Reaching Out

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

Highlights

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Director's Message

Building On Success

Having a plan for the future is a key component of any organization's operations. At the CdLS Foundation, we have established a three-year strategic plan cycle, with each new plan serving as a "road map" for the work the staff does during a three-year period.

As the Foundation wraps up its 2013-15 strategic plan, I'm happy to report successful completion of our goals and objectives. As we transition into a new strategic plan for 2016-18, we also say goodbye to Marie, who was integral in accomplishing our goals, including the creation of a CdLS clinic on the west coast in San Jose, CA, creation of a CdLS Registry, and an increase in Foundation-sponsored family gatherings. I am extremely grateful for her leadership and guidance, and will continue to build on her success.

The 2016-18 plan, approved by the Board of Directors in July, calls for continued increase in research funding, establishment of CdLS clinics in the Midwest and Southwest, expansion of our biggest fundraiser—Team CdLS—into new regions, and creation of publications covering important social and family issues.

Foundation staff does not decide what the program service goals and corresponding objectives of the plan are, rather it's the families who do. This new plan, just like the previous one, was created from input from participants in a strategic planning survey last fall and personal interviews earlier this year. Some program goals of the 2016-18 plan are:

- Ensure high quality medical care that reflects our most current evidence-based diagnostic and treatment procedures for common medical issues in CdLS.
- Equip and empower families to make informed decisions about their child's health, development, education, and future.
- Cultivate an environment that sustains and grows quality and relevant clinical and molecular research.

On the operations side, our goals—which are determined by the Board and staff and support the program goals above— include increasing revenue by 20 percent over three years, and partnering with like syndrome groups in an effort to share resources and create alliances to solicit funding/grant support. If you have questions or comments about the Foundation's strategic plan or its process, don't hesitate to contact me at 800.753.2357 or kbrown@CdLSusa.org.

Kelly

Kelly Brown
Acting 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.

In This Issue

Importance of Research Grants

Guest Author Richard Haaland, Ph.D., CdLS Foundation Research Committee Chair; Vice President, Board of Directors

The CdLS Foundation is proud to announce support of four exciting research projects in clinical and basic science. We hope they will lead to a better understanding of CdLS and its complications, as well as improved care and quality of life for everyone with CdLS. The CdLS Foundation selected the following research projects for funding:

Dr. Siddharth Srivistava, of Johns Hopkins Hospital, will use a picture exchange communication system to teach communication skills to nonverbal individuals with CdLS and their families. Dr. Srivistava will evaluate communication before and after the program to determine if the picture exchange communication system could be used to improve communication for all individuals with CdLS.

Dr. Jason Mills, of the Children's Hospital of Philadelphia, will use laboratory techniques to treat stem cells collected from children with CdLS in order to grow tiny structures with similar properties to developing brains. Dr. Mills hopes to use these laboratory-developed structures to identify the specific molecular signals that result in brain development problems among children with CdLS.

Dr. Lynne Kerr, of the University of Utah Medical Center, will use a special test to identify dysfunction in the part of the nervous system that controls unconscious body functions. This test will be used to help identify specific nervous system issues individuals with CdLS have and could help determine the best way to reduce complications caused by these problems.

Genetic Counselor Sarah Noon, of the Children's Hospital of Philadelphia, will examine blenderized diets and their use in feeding individuals with CdLS through feeding tubes in place of formulas. Ms. Noon will evaluate various combinations of blenderized diets being used by caregivers to look for the best way to improve nutrition and food tolerance for children with the syndrome.

All grant recipients are required to present their finding at a forthcoming CdLS Scientific Symposium, as well as write an article for *Reaching Out*.

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Let us know your thoughts about this issue of *Reaching Out*, and what you'd like to see in future issues, by visiting http://bit.ly/4thQtr2015RO.

Reaching Out
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Genotype-Phenotype Correlations in Cornelia de Lange Syndrome

Ian Krantz, MD, and Sarah Noon, MS, CGC, Center for CdLS and Related Diagnoses, Children's Hospital of Philadelphia; members, CdLS Foundation Clinical Advisory Board

Our bodies are made up of billions of cells and within each cell there are chromosomes which are the structures that hold all of our approximately 20,000 genes and genetic information. Genes function as the body's instruction manual telling our body how to grow and develop. We have two copies of each gene as we get one set from our mothers and one set from our fathers. Genes are made up of genetic material, called DNA, and they serve as the blueprint from which proteins are made. Proteins are the basic building blocks of the human body performing specific functions so that our bodies work properly controlling everything from our heartbeat to determining our eye color (Figure 1).

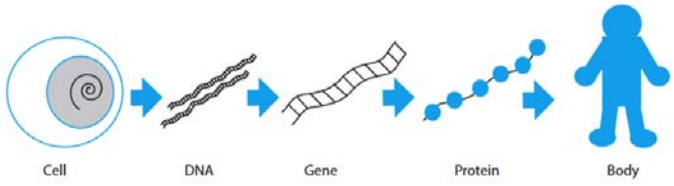


Figure 1. Cells contain DNA material which is organized into genes. Genes make proteins which are the basic building blocks of the body responsible for making our bodies work properly.

Sometimes changes (or mutations) spontaneously occur in genes that prevent them from working properly altering the proteins that are made. As a result, this affects various bodily functions including growth and development. Currently, mutations in five different genes, *NIPBL, SMC1A, SMC3, HDAC8* and *RAD21*, have been associated with Cornelia de Lange Syndrome (CdLS) (and several others, such as *AFF4, TAF1* and *TAF6* that have been associated with clinical pictures similar to CdLS). The association between the presence of a certain mutation or mutations in a specific gene (genotype) and the resulting presence, absence or severity of symptoms or clinical features (phenotype) is called a "genotype-phenotype correlation".

