

Reaching Out

The Newsletter of the Cornelia de Lange Syndrome (CdLS) USA Foundation, Inc.  Third Quarter 2016

Highlights

Director's Message:

Friends, Family and Fun

Spotlights:

Helping the School Help Your Child with CdLS

Anesthetic and Airway Management of Patients with CdLS

Super Siblings

Abigail and Isaac





Guest Director's Message



Friends, Family and Fun

Rich Haaland, Ph.D., Board of Directors President

Thank you to guest contributors, Board of Directors members, Beth Smisloff and Jim Kesting

This statistic may surprise you, but special events and fundraisers generate 38 percent of the CdLS Foundation's annual revenue. Several events have been providing financial support and raising awareness about CdLS for many years, but maintaining these same efforts can be difficult. Two board members, Beth Smisloff and Jim Kesting, have used Team CdLS and the Missouri Golf Tournament to support the CdLS Foundation and raise awareness of the syndrome.

Beth began participating in Team CdLS events soon after her son, Will, was born with CdLS in 1991. "I needed the CdLS Foundation," said Beth. "It was my only lifeline in what felt like a rapidly sinking ship. I knew as a nonprofit agency they relied primarily on donations. They needed my financial support to provide the services and information I desperately needed."

Jim, whose business partner, Doug Gaines, has a daughter with CdLS, considered different ways to raise funds and awareness for the Foundation. "I was not an avid golfer," said Jim, "but I knew a lot of folks that played. Our company had many loyal supporters, so we relied heavily on them and they answered the call."

Finding ways to sustain those efforts for such a long time is challenging, but focusing on the needs of those with CdLS and recruiting other volunteers made it possible.

"You realize you can help by providing funds to secure the resources families badly need," said Jim. "After 27 years of tournaments, I see that we can do much more to help the families and children with CdLS navigate down this rocky road by crushing the many stones they come upon."

Growing and expanding these efforts is vital to sustaining the Foundation. "The challenge is to continue to branch out to different types of events," said Beth. "Finding events the whole family could get involved in, including our kids with CdLS. It also doesn't have to be running. It could be biking, bowling, knitting - anything is possible! The only limit we face is our imagination."

As the Foundation grows, we must find a new generation of people willing to take what Jim, Beth and other event organizers have started, and use new avenues to spread the word about CdLS. If you have an event idea, or would like to participate in special events in support of the CdLS Foundation, please contact Kristi Larson at 800.753.2357, or events@CdLSusa.org.

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

4 Helping the School Help Your Child with CdLS

6 Anesthetic and Airway Management of Patients with CdLS

8 Awareness Day Highlights

9 Rare Disorder Registries Lead to Progress

10 Super Siblings-Abigail and Isaac

11 The de Lange Society Class of 2016



Advocate for Your Child's Medical Home



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

Patient-centered healthcare and medical homes are fast-becoming the current status of healthcare, as opposed to the traditional model of providers and patients, health centers and hospitals, and emergency room and outpatient visits. Each individual (child and adult) is expected to have an identified primary care provider and an established medical "home"

through which all care can be obtained. One outcome has been to have a higher incidence of preventative visits and a lower incidence of emergency room visits or hospitalizations for children¹. Children with chronic or special care needs make up about 18 percent of all of the children in United States². These children use more resources than those without, and this is supposed to have been taken into account in the medical home model.

A recent report suggests that there is disparity, however, with over a third of families with children with special health care needs encountering difficulties, delays and frustrations in obtaining services, especially minorities, immigrants, those in poverty and those with complex emotional, behavioral and/or developmental needs and functional limitations³. In addition, many primary care providers have a lack of time and/or experience in managing children with complex needs in an office setting⁴. There has been debate about whether the medical home should be with the primary care physicians and pediatricians or with subspecialists, but the results have been leaning towards the primary care providers⁵.

All of this relates to you having a child or adult with Cornelia de Lange Syndrome (CdLS) (or you as an adult with CdLS) and your family. Many individuals with CdLS may have complex emotional, behavioral, and/or developmental needs as well as some functional limitations. You should be able to seek out the most qualified primary care provider who can administer care for someone with CdLS in your area. All primary care providers should have access to specialists, even if they are in different cities or other states. In addition, we have members from most specialties on the CdLS Foundation Clinical Advisory Board, each of whom is always willing to speak to the local specialists about ideal care in CdLS.

You will often need to bring the expert information to the primary care provider, or suggest seeking advice from the specialist. The Foundation's Medical Specialty Cards should be helpful with this, as is our website, but each circumstance is unique. Insist that your primary care providers educate themselves. Seek the advice of specialists related to specific body systems if your child is having issues that the primary care provider cannot explain or help improve. And finally do not hesitate to contact the Foundation if you are having any difficulty in accessing the appropriate medical care.

1 [Romaine MA, et al., Med Care 50:262-9,2012]
2 [Traisman ES, Pediatr Ann 44:522-5,2015]
3 [Rosen-Reynoso N, et al., Matern Child Health J 20:1041-53,2016]

4 [Traisman ES, ibid]
5 [Van Cleave J, et al, Acad Pediatr 16:366-72,2016]

Reaching Out
Published Since 1977
(ISSN 1097-3052)

Cornelia de Lange Syndrome
Foundation, Inc.
Incorporated December 1981
email: info@CdLSusa.org
www.CdLSusa.org

BOARD OF DIRECTORS

PRESIDENT . . Richard Haaland, Ph.D., GA
VICE PRESIDENTS
David Barnes, Esq., NH
Wendy Kaplan-Miller, Esq., CA
TREASURER Eric Johnson, CT
SECRETARY Mary Opitz, TX
DIRECTORS
Catherine Caron, NH
Shelly Champion, MA
Michele Churchman, DE
Dennis Drislane, CA
Lynne Kerr, M.D., UT
Jim Kesting, MO
Dianne Lessa, MA
Pat Lyons, NH
Carlos Madrid, Esq., TX
Katherina Nikzad-Terhune, KY
Julia O'Connor, Ph.D., MD
Fran Rissland, GA
Beth Smisloff, NY

PROFESSIONAL STAFF

ACTING-EXECUTIVE DIRECTOR
Kelly Brown

MEDICAL DIRECTOR
Antonie Kline, M.D.

