

# Reaching Out

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



## Highlights

### Director's Message:

*Reaching Out* is YOUR  
Newsletter

### Spotlights:

We Already Have a CdLS  
Diagnosis...Why Follow  
Up With a Geneticist?

Genetic Testing and  
Your Family

### Super Siblings

Kendal and Aaron



## Director's Message



### Reaching Out is YOUR Newsletter

Since 1977, *Reaching Out* has been published to provide families with information, ideas and inspiration, from serious research news to light-hearted stories about those living with the syndrome.

Feedback about *Reaching Out's* articles is critical in ensuring the quarterly publication is valuable to those who read it. Foundation staff can't improve or change the newsletter if it doesn't know what its readers want.

Starting with this issue, there will be a short online survey for readers to provide feedback—both good and bad. It's your opportunity to tell us if we missed anything in an article; what you liked and didn't like; and what you want to read about in future issues. Remember, *Reaching Out* is your newsletter, so it's up to you to let us know what you want it to be.

We look forward to your feedback. You can access this issue's survey at <http://bit.ly/2ndRO2015>. Look for a new survey link after each issue.

Marie Concklin-Malloy  
Executive Director

## Be Prepared

Tear out the CdLS Medical Alert Card on page 15 to be prepared in the event of a medical emergency. By handing this document to emergency medical technicians or emergency room personnel, you save critical time. Keep a copy in your home; give a copy to your child's school or care facility; and keep a copy with your child (in a backpack or carrying case).

### 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 is the point of genetic testing?



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

*What is the point of genetic testing? Once you have received the results of genetic testing, what do you do with the information? How do you know if it will be helpful? What does it all mean? When you have a child with Cornelia de Lange Syndrome (CdLS), these are all questions to ponder, ideally with a geneticist or genetic counselor to help answer them.*

Genetic testing for a condition that is clinically diagnosed, like CdLS, can be helpful in a number of ways: clarification of diagnosis, further indication to obtain additional studies or additional testing, prenatal diagnosis for future children (to the couple, the individual or the siblings), and enrollment in the future clinical trials for treatments or medications.

Testing is expensive, may not be covered by insurance, and can result in vague and unsatisfying answers, so you do not want to embark upon it without careful thought. Although we are not even close to the point of carrying out clinical trials in CdLS, we hope that eventually we will be. And to enroll anyone for clinical trials, a diagnostic “marker” will be needed.

There are five known CdLS genes—*NIPBL*, *SMC1A*, *SMC3*, *HDAC8* and *RAD21*—and different types of testing on those genes (e.g., sequencing, deletion/duplication testing). It may be difficult to know if the correct test has been sent or if adequate testing has been completed. There are several companies that advertise testing for CdLS, but that testing may look for absence of the entire gene, which may be lethal before birth, and thus not even relevant.

We have heard from multiple families with children with CdLS who have started the quest for diagnosis and have had difficulties, such as missing results, incorrect interpretation and faulty conclusions. That is the point at which meeting with a genetic professional can be helpful. In addition to facilitating the actual testing sample, the professional can track down results, explain them and clarify the implications.

Overall, genetic testing can be mired in difficulties, so we would encourage all parents to seek genetic help prior to any sample being sent and, most importantly, upon receipt of results.

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**Cornelia de Lange Syndrome Foundation, Inc.**

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[www.CdLSusa.org](http://www.CdLSusa.org)

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All information contained herein is for the reader's personal interest. Articles on treatments, medications, or procedures, etc. are not guides for self-treatment. Questions should be discussed with your doctor or other appropriate professionals. The CdLS Foundation does not endorse any product advertised and/or mentioned in *Reaching Out*.



## We Already Have a CdLS Diagnosis...Why Follow Up With a Geneticist?



*By Amy Kimball, M.S., Genetic Counselor, Greater Baltimore Medical Center; CdLS Foundation Clinical Advisory Board member*

Most families see the role of the geneticist or genetics clinic as helping to make a diagnosis or facilitating genetic testing. But

there is value in returning to the genetics clinic for a follow-up visit or even seeing the geneticist on a regular basis.

First, geneticists and genetic counselors are considered experts in genetic conditions, not only in making a diagnosis, but also interpreting genetic testing results and management of the condition. For example, while other medical specialists focus on their specific fields, geneticists and genetic counselors focus on the genetic syndrome as a whole and provide an overview, such as: *Are all body systems being addressed? Are early intervention therapies appropriate? Is the family aware of specific recommendations or management guidelines at certain ages based on what we know about Cornelia de Lange Syndrome (CdLS)?*

Because of the expertise in the specific condition, the geneticist can provide anticipatory guidance regarding future concerns, referrals to appropriate subspecialists in the area who may have experience with CdLS, and discuss the benefits of a multidisciplinary clinic.

Additionally, questions regarding genetic testing results can be addressed with the genetic counselor or geneticist at a follow-up visit. These questions include: *What does this result mean? What are the chances of having another child with CdLS? Should additional testing be performed if a result is negative?*

Our knowledge and understanding of the gene changes and genes involved in CdLS are rapidly growing. It is the geneticist or genetic counselor's responsibility to re-investigate what we know about specific test results as researchers learn more about how gene changes cause CdLS. Therefore, questions about genetic testing results

should be revisited periodically, ideally during a follow-up visit with a geneticist or genetic counselor.