Now that there are five genes known to be involved in CdLS certain genotype-phenotype correlations have been observed for each of the genes. Changes in these five genes are found in approximately 65 percent of individuals with a clinical diagnosis of CdLS, with the vast majority being caused by mutations in *NIPBL*.

NIPBL

Individuals with classic findings of CdLS, including characteristic facial features and limb anomalies, are likely to have a change identified in the *NIPBL* gene. However, changes (or mutations) in *NIPBL* have been found in individuals with both classic and mild presentations. The degree of severity depends on the specific type of mutation that occurs and where the mutation falls within the *NIPBL* gene.

A truncating (or frameshift) mutation is one type of mutation that tends to have a more significant effect on the gene that can ultimately block protein production. Therefore individuals with truncating mutations typically present with a more classic or severe form of CdLS.

Missense mutations are a different type of mutation which generally only slightly changes the protein. Therefore, individuals with missense mutations typically present with milder forms of CdLS, since their proteins likely retain some residual function.

SMC1A and *SMC3*

Individuals with *SMC1A* or *SMC3* mutations typically have fewer structural differences, such as a limb difference or heart difference. Such individuals also tend to present with less significant growth restriction than those with *NIPBL* mutations. However, individuals with *SMC1A* or *SMC3* mutations will still typically have intellectual disability that can range from moderate to severe [Deardorff et al 2007].

Subtle facial features in individuals with *SMC1A* or *SMC3* mutations may differ than those observed in "classic" CdLS caused by *NIPBL* mutations and can include slightly flatter and broader eyebrows with a broader and

longer nasal bridge [Rohatgi et al 2010]. Specifically, individuals with *SMC3* mutations often have subtle or absent synophyrs (connecting eyebrows), wider nose with a rounder tip, and a well-formed philtrum (vertical groove between the base of the nose and upper lip).

RAD21

Individuals with mutations in *RAD21* typically do not have major structural differences. Individuals with *RAD21* mutations have milder cognitive impairment compared to those with "classic" CdLS. These individuals typically display growth retardation, minor skeletal anomalies, and facial features that overlap with CdLS. [Deardorff et al 2012].

HDAC8

Individuals with mutations in *HDAC8* have facial features which overlap with CdLS but typically display delayed closure of the anterior fontanel (the opening or "soft spot" on the top of the head in babies which typically closes around one year of age), hooded eyelids, a wider nose, varying pattern of skin pigmentation, and friendly personalities. Growth restriction also tends to be less significantly affected with this gene and a lower frequency of microcephaly (small head circumference) is reported.

In females, the severity of clinical presentation caused by mutations in *HDAC8* is variable, since this gene is on the X chromosome and females have two X chromosomes while males have only one X chromosome and a Y chromosome. Since women have two X chromosomes in every cell, they randomly shut off one copy of the X chromosome (called X-inactivation). Therefore, depending on how many X chromosomes with the mutation versus those without the mutation are inactivated will directly influence the severity of their clinical presentation. (Though *SMC1A* is also located on the X chromosome this X-inactivation process does not apply to the *SMC1A* gene).

***NIPBL* mosaicism**

A recent study led by Dr. Raoul Hennekam in the Netherlands [Huisman et al 2013] has found that mosaicism for *NIPBL* mutations may be found in up to 30 percent of individuals with CdLS who have tested negative in the blood for mutations in the known CdLS genes. Mosaicism means that an individual has a change in a gene which is present in only some but not all of the cells in their body. If an individual is mosaic for a change in *NIPBL*, we may not be able to identify this change by testing only their blood; instead, we may need to test other cells from other tissues such as cheek cells, also called buccal cells.

Depending on the number of cells carrying the mutation and the tissues involved, an individual with *NIPBL* mosaicism can theoretically present with a more mild form of CdLS. However, additional research is needed in this area since only a few patients with *NIPBL* mosaicism have been identified thus far.

What it all means

While clinicians and researchers have reported these genotype-phenotype correlations (summarized in Figure 2) it is important to note that they are generalities and are often true but these "rules" are also frequently broken. We have seen children with severe (truncating or frameshift) mutations in *NIPBL* who have a mild clinical picture (phenotype) and conversely also seen children with mild (missense) mutations in *NIPBL* who have a very severe clinical picture.

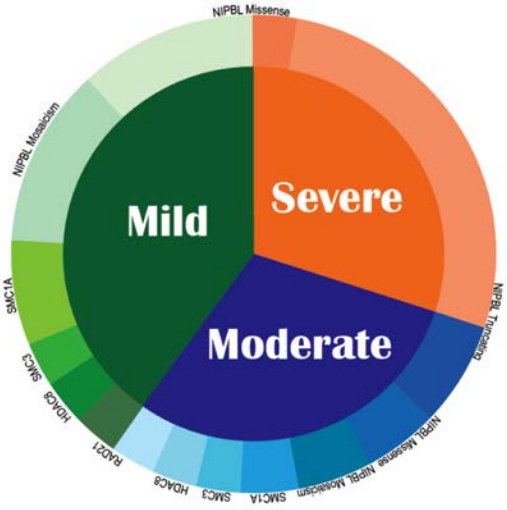


Figure 2. With five genes known to be involved in CdLS certain genotype-phenotype correlations have been observed for each of the genes.

There is still much work to be done to fully understand the correlations between the genotype and phenotype in individuals with CdLS, but there are general principles emerging and clinicians are getting better at using this information to help provide some guidance and prognostic information to families.