FAMILY SERVICE MANAGER
Deirdre Summa, L.M.S.W.

FAMILY SERVICE COORDINATORS
Lynn Audette, L.M.S.W.
Justyna Wawrzonek, L.M.S.W.

DEVELOPMENT MANAGER
Kristi Larson

COMMUNICATIONS COORDINATOR
Brenda Sawyer

GRAPHIC DESIGNER/WEB MASTER
Francesca Scognamiglio

OFFICE COORDINATOR
Susan Salina

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*.



Helping the School Help Your Child with CdLS



By Barbara Galyean, Master of Education in Reading, Educational Diagnostician, Licensed School Counselor

Parents and caregivers of school-aged children with CdLS should strive to provide and encourage strong relationships and clear communication with teachers,

school nurses, administrators, and other personnel which could include support staff such as educational aides, bus drivers, cafeteria workers, custodial staff, etc. that will have contact with your child.

While much of this is developed in your child's Individualized Education Plan (IEP), sharing and clarifying this information directly with school personnel who did not attend the most recent IEP team meeting will help make them aware of their role in your child's success.

The following are some suggestions that may help:

- Arrange conferences as early in the school year as possible with everyone who will be involved with your child so that each person will know what is expected to ensure your child's safety and success. Remember, face to face meetings will help to establish the rapport necessary to insure your child's needs are met.
- Familiarize yourself with school policies and procedures and develop plans for your child during events such as fire drills, disaster drills, lock-downs, early releases, inclement weather days, as well as other emergencies that the school normally anticipates.
- Make certain all educational personnel have written copies of your child's unique needs.
- Share all distinctive traits, behaviors, learning styles, communication methods, medication side effects, feeding, toileting needs, mobility issues, and other medical concerns such as seizures and allergies.
- Provide means to enable school personnel to readily access you with their questions and concerns.

- Consider providing a list of outside sources such as the Cornelia de Lange Syndrome Foundation, personal doctors, therapists, agencies, etc. who can provide information on your child's needs.
- Provide pamphlets, magazines, articles, and any other documentation describing your child's disabilities or medical conditions. Keep in mind that not everyone is the expert on your special child that you are.

A final reminder/disclaimer: school policies and regulations differ from state to state, and from district to district. There are some federal requirements such as those in the Health Insurance Portability and Accountability Act of 1996 (HIPAA) that must be followed nationwide which may require you to sign a consent to access and share your child's personal information. If you have any concerns, you can always contact the CdLS Foundation for guidance.

A Closer Look at Communicating with Your Child's School



We'd like to thank Sarah's mother, Anne, for sharing their story with us.

Sarah has various medical issues including seizures, g-tube feedings, medications that need to be given at school along with venting, she is at high risk for aspiration, and she occasionally

has the potential for needing oxygen and/or suctioning while at school. She also has some self-injurious behavior, and other safety issues, and she cannot be left alone and needs direct supervision. Sarah is nonverbal, and has physical therapy, occupational therapy and speech therapy at school. She's mobile, but uses a wheelchair for easier access around the building. Her cognitive abilities are around that of a one year old.

We've found that the best way to help educate Sarah's teachers is by going through the school's nurse. Medically speaking, we share all of our information with the school team, typically first with the school nurse and she helps



to filter out the information that is important for teachers and staff. Initially, we meet with the nurse, outside of the Individualized Education Plan (IEP) meetings, and go through feeding routine, schedule, medications, etc. That information is shared during her IEP meeting, but it gives the nurse and I a chance to really assess what issues Sarah may have while at school. I also provide some information from the Foundation about CdLS in general.

Being a former teacher, I understand that it can get overwhelming for teachers. So, each school year, I create a one-page sheet with general information about Sarah that I share with her teachers, nurses and other school staff who will be working with her. It includes things that would help her succeed, like routine and repetition, and things she likes and doesn't like. For example, it gives several possibilities of what to do if Sarah gets upset, kind of a roadmap of things to do or not do. It also gives them a list of major concerns that I should be notified about immediately so appropriate action can be taken. Along with this little one sheet page, she of course has her more detailed health care plan. The school nurse also has a training log where she trains all staff members necessary on Sarah's needs and CdLS.

The hardest part is getting it all started when your child first goes to school. The initial worry of, "is this all the information they need? Does everyone involved know what's going on?" can be stressful. When Sarah first started school we had to refocus our thoughts, share all that we knew about Sarah, and tell the care team at the school our greatest fears and concerns. When it comes to safety and medical issues for your child, don't assume anyone else understands or knows what to expect. Because we live it, it is our "normal," not anyone else's. Her healthcare plan and IEP are all a work in progress. There are changes every year, and this kind of communication ensures that her care team is aware of any updates.

WELCOME NEW FAMILIES

California

Megan and son Jacob
born September 2, 2014

Colorado

Sheila and Dave and
daughter Anikka,
born June 15, 2003

Florida

Dayhana and daughter
Alexandra,
born October 3, 2012

Florida

Taylor and daughter LailaKay,
born May 4, 2015

Kentucky

Laura and Thomas and son Ian,
born September 26, 2002

Louisiana

Shannon and Anthony and
daughter LaShalle,
born July 13, 1993

Massachusetts

Jennifer and son Max,
born January 28, 2004

Massachusetts

Meagan and Jeffery and
daughter Samantha,
born August 10, 2009

Michigan

Holly and Jonah and
daughter Gwyneth,
born December 5, 2015

Michigan

Jen and Dustin
and son Kaiden,
born February 2, 2016

New York

Sarah and son Angel,
born September 10, 2009

New York

Kim and Carmine
and son Rocco,
born August 31, 2015

North Carolina

Destiny and Nicholas
and son Christian,
born July 13, 2014

South Carolina

Maria and son Santiago,
born November 1, 2013

Texas

Connie and daughter Leanna,
born June 3, 1984 and
granddaughter Sapphire
born July 24, 2005

Texas

Adriana and daughter Juliana,
born February 4, 2016

Love something in this issue?

