

Reaching Out

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



Summer 2012



Highlights

Director's Message:
2011 in Review

Spotlights:
Anesthesia
Behavior Plans for
School

Super Siblings:
Austin and Katherine





Director's Message



2011: Record-breaking Fundraising Events Made the Difference

Each summer, the CdLS Foundation publishes its annual report, which provides a summary of the financial health of the organization and highlights from the preceding year.

For the CdLS Foundation, 2011 proved to be the “year of the event,” with several new endeavors and record fundraising income making a big impact on the bottom line. From bake sales and bowl-a-thons, to mud runs and formal affairs, events throughout the country raised \$479,490—an impressive \$72,000 more than the previous year and nearly half of the year’s revenue.

Team CdLS, which had runners in the New York City and Chicago marathons, expanded to the Baltimore Running Festival, Saratoga Palio and several half marathons and 5Ks throughout the country. The culmination of these runners’ fundraising efforts was \$158,000.

The other highly successful event was Toast the Chief, which honored the Foundation’s longtime volunteer and outgoing board president David Fowler and his wife Susan. The Boston event generated approximately \$97,000. The occasion will serve as a model for future events honoring exceptional volunteers and leaders.

Additionally, golf tournaments raised a record \$97,000.

Total revenue of \$972,326 was down from 2010, due to loss of government funding and fewer individual donations, typical in a non-conference year. However, the year ended just \$28,000 shy of the \$1 million mark.

Spending remained in check, with just 11 percent of revenue supporting administrative and fundraising expenses.

If you would like to see the complete 2011 annual report—which also highlights several Foundation “heroes”—you can view it online at www.CdLSusa.org. Click on the **Who We Are** tab and scroll down to **2011 Annual Report & 2011 Financials**. Paper copies are also available. Request one by calling 800-753-2357.

Marie Concklin-Malloy

Acting Executive Director

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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.



RADical New Findings for Some With Features Like CdLS

Guest Columnist Matt Deardorff, M.D., Ph.D., geneticist, CdLS Foundation Clinical Advisory Board and Children’s Hospital of Philadelphia

After several years of work, we have finally completed our first efforts to understand how changes in a gene called *RAD21* cause features that overlap, some seen in children with CdLS. This work was guided by our previous findings of changes in the *NIPBL*, *SMC1A* and *SMC3* genes.

Yes, I agree the names of these genes are a bit crazy and cryptic, but the *SMC1A* and *SMC3* genes work together to make a bracelet-like protein called cohesin that is essential to hold a chromosome together. More specifically, a chromosome is actually made up of two exact replicates, called sister chromatids, which are held together at the middle.

The cohesin ring is the structure that holds them together in the middle. Without any cohesin, a cell will die. However, we have realized that even in situations where cohesin is present, but in smaller amounts, the chromosome function is normal, but there are problems with the way genes are turned on and off. It is this problem that results in the features of CdLS.

How does *RAD21* fit in here? Well, *RAD21* essentially functions as a clasp for the cohesin bracelet. Because we knew how it worked in other settings (most information came from yeast), we asked whether it might play a role in causing CdLS. To check this, we tested for changes in many children with CdLS for whom we had not figured out a cause. It turns out that we did not find changes in any children with more “typical” CdLS, but found changes in this gene in three children with milder features of CdLS.

Because we found the change in so few children and we wanted to know that we were not making any mistakes, we checked how these changes affected the way cohesin works in several settings, including cells, yeast and in zebrafish. All of these tests helped to confirm that the changes we found were relevant.

One of the findings that we found to be quite interesting is that these children, while they have growth and facial features that look a bit like CdLS, their intellectual development can be normal. We are hopeful that by understanding more about *RAD21*, we will gain insight about how we might find an angle to improve cognitive performance for children with CdLS.

This work was published in June in the *American Journal of Human Genetics*. Our goal is that this will encourage other researchers who have thought about how this gene works to focus some of their attention on CdLS. As always, we could not have done this work without all of the support and participation that we receive from families. This finding belongs to you as much as it does to our team here in Philly.

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CdLS and Anesthesia

By Aaron Zuckerberg, M.D., Pediatric Anesthesiologist and Critical Care Medicine, Sinai Hospital and member of CdLS Foundation Clinical Advisory Board and Antonie Kline, M.D., Medical Director, CdLS Foundation

One of the guiding principles of pediatric anesthesia is to utilize an individualized approach for each patient. As has been emphasized previously, the anesthetic approach to children and young adults with CdLS is and must be very individualized.

The most important elements for a positive experience are meaningful communication with the anesthesiologist before the procedure and parental presence during induction and emergence from anesthesia. Parents should be clear as to their child's previous experience with anesthetic drugs and actively participate in choosing what approach would be best for their child. Parents should find out from their anesthesia providers what the options are available for their child.

Parents know their children and will often guide the anesthesiologist into choosing the best option. As an example, some children who have asthma have experience with breathing treatments and are usually more comfortable than others are with a mask induction of anesthesia.

Some may not be fearful of needles, and others will only do well with a rapid shot of sedating medicine. The challenge is to successfully obtain IV access in the least traumatic fashion possible. Parental presence is often much more effective at relieving a child's anxiety during these difficult periods with medications. Depending on the institution, parents can bring their child's comfort items with them for both induction and the recovery period.