Continued on page 17

A Closer Look at SMC1A: Katie

We want to thank Katie's mom, Mary, for sharing Katie's story with us.

Katie is 28 years old and was diagnosed with CdLS at three months old, but was nearly 20 years old when she was identified as an individual with the SMC1A mutation. At birth, she was 20 inches long and weighed 8 pounds 6 ounces. Today she is 5' 1" and weighs 150 lbs.

For the first year, it was very difficult for Katie to gain weight because of her severe reflux. At the time there were no medications that worked to control it and the doctors would not perform a Nissen. Like many infants, she had a Nasogastric (NG) tube, but when it proved to be more of an irritant, we switched back to a bottle thickened with cereal. We were dealing with her reflux, and by the age of four or five, we thought she had 'outgrown' it. Learning about "silent reflux," we now know this was a mistake. Today, we manage her reflux with Prilosec.

From infancy, Katie was sickly, and was hospitalized a few times and had chronic sinus infections. Following a particularly serious sinus infection, she had surgery at age 12. Her health improved dramatically after the surgery and she has not been hospitalized since.

Katie started using words when she was about two in conjunction with the use of "signs," words like "cookie" and "more." Despite her language limitations, she does hold a job and is currently working as a greeter at a local hair salon.

Our biggest concern is finding a suitable living arrangement in the future. While we'd love to give her greater independence, we must be constantly vigilant. She went through a period of escaping from the house, and if no one is in the kitchen, she's quick to get into things and make a mess. She likes helping with cooking and wants to help with other chores. She's definitely trainable, and we look forward to her gaining more independent living skills.

A Closer Look at SMC3: Kinsley

We want to thank Kinsley's mom, Sarah, for sharing Kinsley's story with us.

When I was expecting Kinsley, I had numerous sonograms and even a 3D/4D ultrasound, but CdLS was never detected. My obstetrician-gynecologist said, "She's just a petite baby, like her momma." When Kinsley was born at 36 weeks, she weighed 4 pounds 10 ounces, was 16.5 inches long and had a cleft palate (which was repaired this past April).

We knew Kinsley had a syndrome and were relieved when we finally knew its name. Testing was done in December 2013, and results came back February 2014. She wasn't clinically diagnosed; the genetics doctor wanted testing before fully diagnosing her.

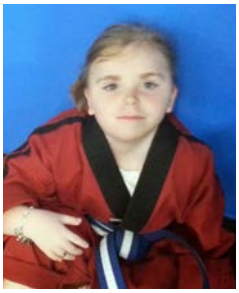
Kinsley, who is four years old, has asthma, GERD, a tethered cord, microcephaly, short stature, micrognathia, and vesicoureteral reflux (kidney reflux). She has a gastrostomy-button and eats some foods (she loves macaroni and cheese, Spanish rice, Cheetos, scrambled eggs, and bean and cheese tacos), but has oral aversion to many items (she will not touch Jell-O, pudding, applesauce, whip cream, or mashed potatoes).

As with many kids with CdLS, she has sensory issues. She hates socks and shoes, unless she can chew on them. She likes bead necklaces, and she likes to turn her tricycle over and spin the tires and lick them. She loves music and will sing along and dance to every show on the Disney Jr. channel.

Our biggest challenge has been keeping her well, but also not keeping her in a bubble. All kids need to play, but we have to be very careful.

I've learned never to be surprised at what she can do or can't do; every day Kinsley amazes us with something different.

A Closer Look at RAD21: Baylee



We want to thank Baylee's mom, Cheryl, for sharing Baylee's story with us.

My first thought after I got Baylee's diagnosis last December was "finally." Finally, we know what it is; now we know what to do and where to go from here.

Baylee's doctor diagnosed her with a photo and an app on her phone. Baylee had some of the characteristics of CdLS, but not the normal facial features, so testing was done to confirm. Of course, I went home and Googled CdLS. I read the information and was thinking, "check, check, check and check." Long eyelashes, cleft palate, hyper-focused. Baylee didn't talk until age three. We used sign language for most things, and she would use the word "mom." She lost the use of sign after she started school since they focused on her verbal skills. We still go to one-on-one speech therapy.

Moving forward, I hope to ensure that she has what she needs, whether it's the correct pencil or the correct desk height. But I don't want to over do it either. I have noticed that she has problems with writing, and her hands get tired. I have discussed with her school about her condition the best I can as this is really new and a lot of unknowns.

At seven years old, I don't treat my daughter any different. I have the same expectations for her as I do my oldest child, and I know she will do it in her own way. But she will do it. Baylee is in karate, and has become stronger than when she started. Her instructor does not treat her differently, nor expect any less from her than any other student.

We are still learning and dealing with her diagnosis. There isn't a lot of information with her specific gene change, but everyday we learn and get through.

A Closer Look at HDAC8: Cade

We want to thank Cade's mom, Jennifer, for sharing Cade's story with us.

We did not receive a CdLS diagnosis until Cade was 11 years old, and by that time we'd done extensive testing of all kinds. We got our diagnosis through whole exome sequencing, and it really was the last option for us to find out what was cause was for all of his issues. We were actually very happy to finally have an answer or underlying reason for his "laundry list" of problems. I felt like we finally had a place to belong.

Cade, who is 13 years old, takes 15 different medications daily. He talks to people when he feels comfortable, but he does have social language issues and has been in speech therapy since 14 months of age. He gets speech therapy and occupational therapy (OT) at school. He was late on all developmental milestones and started our state's early intervention program when he was five months old. Currently he sees specialists in ophthalmology, endocrinology, immunology, sleep/pulmonology, psychiatry, psychology, and gastroenterology. In the past he's also seen neurology, ear, nose and throat, and dermatology.