Want to see **something else** in here?

Let us know your thoughts about this issue
of *Reaching Out*, and what you'd like to see in
future issues, by visiting
<https://www.surveymonkey.com/r/RO32016>



Anesthetic and Airway Management of Patients with Cornelia de Lange Syndrome

By Yvon Bryan, M.D., Anesthesiology-Pediatric Anesthesia
at Wake Forest Baptist Medical Center

Considerations for Physicians

Children with CdLS tend to have a variety of procedures performed during their lifetime. Depending on the procedure being performed, different anesthetic techniques may be chosen. These recommendations are based on responses to a survey on the airway and anesthetic management of children with CdLS. The survey primarily dealt with response to medications, intubation, aspiration, ventilation, oxygenation, and emergence.

Preoperative

- It is important to discuss the medications that the child is already taking, if any.
- Certain medications, such as benzodiazepines (midazolam), and/or psychiatric medications, can cause excitation and/or aggression, so beware of paradoxical responses.
- The responses to medications are unpredictable among children with CdLS. Certain patients may be sensitive to narcotics.
- The cognitive function of children with CdLS ranges from age-appropriate to almost complete cognitive dysfunction. Therefore, expect a spectrum of behavioral issues in spite of their age.
- Be careful with the limbs of the child due to deformities of the upper and lower extremities leading to difficult IV access.
- Be careful of aspiration due to the high risk of GERD.
- Due to the high incidence of a high-arched palate, small mouth opening, and micrognathia expect difficult intubation.
- Use a smaller sized endotracheal tube or supraglottic airway device.

- Expect difficulty with the insertion of the airway device.
- There is a high risk of problems with conventional devices; therefore, consider using an alternate device, such as a flexible fiberoptic bronchoscopy (FFB), or supraglottic airway device.

Intraoperative

- A choice of intravenous or inhalational anesthetics is possible.
- Beware of increasing CPAP during bag mask ventilation due to an increase in the incidence of GERD.
- Consider using flexible fiberoptic bronchoscopy (FFB), with or without a supraglottic airway, due to the greater maneuverability leading to an easier insertion of the endotracheal tube.
- Be careful of the risk of aspiration.
- Consider using alternate supraglottic airway devices for a better seal.
- There is a higher risk of desaturation which may lead to bradycardia or cardiac arrest.
- Standard anesthetic agents and medications may be used; however, expect variability in the response to the medication.
- If multiple procedures are being done, it is important to decide whether all procedures should be done under one anesthetic or if different anesthetics should be given for each procedure.
- There is a lack of evidence for there being one safest anesthetic technique.

Postoperative

- There is variability in how children with CdLS emerge from sedation and/or general anesthesia.
- Delayed emergence can occur and may last up to one week.
- There is sometimes a change in the diet patterns of the child.



- Amnesia has been known to occur after sedation and/or anesthesia in some children with CdLS.
- Watch for self-mutilating behavior in response to being sedated and/or put under anesthesia.
- Beware of breathing problems postoperatively as there is a higher incidence of lost airway/reintubation and subsequent cardiac arrest in this population.
- Reintubation and/or postoperative ventilation is sometimes required.
- Consider bridging the extubation with an airway exchange catheter.
- Most of the postoperative problems occur shortly after the procedure; therefore, overnight stay is not always necessary.

A Closer Look at Anesthesia Guidelines for Professionals: Micah's Story



We'd like to thank Micah's parents, Eric and Sarah, for sharing Micah's story with us.

Micah had anesthesia when he was around 18 months old, and he was given Sevoflurane. A few hours after the procedure, once we were already home, he suddenly spiked a temperature of 104 degrees or more, and became very pale and listless. While he fortunately returned to fairly normal relatively quickly after we gave him Tylenol, and without needing to go to the hospital, we have always been concerned about him getting anesthesia moving forward. We have found that some anesthesiologists are unfamiliar with the risks associated with anesthesia in individuals with CdLS, while others are very thorough and cautious. Unfortunately, you may not always get the latter.

When Micah had a hearing test under anesthesia recently, I was sure to raise my concerns about Sevoflurane, his history, and the slight risk of malignant hyperthermia in individuals with CdLS. The anesthesiologist underplayed my concerns, but finally did agree to give him Propofol instead of Sevoflurane, and then proceeded to say he wanted to give him Versed prior to the procedure. I recalled reading that some individuals with CdLS have a difficult time with Versed and mentioned this to him, but he reassured me he hadn't seen any issues with it. I was quite concerned about giving Micah Versed, so when the anesthesiologist stepped out I immediately called the Foundation and spoke to Deirdre Summa. She quickly sent me the Best Practices for Physicians and Families regarding anesthesia, which mentioned that there can be a paradoxical effect with medications such as Versed. I was able to share this with the anesthesiologist, and he agreed to not give Micah Versed. I was so thankful that I had access to these materials in such a short timeframe, and plan to take copies next time Micah has anesthesia.

2016 CALENDAR

August 13

Northeast Region
Family Gathering
New Castle, NH

September 17

Golf Tournament
Tekoa Country Club
Westfield, MA

September 18

The Saratoga Palio
Saratoga Springs, NY

October 9

Bank of America
Chicago Marathon
Chicago, IL

October 10

28th Annual CdLS Charity
Golf Tournament
Sunset Hills, MO

October 16

2016 Baltimore
Running Festival
Baltimore, MD

October 21-22

CdLS
Foundation Board of
Directors Meeting
Seattle, WA

October 22

West Region
Family Gathering
Seattle, WA

November 5

Multidisciplinary Clinic for
Adolescents and Adults
Baltimore, MD

November 6

TCS New York City
Marathon
New York, NY



Awareness Day 2016 Bring Families Together



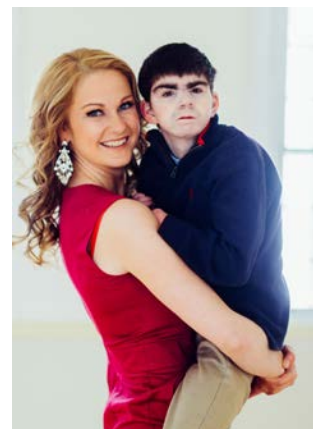
The Foundation is always striving to increase awareness about CdLS. Whether it's posting on Facebook, wearing custom purple sneakers, or hanging up a CdLS towel in a pub in the British

Virgin Islands; we are all hoping to catch people's eye. As we do so, we continually reach more families, provide information and support, and also increase interest in furthering research to understand CdLS and develop treatments for individuals with CdLS. Here are just some of the ways you raised awareness of CdLS this May.