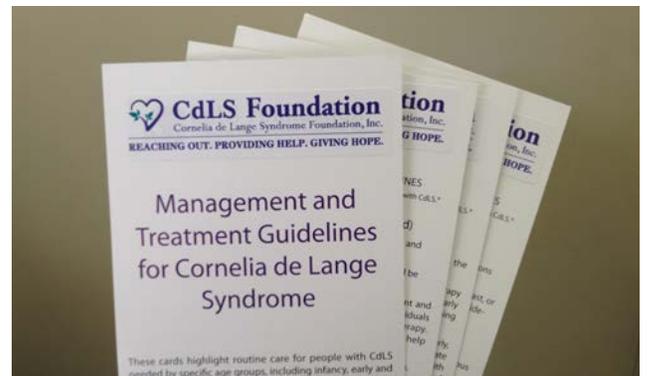
In addition, geneticist or genetic counselor can assess how the family is doing overall and if they would benefit from support resources in the community. This may include facilitating contact with other local families or identifying ways in which the CdLS Foundation may be helpful, given the current issues faced by the family. The genetic counselor can address concerns about the chance of having a second affected child and share options for testing or monitoring any future pregnancies. This may also include questions about future pregnancies for other relatives, such as siblings.

Most importantly the geneticist is a resource and someone to ask the many questions that arise when caring for an individual with CdLS: *What is typical? What should we expect? How should we address this issue?*

To find a clinical geneticist in your area, go to [www.acmg.net](http://www.acmg.net), and search 'find genetic services,' or talk to your primary care physician.

## Management and Treatment Guideline Cards

A hallmark service of the CdLS Foundation is its detailed materials for parents, professionals and the public. The treatment guideline cards highlight routine care for people with CdLS at different ages, including infancy, early and late childhood, adolescence, and adulthood. There is no charge. Please contact Deirdre Summa at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org) or call 800.753.2357 to request your cards.





## A Closer Look at Genetics Follow Up: Micah

*We want to thank Sarah, Micah's mother, for sharing Micah's story.*

We had taken Micah to a local geneticist due to concerns we had a few weeks after he was born. The local geneticist asked questions and examined Micah and told us, he “may” have CdLS. She recommended seeing if insurance would cover the genetic testing, which wouldn't necessarily tell us whether he had CdLS or not, or see Dr. Antonie Kline, the CdLS Foundation's Medical Director, in Baltimore. We opted to see Dr. Kline.



We have not visited a geneticist/genetic counselor every year, but we have seen them at events and/or been in contact with them via email more frequently. We consider Dr. Kline and Amy Kimball as Micah's geneticist and genetic counselor. We see them because they are CdLS experts and Baltimore is reasonably close to where we live. However, it is still an hour and a half away, so that is why we don't have more regular visits. When we have had follow up questions, we've emailed or called and they have been great about getting back in touch with us. If it is something that requires more than a phone call or email, we'd schedule an appointment and see them.

It is so reassuring seeing specialists who have seen many individuals with CdLS. They can help us know whether Micah is on track for Micah. There is no comparing him to typical children or with children with different syndromes. If we are having a particular issue, they can help troubleshoot, recommend specialists, and/or help come up with a plan of action. In addition, they are up to date on the latest research and findings.

## Participate in Research: Epilepsy Characteristics in CdLS

Researchers from Johns Hopkins University School of Medicine are recruiting individuals with CdLS for a study on epilepsy. They are looking for children/adults with a diagnosis of CdLS and epilepsy (defined as two or more lifetime seizures). The study involves completion of a short online survey and submission of certain medical records (copies of latest brain MRI, EEG, and neuropsychological testing, if available).

The purpose is to better understand epilepsy within CdLS, anti-seizure medications that are helpful in CdLS, and factors that contribute to epilepsy severity.

The study is titled “Epilepsy Characteristics in Cornelia de Lange Syndrome.” It is approved by the Johns Hopkins University School of Medicine IRB (NA\_00093750). The Principal Investigator is Marco Grados, M.D., Associate Professor, Department of Psychiatry and Behavioral Sciences, Johns Hopkins University School of Medicine.

To participate, please contact Drs. Sid Srivastava (sid@jhmi.edu) or Marco Grados, 443-287-2291; mjgrados@jhmi.edu.

*This research study has been approved by the CdLS Foundation Research Committee.*

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## 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.

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



## Genetic Testing and Your Family



*Sarah Noon, M.S., Children's Hospital of Philadelphia, CdLS Foundation Clinical Advisory Board member*

The first gene known to be associated with Cornelia de Lange Syndrome (CdLS), *NIPBL*, was discovered in 2004. Since then, four more genes have been identified:

*SMC1A*, *SMC3*, *HDAC8* and *RAD21*. Changes in these five genes are found in approximately 70 percent of individuals with CdLS, with mutations in *NIPBL* being identified in at least 65 percent of individuals, and the other genes being responsible, collectively, for about 5 percent. This leaves approximately 30 percent of individuals with CdLS in whom an identifiable underlying genetic etiology has not been found.