In the past – for four years – he had Intravenous immunoglobulin (IVIG) infusions every four weeks. At this point, his immune system is better, and he is on daily prophylactic antibiotics, but it is concerning to me how fast things can change. Education issues are getting harder as well.

I feel like we'd gotten so many answers, before we even had a diagnosis. Since we did the sequencing, we do know that I passed on the gene change. I do have concerns that my daughter may also carry the mutated gene.

I think the most important thing for any family facing challenges to do is to find support. It's important to find support locally even though the people you find will most likely not be dealing with the exact same things you may be going through. As a parent, you need to nurture yourself so you can be a better nurturer for your family.

A Closer Look at NIPBL: Maya

We want to thank Maya's mom, Rachel, for sharing Maya's story with us.

When Maya was first born, she did not have the same features she does today. Her nose was triangular and upturned and she had a small head, but her eyebrows were perfect. In the delivery room, the doctor dismissed my worries over her low birth weight (4 pounds, 12 ounces), but a geneticist did come see Maya upon the neonatologist's request. He told us there was something he wanted to test for.

It wasn't until the next day that the pediatrician told us that she was being checked for Cornelia de Lange Syndrome (CdLS). My gut told me something was going on, so I calmly prepared myself for it. At this point, I knew nothing about CdLS and did not wish to. While we waited the weeks for the test to come back, Maya's eyebrows grew in thicker and together, preparing me even more for this certain diagnosis.

Weeks went by and we received a call that Maya's results were in. We were told that she had a certain variant on NIPBL that had never been found to be associated with CdLS. After testing for the mutation in my husband and me, it was determined that the variant was new to Maya and the diagnosis made. This was actually the first time I could handle researching CdLS and learning about what had stolen so much from my baby. It was then that I came to terms with what we would battle for the rest of her life.

So far, Maya's biggest issue is eating. She will take tastes of food and sips of water, but her main nutritional source is her G-tube. I worry that she may never eat fully by mouth, but I also am thankful that when Maya could not do this herself, there was a way to feed her. No parent envisions his or her child having a feeding tube, but it's been our biggest blessing. Maya has grown so much since it was placed. At just over a year old, she can now say "mama," "dada," and "bye bye." Some days she babbles lots, and some days she doesn't say a word. She still isn't crawling or pulling up, and has no limb differences.

CdLS 411: Malrotation

What is malrotation?

Malrotation occurs when the intestine does not fold or rotate properly in early fetal development. Malrotated intestines may "flop" around since they are not properly attached to the abdomen wall. Malrotation is a malformation that is present at birth; however, symptoms can show up at any age.

What is volvulus?

Volvulus is a type of malrotation where the intestines twist around one another. As this happens, the duodenum may be twisted shut, which can induce vomiting or pain. If the intestine twists further, it may compress the blood supply. This can cause severe pain as the blood supply to the small intestine is cut off. The twists can also cause bowel obstruction by blocking the gastrointestinal tract.

Symptoms of Malrotation and Volvulus

- Vomiting
- Pain
- Swollen abdomen
- Bloody or loose stools
- Rapid heart rate and breathing
- Little or no urine
- Fever

Malrotation is a Medical Emergency

If the twisting cuts off blood supply to the intestine for too long the intestine will die. The entire small intestine is usually involved, and when this much bowel is lost, most children require intravenous nutrition for the rest of their lives.

If volvulus is not present, surgical correction is usually done within a few days. If volvulus is present, surgery should occur immediately. The surgeon will untwist the intestine and try to determine if any intestine has died. Once dead intestine is removed, the remaining intestine is attached to the back of the abdomen to prevent future volvulus.

Information taken from CdLS CAB Member Carol Potter's article: "GI Distress, Malrotation, Volvulus, Bowel Obstruction, Pancreatitis."

Genitourinary Issues in CdLS

By Ming-Hsien Wang, MD, Spiegel/Nichols Assistant Professor of Pediatric Urology, Director of Pediatric Urology Fellowship, Johns Hopkins Medical Institutions, Kathy Lue, MD, Research Assistant in Pediatric Urology, and Timothy Baumgartner, MD, Research Fellow in Pediatric Urology.

Genitourinary (GU) manifestations are commonly found in patients with Cornelia de Lange Syndrome (CdLS). These can encompass anatomic and functional anomalies of the kidney, urinary tract, reproductive organs, and external genitalia. Reflux of urine into the kidneys, improper drainage of the kidneys, and abnormal development are the most common abnormalities of the urinary tract, and can lead to a decline in renal function and recurrent infections. For males, undescended testis is the most common anomaly of their external genitalia, which can affect fertility and increase their risk of developing testicular cancer. Hypoplastic (underdeveloped) genitalia and hypospadias, where the urethral opening is located on the underside of the penis rather than at the tip, can also be present. Similarly, females with CdLS can have small external genitalia, abnormal uterus, and ovarian atrophy (decreased size/function).

The first step in management is a detailed family, medical, and surgical history. This can aid in identifying genetic or pre-existing risks factors for development of a concomitant GU disease. A review of systems should focus on a detailed analysis of urinary and bowel habits, nutritional habits, and abdominal or groin pain. The physical exam should assess the abdomen for fullness or masses, curvature and size of the penis, position of urethral opening, circumcision status, asymmetry or discoloration of the scrotum/labia, location and ability to examine the testicles, location and patency of the vagina, patency of the anus, and lower back anomalies. As an infant, ultrasound of the bladder and kidneys should be completed to help detect any GU anomalies which may prompt further imaging. Ultrasonography is an ideal initial study as it is non-invasive, cost effective, and does not expose the child to any radiation.