As children awaken in an unfamiliar environment, they may become agitated and upset. The remaining effects of the anesthetic may worsen the child's discomfort by clouding their perceptions and sensations. The presence of a parent and the child's favorite blanket, music tape, or video can be very effective at easing children through these periods. Parents should find out ahead of time what to bring on the day of the procedure.

Unfortunately, much of what we know about the interaction of anesthesia with individuals who have CdLS is based on anecdotal experience. In pediatric anesthesia textbooks, the anesthetic consideration for those with CdLS is limited to concerns about their airways and difficulty with IV access. Children with CdLS often have short jaws and necks, which increase the difficulty of maintaining an open airway and placement of a breathing tube. Not infrequently, a bronchoscope (an airway telescope) is required to safely place the breathing tube. Many patients with CdLS have shortened upper extremities and limitation of movement at the elbows, which limit the areas available for IV placement when they are awake. Often, following sedation, these contractures may be modestly relaxed and IV placement is easier. One textbook has stated that "patients with Cornelia de Lange Syndrome may have decreased anesthetic requirements." This certainly has not been our experience. Although individualized, children with CdLS seem to require more anesthetic per body weight than average in order to maintain an adequate plane of anesthesia.

Several medications often used in pediatric anesthesia are the Benzodiazepines: Midazolam (Versed) and Diazepam (Valium). Benzodiazepines are commonly used as a preoperative sedation in an effort to minimize anxiety and ease IV insertion. Depending on a child's age, these drugs can be given orally, nasally, as a suppository, or as an injection. These drugs work by binding to certain specific areas, called GABA receptors, which decrease the general activity of the brain, usually producing a state of calmness, sedation and amnesia.

Unfortunately, some people respond to Benzodiazepines in a paradoxical manner, becoming disinhibited (free of inhibitions), agitated, emotional, excited, or violent. It is estimated that one percent of healthy adults and up to five percent of healthy children will develop these paradoxical reactions to Midazolam. The exact cause of this reaction is unknown. There may be subgroups of patients who have abnormal GABA receptors that may predispose them to this abnormal response. To date, there is no data to determine whether the GABA receptor in CdLS is similarly affected.

Some recent information has been obtained, however, from a Foundation-supported study carried out by two dentists, Douglas Clemens, D.M.D, a CdLS Foundation Clinical Advisory Board member in private practice, and



Ellen Alpano, D.D.S., from the University of Maryland Dental School.

Dr. Alpano collected records and hospital charts on patients with CdLS who had undergone anesthesia for dental procedures. Based on hospital records, of those individuals with CdLS who received a pre-medication for anesthesia, 80 percent (four of five patients) who received Midazolam had an adverse post-operative event, compared to 28 percent (two of seven patients) who received a different medication as a pre-medication.

Thus, an alternative medication to Midazolam for anesthesia should be considered in patients with CdLS, but further studies need to be carried out since these numbers were very small [Kline AD, et al. Cornelia de Lange syndrome 4th biennial scientific and educational symposia abstracts. *Am J Med Genet Part A* 152A:2683-94, 2010].

The common risks of an anesthetic can involve complications of breathing, heart function, recall, and allergic reactions to the medications. These risks are usually far less than the risks of the car ride to the hospital. The major areas of increased risk for the patient with CdLS are airway and injury when emerging from anesthesia. As mentioned, some children with CdLS have an abnormal structure of their airway which increases their chance of developing some form of airway obstruction either during or after the procedure, as well producing an increased difficulty when placing the breathing tube.

Some children with CdLS awaken from their anesthetic in an aggressive state, which puts them at risk for injury from contact with bed rails and other medical equipment. Since history is usually very predictive, alerting the care team if this has happened previously will allow them to minimize the chances of injury by the use of pads on the hard surfaces of the bed.

All of this information should be reviewed with the anesthesiologist prior to any procedure performed under anesthesia. We are always happy to speak directly with the professionals, if indicated, as well.

New Research on Anesthetics and Airways Underway

Dr. Yvon Bryan, an anesthesiologist at Wake Forest School of Medicine in Winston-Salem, NC, and his research team are currently studying airway-related problems in individuals with Cornelia de Lange Syndrome (CdLS).

At present, there are few studies published regarding airway and anesthetic techniques in children with the syndrome. Results from Dr. Bryan's research survey will supplement current studies and facilitate the formation of specific airway and anesthetic techniques to reduce problems with oxygenation (getting oxygen into the body), ventilation (removing carbon dioxide) and intubation. This may be especially helpful when children require sedation and/or general anesthesia for repeated procedures or surgery.

If your child has received sedation or general anesthesia for a procedure or surgery, considering taking a few minutes to complete the survey at <https://www.wakehealth.edu/CDLS-Survey>.

Dr. Bryan's results and recommendation will be published in a future issue of *Reaching Out* at the conclusion of his study.

For questions, please contact Lauren Hoke at lhoke@wakehealth.edu.

This study has been reviewed and approved by the CdLS Foundation Research Committee.

You've probably seen funny looking blocks like this one on everything from books to orange juice containers these days. They are called Quick Response (or QR) codes. If you have a smartphone, simply download a QR code reader app, scan a block and you'll be taken to a Web page—in this case, the CdLS Foundation Web site.