CdLS Awareness Event Highlight

Since January, Maddi Shaw (Miss Vermont International) has made 13 appearances—speaking at churches, elementary schools, local pageants, non-profit organizations, and more. One simple saying she asks listeners to remember is to “stay informed and be loving.”



“It encourages us to learn about others and in turn treat them with respect and love. In all of these different environments I have been able to tell my story, sharing what it's like growing up with a brother who has CdLS, and educating them about the syndrome, offering suggestions of ways everyone can interact with individuals who have disabilities.”



For National CdLS Awareness Day, Maddi hosted an event in Brattleboro, VT, where Foundation staffers Francesca Scognamiglio and Justyna Wawrzonek attended to raise awareness in the community. Maddi shared her story and educated the crowd, who generously donated \$1,053 to the CdLS Foundation.

Learn more about Maddi's experience as Miss Vermont International and her efforts in raising awareness of CdLS by visiting: <http://supportcdlsvt.com>.



Rare Disorder Registries Lead to Progress



*By Angela Van Veldhuizen, R.N., B.S.N., and Jeremy Morgan, M.S.,
Coordination of Rare Diseases at Sanford (CoRDs)*

One of the primary challenges faced by the rare disease community is the overall lack of information available to patients, families, physicians, and researchers. This information is vital in the process of identifying effective treatments for rare diseases, since 1 in 10 Americans or 30 million people in the United States have been diagnosed with a rare disease, with over 50 percent of rare diseases affecting children. The overall mission of Coordination of Rare Diseases at Sanford (CoRDs) is to accelerate research into rare diseases. In order to meet this mission, CoRDs has three goals: first, to assemble contact registries; second, to generate standardized datasets through common data elements (CDEs); and third, to curate disease-specific databases on rare diseases.

CoRDs Registry
Coordination of
Rare Diseases at Sanford



The CoRDs registry was primarily developed to meet an unmet need for rare diseases since only about 20 percent of rare conditions worldwide are represented by a registry. The most basic definition of a registry is the collection of contact information of a group of people with similar characteristics. We are able to collect and warehouse this information so that researchers may contact us about potential participants for research studies and clinical trials. If a researcher would like to contact CoRDs participants, CoRDs will reach out to participants on behalf of the researcher. Therefore, participants have complete control over the decision to participate in a study or not.

To generate standardized data sets, CoRDs collects the required CDEs set by the National Institutes of Health Office of Rare Diseases Research. Doing this ensures that data across all rare disease registries are defined in the same way, using the same standards, and the same

vocabulary. This helps to facilitate harmonization as well as analyses and studies within a specific disease, and across multiple diseases. We are then able to integrate the data back into the Global Rare Disease Repository for data standardization.

Lastly, CoRDs partners with patient advocacy groups, like the CdLS Foundation, to establish patient registries and conduct natural history studies. As discussed above, having a patient registry increases the likelihood that researchers will be able to move forward with clinical trials for a specific disease. A natural history study is performed to better understand the course of a disease from its onset throughout its progression, which allows for better targeting of treatments for the disease.

Right now there are 76 participants enrolled in the CoRDs registry with CdLS. CoRDs encourages everyone who has not already done so, to go in and sign up to be part of this registry. You can contact CoRDs anytime with questions or concerns at 1.877.658.9192, Monday through Friday 8:00 a.m.-5:00 p.m. (CST), or by emailing CoRDs@sanfordhealth.org.

Claim Your Spot for the Multidisciplinary Clinic for Adolescents and Adults at Greater Baltimore Medical Center!

This Foundation-sponsored, free clinic occurs twice yearly at Greater Baltimore Medical Center (GBMC) in Baltimore, MD. Foundation Medical Director Antonie Kline, M.D., director of Pediatric Genetics at the Harvey Institute for Human Genetics at GBMC, has led these free clinics since 2001. Any individual age 12 or older with CdLS can attend with their family. Clinics occur each spring and fall and can accommodate up to eight families. A Family Service Coordinator from the Foundation always attends to provide support.

Participants receive one-to-one consultations with specialists from pediatric ophthalmology, gynecology, pediatric dentistry, genetics, gastroenterology and nutrition, psychiatry and behavioral psychology.

The Foundation provides up to \$500 in travel reimbursement to help defray expenses associated with a family's trip to a CdLS clinic. Families are reimbursed for the expenses AFTER they have been incurred. Travel support for clinic is available to a family only one time. Covered expenses are hotel, gas, and airfare, bus or train tickets.



Super Siblings – Abigail and Isaac



Thank you to Melissa, Abigail's mother, for sharing their family's story with us.

Ten-year-old Abigail was studying formal letter writing in school. Their class assignment was to write a persuasive formal letter to a business with a "suggestion." She chose to write a letter to her dad's employer, Blue Ribbon Builders, suggesting that

they build a large playhouse and auction it off with the proceeds going to the CdLS Foundation. This donation would be in honor of her brother Isaac, age 14, and all other individuals with CdLS.

"I knew it could really help the people who have CdLS, and their families," said Abigail. "I know my brother, Isaac, doesn't understand what this money means, but it's so great to know it will help so many others."

Abigail received a response from the company, along with a \$500 check written to the CdLS Foundation, and was very excited.



"I thought I would just get a letter back thanking me for the idea to build a playhouse and auction it off for the CdLS Foundation," said Abigail. "I never expected to get a \$500 check for the CdLS Foundation!"

Share your Mailbag or Super Sibling Story!

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

Mailbag –Tami Reaching Out With Music

We look forward to each issue of *Reaching Out*. In addition to the technical information, we enjoy reading about the kids that are featured, as they often have much in common with our child. Thanks for giving us this opportunity to share a bit about our little treasure, Tami, age 10.