A number of these exams can be accomplished at home during routine care for the patient. If hypospadias is a concern, observing the patient while voiding can help locate the urethral opening. A testicular exam is usually best accomplished in a warm bath if presence of testes within the scrotum is questionable. Any concerns should be brought to the attention of your primary care physician.

If an undescended testis is diagnosed at birth, it is observed for positional changes during the first 6 months of life to allow for spontaneous testicular descent. After six months of age, surgical treatment by a pediatric specialist is encouraged. Since hypospadias does not routinely affect the patient's ability to empty their bladder, surgical repair is not recommended until at least six months of age and may require multiple procedures depending on its severity. If renal or bladder anomalies are detected, the patient may be placed on antibiotics to prevent infections or require future interventions.

By increasing awareness of the GU manifestations that can present in CdLS, early detection and appropriate management can be better achieved. There are many other, more rare diagnoses related to the GU system that the patient may develop. Therefore, it is imperative to be vigilant to any change in the patient's exam or symptoms which may warrant a formal evaluation with a board certified pediatric urologist.

A Closer Look at Genitourinary Issues: Kailani

Six-year-old Kailani was almost two years old when she was diagnosed with her first urinary tract infection (UTI). Although, her urologist, pediatrician, and ENT, agreed that since she had been treated for so many ear infections since around six months of age, that we may have missed some UTIs and been unknowingly treating them.

We first noticed that there was a problem when Kailani was sleeping all day and night and refusing food and fluid (she was able to get nutrition through her g-tube). She seemed very confused and lethargic when she was awake, and she had a high fever, and that's what landed us in the hospital for a week and seeing a urologist. Once she had the UTI diagnosis I knew what to look for when I suspected an infection. Aside from the extra sleep and fever, the smell of her pee was a good indication (gross, but very helpful).

The first step in treatment was an antibiotic, in hopes she wouldn't get another UTI. After her first renal ultrasound, we started to see Dr. Canning at the Children's Hospital of Philadelphia (CHOP). She was diagnosed with stage two vesicoureteral reflux (VUR) in her right kidney. She was then put on daily antibiotics. We were told 90 percent of children grow out of their low grade VUR – including some individuals with CdLS.

Daily antibiotics worked well for Kailani, and for a few years we only averaged about two or three febrile UTIs/kidney infections. But when she was four, her UTIs became more frequent. We changed medications and repeated a Voiding Cystourethrogram (VCUG) that showed the VUR was actually in both kidneys. A dimercaptosuccinic acid (DMSA) scan showed scar tissue from years of infections. In October 2013, she had a deflux implant and dilation of her urethra, which ultimately did not work. Her infections were just as frequent after these procedures.

Kailani had the deflux implant removed and an open bilateral re-implantation of her ureters in May of 2014. After the surgery it came to our attention that she could not empty her bladder; she is now catheterized every three hours. We recently had a video urodynamic study done and she is no longer having any kidney reflux, which is great news. She is still on a daily Bactrim medication as a precaution, but hopefully she will be off of it soon.

Her surgery last May was difficult on our little family. The recovery was rough and learning how to catheterize Kailani—and getting her used to it—was a difficult time. Her big sister, Adalei, stays right by her side holding her hand during "cath time."

If dealing with VUR, being very proactive about getting a urinalysis is so important. As soon as you suspect a UTI, call your pediatrician. For anyone dealing with intermittent catheterization: it gets easier. Having a six year old in diapers requiring that kind of attention has its challenges. It was scary at first, but after a year, it's become our "normal."

CdLS Registry: Enroll Today!

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

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 CoRDS registry specifically houses basic contact and clinical information on any individual who chooses to enroll and who has been diagnosed with a rare condition.

For those of you interested in participating in this registry, visit http://bit.ly/CdLSReg to begin the process.

Anesthetic and Airway Management of Patients with Cornelia de Lange Syndrome

Considerations for Families

Most children with Cornelia de Lange Syndrome will have to undergo sedation and/or anesthesia at some point in their lifetime. Whether it's for a dental cleaning, imaging study or surgical procedure, this page should help give you an overview of what to expect for your child's anesthetic/ airway management.

My child's doctors tell me that he/she will be undergoing anesthesia for a surgery/ procedure. What is anesthesia and what exactly will happen?

We will explain by dividing the process into preoperative, intraoperative, and postoperative

Preoperative

Sedation medicine will be given to prepare/calm child before the procedure. These may be given orally or intravenously.

- Let the anesthesiologist know who your geneticist is, or any other doctor that knows the medical needs of your child. Let the doctor know if your child has cognitive and/or behavioral issues that may lead to a lack of cooperation.
- Some hospitals may allow you to accompany your child into the operating room to calm him/her down until he/she is put to sleep if you think that would be beneficial.
- Notify the anesthesiologist if your child has had previous complications when sedated and/or put under anesthesia, and what those complications were.
- If you are aware of any specific medications that your child is sensitive to, let the anesthesiologist know.
- If you are aware of any specific medications that you know your child benefits from, such as something to reduce postoperative nausea and vomiting, let the anesthesiologist know.
- Notify the anesthesiologist if your child has GERD (gastroesophageal reflux disease), the movement of contents from the stomach back to the esophagus.
- Notify the anesthesiologist if your child has a history of pneumonia.
- Notify the anesthesiologist if your child has very small facial features and/or cannot open his/her mouth easily.
- Let the anesthesiologist know if your child has a challenging airway and/or a history of difficult intubation (the placement of a breathing tube).