Behavior Plans in Your Child's Individualized Education Plan

By Roger Woerner, B.S., SpEd., CdLS Foundation Educational Advisory Group

There are a couple of things about me you just can't change. I will live in Texas as long as I'm breathin', and I love working with children who have special needs. I have been an EBD teacher for a number of years. What's that you ask? Well, if you've been to meetings with professionals who work with children who have special needs, you know we like to throw around letters of the alphabet. EBD means Emotional and Behavioral Disorder.

Let's get this started with some sayings for you to remember. First, the barking dog gets the bone. That means if the "pros" (as I will call school personnel), say anything you don't understand or you disagree with, it's time to say, "hold your horses."

Two things have been proven: 1) Every child can learn; we are simply talking about the time it will take, and 2) if the pros are seeing a behavior, then there's a reason for it. The reason why a behavior is happening is sometimes the hardest thing to figure out. Often the parent's input gives the pros insight to why the behavior is occurring. Are the pros saying "this child IS a problem" or "this child HAS a problem." There's a big difference between the two statements.



A Behavior Improvement Plan (BIP) is a serious process to add to a student's Individual Education Plan (IEP). It says that the student has a behavior problem that is interfering with his/her educational progress. You are stating that the child's behavior plan is part of their educational needs for success in school. This will be reviewed and updated each year and follow him or her from teacher to teacher and school to school. A BIP must be followed across all educational settings with every "pro" who works with your child. This is not to say the child is bad, but that they have

special needs and there is a protocol to follow, if needed.

Before a BIP is implemented, a Functional Behavioral Analysis (FBA) must be performed by a qualified person (school psychologist). And before a FBA is performed, hopefully you have worked extensively with the teacher and tried some interventions. Interventions are ways of redirecting an undesired behavior. To change a behavior you must have the patience to teach and reteach the desired behavior, at home and school.

This is hard to implement at home with one child, so you can imagine a teacher having the time and patience to work with a student with a BIP.



Be supportive of the teacher. The teacher and parent must form a partnership to come up with meaningful incentives and consequences for the desired behavior. By saying meaningful, I mean meaningful for the child not the teacher or parent. Interventions should be tried (with a little modification) for six weeks before it can be judged as successful or not. The following is a list of information to document during this process:

- Are there antecedents to the behavior? (what happens before the behavior begins?)
- If so, is it predictable or is there a consistent pattern?
- What's the environment of the behavior? (where does it occur and not occur, e.g., with certain people, places, level of noise?)
- What does the child hope to get from exhibiting this behavior? (attention, timeout, etc.)
- What behavior can be replaced to gain the desired results?

If the interventions fall short, then a new FBA must be preformed. This also takes weeks and a lot of paperwork. You will be contacted by several school officials asking to meet with you and asking for permission for several



things, such as contacting your doctor, observing your child, and testing your child.

Upon completion of the FBA, your child’s IEP team will decide what action will be best for your child. For a BIP to become legally part of a student’s IEP it must:

- be stated in a positive manner and describe the behavior you want to see;
- be measurable (a certain percent of the time or how many minutes); and
- define the environment where the said behavior should be seen (in the classroom, the gym, etc.).

The BIP along with the FBA can be very useful tools, when used correctly with the parent’s partnership with the teacher. Gathering useful information during the process can mean the difference between correcting an unwanted behavior or not.

In addition to being an EBD teacher in Texas, Roger is the father of Michael, 24, who has CdLS.

A Closer Look at Behavior Plans

Behavior problems are an issue most parents with young children face; however, for parents of children with Cornelia de Lange Syndrome (CdLS) it can be a constant battle. Oftentimes, children with CdLS struggle with behavior. They may be aggressive, exhibit self-injurious behaviors and can also be hyperactive. This may lead not only to troubles at home, but also in school.



Isaac at the National Family Conference

Vanessa, mother to six-year-old Isaac, is very familiar with this topic. Isaac started having behavioral issues at age two and a half—around the time her second child was born. She began to notice that Isaac was aggressive and showing self-injurious behaviors like hitting, nail biting, and hair pulling. To this day, his behavior is an ongoing struggle.

“Isaac is non-verbal, although he is learning sign language. Due to his lack of communication, it’s very difficult to determine what the problem is when he begins to misbehave,” says Vanessa.

Luckily, Isaac has a paraprofessional who has known him since he was eight months old. Isaac has also had regular IEPs (Individualized Education Plan) since he was in preschool. With the combined effort of Isaac’s family and his paraprofessional, a behavior plan was implemented that has proven to make a difference.

Vanessa says that Isaac’s paraprofessional has been able to “grow with him,” which has helped in dealing with his behavior problems. She says that providing Isaac with the same paraprofessional for so long gives him consistency, which is critical in his behavior plan.

Sometimes Isaac’s behavior can be dealt with by simply giving him a timeout, and other times it requires complete removal from the situation. One element of his behavior plan is a one, two, three counting sequence. When Isaac begins to misbehave, he gets until the count of three to start behaving or else he gets a time out.

This counting sequence is usually put in place when Isaac’s behavior isn’t severe. When Isaac acts out physically, it leads to an immediate timeout. Vanessa says that a lot of Isaac’s behavior is attention seeking; therefore, removing him from the situation is one of the most effective ways to deal with his behavior.