Recently a few researchers have seen children with CdLS in whom a mutation was not identified in the blood, but was found in other tissue samples, such as cheek cells. This indicates that some individuals do not have the gene change in all of their cells, but rather in a subset of cells. This is called mosaicism. It is unclear at this time what percent of individuals with CdLS have a mosaic mutation as the underlying cause. And evidence suggests there are likely additional genes to be found that cause CdLS when mutated.

### Genetics 101

As a brief background on genetics, we all have around 20,000 genes. We have two copies of each gene, with one set being inherited from our mothers and the other set from our fathers. A dominantly inherited condition, like CdLS, results from a change in only one copy of a set of genes. We say the change is “dominant” to the normal functioning other copy of the gene. This means that even though you have a copy of the gene that is functioning fine, the changed (or “mutated”) copy still has an effect on that individual – and hence is dominant.

Dominant changes can happen on the sex chromosomes (X and Y chromosomes) or on the non-sex chromosomes

(called autosomes). Mutations in the *NIPBL*, *SMC3*, and *RAD21* genes are associated with an autosomal dominant inheritance pattern. *SMC1A* and *HDAC8* are associated with X-linked dominant inheritance pattern. Women have two copies of the X chromosome and men have one copy each of the X and Y chromosome. Since women have two X chromosomes, the risks for future pregnancies are determined by the carrier status of the mother—although in almost all cases of CdLS caused by an X-linked gene, they have been new changes and are not carried in the mother.

Genetic testing is performed most often through a blood test and the vast majority of mutations identified are *de novo*—meaning that the mutation occurred spontaneously in the egg or sperm that went to form that child. As mentioned above, if a mutation is not identified in the blood of an individual with CdLS, it is recommended that a different tissue be tested (cheek, saliva or skin biopsy) to rule out a mosaic mutation.

### Germline Mosaicism

If both parents are clinically unaffected, then the risk to have additional children with CdLS is estimated to be approximately one percent. This is due to “germline mosaicism.” In germline mosaicism, the mutation in the gene that causes CdLS arose in a precursor cell that went on to form a group of eggs or sperm. In these rare cases, the change then exists in multiple sperm or eggs (but not in other tissues of the parents, so their blood testing will be negative and they will not have features of CdLS), but are at risk (as high as 50 percent) to have other children with CdLS.

In future pregnancies, serial ultrasound examinations may be performed to follow overall growth and the development of the heart, limbs, palate and other structures affected in individuals with CdLS. In some pregnancies, the maternal serum marker, PAPP-A, may be reduced and the fetal nuchal translucency (NT) may be increased, though these findings are not specific to CdLS.

If a mutation has been previously identified, prenatal diagnosis of future pregnancies can be performed by chorionic villus sampling (at 10-12 weeks gestation) or an amniocentesis (15-18 weeks gestation). Preimplantation genetic diagnosis (PGD) is also available for families in which the causal mutation has been identified. PGD is a procedure in which embryos are created outside of the



body through in vitro fertilization (IVF) and one cell (in the first few stages of division) is biopsied to determine if the genetic change is present. Then, there is implantation into the mother's uterus of embryos that do not carry the mutation.

## Second Generation CdLS

Rarely, a mildly affected individual with CdLS will have children, and in those instances the parent has a 50 percent chance of passing on the affected gene (see below).

For individuals with CdLS who are interested in having families of their own, the recurrence risk is largely dependent on the underlying gene change present in the individual. For those who have a mutation in the *NIPBL*, *SMC3*, or *RAD21* gene there is a 50 percent chance to pass on the mutation to any child. Since we all have two copies of every gene, there is a 50 percent chance to pass on the copy of the gene with the mutation and a 50 percent chance to pass on the copy of the gene without the mutation.

If a child inherits a CdLS gene mutation from a parent, the child will inherit the same mutation as the parent and will be similarly affected. For those who have mutations in either of the X-linked genes, *SMC1A* or *HDAC8*, females have a 50 percent chance to pass the copy of their X with the mutation to their children, while males will pass on the copy of their X with the mutation to all of their daughters but none of their sons (who inherit the father's Y chromosome and not the X chromosome). Unaffected siblings of individuals with CdLS are not at an increased risk to have children of their own with CdLS.

## Conclusion

For individuals interested in expanding their family or who are interested in pursuing prenatal genetic testing, it is recommended to meet with a geneticist or genetic counselor before pregnancy to discuss recurrence risk as well as the benefits, limitations, and possible uses of genetic testing for you and your family.

## A Closer Look at Genetic Testing: Claire



*We want to thank Kimberly, Claire's mother, for sharing Claire's story with us.*

It took us 17 months and a roundabout way to receive a diagnosis. Claire presents more mildly and was misdiagnosed by two different genetic teams at major hospitals. We ended up 2,000 miles from home meeting with an endocrinologist specializing in a different syndrome who referred us to Dr. Ian Krantz at Children's Hospital of Philadelphia (CHOP). Two days later, we were at CHOP, where she was clinically diagnosed with CdLS.