Intraoperative

What happens during your child's procedure under anesthesia:

- Some children are sensitive to certain medications. Ask the anesthesiologist what medications he/she plans on using.
- If your child has challenging airway anatomy, it may be difficult to visualize the voice box. Ask the anesthesiologist which instruments he/she plans on using.
- Ask the anesthesiologist if he/she thinks it will be difficult to place a breathing tube and the reasons why.
- Often times, a smaller breathing tube is needed to intubate children with CdLS. Ask the anesthesiologist if he/she plans on using a smaller than expected tube.
- Ask the anesthesiologist if he/she plans on using any specialized devices, such as a laryngeal mask airway, or a flexible fiberoptic bronchoscopy.
- Ask the anesthesiologist if he/she expects any complications while intubating, such as a decrease in oxygen levels or an increase in carbon dioxide levels.
- Ask the anesthesiologist if he/she expects any major complications during the procedure, such as a respiratory arrest and/or a cardiac arrest.

- Aspiration is a high risk for children with CdLS, especially those with GERD, so ask your anesthesiologist what precautions he/she plans on taking to prevent it.
- If the anesthetic technique your anesthesiologist uses seems to work for your child, take note of it so that you can inform anesthesiologists in the future.
- Likewise, if anything did not go well with your child's airway and anesthetic management, take note of it as well so that you can ensure it does not happen again.

Postoperative

The emergence (waking up) from anesthesia and what happens after the procedure is over.

- Compare your child's general temperament (how they generally act everyday) to their temperament upon waking up.
- Children with CdLS seem to be unpredictable in how they wake up from sedation or general anesthesia. Note what medications were used and how your child woke up from them.
- In addition, some children wake up slower than expected or some wake up quicker than expected when coming out of sedation or general anesthesia.
- Note if your child woke up calm and aware, or if your child woke up disoriented, agitated, crying, etc.
- Your child may be treated with medication if the wake up did not go well.
- Look for breathing problems after the procedure, which would include problems with oxygen levels, problems with carbon dioxide levels, respiratory depression, or delayed extubation (the removal of the breathing tube).
- Most of these problems happen soon after surgery if they are going to occur; therefore, overnight stay is not always needed.
- Make sure the nurses in the room that your child is taken to after the surgery know that your child has CdLS and that there is an increased risk of a postoperative breathing problem postoperatively.
- Keep track of your child's airway and anesthetic management for every procedure. This will give you an idea of what works and what doesn't work for your child.

Contact the CdLS Foundation with any information about your child's experience with sedation and/or anesthesia.

Created by Dr. Yvon Bryan, pediatric anesthesiologist, Wake Forest Baptist Health Medical Center, Winston-Salem, NC; 2013 CdLS Foundation research grant recipient.

The Anesthetic and Airway Management of Patients with CdLS guide was created as a result of a 2013 CdLS Foundation research grant. **We also have a specific version of this targeted for medical professionals, which you can give to those on your child's medical team.** If you would like to receive a version specific for professionals, please contact Deirdre Summa, at familysupport@CdLSusa.org, or call 800.753.2357.

Stronger Together in Orlando

Join us for the CdLS Foundation National Family Conference, June 23-26, 2016, in Orlando, FL. Conference provides education and support to families of individuals with CdLS. Attendees receive free head-to-toe consultations with experts from a range of medical and educational fields; attend workshops on legal concerns, educational issues and medical/behaviors challenges, and have opportunities to meet other families facing similar challenges.

FAQs

When can I register for conference?

Registration for conference is scheduled to open February 15, 2016. The early bird registration and scholarship request deadline is March 25; the final deadline is May 27. Registration is available online or through a paper form. Conference can accommodate up to 150 families. Once that number is met, registration is closed.

What does registration cover?

The registration fee covers all workshops and sessions, medical and educational consultations, and the following meals: Thursday night ice cream social; Friday breakfast, lunch and dinner; Saturday breakfast, lunch and dinner; and Sunday continental breakfast. It does not cover hotel or travel expenses.

REGISTRATION FEES

	Before 3/25	After 3/25
Adult (18+)	$330	$380
Person providing childcare	$230	$260
Children 3-18	$180	$205
Children 2 and under	No change	No change
Person with CdLS	No charge	No Charge
One-day professional	$110	$110

Where are we staying, and when can I book my room?

The conference takes place at the DoubleTree by Hilton at Sea World (www.doubletreeorlandoseaworld.com), just 15 minutes from the Orlando International Airport. Attendees are responsible for booking their own rooms. Hotel reservations will be accepted beginning December 3, 2015. We will provide you with a code to get the reduced room rate of $112/night plus tax. The reduced room rate is only for people registered for conference. All non-registered guests using the conference room rate will be removed from the room block and be required to pay the regular room rate.

What are my childcare options?

Children with CdLS (any age) can attend **Jindi's Place** at no cost. Activities include arts and crafts, movies, games, toys, reading, and a quiet room. Arrangements can be made for tube and bottle feedings.

Programs for siblings on Friday and Saturday:

Tiny Tots: for siblings aged six months to seven years. This program closes down during lunch each day (parents are responsible for picking up their child during this time).

Kids Explore!: for siblings ages five to 17. Participants go off-site. Lunch is provided. Individuals with CdLS over the age of five can participate in *Kids Explore!* if they have a paying chaperone over the age of 18 to accompany him/her.

The cost is $125 per child for two days and $80 for one day, with discounts available for each additional child. There is a mandatory meeting Thursday at 7 p.m. for those using any of the childcare programs.

More details on conference, as well as registration links, amusement park discounts, and transportation information will be on the conference web page, www.cdlsusa.org/what-we-do/biennial-cdls-conference.htm.