Tami (left), and sister Kendra (right) share a moment with American blues slide guitarist, Robert Logan Reynolds III, on a pair of Pfalzgraf family homemade guitars.

With the numbers tracking along the lowest percentiles for the entire pregnancy, we knew there might be challenges ahead. When she was born in 2006, Tami was very small and did not progress like a typical baby. We finally received our diagnosis at 18 months. Things have gotten better since. In addition to the ongoing health and physical related supports provided by Tami's team of specialists at Children's Hospital Los Angeles, we found a neuropsychiatric evaluation to be particularly valuable in helping us understand how Tami's neurological deficits manifest in undesirable behaviors.

Communication continues to be our biggest challenge. Recognizing early that Tami was not going to have intelligible speech anytime soon, we invested in the "Signing Time," American Sign Language program, and learned the basics at home as a family. We've learned that navigating the public school system is uniquely challenging for a non-verbal child with cognitive limitations, especially if the child does not qualify for deaf/hard of hearing services. Our private Speech Language Pathology and Alternative/Augmentative Communication device consultants continue to attend



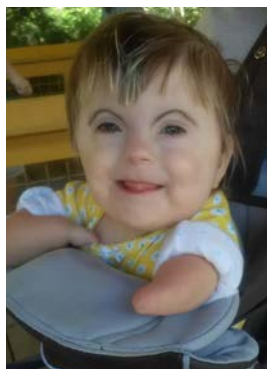
Tami's Individual Education Plan (IEP) meetings to keep everything on track. Tami's vocabulary has grown to 300 signs now.

Tami loves music, especially guitars. When Tami was five, her first guitar was pieced together, using an old cigar box and some other junk. Oddly enough, a movie of Tami playing that guitar was used in an IEP meeting to show that Tami's display of rhythm and beat proved that she has some math skills that those "standard" block-stacking tests didn't reveal and that she couldn't tell the non-signing assessor about. Over the years, a few of those "junk guitars" have found their way into the hands of a few blues guitarists, who tell the story of the little girl with CdLS who can't talk, and her junk guitar, each time they play one of their "twangy" songs on one.

Parents: it is ever so critical to give your child a voice to express their potential, regardless of what form that voice takes on.

The Pfalzgrafs
Acton, CA

On the Cover: Bella Marie



Bella Marie was born one of the smallest kids with CdLS, at 1 lbs 12 oz, she has come along way to 11 lbs 6 oz. She received her preliminary diagnosis of CdLS the day after she was born and her genetic diagnosis at 2 months old. Bella will be 3 in July and is currently working on walking alone and learning how to use her new prosthetic arm.

Bella is very curious about everything and loves being and doing anything outside. She loves watching Minnie Mouse with her mommy and every kind of car show with her daddy. She recently got a new friend, a cocker spaniel, named Roxy Ann. Bella also shows a true love for music and all kinds of books.

The de Lange Society

Class of 2016

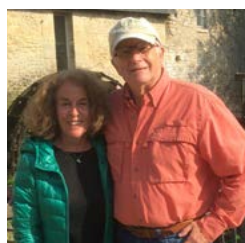
Lynn Audette, Tolland, CT



Lynn Audette has been a CdLS Foundation staff member since 1996. Since joining the Foundation, Lynn and her family have actively volunteered at numerous Foundation events, including family gatherings, conferences and other awareness opportunities. She

facilitated the creation of *When Angels Take Flight*, among other publications, and has been integral in expanding services for families throughout the years. In her role as Family Service Coordinator, she has visited families in almost every state, welcoming them as they meet other families for the first time.

Linda and Ron Berger, M.D., FACS, Torrington, CT



Linda and Ron Berger have been actively involved with the CdLS Foundation since 1976, before it was even an incorporated Foundation, when they became friends with Julie and Frank Mairano, who had a daughter with CdLS. Linda has helped coordinate

with consultations at the CdLS Foundation National Family Conference for the past several years and received the John King Award in 2010 for her long-term efforts on behalf of the Foundation.

Ron has served on the Clinical Advisory Board for the Foundation since 2003. He has written numerous articles for Foundation publications regarding eyes and eye-care in CdLS. Ron has also attended numerous CdLS national and international conferences and through consultations, provided countless families with valuable information to bring back to their medical care team.

Continued on page 12



Michele Churchman, Dover, DE

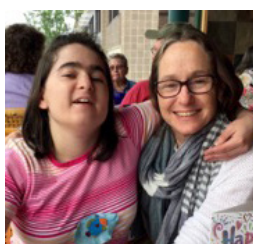


Michele has actively worked to raise awareness of CdLS in her community, including writing to Congress, appealing to Rotary clubs, and manning tables to raise awareness at local events.

Michele Churchman has served on the Board of Directors since

2011; 2016 is her final year on the Board. She's been an Awareness Coordinator since 2002, and has served on the Public Affairs Committee since 2008, providing guidance in the Foundation's outreach efforts in awareness, branding and public relations. Her son, Joshua, passed away in 2013, and Michele's desire to raise awareness about CdLS and the Foundation continues in Delaware and her local community.

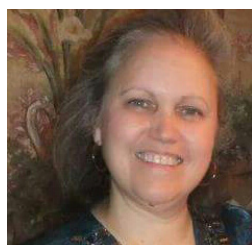
Kari Cunningham-Rosvik, APRN, Seattle, WA



Kari Cunningham-Rosvik served on the Board of Directors from 2008 to 2013. She and her husband, Geir Rosvik, have been involved with the Foundation since their daughter, Iselen, was diagnosed in 1995. Kari has served as a Regional Coordinator since

2008, and received the Sue Anthony Award in 2010 for innovation, commitment, integrity, and enduring contributions to families and people with CdLS. This award is given to those who have developed a creative approach to improve the quality of the services the Foundation provides, and its ability to fulfill its mission. Kari served as Program Council Chair on the Board of Directors from 2009 to 2013.