While at National Family Conference, Vanessa was pleased to learn that removal is an effective element of a behavior plan for a child with CdLS. She also learned that including “social stories” before an event with high activity and many people—such as a birthday party—can help Isaac know ahead of time what happens at such events and how to behave appropriately while attending the event.



CdLS Doesn't Stop These Adults from Living Full Lives

Attendees at the National Family Conference were treated to a very special session, *Grow with Me*, featuring the stories of five adults with CdLS—four of them told in their own voice. While each presenter's story was different, the one similarity was that each person was determined to lead a normal life and achieve his or her goals.



Molly, Emily, Rachel, Valerie, and Maegan

Rachel, 27

Rachel has earned her high school diploma, Associate's Degree and driver's license. She said that working, going to college, setting goals, counseling, and socializing all made her transition into adulthood less stressful.

For Rachel, friends and employers were very important in helping her grow into an adult. She said she recognized that the journey towards adulthood is not always easy for someone with a disability; however, she felt that the people surrounding her have provided important support and opportunities to help her continue to move forward and grow.

Valerie, 33

Valerie has been able to grow and mature tremendously. She never used to speak to anyone outside her immediate family. As Valerie got older, she got a job and moved into her own apartment, but still rarely spoke to anyone.

She met another adult with learning disabilities and the two developed a very close friendship. That friendship helped her overcome her shyness, and she began talking in front of people for the first time, including speaking publicly at many CdLS Foundation events.

Alex, 21

Molly, mother of Alex, spoke on her son's behalf, as he cannot communicate verbally. Molly said that in order for transition to be effective, young adults with disabilities should have the same opportunities as their peers to participate in society through work, school and social activities.

Alex has several jobs, including working at a dog treat bakery and library, and owns a vending machine that he is responsible for sorting items for and replenishing. Molly is looking at having Alex attend the local community college to audit a class or two and hang out in the cafeteria with others his own age.

Maegan, 38

Maegan's major transition event was being diagnosed with CdLS in her late 20s. She had the same normal kind of hopes, like a job, living in her own apartment and finding friendships.

Once she learned she had CdLS, struggles she had experienced for much of her life—like difficulty in school, voice problems and trouble staying on task—made sense.

Maegan did finally get her own place and said she has learned not only about her own personal habits and quirks (like not wanting to talk on the phone or forgetting to refill prescriptions), but also some important life lessons—like patience and self-determination. She also found community supports and job assistance and is an advocate for herself and others with disabilities.

Emily, 19

One major transition that Emily, the youngest presenter, discussed is her upcoming transition from high school to college. Emily is attending Faulkner University in the fall and is ecstatic about it. She has already taken several big steps for her transition, such as taking placement tests, registering for classes and learning more about college life.

Emily's successes in leadership programs and speech competitions have helped her gain confidence that will help with the transition from high school to college. Although Emily recognizes that she has challenges ahead of her, she is more than ready to move forward in her life. Emily ended her presentation with an inspiring quotation: "I have CdLS, but CdLS will never have me."



Special Individuals Honored at Conference

As with past family conferences, several awards were given out at the Saturday night banquet. The awards recognize and show gratitude to individuals who dedicate their time and effort to the Cornelia de Lange Syndrome (CdLS) Foundation.

Sue Anthony Award: The Sue Anthony Award is given to someone who displays innovation, integrity and commitment. This year, it went to two people: husband and wife Mary Opitz and Brian Luyt.

John King Award: The John King award is given to an individual whose long-term efforts on behalf of the Foundation have made a great impact. The award was given to Marjorie Goodban, Ph.D., a speech-language pathologist who has been working with children with CdLS since 1984.

Mark Etcheberry Award: The Mark Etcheberry award is given to someone who has dedicated his or her efforts to raise awareness of CdLS and the Foundation. The award was given to Ken Fouts, who rode his Harley from New Hampshire to California in honor of his grandson Adrian.

Special Recognition Award: The special recognition award was given to Emily Turner. As part of Emily's Girl Scout Gold Award requirements, she organized a CdLS family gathering in her home state of Alabama.

Outstanding Philanthropy Award: This award recognizes an outstanding individual or organization whose long-term contributions of financial support, time, and talent have made a significant impact for individuals with CdLS. Honorees were Sue Leone, John and Mary Fowler, and Andy and Wendy Miller.

Doctor Laird Jackson Award: The award recognizes significant medical or scientific contributions towards understanding CdLS. This year the award was given to Dr. Alex Levin, an ophthalmologist and long-time member of the CdLS World Scientific Advisory Council.

In addition, former executive director Julie Mairano received a Lifetime Achievement Award from the CdLS World Federation.

Corrections

The following are corrections and clarifications in regards to statements made in the Spring 2012 article *Making Sense of Sensory Integration in CdLS*.