Mailbag

– Jack –

My son, Jack, and the five other students in his sixth grade special education class walked nearly 400 miles this past school year. Throughout the year, they took turns walking on a treadmill during class, starting at five minutes and working their endurance up to 15 minutes. The students tracked their miles on a map of Wisconsin, and learned about the cities that were on the path. They would then visit the community's website to discover places of interest.

This exercise helped immensely with challenging behavior, and they plan to continue the program this year as well. I asked the school to include walking on the treadmill in his IEP, twice a day: morning and afternoon. Jack lost some weight, which was great because his height has plateaued and even with a healthy diet, he wasn't getting as much exercise as he really needed. He has increased in muscle tone and hand-eye coordination during these treadmill sessions while listening to music or watching a movie on his school iPad.

The students in the class have been together since Kindergarten and are now in 7th grade. They all are comfortable with each other and have learned to accept each other's differences. This summer, Jack worked with a tutor at our local library working on math, reading and life skills such as handling currency. His tutor just happened to be his teacher from 3rd-5th grade

Share your Mailbag or Super Sibling Story!

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

Super Siblings: Colin and Amelia

Hi! My name is Amelia and I am nine years old. My brother, Colin, is twelve years old and has CdLS. My brother likes playing with Legos and Tinkertoys, listening to music, and loves taking stuff apart with a screwdriver. He really likes snakes, but is happy with our puppy, which he lets kiss his face. We like to build forts together on the weekends. I admire Colin because he is like no one else and he is good at swimming and breakdancing.

We go to different schools, but I wish I could go to the same school as my brother. I worry he gets teased at his school because when we went to summer day camp together some kids teased Colin, so I would tell the kids to stop. I wish I could make sure that doesn't happen at his school.

My friends are curious about Colin and ask me about him. I tell them that he has CdLS. When they ask, "What is CdLS?" I tell them that his brain doesn't work very fast and it takes him longer to do things. He works really hard at trying new things and I am helping him learn to read. I am proud of him. Sometimes I get really frustrated when he copies me, but I love Colin no matter what.

Grandparent Greeters

Grandparent Greeters are a vital group of CdLS Foundation volunteers who reach out to new grandparents or those who want to connect with other grandparents in their region. Two of our Grandparent Greeters are bilingual and available to speak with Spanish-speaking grandparents.

If you would like to be a Grandparent Greeter in your area, or would like to be put in touch with one in your region, contact Lynn at families@CdLSusa.org or call 800.753.2357.

Meet Grandparents Karen and Kate

Eric will be 20 years old in August. His Grandma Kate (pictured below, right) and I both live in different directions, seven hours from Eric's home in North Carolina, though we would love to live closer to be more involved in Eric's life. When we visit him, or he comes to see us, he always has a big smile on his face. He will sign for "Grandma" and "Grandpa." Even though he has no grandfathers living, he still remembers them and signs for them.

When he visits Grandma Kate's home in Florida, he loves to ride in the golf cart around the property. One time he even started it up himself and was driving it around before he was caught! Visiting my home in Pennsylvania, he loves to turn the kitchen fan on and off and play with the umbrellas on the deck.

At his house, Eric enjoys going to McDonald's, Bo Jangles and the Kangaroo convenience store, where all the staff knows him. He likes to visit the mall, the pet store, and Barnes and Noble. Nothing keeps his attention, so Eric always has us constantly on the go. At home he gives the sign of "boat" to watch The Deadliest Catch or the horse sign to see a National Geographic show while he relaxes.

Eric has a wonderful family with parents Chris and Amy and siblings Elizabeth and John. If his parents are gone, we can always count on Elizabeth and John to help with sign language. Eric has attended three CdLS Foundation National Family Conferences with his family, one of these being with the two grandmothers. Eric's cousins, Laura and Faith, have run the Chicago Marathon for Team CdLS, and I've presented at a medical conference in Harrisburg to educate hospital staff about CdLS.

On the Cover: Mega

Mega was born in October 2014 in Atlanta, GA. I learned to feed him in the NICU, so he wasn't on a feeding tube for long after he was discharged. He has been eating fine ever since.

Mega is a mama's boy and loves to cuddle with me; I am the only person that can calm him down when he's upset. He loves music. He gets quiet and just gazes when I play music that he likes. He is now rolling over and won't stop. I try to use my hands as a barrier to help him try to push off but he is not sitting up yet. Mega is also beginning to "coo" and loves the sound of his own voice

He loves to look at lights and enjoys baths, but dislikes being cold. He is a major people person and seems to like everyone he meets.

2015-16 CALENDAR

October 3
Midwest Region Family Gathering
St. Paul, MO

Maddy's Run 5k
Salt Lake City, UT

October 11
Team CdLS
Bank of America Chicago Marathon
Chicago, IL

October 17
Team CdLS
Baltimore Running Festival
Baltimore, MD

Wisconsin Family Gathering
New Berlin, WI

October 23-24
Board of Directors Meeting
Orlando, FL

October 24
Southeast Region Family Gathering
Orlando, FL

Utah Family Gathering
Salt Lake City ,UT

November 1
Team CdLS
TCS New York City Marathon
New York, NY

December 5
West Region Family Gathering
Escondido, CA

February 14, 2016
Skechers Performance Los Angeles Marathon
Los Angeles, CA

Board Corner

Bob Boneberg Esq., President, Board of Directors

I am writing my last message as president with great appreciation and profound humility. CdLS Foundation Board members serve a maximum of two consecutive three-year terms and, as my six years end in December, it is appropriate to thank those who have made my Board service so very special to me.