Luanna Fielder, Pearland, TX



Luanna "Lu" Fielder and her family have made it a priority to raise awareness within their business, Fielder Electric Supply Co., encouraging her coworkers, friends and neighbors to become informed about CdLS and the Foundation. They continue to

be active on behalf of their daughter, Jaclyn, age 25, who has CdLS. Since 2013, Fielder Electric, has been

a Team CdLS Sponsor, and has also held a Dress Down Day, where employees dress in costume and decorate their cubicles as a fundraiser for the Foundation. From 1997 to 1999, Lu served as an Awareness Coordinator, and continues to be a support to other families on social media, and is a resource for Texas families should they need someone to call.

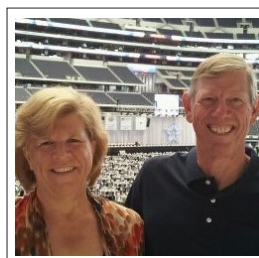
Elizabeth Fouts, Ph.D., Manchester, NH



Elizabeth Fouts, a professor at Saint Anselm College, has been a part of the CdLS Foundation since 1994 she attended her first family gathering in 1996 with her son, Adrian, who has CdLS. She has been an Awareness Coordinator since 1997, and has helped many

Spanish speaking families during her time as a Foundation volunteer, providing translations of numerous publications and articles. Elizabeth served on the Board of Directors from 2008 to 2010, and served on the Foundation's Clinical Advisory Board for many years. She ran a marathon on behalf of the CdLS Foundation, and her family has hosted Adrian's Ride and a family gathering in New Hampshire, among many other involvement activities with the Foundation.

Brian Luyt and Mary Opitz, Plano, TX



Brian Luyt and Mary Opitz first contacted the Foundation in 1992 prior to a CdLS Conference in Arizona. Their daughter, Katie, age 29, has CdLS. They were the host family for the 1999 CdLS National Family Conference in Texas, and joined the newly formed CdLS

National Conference Planning Committee in 2010 and continue to serve on the committee. Brian served on the Foundation's Board of Directors from 2001 to 2007, and ran a Texas golf tournament from 2003 to 2007 to raise money for the Foundation. Mary has been on the Board of Director's since 2011, and is in her final year on the Board. She has also been a member of the Program Council, where she has served as chair since 2014, guiding Foundation services. The couple received the Sue Anthony Award in 2012 for innovation, commitment, integrity, and enduring contribution to families and people with CdLS.



Donations from 02/25/16- 06/01/16

Gifts that Count - In Honor/Celebration

A.J. Staggs

Charles and Kortni Staggs
Erin Crunk
Jina Homan
Kristine and Hayden Adams
Larry Kozlowski

Aaron Bailey

Linda and Greg Bailey

Aaron Young

Angela and Tim Young

Abigail Busk

Jennifer and James Busk

Adam Hasecke

Jane McGraw and Rick Stoerker
Pam Barcalow
Susanne and Larry Rowe

Adam Jackson

Emerson and Marilyn Prange

Alexandra Boteler

Linda and Dawn Boteler

Alexys Spencer

Christina Thode

Alisha Palermo

Jackie and Joe Palermo

Alyssa Pletl

Donna Pletl

Amber Gaines

Barbara and Doug Gaines
Elizabeth and Donald Doherty
Joyce and Jim Kesting

Amy Haaland

Richard Haaland, Ph.D.

Andrew Patitucci

Nancy Tossona

Angie Maglione

Jose and Yadir Maglione

Aryanna Gage

Sarah Griffith

Aubrey Garigen

Jackie Biddle
Louanne and John Strauss

Barbara and Norm Winnerman

Anne Cahn
Lynn and David Wells

Brady Kelton

Audy Neal

Benjamin Miller

Jewish Community Foundation
Orange County
Mindy and Jeffrey Miller

Breeze Davis

Elaine and Ron Davis

Brenda and Andrew Sawyer

Barbara and Norm Winnerman

Brenden Keating

Patricia and George Zirm

Brett Fields

Kathy and Russell Fields

Brian Drach

Alice Hultmann

Brianne Prada

Karen and Larry Prada

Brynnlee Beekman

Diane Adams
Karen and James Morton
Valerie and Adam Skiles
William Shaffer

Caitlin Igoe

MaryAnne and Stephen Igoe

Caleb Wherry

Donald Wood

Camden Robbins

Catherine Belcher
Debbie and Dennis Lark
Diane Claar
Teri and JD Robbins

Charles Gladson

Susan Hartman

Charlotte Weiss

Harriet Weiss

Christian Musumeci

Joanne and Franklin Morgan

Colin Bell

Jean and David Bell

Colt Highberger

Carla and Marty Highberger

Connor Myrthel

Willing Workers of
Paradise Valley

Daniel Toubes

Sharon and Mitchell Weiss

Danielle Bauer

Diane and Russell Bauer

David Podell

Diane Fromhartz

Denise MacDonnell

Audrey Hayslett

Destiny Caruso

Pat Heuring

Devin Miller

Lynnette and Dean Miller
Tammy and Jay Schaab

Dianne Haaland

Richard Haaland, Ph.D.

Douglas Canning

Maria and Maury Paslick

Dylan Taggart

Nancy and Richard Troast

Edmund McCurdy

Stephanie and Edmund
McCurdy

Ella Grace Musial

Christine Leddy
Constance and Richard Lytle
Daniel Major
Farrah Kennedy and
Timothy Peterson
Peggy and Lonnie Harrington