- The correct credentials for Christine Ackermann are M.Ed., O.T.R. /L.
- The preferred terminology is Sensory Processing rather than Sensory Integration.
- “Occupational and physical therapy, provided by qualified professionals who have received post-baccalaureate training in sensory integration theory and treatment, can help your child.” **Clarification:** There are qualified therapists with and without post-baccalaureate training; the important thing should be qualified professionals who have received training in sensory integration theory and treatment.
- **Clarification:** In regards to *The Sensory Profile*, by Winnie Dunn, it is just one tool that may be used, but is not the only tool. Therapy will begin with an evaluation by a qualified occupational or physical therapist, which may include a sensory assessment tool, such as the one by Winnie Dunn. As part of the evaluation, the therapist will observe how the child responds to different stimuli in his or her environment.
- “From this information, the OT creates a “sensory diet” created to meet specific sensory needs.” **Clarification:** This is not always the case; some children need a specific sensory diet, and others benefit from sensory strategies/tools without specific timed interventions.
- “The OT should write this sensory therapy into the student’s Individualized Education Program (IEP) each year it is needed. **Clarification:** Sensory strategies are used regularly without being in the IEP; additionally, sensory diets may change throughout the year as well.



Super Siblings: Austin and Katherine

Eleven-year-old Katherine, has already taught her brother Austin, 14, several quintessential life lessons. “Having a sister with CdLS has made me more aware. It has made me more humble, more mature and more of a leader,” says Austin, who lives with his family in the Atlanta suburbs.



And although he knows there are challenges of having a sister with a disability, he says he chooses to see the positive and use the lessons he’s learned from his sister as a way to better himself.

One of those ways is volunteering in the CdLS child care program at the National Family Conferences in 2010 and 2012. Austin says that he thoroughly enjoyed the experience, especially playing with the kids.

“I do child care because it gives me experience, and also allows me to see different stages of CdLS from mild to severe,” he says. Austin says that one of the biggest lessons he’s learned by participating in child care is “be patient and show love and compassion.”

Austin believes it’s very important for every family touched by CdLS to participate in Foundation events, like conference. He understands that not all families can make every conference or every event; however, he feels if each family were to contribute something, whether it’s raising awareness, making a donation or attending an event, it could make a great impact.

He says that the CdLS Foundation is “one big family” and being active in events such as conference 2012 “brings everyone closer together.”

Study Examines Sleep Patterns in CdLS

Dr. Stacey Ishman invites individuals with Cornelia de Lange Syndrome (CdLS) and their parents to take part in a research study in order to characterize sleep patterns in people with CdLS.

Participants will be asked to complete a questionnaire regarding typical sleep patterns. The information gained from this study will improve recognition of sleep disturbances in the CdLS population and benefit future patients.

The questionnaire can be accessed and submitted electronically through <http://bit.ly/Mztf97>.

If you have any questions or would like to receive a paper copy, contact Dr. Stacey Ishman at researchsleep@gmail.com or call 410-502-3225.

This study has been reviewed by the CdLS Foundation Research Committee.

WELCOME NEW FAMILIES

Illinois

Anna and Leonard and son Cole, born February 28, 2009

Ohio

Jessica and daughter Josie, born September 14, 2009

Georgia

Rachel and Brian and daughter Elizabeth, born September 13, 2009

Pennsylvania

Stephanie and daughter Dondriah, born October 13, 2011

Michigan

Audrey and Aaron and daughter Evelyn, born March 7, 2012

West Virginia

Bethany and Phillip and son Eli, born December 3, 2011

North Dakota

Sarah and Brian and son Regan, born June 17, 2011

Wisconsin

Ronald and son James, born July 2, 1964



Mailbag – Christopher –

Christopher, otherwise known as CJ, was born in May 2004 to parents Christopher and Ali in Mauston, WI. You may not know of Mauston since it is a very small town in Wisconsin. At first, CJ seemed like a normal, happy, healthy, baby boy. But shortly after his birth, it was discovered that he was blind in one eye. CJ and his parents had to endure many tests, many doctors, and still no answers. Finally, after being examined by a group of specialists who reviewed his medical records, he was diagnosed with an uncommon condition known as CdLS, which stands for Cornelia de Lange Syndrome.



Some of the characteristics of this condition are: long thin eyebrows that meet in the middle, small hands, lazy eye, and that is just to name a few. CJ does carry all of the characteristics that were just mentioned, as well as a significant learning disability and repetitive speech. In spite of his condition, CJ is the most adorable, polite, caring, and affectionate little boy I have ever met!

Ever since CJ was a young toddler, he has found an incredible interest in music and he is talented too! As long as I can remember, he has always loved the drums. He will use anything that can become a drum or drumstick, and just play it constantly. When he was not banging on drums, you could find CJ playing with one of his many train sets because he adores them. Likewise, he has always had a fascination with fire trucks, ambulances, and police cars. It could be because his father was a part-time firefighter, or just simply because of the bright lights and noises. Also, CJ has always been a musical and bubbly little boy. He is very delightful and enlightening to be around!

Furthermore, CJ loves his family members dearly. He will go out of his way to just tell someone that he loves them. Whenever, he comes to town he is overjoyed to see everyone again! He is especially close to my younger brother, Christopher, who shares the same name. CJ calls my brother ‘Chrifer’, and me, ‘Stina’. It is adorable how much he cares for all of us!

All in all, CJ is one special young boy. He is unique, and loving, and his caring can fill anyone’s heart. He has love for everyone and everything, no matter what!