Claire's diagnosis was in April 2005, a year after the *NIPBL* gene was discovered. Dr. Krantz asked if he could include her in his study and obtained a blood sample. She tested negative for *NIPBL*, but since they had her sample in the lab, her sample was tested each time a new gene was discovered.

I believe it was in the spring of 2010 that we received a phone call from CHOP saying they thought they had a gene match (it turned out to be *HDAC8*) for Claire and requested blood samples from my husband and I to see if the mutation was new to Claire, or inherited. It was new.

Doctors obtained additional information from us at the CdLS Foundation National Family Conference that summer in Dallas, and in the fall, Dr. Matthew Deardorff from CHOP obtained a tissue sample from Claire while traveling for work in our home state of California.

Fast forward to 2015 and you can often request genetic testing through insurance. More information about your child medically is usually a really good thing; it gives you a more complete picture. Sometimes people don't want to know, but in this case, knowing opens up avenues for future treatment that might otherwise not be there.

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## 15 Ways to Support the Foundation in 2015



**CdLS Foundation**  
Cornelia de Lange Syndrome Foundation, Inc.

Want to help raise awareness and/or support the Cornelia de Lange Syndrome (CdLS) Foundation this year? The Foundation has come up with 15 ways to do so. Take a look

and pick out ways you can get involved, and we'll help you along the way.

You could...

1. Educate 15 people about CdLS by sharing a CdLS bookmark or brochure (just call and we'll send you some).
2. Ask a local organization or club to give you 15 minutes to do a presentation on CdLS at one of its meetings.
3. Commit to a monthly gift of \$15.
4. Help the Foundation celebrate the 15th anniversary of Team CdLS by participating in an existing event or starting your own team to run in a local race.
5. Invite 15 friends to a CdLS Foundation event or gathering. If there's not one near you, consider hosting one.
6. Ask friends and relatives to donate \$15 to the Foundation in lieu of a birthday or anniversary gift.
7. Volunteer 15 hours of your time with the Foundation, either at an event or virtually.
8. Invite 15 friends to plant hope by participating in Cornelia's Garden this spring.
9. Coordinate a dress down day at work and ask at least 15 people to participate.
10. Retweet a CdLS Tweet to 15 Twitter friends. (Find us on Twitter and Instagram @CdLSFoundation)
11. Send the CdLS Awareness video link (<http://bit.ly/CdLSvid>) to 15 friends.
12. Bring 15 bookmarks to your local library to leave at the front desk.

13. Invite 15 friends to "like" the CdLS Foundation Facebook page.

14. Pick 15 days throughout the year and commit to raising awareness in different ways.

15. Ask community businesses you support (like a salon, coffee shop, boutique) to donate 15 percent of its proceeds to the Foundation for one day.

Maybe you have your own ideas for getting involved this year. We'd love to work with you! Email Brenda Shepard ([bshepard@CdLSusa.org](mailto:bshepard@CdLSusa.org)) to get started on your plans to support the CdLS Foundation in 2015.

## Austin's Day



Earlier this year, Chico's of Sonoma, CA, held a special day in honor of Austin Staggs. The clothing store donated 10 percent of its sales to the CdLS Foundation—more than \$400. The Staggs were on hand to celebrate "Austin's Day," along with friends and family.

Austin's grandmother, Barbara (pictured far right), her daughter Melissa (center), and grandson were on hand to celebrate, along with friends and family.

"The whole day was amazing. So many people came in to talk to us and buy clothes. We gave out lots of information on CdLS, and the sales girls who worked that day also donated their bonuses," said Barbara.

Another grandmother, Linda (pictured far left), and great grandmother (pictured second from left) to Henry (not pictured), who has CdLS, traveled an hour and a half to attend the event.



# Team CdLS celebrates 15 Years



Cornelia de Lange Syndrome Foundation

The Foundation's goal is to have 15 races around the country to celebrate the 15th anniversary of the program. Since our first runner at the Chicago Marathon in 2000, the team has raised over \$1 million to provide program, services and support to families at no charge to families.

If you are interested in joining Team CdLS, you can run/walk in an existing race in your community, start your own Team CdLS event, or join our established teams at races throughout the country. Visit <http://cdlsusa.org/events/team-cdls.htm> for a schedule of runs.

Biking, skiing, swimming, volleyball, and other sporting events are also welcome additions under the Team CdLS umbrella.

# Make this your year for a 5K

The Team CdLS 5K Club—in its second year—saw a lot of smiling faces crossing finish lines around the country in 2014 at races in Kentucky, Iowa, North Carolina, Minnesota and Connecticut. Raising over \$3,000 for the Foundation.

Joining the Team CdLS 5K Club makes running or walking a 5K easy and fun. We'll help you find a race near you, or work with you to reach the finish line of a race of your choosing. Email [TeamCdLS@CdLSusa.org](mailto:TeamCdLS@CdLSusa.org) or call 800-753-2357 to learn more.



Thank you to Team Shelby, who ran a 5K and raised over \$2,000 for the CdLS Foundation this year.

