro Research, BMO Capital Markets, Brean Capital, LLC, Craig-Hallum, KeyBanc Capital Markets, Donald A. Sadoski, DMD, the Teixeira Family, Schnitzer, Samuel Adams and Insurance Recovery Group.

### Did you know...



0.5% of the price of your eligible AmazonSmile purchases is donated to the CdLS Foundation.



Register for an iGive button and iGive donates a percent of your purchase (varies by store) to the CdLS Foundation.



40% of every digital or print subscription is donated to the CdLS Foundation.

**...when you shop, they give!**

To find out more about these options, visit [donate.cdlsusa.org/shop.htm](http://donate.cdlsusa.org/shop.htm)



## Do Something Today, to Make a Difference Tomorrow



**CIRCLE of CARING**  
CdLS Foundation

Recognizing the CdLS Foundation's tomorrow depends on today's conscientious planning, David and Olivia Harvey made an important commitment to the future of the Cornelia de Lange Syndrome (CdLS) Foundation. They joined the Foundation's legacy society, Circle of Caring. Circle of

Caring was created to recognize and thank family and friends who have included the CdLS Foundation in their estate plans or made a planned gift to the CdLS Foundation.

Through their personal involvement with the Foundation, the Harveys understand the growing need for services, programs, education and research that ensures a better tomorrow for individuals and families affected by the syndrome. David stated, "At the 2012 conference, a seminar presented by Protected Tomorrows, detailed the importance of estate planning for ourselves and our three children, especially Katherine with CdLS. It became very obvious that my wife and I had put this off way too long. It prompted us to begin the process of setting up wills, trusts and powers of attorney to protect our children and estate."

During meetings with their attorney, David and Olivia also realized they had a desire to take care of the organization that made a difference in their lives – the CdLS Foundation. Their family's connection to the CdLS

Foundation would be for a lifetime. So after considering their personal circumstances and the needs of their heirs, their estate plans included a planned gift to the CdLS Foundation.

Thank you, David and Olivia for your personal belief and trust in the CdLS Foundation by providing a future gift that will impact the lives of others.

*The most common way of making a planned gift is by designating the CdLS Foundation as a beneficiary in your will. Other options are designating the organization as a beneficiary of your retirement plan or life insurance policy. There are also other, more complex ways of setting up a planned gift. Your attorney or financial planner may have suggestions that are best suited to your unique needs.*

*If you have already included CdLS Foundation in a bequest or other planned gift, we hope you will let us know. Please contact Gail Speers at 800.758.2357 or [gspeers@CdLSusa.org](mailto:gspeers@CdLSusa.org).*

## 21<sup>st</sup> Century Conference Fund

During 2014, your gift to the 21st Century Conference Fund will be matched by an anonymous donor. That means every dollar you give becomes two. Please take advantage of this opportunity to help the Foundation make twice the progress toward its goal.

Here's how:

**Online:** Go to our secure Donate page and choose Match it! as your donation type in the drop down menu.

**Mail:** Send a check to the CdLS Foundation at 302 West Main St., #100, Avon, CT 06001. Be sure to write Match it! in the memo line.

**Phone:** Call 800.753.2357 and use your credit card.



### MATCH IT! CHALLENGE

**I want to support the future of the National Family Conference**

Enclosed is my tax-deductible gift of:

- Other \$ \_\_\_\_\_  \$500  \$250  
 \$100  \$50  \$35

- I have included the CdLS Foundation in my will or trust

- 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: \_\_\_\_\_  
Email address(es): \_\_\_\_\_

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

Every dollar is matched, up to \$10,000.



## Board Corner

*Bob Boneberg, President, Board of Directors*



One of the pleasures and privileges of being a member of the Foundation's Board of Directors is the opportunity to meet so many people who are members of the larger CdLS family. Our Board members are themselves members of this family. Board members come from many walks of life and from all across the

country. Many have served on Foundation committees before becoming Board members. We have different backgrounds, and a variety of skills and talents, but are united in our commitment to improve the Foundation. Whether it be family services, or research, or professional support, or any of the many aspects of the Foundation's activities, we try to assess and improve.