Kristina, age 13
CJ’s cousin, IL

Submit your Mailbag or
Super Siblings Story!
Send your story and photo to
bshepard@CdLSusa.org.

Foundation Accepted into Federal Rare Disease Registry Program

The Cornelia de Lange Syndrome (CdLS) Foundation was one of just 34 organizations from around the country accepted into the National Institutes of Health’s (NIH) Global Rare Disease Patient Registry and Data Repository (GRDR) pilot program.

The GRDR strives to aid in research, clinical trials, drug developments and therapeutics for people with rare conditions, such as CdLS. The goal of the GRDR is to provide a sustainable resource for the organizations accepted into the program.

“We are thrilled to be part of this important new program,” said Acting Executive Director Marie Malloy. “A national registry of CdLS data lets researchers compare and evaluate data among people with CdLS, as well as other similar conditions. In the long-term, the longitudinal data collected through this registry will help doctors understand more about CdLS over a person’s lifetime.”

Families will be contacted in the coming months about participating in the registry.



Golfing and Giving

New England families and friends of the Cornelia de Lange Syndrome (CdLS) Foundation hit the links at Ipswich Country Club in Ipswich, MA, on May 21 to celebrate the 20th Annual CdLS Charity Golf Tournament.

The noon shotgun start was only the beginning of a day filled with opportunities designated for supporters to challenge their skills on a course designed by Robert Trent Jones, Sr., try their luck at games of chance and bid on both live and silent auction items.



New England Golf participants

Returning sponsors included RBC Capital Markets, KeyBanc, and Macquarie Capital. New sponsor, Susquehanna Financial Group, joined in the effort this year as well. Individuals with CdLS were the real winners, as more than \$64,000 was raised at this event thanks to the efforts of the golf committee, supporters and volunteers that helped that day.

Less than a month later, another enthusiastic field of golfers gathered at Crane's Landing Golf Club in Lincolnshire, IL, to celebrate and support individuals

with CdLS. Among our guests was 15-year-old Nick, a young man with CdLS, who rallied the golfers at the start, addressed the guests at dinner and provided a wrestling demonstration with the assistance of his coach.

Although temperatures reached the mid 90s that day, it did not distract the golfers from helping the Foundation raise more than \$26,000 to benefit the 2012 National



Nick and friends at Crane's Landing

Family Conference, which started the following day.

On October 8, the 24th Annual CdLS Charity Golf takes place in Eureka, MO, organized by Madison County Wood Products and Pallet Logistic employees.

Throughout the years, dedicated sponsors and golfers have shared the gratification of raising more than \$386,000 to help people with CdLS. Registration for this event is available online at www.calendar.cdlsusa.org or by calling the Foundation at 800.753.2357.

2012 CALENDAR

September 15

3rd Annual Brew & BBQ
Atlanta, GA

Northeast Region Family Gathering
Dresher, PA

Midwest Region Family Gathering
N. Platte, NE

September 16

The Saratoga Palio
5K and half marathon
Saratoga Springs, NY

Maddy's Run 5K
Salt Lake City, UT

October

Southeast Region Family Gathering
Kentucky
Date and location TBD

October 7

Bank of America
Chicago Marathon
Chicago, IL

October 8

24th Annual CdLS Charity
Golf Tournament
Pevely Farms Golf Club
Eureka, MO

October 13

ING Hartford Marathon
5K, half marathon and marathon
Hartford, CT

Baltimore Running Festival
5K, half marathon and marathon
Baltimore, MD

California Family Gathering
Escondido, CA

November 4

ING New York City Marathon
New York, NY

November 10

Florida Family Gathering
Fort Myers, FL

November 11

Sarah's Bowl
Batesville, IN

November 18

Philadelphia Marathon
8K, Half and full marathon

December 1

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

December 8

CdLS Multidisciplinary Clinic
for Adolescents and Adults
Baltimore, MD



Event Recap

Lapel Village Fair CdLS Benefit 5K



Scott and Tammy Hersberger of Lapel, IN, organized the 13th Annual 5K to benefit the CdLS Foundation.

Down & Dirty in Philly



The Feehan and Stone families came out in full force to get “dirty” for CdLS in July.

Awareness Runs for Danika



Meliza Carter and her family organized runs in both Olympia, WA, and Waikiki, HI.

Kristian's Bowl



The First Annual Kristian's Bowl in Manitou, KY, was a “strike” of success in June.

Behind Team CdLS is an Even Bigger Team

One that includes you. This fall, Team CdLS runs in races throughout the country, including the Bank of America Chicago Marathon, ING New York City Marathon, Baltimore Running Festival, ING Hartford Marathon, and Philadelphia Marathon. You can support Team CdLS and the Foundation by joining our Phantom Marathon team.



Since 2008, this virtual team of parents, relatives and friends has raised thousands of dollars for the Foundation simply by asking people to support their “run.”

No sneakers, no sweat, no stretching.

Ready to join the team?

There are two ways to get started.

If you prefer contacting friends and family by mail, request a Phantom Marathon Runner Kit. The kit has everything you'll need to ask 10 people for support of your “run” (you can request more kits). Request your kit on our web site at www.cdlsusa.org/events/phantom-runner.htm or call 800-753-2357.