During the time that I have been privileged to serve on the Board, I have been helped and guided and inspired by many, many people beginning, of course, with all the past and present members of the Board who volunteer to serve. I have tried to learn from, and be guided by, each of you. Also, I thank the Foundation staff who helps the Board as they help so many others, with skill, insight and patience. Thank you also to the many doctors, scientists, social workers, and other professionals who not only address the problems of today, but also help the Board think about the best way to meet the challenges of tomorrow. Thank you very much also to the friends and supporters who help the Foundation in many ways, seen and unseen. Truly, without your efforts, the Foundation would not be what it is today.

And, most especially, thank you to all those who have been diagnosed with CdLS and their families and friends. I have met and spoken with you at family gatherings and conferences and at places in between. Thank you for your advice, suggestions, your good humor and your support. You are our Foundation.

I know that you all will continue to guide and support the Foundation and its Board, and I look forward to seeing how the Foundation grows and thrives in the years to come.

I look forward to seeing you again just around the next corner.

All the best,
Bob Boneberg

2015 de Lange Society Inductee: Phyllis Musumeci

Phyllis Musumeci lives in Viera, FL, and has been involved with the CdLS Foundation for more than 20 years. Her son Christian, 23, has CdLS.

When Phyllis first began volunteering for the Foundation, it was because of those who reached out to her. She officially became a Regional Coordinator in 2002, after raising awareness and support for the CdLS Foundation and families in her region for many years before that.

“I really liked the way the Foundation operated,” said Phyllis. “When I first joined, someone reached out to me, and I found that very helpful, so I wanted to do the same for other people.”

Some of Phyllis’ favorite memories as a volunteer involve spending time with other parents.

“Talking with other parents, meeting people at picnics, just knowing that you have this in common you’re able to connect much easier with people,” she said. “I never felt like I did that much, and always wished I could do more. It was a surprise and an honor to be asked to join the de Lange Society.”

To other parents who may be thinking of becoming a Foundation volunteer, Phyllis says, “give it a try.” She continues, “It’s very rewarding to be able to reach out to others. It’s natural to be very nervous at first, but when you realize you’ve been in their shoes and know what they’re going through, it makes it much easier to talk to them.”

Give at Work

Workplace giving is an easy way to support the CdLS Foundation. Once you sign up for your employer's workplace giving program, a set amount (you decide) is deducted from each paycheck. Those funds are then sent to the Foundation, usually quarterly. Some companies have their own workplace giving program, while others participate in the following programs:

Combined Federal Campaign: Federal employees and military personnel can support the Foundation through the annual Combined Federal Campaign (CFC), which runs September 1 through December 15. The Foundation's CFC number is 11777.

United Way: If your company participates in a United Way campaign, you'll need to write in "Cornelia de Lange Syndrome Foundation" as we do not have a United Way identification number.

WELCOME NEW FAMILIES

California
Kelly and Ken and son Johann, born November 12, 2012

Florida
Fatmira and Herion and daughter Bora born on January 11, 2014

Cristiana and daughter Amy, born November 25, 2010

Georgia
Courtney and son Dakota, born March 27, 2015

Kansas
Kristen and son Demetri born on February 27, 2009

Angel and son Anthony born June 9, 2005

Louisiana
Brittney and Dustin and son Cohen, born February 26, 2013

Missouri
Sara and Casey and son Simon, born August 22, 2010

Nikki and Dustin and son Gavin, born March 10, 2013

Nebraska
Rebecca and son Alexander, born December 18, 2014

Ohio
Scarlett and daughter Savannah born January 8, 2015

Pennsylvania
Sue and daughter Julia born November 18, 1995

Texas
Crystal and daughter Peyton, born July 9, 2012

Genotype-Phenotype Correlations in Cornelia de Lange Syndrome cont.

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Donations from 05/16/2015- 9/10/15

Gifts that Count - In Honor/Celebration

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Alice DeLucia

Andrea Brennan
Amy and James Brennan

Anissa Reynolds
Dora and Ted Arenas

Austin Cadena
Molly Brantley

Brianne Prada
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Our Deepest Sympathy

Evan Michael Prince
December 2,1994 – June 6, 2015
Son of Angie Anders-Mutter and Richard Mutter, Jr.
1915 Highland Ave
Shreveport, LA 71101

Ethan Walters
October 9, 1996 - June 17, 2015
son of Candace Kelly and step-son of Shane Kelly
19221 Merlot Ave
Baton Rouge, LA 70817

Matthew (Matt) Jones
October 4, 1990 - July 1, 2015
son of Barb Jones
2925 N 19th Ave Unit 100
Phoenix, AZ 85015-6003

Asher Chmiel
May 25, 2001 - August 18, 2013
Son of Theresa and David Chmiel
6381 Keating Rd
Pensacola, FL 32504

Walking to Remember

This summer, Tia Marie's sister, Angelei, and her friends at camp held a walk in memory of Tia Marie, who passed away in 2015.

My family and I, especially Tia Marie's sister, Angelei, are extremely thankful for Columbus Square Park raising awareness for our CdLS family in honor of our angel, Tia Marie, gone but never forgotten. She will forever be with us.

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
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Card #: _____
Expiration: _____ Security Code: _____
Print name on card: _____
Signature: _____
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A little at a time goes a long way

Monthly donors help us ensure that our mission can thrive year-round. We have big plans for the future, and hope to continue pushing forward with new initiatives, such as more gatherings, more clinics, and more professional outreach across the U.S.

It's simple and easy to set up, and provides a consistent and reliable source of funding that allows us to plan ahead, enabling the CdLS Foundation to improve our support services around the country. A little each month goes a long way to support the CdLS Foundation, and we hope we can count on you for support.

To get started, call Kelly Brown at 800.753.2357, or email kbrown@CdLSusa.org.