# Thank You 2015 Team CdLS Sponsors



# 10 years of "One Love, One Heart"



For 10 years, the Pomfret family of Decatur, GA, has hosted the One Love, One Heart 5K Walk/Run in honor of their daughter, Maya, and all individuals with CdLS. The 5K, held this past December, raised \$11,325 to benefit the CdLS Foundation—bringing the 10-year total to more than \$158,000.



## Super Siblings: Kendal and Aaron

*Kendal is a junior in high school and sister to Aaron, who is 35 and has CdLS. Kendal shared her experiences volunteering at CdLS National Family Conferences.*

I've attended eight conferences and volunteered at four. I have a special place in my heart for kids with CdLS, and I love working with kids in general, so I started working in childcare at conferences as soon as I was old enough.



This most recent conference, I also volunteered with doctor consultations, where a lot more needed to be done, and it was very busy—signing people in, taking them to the doctors, running errands. I also got to go into the consults with my parents and talk about my brother. I liked hearing information straight from the doctor.

I've made many connections at conferences. There's a sibling of an individual with CdLS who I see at conferences and have built a friendship with over the years. We're the same age and have known each other for six years. This past December, we got to travel to see one another and hang out together.

At last year's conference, I was working with a little boy and found out it was his family's first time attending. They live two hours away from where I live in Florida, so my family connected with them. Being able to share my experiences with my brother with them was awesome.

**Share your Mailbag or Super Sibling Story!**  
Send your story and photo to  
[bshepard@CdLSusa.org](mailto:bshepard@CdLSusa.org).

## Mailbag – Josie –



Josephina Mae was born in October of 2008 in Springfield, IL. She weighed in at 2 lbs. 1.3oz. and 13.5 inches long, and spent 34 days in the NICU. Since Josie wasn't one of the NICU's "sicker" babies, they let her go home at 3 lbs. 3.3oz, and she proudly hasn't spent a night back in the hospital since that day.

I was at about four months into my pregnancy when I was told something was not right. I had an amniocentesis done and everything surprisingly came back fine. The doctors knew something wasn't right but had to wait until she was born to really know what condition she had and what was next. It was two days after her birth we received the diagnosis of CdLS.

CdLS to me is just her medical condition. It does not define her abilities in life. To me it's just an easy way to explain why she is small and why she has her arm differences. Josie has mild hearing loss, for which she has hearing aids, but they haven't been worn in forever because she pulls them out now. She also has AFO braces for walking.

Josie has completed two years of preschool and is currently in Kindergarten, where she has made numerous friends and memories. I'm sure that if she was verbal she would tell me all about her days. Thankfully her wonderful teachers and friends tell me when they see me, and they capture those special moments through pictures.

My beautiful daughter Josephina Mae is a blessing. She makes everyone she meets smile. She is and always will be my little toughie. She has proved without a doubt that she can survive and succeed in this world. We may have a different lifestyle than most, but it's ours, and just the path that God wanted us to have.

Deanna  
Falls City, NE



## Logo Usage



The CdLS Foundation logo is unique and memorable. Guidelines are in place to help maintain consistency and coherence in communicating the CdLS Foundation's identity and personality to all audiences. The use

of our logo represents an endorsement by the CdLS Foundation in the eyes of our families and supporters and is one of the reasons there are specific criteria for use.

Approval of any design incorporating the CdLS Foundation logo is required.

We are here to work with you. If you are interested in using our logo for awareness or fundraising efforts benefiting the CdLS Foundation, contact Brenda Shepard at [bshepard@CdLSusa.org](mailto:bshepard@CdLSusa.org), or call the Foundation office at 800.753.2357 to discuss your efforts.

## In lieu of gifts...

Alyssa's 30th birthday raised \$1,670 for the CdLS Foundation. You too can ask friends and relatives to donate to the CdLS Foundation in lieu of a birthday or anniversary gift, and the CdLS Foundation can supply customized donation cards for your celebration. By suggesting the option to donate "in lieu of gifts," you support the many services the CdLS Foundation provides for families around the country, at no cost to them. Call 800.753.2357 for information.

## 2015 CALENDAR

**April 24-25**  
Board of Directors Meeting  
Windsor Locks, CT

**April 25**  
Faith Hope Charity Bowl for  
Kennedi  
Lexington, KY

**May 2**  
Southeast Region Family  
Gathering  
Randleman, NC

**May 18**  
23rd Annual CdLS Charity  
Golf Tournament  
Ipswich, MA

**June 13**  
Brew & BBQ for CdLS  
Bloomfield, CT

**August 24**  
27th Annual Golf  
Tournament for CdLS  
Sunset Hills, MO

**September 9-12**  
CdLS World Conference  
Lisbon, Portugal

**September 20**  
Saratoga Palio  
Saratoga Springs, NY

**October 11**  
2015 Bank of America  
Chicago Marathon  
Chicago, IL

**October 17**  
Baltimore Running Festival  
Baltimore, MD

**October 23-24**  
Board of Directors Meeting  
Orlando, FL

**November 1**  
2015 TCS New York City  
Marathon  
New York, NY

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

Plant hope today! Email Nicole Dalto at [nicole@CdLSusa.org](mailto:nicole@CdLSusa.org) or call 800.753.2357 to get started.