Ellie Westrate

Jeanne Westrate

Emmalee Ellis

Susan Heyer

Eric Loftus

Amy and Chris Loftus
Karen Szukalski

Eric Solomonson

Gordon Gnasdoskey

Ethan Atkin

Shruti and Ian Shah

Frank Leone

Marcia Matten

Gabriel Sloan

Linda and Don Ziglar

Gavin Bartelt

Garry Bartelt

Grace Pena

Beatriz and Domingo Pena

Gracie Fry

Dr. and Mrs. Michael Hoog
Linda and Bruce Fry

Hayes Ganno

Linda and Bill Ganno

Hayley Turbyfill

Carol and Glenn Zeliff

Hope Kurth

Anita Kurth
Delores and Rod Flynn
Janet Wolf

Hunter Barrett

Gary Hartman

Isaac Smude

Karen Smude

Isabelle Skelonc

Kathy and Douglas Tompsett

Iselen Rosvik

Audrey and Mike Weymiller
Kari Cunningham-Rosvik and
Geir Rosvik
Regina Bowman

J.D. Cook

Jane and James Cook

J.D. Diaz

Jessie Sierra Diaz and Jose Diaz

J'Erica Jetson

Pastor and Mrs. Raymond
Jetson

J.T. Picazio

Linda and Tom Picazio

Jack Barnes

Mary Waldo
Valerie Barnes

Jaclyn Fielder

Luanna and William Fielder
Wendy Franty

Jacob Magnus

Teresa Magnus

Jason Shufelt

Sharon Shufelt

Jaxon Schweickhardt

Joyce Anderson

Jessica Smith

Bethel Southern
Methodist Church
Debbie and Randy Smith
Emma Smith
Greg and Carolyn Smith
Patty and Mike Smith
Ronny Smith
Vickie McKorey

Jessica Ward

Clave Holdings, LLC
Deborah Rasleigh
Victoria Woods

Jonathan Swanson

Julia and Peter Swanson

Joy Zerbarini

Jean and James Crowley

Julie Champion

Shannon and Thomas Noonan

Kaitlyn Kuepferle

Nancy and Larry Kuepferle

Karly Diller

Dori and Ray Diller

Katherine Harvey

Lyn Tucker



Kathleen Moriarty
Christine and Dave Bedell

Katie Kavouras
Beverly Kavouras
Patricia and John Kavouras

Katie Luyt
Robbin and Bruce Bailey

Kiara Drake
Katherine King

Kolten Griffin
Belinda and John Waller
Jack's Place LLC

Laura Dupuis
Brooke Bender
Celeste Mendez
Julie Dupuis
Livia Casillas
Scott Cooper

Lauren Lewin
Jacqueline Lewin

Leonard Furman
Jasmin Furman

Levi Fanning
L.S.J. Properties

Lillian Drury
Carol and Robert Drury

Lillian Choong
Lil and Lonnie Williams

Lillie Kellam
Karyn and Monte Kellam

Logan Fowler
Meredyth Armitage

Lori Etlicher
Barbara and Paul Etlicher

Luca Barone
Chris Marinaccio

Lukas Schlamminger
Monika and Stephan
Schlamminger

Madalyn Gilchrist
Judy Hawley

Madelyn Ryther
Sandy and Brian Durnell

Maegan Lowrey
Bolling Lowrey
Tricia Vogel

Maggie Johnson
Jennifer and Glenn Johnson
Sarah Johnson

Makayla Geary
Mary Geary

Mandy Drexler
Mary Drexler
Susan and Charles Drexler

Manny Garcia
Eileen and Brian O'Rourke

Maria Sanchez
Juanita and Guillermo Sanchez

Mary Hodina
Georgia Hodina

Matthew Chevette
Marv and Fran Silverman

Maya Pomfret
Janice and Eric Duff
Timothy Roenigk, D.M.D.

Meaghan O'Keefe
Martha-Jane and
Richard Moreland

Michael Cataline
John Cataline

Michael Friedman
Ida Friedman
Sandra and Charles Horberg

Molly Wagner
Kathleen Browne

Monte Telischak
Carol and Ron Telischak

Natalie McClain
Kathleen and Kenneth McClain

Nicholas Arroyo
Barbara and Alex Kalb

Nicholas D'Angelo
Marie, Dennis and
Christine D'Angelo

Nicholas Turman
Anne and Marvin Bertsch

Nicole Miller
Eric Miller
Toshiye Erickson

Nikhil Menon
Gopal and Sumita Menon

Noah Thornberry
Phyllis Mattingly

Olivia Hoogland
Walter Marks

Olivia Roberts
Albarran Law Offices
Albert and Sara Hancock
American Tradesmen, Inc.

Anthony Ricchetti
Bridget Salvi
Carol Battaglia
Cindy Iwanski
Dan Lodesky
Debbie and Glenn Slater
Diane Behnke
Gina and Douglas Roberts
Joan and Wallace Dunn
Julie and Christopher Stride
Kathleen and Raymond Collins
Kerry Forman

Leia and Michael Betar
Libertyville Lincoln-Mercury
Sales, Inc.

Lois and Glen Braden
Lyyli Lynch

Mary and John Scully
Nancy Waites
Nutschnig & Cavanaugh
Patricia Botsios
Ronald Sandler
Suzanne and Robert McDonald
Taylor Enterprises, Inc
Tia Boswell

Paige Olson
Dawn and Steve Olson
Edward and Jean Connon

Payson Rouse
OneHope Foundation

Phoebe Taylor
Susanne and Darren Taylor

Riley Rissland
Michael Castellana
Francesca and Karl Rissland
Michelle and Robert Scobie

Robert Brough
Barbara Brough

Robert Carter
William Carter

Ryan Rockers
Michelle and Bernard Rockers

Ryan Lenth
Marilyn and James Lenth

Rylie Kohn
Jeanne Keller

Samuel Miller
Bobbi and Stephen Paris
Wendy and Andrew Miller

Sarah Siegel
Jaime and Timothy Siegel
Joan and Robert Busteed
Jodi Warren
Mary Siegel

Sarah Spaulding
Wanda and Laurier Turcotte

Sarah Suttman
Ellen and Thomas Heile
Laurie and Dwight Krieg
Shirley and Paul Suttman

Siena Renteria
Bob Barber

Sydney Hoffman
Christina and Matthew Evans
Debi Marks
Jennifer Sivak

Tanaya Molzan
Jean and Lawrence Todesca

Taryn Christie
Rosemary Christie

Thomas Scruggs
Lisa Ratliff

Tristen Black
Ginger Black

Tyler Macy
Barbara and Timothy Macy
Mary Ann and Raemon Polk
Rogene and Richard Moss
Stephen Moss
Susan Macy