Perhaps you know someone who might be a strong addition to the Board. We are always looking for talented people who wish to make a difference. One does not need to be a relative of someone diagnosed with CdLS to be a Board member. In fact, we believe that non-family members can make unique contributions to the Board, and we most warmly welcome them aboard. Of course, we look for diversity of background and experience. We do not want to hamper the Board's perspective and creativity by having, for example 12 accountants from Albany, or 12 bakers from Bakersfield, or 12 chemists from Charlotte.

What we do want and need and welcome is the next generation of able leadership. If you know of someone that you would recommend for the Board, please let us know.

Best wishes,  
Bob Boneberg  
president@CdLSusa.org

## Center for Excellence Profile: University of California, Irvine

The Cornelia de Lange Syndrome (CdLS) Foundation designated the University of California, Irvine, as a Cornelia de Lange Syndrome Center for Excellence for a second five-year period. This designation recognizes continued excellence and outstanding achievement in research work related to CdLS by five UC Irvine faculty.



"The research overseen by Anne Calof, Ph.D., (pictured above) Arthur Lander, M.D., Ph.D., Thomas Schilling, Ph.D., Kyoko Yokomori, Ph.D., Ali Mortazavi, Ph.D., and others continues to further what we know about CdLS and, in turn, brings new hope to families," said Marie Malloy, Executive Director of the CdLS Foundation.



Calof and Lander lab members working on Cornelia de Lange Syndrome

Dr. Calof who is professor of Anatomy and Neurobiology and Developmental and Cell Biology, together with her husband and collaborator Dr. Lander (Donald Bren Professor of Developmental and Cell Biology and Director for the Center of Complex Biological Systems), became involved with the CdLS Foundation and research on the syndrome in 2000, when Dr. Calof gave birth to daughter Isabel, who was diagnosed with CdLS. The couple worked together with researchers at Children's



Hospital of Philadelphia to help identify the gene whose mutation is responsible for the majority of cases of CdLS, *NIPBL*. The discovery of the *NIPBL* gene has led to the development of tools for molecular diagnosis of CdLS and has spawned a large body of biomedical research on CdLS and related syndromes.



As part of this effort, Drs. Calof, Lander (pictured above), and Thomas Schilling (Professor of Developmental and Cell Biology at UCI) have developed mouse and zebrafish models of CdLS that are being used to test methods for treating and/or helping to prevent changes such as cardiovascular disorders, changes in limb and hand structures and neurological problems that affect individuals with CdLS. The effort has expanded in the past few years to include Drs. Yokomori and Mortazavi, whose work on genomics is helping to increase understanding of how changes in *NIPBL* expression alter the expression of genes throughout the body.

## On the Cover

Dustin is an extremely happy young man who lives in Ames, IA. Born in 1993, Dustin graduated from Gilbert High School in 2012. He enjoys listening to music, and playing with a variety of toys, especially Legos. Some of his favorite activities include bowling, swimming, going for walks outside, and camping with his family. Some of his favorite memories include going to the CdLS Foundation Conference in Chicago, local CdLS Family Gatherings at Lake Okoboji, IA, the Victory Junction Gang Camp in Randleman, NC and vacationing with his family at the Outer Banks in North Carolina.



## 2014 CALENDAR

**July 12**  
Nebraska Family Gathering  
Lincoln, NE

Lapel Village Fair CdLS  
Benefit 5K  
Lapel, IN

**July 27**  
Merrell Down & Dirty  
National Mud Run Series  
Warminster, PA

**August 23**  
Midwest Foundation  
Sponsored Family Gathering  
Anoka County, MN

**September 15**  
26th Annual CdLS Charity  
Golf Tournament  
St. Louis, MO

**September 21**  
Saratoga Palio  
Saratoga Springs, NY

**September 27**  
Northeast Foundation  
Sponsored Family Gathering  
Whitinsville, MA

5th Annual Brew and BBQ to  
benefit CdLS  
Alpharetta, GA

**October 4**  
Maddy's Run  
Salt Lake City, UT

**October 12**  
Bank of America  
Chicago Marathon  
Chicago, IL

**October 18**  
Southeast Foundation  
Sponsored Family Gathering  
Sweetwater, TN

Baltimore Running Festival  
Baltimore, MD

**October 24-25**  
Board of Directors meeting  
Texas

**November 1**  
Multidisciplinary Clinic  
for Adolescents and Adults  
with CdLS  
Baltimore, MD

**November 2**  
TCS New York City Marathon  
New York, NY



# The Individualized Education Plan cont.

## Reviews and reevaluation

The child's IEP is reviewed by the IEP team annually, or more often if the parents or school asks for a review. If necessary, the IEP is revised. Parents, as team members, must be invited to attend these meetings. Parents can make suggestions for changes, can agree or disagree with the IEP goals, and agree or disagree with the placement.