*planting hope for CdLS*





## Now Accepting Nominations for 2015 de Lange Society



The CdLS Foundation is accepting nominations for the third class of the de Lange Society. Members bring creative vision, innovative ideas, vital assistance and leadership to the Foundation. Their contributions enhance the quality of service and

programs offered to individuals with the syndrome and their families.

The following criteria provide guidelines and qualities that the CdLS Foundation believes exemplify a strong sense of volunteerism:

- Demonstrates exceptional leadership, creativity, vision, and commitment to the CdLS Foundation.
- Has made significant overall contributions to the Foundation over a minimum of 20 consecutive years.
- Is recognized by his or her peers as contributing above and beyond standard performance.
- Advocates and advances the Foundation's mission and goals.
- Promotes positive character traits, such as caring, citizenship, fairness, respect, responsibility, and trustworthiness.
- Exemplary performance in response to an important organizational need.
- Inspires others by providing guidance, teamwork, diversity, integrity, and innovation as they strive for excellence.

If you know someone who you feel meets this criteria, visit [bit.ly/2015deLange](http://bit.ly/2015deLange) to nominate them for a spot in the 2015 de Lange Society.

To view a list of the class of 2013 and 2014, go to <http://cdlsusa.org/getinvolved/delange.htm>.

## Volunteer With Us

A core component of the Foundation is its volunteers. Parents, relatives and friends around the country give their time and energy to help improve the lives of people with CdLS. Awareness and Regional Coordinators are two types of our core volunteers.

### *Awareness Coordinator Program*

Awareness Coordinators (ACs) are volunteers who raise awareness about CdLS through community activities/ events, outreach to professionals, and contact with the media. ACs are passionate about increasing awareness of the syndrome and are asked to:

- promote awareness in their communities and support others who are interested in planning a CdLS presentation or an event;
- be available to media working on news features about CdLS; and
- provide the Foundation with updates on awareness activities for inclusion in the Foundation's newsletter and Web site.

### *Regional Coordinator Program*

Regional Coordinators (RCs) are parents or caregivers of individuals who have CdLS. The program assures that families are provided with the emotional support and essential information needed when caring for an individual with CdLS. By providing one-to-one contact and matching families who have experienced similar issues, the stress and isolation an individual family may feel will be reduced. RCs are asked to:

- write, telephone or meet in-person newly referred families;
- maintain a network of existing families in their areas and match families by area and/or similarities;
- refer families to known community resources; and
- host or support regional gatherings

If you're interested in becoming an RC, contact Deirdre Summa at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org) or 800.753.2357. If you would like to become an AC, contact Brenda Shepard at [bshepard@CdLSusa.org](mailto:bshepard@CdLSusa.org) or 312-912-7198.



## Board Corner

Bob Boneberg Esq., President, Board of Directors



As I begin my sixth and last year of service on the CdLS Foundation Board of Directors (the past two as president), I am reflecting once again on the balance that the CdLS Foundation must try to find in three points in time: today, tomorrow and beyond.

When I think of today, I focus on the Foundation’s annual operational activities. Every day the Foundation opens its doors and engages in all the programs and activities that are described in *Reaching Out* and on the website, including working with the hundreds of families that contact the Foundation every year. From the board’s perspective, we try to ensure that each year the Foundation has the funds and staff to engage in its activities and, further, that the Foundation provides its services in an effective and efficient manner.

“Tomorrow” may best be thought of as the next three years that encompass the periods of the Foundation’s strategic plans. As the perspective shifts from the immediacy of today, the questions for the board change also. What change in emphasis in programs should we make? How do we find the correct balance among, for example, family support, regional clinics, professional awareness, and research? What are the implications for our budget and staff if we change our programs? How has our fundraising changed over time and how should we raise money to ensure that we can provide the programs that our friends and supporters want us to provide?

Lastly, I think about the long-term future for the Foundation. Where should the Foundation be in 10 or 20 years? Should we have the same mix of programs? Should some programs and activities be phased out? Are there new things that we should be doing? The extended long-term is where dreams and visions are tempered with our history and experience, so that we can begin to develop those long-range plans that make the best sense for everyone.

If you think that there is a better way for the Foundation to operate, or have thoughts on the future, please let me know by emailing [President@CdLSusa.org](mailto:President@CdLSusa.org).

## Se buscan coordinadores bilingües voluntarios

Una de nuestras metas para el año 2015 es la provisión de mejores recursos y apoyo para las familias hispanohablantes que se ponen en contacto con nosotros. Si usted es bilingüe en español e inglés, podrá ser un recurso fundamental para esas familias en todo el país. Si desea obtener más información sobre cómo convertirse en Coordinador hispanohablante voluntario para la Fundación SCdL, póngase en contacto con Deirdre Summa enviando un correo electrónico a [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org).