If you prefer to do things on the Internet, create an online fundraising page at First Giving for free. Simply go to www.firstgiving.com/TeamCdLS/2012-team-cdls-phantom-runner. First Giving guides you through the process of creating your individual page, which you can then email to your contacts.

For questions or more information, contact Kellie at 800-753-2357 or specialevents@CdLSusa.org.



Donations from 3/31/2012-6/30/2012

Gifts that Count - In Honor/Celebration

Aaron Birdwell's 16th Birthday

Mrs. A. Jetton
Mrs. D Waldon
Mrs. N. Cox
Mrs. B. Ogle
Mrs. L. Wasserbeck
Mr. Charlie Dunn

Adam Jackson

Dorothy and Daniel Steimke

Allie Boteler

Ruth DeBelo

Mandy Drexler's Birthday

Tanya Ruhe

Andrew Schwalbe's Birthday

Rosanne and Frank Fidler

Charles Gladson

Debra Fleeger
Richard Hebson
Alan Kessler

Charlie Newcomb

Carol Heitman

Colin Bell and Rilette Hugo on

their birthdays
Sally and Charles Simpson

Colin Bell's 9th Birthday

Jane and David Bell
Rilette and Jerry Hugo
Cheryl and James La Roe

Conrad Hersberger's Birthday

Laura and Michael Doyon

Conrad Clemens' Bar Mitzvah

Mollie Herman
Ronna Jablow

Hollis Wein's Birthday

Lois Braverman

David Podell's Birthday

Diane Fromhartz

Dean Modler's Graduation

Elizabeth and Kevin Hardy

Doris Patitucci's 80th Birthday

Edith and Charles DiAddezio
Mary Tossona
John Zappile

Doug Clemens' Birthday

Michelle and Christopher
Canning
Joanne Kennard
Maria and Maury Paslick

Ella Grace Musial's 4th Birthday

Elaine and John Fuerst
Virginia Lambert

Ellen Braverman and Robert

Fishel's 25th Anniversary
Diane and Michael Friedman

Emma Pietrafesa's 6th Birthday!

Lisa Chen

Gracie Fry

Cheryl and Frank Andrews
Linda Chatham

Miss Hannah Ashley Kimball's

15th Birthday
Barbara Davis
Arthur Dowdle
LuAnn and Don Garmon
Julaine Gentry
Jack and Margaret Joyner
Deborah and Max Kimball
David Lewis
Bettie Lewis
James McAlpin

Hannah E. Moore

Patricia and Brian Moore

Jack Pollard's 12th Birthday

Mary and David Battease

Jared Linsk

Dina and Jim Garber

John Cataline's Retirement

Elizabeth and Richard Brooke

Josh Ackles

Carole and Richard Chapman

Josie Schreder's Birthday

Pat and James Kuta

Kaity Kuepferle

Nancy and Larry Kuepferle

Katie Luyt

Robbin and Bruce Bailey

Koel Bird

Susan Stelzer

Logan Fowler

Mary and John Cavalieri

Matthew Rodgers

Donna and Todd Rodgers

Michael Cataline

Elizabeth and Richard Brooke

Michael Viola's Birthday

Kristine Evina
Lisa Paragano
Noble Woodall

Mikayla Needlman

Phyllis and Joel Needlman

Molly Wagner's 24th Birthday

Kathleen Browne
Erin Wagner

Manny Tutuer's Birthday

Diane and Michael Friedman

Ollie Wilson

Glenda Wilson

Rebecca Davis

Kathy and Dan Clancy
Trisha Trembl

Robert Ackman

Ronald Goss

Ryan O'Connell

Robert Malouf

Steven Siegel's Birthday

Susan Hartman

Stuart Kaufman's 80th Birthday

Howard Braverman
Lois Braverman
Diane and Michael Friedman
Marine and Bud Grant
Gloria and Larry Green
Fran and Nate Karlin
Andrea and Marc Needlman
Betty Schraiber
Evie and Jack Solway
Helene and Milt Wagner

Mr. and Mrs. Paul Frank

Lois Braverman

Sydney Sachs' Graduation

Andrea and Marc Needlman

Taylor Nelson

Jane and Stephen Nelson

Will Smisloff

Elaine and Thomas Whalen

In Memory

Aimee Langlois

Frances Burke

Alex Gustowarow

Gina Auriemma
Diane Glantz
Carol and Juergen Gustowarow
Elizabeth and Alexander
Gustowarow
Caroline and Roger Knauss
Donna and John Murphy
Jan Northington
Jean Parsons
Ruth and Ronald Roshong
Lynn Saal
Inge Wiley

Allison Tryba

Susan and Lyle Tryba

Ann Galanek

Michael Mancini

Anna Gonella

Maria and Bozo Batkovic
Laura Del Carlo
Bianca Caserza
Patricia Costello
Marie and Allan Levy
Mercer Limited - Canada
Barbara and John Molinari
Susan and Raymond Molinari
Diane O'Neill
Alice Perin
Katherine and Jeffrey Whitman