If parents do not agree with the IEP and placement, they may discuss their concerns with other members of the IEP team and try to work out an agreement. There are several options, including additional testing, an independent evaluation, or asking for mediation (if available) or a due process hearing. They may also file a complaint with the state education agency.

The child must be reevaluated at least every three years. This evaluation is often called a "triennial." Its purpose is to find out if the child continues to be a "child with a disability," as defined by IDEA, and the child's educational needs. However, the child must be reevaluated more often if conditions warrant or if the child's parent or teacher asks for a new evaluation.

## Contents of the IEP

The IEP is a very important document for children with disabilities and for their educators. Done correctly, the IEP should improve teaching, learning and results. The following are the contents of the IEP.

- Current performance – The IEP must state how the child is currently doing in school (known as present levels of educational performance).
- Annual goals – These are goals that the child can reasonably accomplish in a year. The goals are broken down into short-term objectives or benchmarks. Goals may be academic, address social or behavioral needs, relate to physical needs, or address other educational needs. The goals must be measurable – meaning that it must be possible to measure whether the student has achieved the goals.
- Special education and related services – The IEP must list the special education and related services to be

provided to the child or on behalf of the child. This includes supplementary aids and services that the child needs. It also includes modifications (changes) to the program or supports for school personnel, such as training or professional development, that will be provided to assist the child.

- Participation with non-disabled children – The IEP must explain the extent (if any) to which the child will participate with non-disabled children in the regular classroom and school activities.
- Participation in state and district-wide tests – Most states and districts give achievement tests to children in certain grades. The IEP must state what modifications in the administration of these tests the child will need. If a test is not appropriate for the child, the IEP must state why the test is not appropriate and how the child will be tested instead.
- Dates and places – The IEP must state when services will begin, how often they will be provided, where they will be provided, and how long they will last.
- Transition service needs – Beginning when the child is age 14 (or younger, if appropriate), the IEP must address (within the applicable parts of the IEP) the courses he or she needs to take to reach his or her post-school goals.
- Needed transition services – Beginning when the child is age 16 (or younger, if appropriate), the IEP must state what transition services are needed to help the child prepare for leaving school.
- Measuring progress – The IEP must state how the child's progress will be measured and how parents will be informed of that progress.

*To read the full article, or for more information about the IEP, including IEP team information, visit <http://www.cdlsusa.org/request-information.htm> and request a copy of the CdLS Foundation's Education Handbook, or call the CdLS Foundation at 800.753.2357.*



Donations from 2/27/2014-4/30/2014

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Chuck Shook

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Tia M. Rivera  
Angelo Santoro

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Cathy Yocum  
  
Dré Hephill  
Board & Staff of the  
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MaryBeth Sicina  
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Carmel School  
Suzanne Raap



### Our Deepest Sympathy



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

Melissa Anne Wood  
February 3, 1983 – March 3, 2014  
Daughter of Mary and the late  
Walter Wood  
7809 W Wethersfield Rd  
Peoria, AZ 85381-5262

La Fundación CdLS envía correos electrónicos en español trimestralmente con actualizaciones sobre acontecimientos y artículos traducidos. Si conoce a algún familiar o amigo al que le podría interesar recibir este correo electrónico, póngase en contacto con Michelle Kiely enviando un correo electrónico a la dirección [familyservice@cdlsusa.org](mailto:familyservice@cdlsusa.org) o llamando al 800.753.2357.

Cornelia de Lange Syndrome Foundation, Inc.  
302 West Main Street, #100  
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## Where will your sneakers land in 2014?

5Ks, and mud runs, and walks - Oh My! You don't have to run a marathon to join Team CdLS. With so many options to get involved, there's something for everyone. Get some friends together and join a local 5K run or walk. Do you prefer a bike ride instead? Sign up for a bike race with Team CdLS. However you like to be active, we want to hear from you.

Email [TeamCdLS@CdLSusa.org](mailto:TeamCdLS@CdLSusa.org) or call Brenda Shepard at 800.753.2357  
for more information or to find a race near you.



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