### WELCOME NEW FAMILIES

#### California

Sophia and John and daughter Brooke, born July 19, 1998

Adriana and Javier and daughter Maileni, born January 31, 2013

#### Colorado

Bonnie and Seth and daughter Emma, born July 29, 2014

#### Florida

Cristiana and daughter, born November 25, 2010

#### Nebraska

Sarah and son Tripp, born September 22, 2014

#### New Jersey

Coretta and son Keyshawn, born July 9, 1986

#### Texas

Cheryl and daughter Baylee, born October 2, 2007

Aly and Tyler and son Samuel, born October 10, 2013

#### Virginia

Rebekah and Antonio and son Jeriah, born April 22, 2014

#### Washington

Brenna and daughter Aelycia, born July 2, 2014

#### Wisconsin

Tanya and son Anthony, born August 1, 1999



## A Closer Look at Genetic Testing: Claire’s Story cont.

It’s really useful to know what gene change your child has. As researchers gather larger subsets in each gene, it will improve the quality of information about what your child may be like, what complications might come up, and also give you specific information on how to treat your child’s medical, emotional and academic needs. Anything you can do to tailor treatments to enhance the quality of life for your child, in my opinion, is a huge advantage.

Not all of our questions were answered by testing, but knowing does explain some differences we see with Claire. Something that affects Claire may not be common to all people with CdLS, but it may be common to those with the *HDAC8* change.

Knowing that it happened at conception also helps the “what could I have done differently?” question in the back of my mind. It releases me from any guilt because it’s the way my daughter was created. It let’s me take a deep breath and give my child the best life I can.

## What is the point of genetic testing? cont.

**Testing is available at the following locations:**

### **Children’s Hospital of Philadelphia**

The CHOP research lab will soon have screening available for the five known CdLS genes. The research team also has a particular interest in individuals who have a diagnosis of CdLS but have had negative testing for the five genes.

Sarah Noon, M.S., (215) 590-4248;  
noons@email.chop.edu.

### **University of Chicago Genetic Services Laboratories**

Clinical lab providing testing for all five known genes. (888) UC-GENES; ctan@bsd.uchicago.edu.

### **Courtagen**

Commercial lab providing testing for all five known CdLS genes as part of their neurodevelopment disorder panel. Financial assistance is available to qualified families. 877.395.7608; www.courtagen.com.

### **GeneDx**

Commercial lab testing for NIPBL and SMC1A. 301-519-2100; www.genedx.com.

## Got clinic?

Don’t miss out on the opportunity to meet with specialists from a range of medical disciplines. The current schedule for CdLS Clinics is as follows:

- **The CdLS Clinic at Santa Clara Valley Medical Center (San Jose, CA)**  
August 10, 2015  
Insurance authorization required.
- **Multidisciplinary Clinic for Adolescents and Adults at Greater Baltimore Medical Center Clinic (ages 12 and up)**  
November 7, 2015
- **Center for CdLS and Related Diagnoses at Children’s Hospital of Philadelphia**  
Meets once monthly.  
Insurance authorization required.



**The CdLS Foundation offers one-time travel reimbursement up to \$250 to assist families with travel.**

For more information on CdLS clinics, contact Deirdre at [familysupport@CdLSusa.org](mailto:familysupport@CdLSusa.org) or 800-753-2357.

# MEDICAL ALERT CARD

CdLS Foundation: 800.753.2357

Name: \_\_\_\_\_ has  
**Cornelia de Lange Syndrome (CdLS)**  
Updated \_\_\_/\_\_\_/\_\_\_

## Medical Conditions Associated with CdLS

*Check all that exist*

### Life Threatening Conditions

- GI problems: bowel obstruction, volvulus, reflux leading to aspiration pneumonia
- Swallowing difficulties/choking
- Small airway (anesthesia risk)
- Seizures
- Cardiac problems

### Other Notable Characteristics

- Speech and hearing problems (may not be able to understand/respond to questions)
- Neurological, behavioral problems
- Slow growth and small stature
- Developmental delays

-----fold here-----

### Personal Details

Blood Type: \_\_\_\_ Age: \_\_\_\_ Sex: \_\_\_\_ DOB: \_\_\_\_\_

Address: \_\_\_\_\_

Phone(s): \_\_\_\_\_

### Emergency/Medical Contacts

Name: \_\_\_\_\_ Relation: \_\_\_\_\_

Ph# 1: \_\_\_\_\_ Ph#2 \_\_\_\_\_

Address: \_\_\_\_\_

Name: \_\_\_\_\_ Relation: \_\_\_\_\_

Ph# 1: \_\_\_\_\_ Ph#2 \_\_\_\_\_

Address: \_\_\_\_\_

-----fold here-----

Doctor: \_\_\_\_\_ Ph#: \_\_\_\_\_

Specialty: \_\_\_\_\_

Doctor: \_\_\_\_\_ Ph#: \_\_\_\_\_

Specialty: \_\_\_\_\_

Doctor: \_\_\_\_\_ Ph#: \_\_\_\_\_

Specialty: \_\_\_\_\_

Ins. Co. \_\_\_\_\_

Policy: \_\_\_\_\_

Medicaid # \_\_\_\_\_ Medicare # \_\_\_\_\_

- ( ) Behavior Issues: ( ) Anxiety ( ) Aggression
- ( ) Self-injurious ( ) Hyperactivity
- ( ) Bowel Obstruction: ( ) Surgery: date(s)\_\_\_\_\_
- ( ) Cleft Palate
- ( ) Small Airway (may require pediatric size tube)
- ( ) Constipation: ( ) Occasional ( ) Often
- ( ) Dental: ( ) Many Cavities ( ) Crowding
- ( ) Developmental Delays: ( ) Mild ( ) Mod ( ) Severe
- ( ) Ear Infections (frequent): ( ) Tubes
- ( ) Genitalia Abnormality: ( ) Hypospadias ( ) Micropenis
- ( ) Feeding: Prone to Choking ( ) NPO
- ( ) Feeding Tube
- ( ) Food Intolerance: ( ) Lactose ( ) Gluten
- Other \_\_\_\_\_ Special Diet \_\_\_\_\_
- ( ) Fundoplication Surgery: date(s)\_\_\_\_\_