Anne and Michael Gonella

Rozella Cole
Philip Schneider

Anna and Andrew Dunn

Jane and David Bell

Carol Snyder

Karen and Larry Prada

Clara Noll

Barbara Ann and Duane Fuller

Collin Murray

Teresa and Ronald Lund

Darlene Lillian Collins

Mary Opitz and Brian Luyt

Fletcher Presgraves

Board and Staff of the CdLS
Foundation
Joanne Maher

Forrest McKerley

Bessy Jones

Gary Hartman

Steven Siegel

George Carter

Patricia and Joe Phillips

George Eckerson in celebration

of Brendan Eckerson
Joann Atlas
Donald Benoit
Peter Burtis
Penny and George Leonard
Local Union No. 373 U.A. -
Plumbers and Steamfitters
PA Interfaith Community
Programs, Inc
Stanley and Marcia Polhemus
Mary and Kenneth Reiver
Susan and Robert Tremper
Teresa Wilson
Cheryl and John Wirth

Jennifer Rebenack

Normarie and Paul Rebenack

Jindi Van Buren

Gloria and Warren Van Buren

John Viola Sr

Margaret and Salvatore
Napolitano

Justin Rinn

Frances Edwards
Kathryn Trakas
Rebecca Wales

Katie Lombardi

Laura Alfano
Theresa Lombardi

Kerry Myrthel

Willing Workers of Paradise
Valley

Manuel Garcia

Tim Cummings
Lev Dassin
Liana Davila and Michael Garcia
Karen Dickson
Barbara and David Eichenberger
Linda Frazer
Laura and Manuel Garcia
Louise Gould
Norma and Ronald Hanstein
Randy Hensley
Eric Houghton
Hazel Kirk
Susan and Frank Leone
Gilbert and Barbara Luctman
Cynthia Nilson
Kathryn Schultz
Carlle and Bob Steele
Carolyn and Leonard Willey

Robert Bublely

Ellen Braverman and Robert
Fishel

Liddy Horsey

Karen Manning and
Thom Horsey
Mary Manning
Jill and Andrew Stefanovich

Michael Gonella

Mary Chory

Michael J. Donziger

Myrna and Alan Kaplan

Natalie Nikzad

Katherina and Jon Terhune

Pat Finley

Angela and Wayne Wrolstad

Robert Henderson

Cecelia Henderson



Sara Peracchio
Amelia Varca

Wendell House
Connie and Scott Kelton
Mary and Mark Kelton

Stephan Knapp
Betty and Michael Obert
Kathy and Gene Van Buren

William Dunn
Jane and David Bell

Tommy Farr
Donna and Barton Bovee

Flexibility. Because you're not actually making a gift until after you die, you can change your mind at any time.

Tax relief. Your estate is entitled to an estate tax charitable deduction for the gift's full value.

Beneficiary Designations

Assets, such as retirement plans, life insurance and insurance annuities, are not controlled by the terms of your will. These assets instead require separate beneficiary forms. The beneficiaries of these assets can be easily modified at any time to meet your changing needs:

- IRAs and retirement plans
- Life insurance policies
- Insurance annuities

If you are interested in leaving a legacy with the Foundation, call 800-753-2357

The information above is not intended as legal, tax or investment advice. For such advice, please consult an attorney tax professional or investment professional.

Plan Now to Make a Difference in the Future

While making plans for your estate, consider including a legacy gift to ensure that the CdLS Foundation remains the vital organization that families of children with CdLS need.



CIRCLE of CARING
CdLS Foundation

A planned gift not only demonstrates commitment to the mission of the Foundation, it ensures the long-term strength of the organization. Long after they are given, planned gifts positively impact the lives of children and adults with CdLS and those who care for them.

Bequests

To make a charitable bequest, you need a current will or revocable living trust. Your gift can be a percentage of your estate, or a certain amount of cash, securities or property. After your death, the CdLS Foundation receives your gift. Called a charitable bequest, this type of gift offers these main benefits:

Simplicity. Just a few sentences in your will or trust are all that is needed. The Foundation can provide you with specific wording.

Our Deepest Sympathy

Fletcher Presgraves
October 5, 2006 – May 28, 2012
Son of Julie and Darian Presgraves
316 Kidwell Ave
Centerville, MD 21617-2027

Azure Haskell
March 17, 2012- April 10, 2012
Daughter of Hope Haskell and Andrew Annese
63 Ford Dr. Waterville, ME 04901



CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.
302 West Main Street #100
Avon, CT 06001-3681
800-753-2357 • www.CdLSusa.org

Yes, I want to help people with CdLS.

Enclosed is my tax-deductible gift of:

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

I have included the CdLS Foundation in my will or trust



Please remember the CdLS Foundation in your will.

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

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Together, we can make great things happen.

You can help the CdLS Foundation expand its resources and improve the free services and programs that directly affect the growing needs of families, including your own.

Today, the assistance available to you and your family is sustained by just 25 percent of the families we serve. With your support, you can strengthen the services that we provide, including:

- regional gatherings and biennial conferences that allow you to connect with other families;
- professional packets for your child's doctors and teachers;
- clinical studies and continued research on CdLS;
- toll-free help lines to call when you have questions; and
- quarterly newsletter you are reading right now.

Imagine the great things you can help us achieve with your contribution.

Why is your participation so important? This is *your* Foundation.

Please consider a donation today. You can use the coupon and envelope included in this newsletter; call 800-753-2357 with a credit card; or go to www.CdLSusa.org and click the **Donate Now** button.

Thank you.