Vivian Comeau
Elizabeth and Bryan Mitchell

Wilder Thompson
Jacqueline and Carl Greiner

Will Smisloff
Beth and Mark Smisloff

William Messer
Pat and Eddie Roncales

Zachry Dowdy
Beverly Lyons

In Memory

Agnes Davis
Debbie and Randy Smith

Aiden Bard
Nikki Zite
Sharon and Jay Zite
Sherry Brick
Shirley and Martin Lubowich
Stacy Bard

Aimee Langlois
Frances Burke

Alex Black
Molly and Garth Black
Monica and Glenn Whiteside
Nancy Hubley

Alison Wells
Ruth Gruber

Allison Tryba
Jan and Les Tryba
Veronica and Ernest Tryba

Andrew Schwalbe
June Cigdemian
Marcia Haglund



Angela Damiani
Janice and Terry Krefting
Mary Wrolstad

Angela Shaw
Kay and Craig Olson

Annabelle Schwindler
Chris Bronkella
Diane Bassemier
Janet Scherr
Lindsay and Jason Calhoun
Norma James
Patrick Little
Sara and Michael Boehman

Betty Hillen
Emma Smith
Greg and Carolyn Smith
Ronny Smith
Patty and Mike Smith
Vickie McKorey

Christopher Bennett
Elizabeth Kidd

Christopher Dugosh
Melanie and Richard Dugosh

Claudia Tionson
Catherine and Jesus Zermeno

Daniel Keating
Beverly A. Hockley
Debra Glorius
Larry Keating
Laurie and Rudy Keating-Duttry
Michael Fake

David Lindholm
George R. Lindholm, M.D.

Norma James
Rhonda Leach

Emma Kissock
Norma James

Ervin E. Molzan
Carol Zichi
Juliana McGovern
Robert Murphy
Ethan Walters
Candace and Shane Kelly
Penelope Keating

Gannon Thomas
Carol and Robert Thomas

Gerard Joyce
Patricia Joyce

Gipsy Stamper
Mary and Harold Bolling

Gordon Tandy
Karen and Larry Prada

Helen Schairer
Margaret Walters

Jaelah Giles
Stacy and James Arellano

Jamal Rich
Mindy Lake

James Walters
Sandra McCurdy

Jasmine M. Querubin
Board and Staff of the
CdLS Foundation
Peter Ivanovich

Jenna Kraemer
Carleen Celusnak
Donna and Paul Nozar
Janet Morris-Harold and
Charles Harrold
Mr. and Mrs. George Angell
Mr. and Mrs. Richard Stone
Sandra and Clyde Lapish

Jeremiah Caudillo
Linda Gray

Jindi Van Buren
Kathy and Gene Van Buren

Keel Salmons
Norma James

Lisa Mairano
Maureen and Paul Schuler
Maribeth Markowski
Norma James

Liddy Horsey
Jill and Andrew Stefanovich
Karen Manning and
Thomas Horsey

Megan Pappas
Board and Staff of the
CdLS Foundation
Jean and Paul Wittmer
Sandra Richardson
Steve Gent

Meredith Clark
Norma James

Michael Rico
Marina Alvarez and Luis Torres

Michael Gonella
Carole and Richard Lee
Shauna and Daniel Valenzuela

Michael Swart
Beth and Mark Smisloff

Muriel Tuteur
Ida Friedman
Sharon and Mitchell Weiss

Natalie Marks
Davidson Urology

Nikki Rauscher
Dawn and Rolfe Rauscher

Nylah Cherry
Amie Neal

Ryan Payne
Timothy and Myra Payne

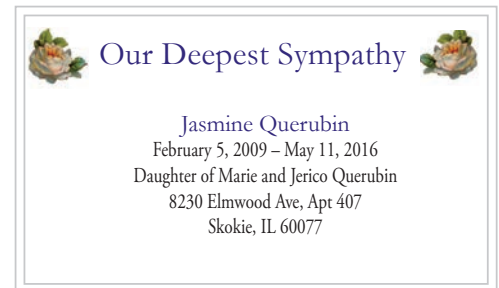
Sean Brisbane
Nancy and Mike Brisbane

Steven Baker
Donna and Robert Baker

Tanner Jencik
Norma James

Victoria Patisaul
Kimberly and Charles Patisaul

Will Kurth
Janet Wolf



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 would like to sponsor a 2016 Team CdLS Runner!

Name: _____

Enclosed is my tax-deductible gift of:

☐ Other \$ _____ ☐ \$500 ☐ \$250
☐ \$100 ☐ \$50 ☐ \$35

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



☐ Please Charge \$ _____ to my credit card

☐ Once ☐ Monthly for _____ months

Charge my gift to: ☐ VISA ☐ MC ☐ AMEX

Card #: _____

Expiration: _____ Security Code: _____

Print name on card: _____

Signature: _____

Email address(es): _____

Donate online at www.CdLSusa.org.

One hundred percent of your contribution is tax deductible.

Cornelia de Lange Syndrome Foundation, Inc.
302 West Main Street, #100
Avon, Connecticut, USA 06001

Return Service Requested

NON-PROFIT
U.S. POSTAGE
PAID
HARTFORD, CT
PERMIT NO. 751

Sponsor a Team CdLS Runner



Meet sisters, Jenna and Ally, running in the Chicago Marathon for Team CdLS:

Sisters Jenna and Ally of Ballston Spa, NY, are running in the Chicago Marathon this year on behalf of their close family friends, the Smisloff family, whose son, Will, has CdLS.

“Throughout the years I’ve seen first hand what a blessing the CdLS Foundation has been for them. Running for Team CdLS is very motivating; it gives all the training hours purpose,” said Jenna. “Knowing that I’m doing it for such a great cause will get me through some of the tough long runs and will help me get to the finish line in October.”

“Since the CdLS Foundation is something very close to me, it makes running my first marathon for the organization feel a lot more meaningful,” said Ally. “As a result, running becomes much less about myself and more about raising money for a Foundation that does so many amazing things for families affected by the syndrome.”

Show your support for Jenna, Ally and other runners on Team CdLS, by sponsoring a runner on Team CdLS 2016. Help them continue to make their miles more meaningful as they run on behalf of individuals with CdLS around the country.

Visit <https://www.crowdrise.com/CdLSFoundation#projects> to find a race and a runner to support.