- ( ) Gastroesophageal Reflux: ( ) Mild ( ) Severe
- ( ) Hearing Loss: ( ) Wears Aides
- ( ) Heart Defect: Type(s)\_\_\_\_\_
- ( ) Hip Problem(s): Type(s)\_\_\_\_\_
- ( ) Malrotation: ( ) Surgery: date(s)\_\_\_\_\_
- ( ) Pain Tolerance (unusually high)
- ( ) Pneumonia: date(s)\_\_\_\_\_
- ( ) Ptosis: ( ) Surgery: date(s) \_\_\_\_\_
- ( ) Renal Disorder: Type(s)\_\_\_\_\_
- ( ) Seizures: ( ) Frequent ( ) Rare ( ) On Meds
- ( ) Sinus: ( ) Infections ( ) Polyp(s)
- ( ) Tear Duct Frequently Clogged
- ( ) Upper Limb Differences
- ( ) Vision Impaired: ( ) Glasses ( ) Contacts
- ( ) Other (not necessarily related to CdLS): \_\_\_\_\_

fold here

### Allergies

- ( ) Antibiotics: ( ) Penicillin ( ) Sulfa ( ) Tetracycline
- ( ) Other \_\_\_\_\_
- ( ) Pain Meds/Anesthesia: ( ) Codeine ( ) Demerol
- ( ) Lidocaine ( ) Morphine ( ) Novocaine
- ( ) Other \_\_\_\_\_
- ( ) Environmental \_\_\_\_\_
- ( ) Insect Stings \_\_\_\_\_
- ( ) Foods \_\_\_\_\_
- ( ) Latex ( ) X-Ray Dyes
- ( ) No known allergies

### Medications

Medication	Dosage	Frequency	Reason
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____
_____	_____	_____	_____





## 2015 National CdLS Awareness Day - May 9

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

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



**Don't forget to wear purple for CdLS!**



Donations from 12/01/2014-03/06/2015

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## On the Cover



Jayden is 11 years old and in the sixth grade. He is in a Life Skills classroom and is doing awesome thanks to his wonderful teacher. Jayden was the manager for his school basketball team and loved helping out and being part of the team. He also played in the last game of the season. He was

so nervous, but he got through it with the help of his teammates and coach. I know it's a moment he will never forget.

In his free time, he loves playing on his Xbox 360, watching the San Antonio Spurs and listening to hip-hop music. Jayden is very set in his ways and must follow the same routine every day. He is very funny, smart and curious, and he has everyone wrapped around his little finger because of his charisma. He is a great role model to his little brother Tristan, who is his best friend. They have a bond that can never be broken.

Jayden amazes our entire family each and every day. Sometimes he needs a gentle push, but he always gets through it with a smile. As his mother, I am beyond blessed to have him as my son.

**Our Deepest Sympathy**

<p><b>Michael (Mikey) Austin</b>          November 30, 1974 – December 27, 2014          Son of Judith Stilwell          25501 Trost Blvd # 7-55          Bonita Springs, FL 34135-6422</p>	<p><b>Nikki A. Rauscher</b>          March 1, 1996 – February 2, 2015          Daughter of Dawn and Rolfe Rauscher          1198 Mt Hope Ave          Oneida, NY 13421-7036</p>
<p><b>Julissa Sherri Raquel Fiol</b>          June 5, 2007 – January 6, 2015          Grandchild of Margaret and Roger Scruggs          810 Chopping Rd          Mineral, VA 23117</p>	<p><b>Addison Hood Weatherly</b>          November 15, 2012- October 3, 2014          Daughter of Michaela Hood          1502 Brush St          Bridgeport, TX 76426</p>

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

**Yes, I want to help people with CdLS.**

Enclosed is my tax-deductible gift of:

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

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

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There's a lot of "small" talk when it comes to Cornelia de Lange Syndrome (CdLS): small stature, small head size, small hands and feet... We're here to tell you though, that people with CdLS may be small, but they are **mighty**. Many face big medical and physical challenges with equally big courage. The CdLS Foundation is on their team, ready to jump into action when needed. But we can't do it alone.

## **BE MIGHTY**

You can make your donation to the CdLS Foundation mightier by committing to monthly giving. That's because a small monthly donation equals a big gift over time—for example, just \$25 per month is \$300 annually.

Contact Kelly Brown at [kbrown@CdLSusa.org](mailto:kbrown@CdLSusa.org), or call 800.753.2357 to get started.