

Reaching Out

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



Highlights

Director's Message:

A logo says a lot

Spotlights:

Understanding Seizures/
Epilepsy

Behavior Scale Can
Help Clinicians

Super Siblings:

Jillian and Delaney



Director's Message



A logo says a lot

You may have seen the logos below on our Web site or in a thank you letter, but do you know their importance?

Only organizations that demonstrate financial and programmatic transparency are allowed to utilize these logos—and not many nonprofits earn the right to do so. In fact, only 1,668 charities nationwide have earned the gold seal from GuideStar—and the Cornelia de Lange Syndrome Foundation is one of them. To earn the gold seal, an organization must openly share its financial data and program goals. It sounds simple enough, but many nonprofits are not willing to share this information publicly.

Charity Navigator, which is known as the nonprofit industry's watchdog, awarded the Foundation its third consecutive four-star rating. This top rating is based on the organization's financial health, accountability and transparency. In layman's terms, Charity Navigator thinks the Foundation is doing a great job with the money our donors entrust us with. Only 11 percent of the charities nationwide have received three consecutive 4-star ratings, meaning the CdLS Foundation is doing phenomenal work.

The Foundation also has earned a Best in America seal from the Independent Charities of America (ICA). The purpose of the ICA is to recognize charities "that meet the highest standards of public accountability and program effectiveness."

And this past year, the Foundation earned a spot as a Top-Rated Charity through Greatnonprofits.org. Organizations can only gain this four-star rating through positive reviews by community members, volunteers and donors.

So what does all of this mean? When you see these logos, you know that your gifts are in good hands.

Marie Concklin-Malloy
Executive Director



Our Mission
The Cornelia de Lange Syndrome Foundation is a family support organization that exists to ensure early and accurate diagnosis of CdLS, promote research into the causes and manifestations of the syndrome, and help people with a diagnosis of CdLS, and others with similar characteristics, make informed decisions throughout their lives.

In This Issue

- 4 Understanding Seizures/
Epilepsy
 - 5 A Closer Look at Seizures
 - 6 Behavior Scale Can
Help Clinicians
 - 7 A Closer Look at Behavior
- 8 Super Siblings:
Jillian and Delaney
 - 9 Mailbags
 - 10 Event Highlights
 - 12 Save the date:
CdLS Foundation National
Family Conference



A Heartwarming International Meeting



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

Marie Malloy and I had the privilege of attending the international Cornelia de Lange Syndrome (CdLS) Federation meeting in Buenos Aires, Argentina, in November. Since that part of the world was heading into summer, the jacaranda trees were in bloom and the city had a festive, buoyant atmosphere.

It was an absolutely heartwarming meeting, not only because most of the families had never attended a CdLS meeting before, but also because the international community that came in support of the meeting had such a strong, generous presence.



The 52 families in attendance hailed from Argentina, Brazil, Chile, Ecuador, Paraguay, Peru, and Uruguay. CdLS World Federation members and professionals came from Australia, Denmark, England, France, Germany, Holland, Italy, Portugal, Spain, Switzerland, and the United States.

For two-and-a-half days, there were medical, educational and supportive talks, followed by consults with professionals representing genetics, gynecology, behavioral psychology, pediatric dentistry, speech pathology, and education.

Much like our U.S. conferences, the talks discussed reviews of medical and dental recommendations at different ages, behavioral concerns and management, puberty issues, adult concerns, prenatal findings, and the genetic basis of CdLS with discussions of the five current genes associated with the cohesin ring.

New information included the possibility of a sixth gene, as well as the presence of mosaicism in CdLS, in which not all of the cells in the body have a change in a gene associated with CdLS.

There were also several talks about countries building a hospital or chronic care structure for individuals with CdLS and other conditions, especially as they get older.

The group felt like one giant family. The meeting ended with a photograph of all of the children attending in the middle of the conference room and doting family members and professionals alike crowding in to take the picture.

We're looking forward to the next international meeting in Lisbon, Portugal, in 2015.

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 . . . Robert Boneberg, Esq., NJ
- VICE PRESIDENTS
- David Harvey, GA
- Richard Haaland, Ph.D., GA
- TREASURER David Barnes, Esq., NH
- SECRETARY Wendy Miller, Esq., CA
- DIRECTORS
- Eileen Ahearn, M.D., Ph.D., WI
- Catherine Caron, NH
- Shelly Champion, MA
- Michele Churchman, DE
- Dennis Drislane, CA
- Dianne Lessa, MA
- Maegan Lowrey, NC
- Carlos Madrid, Esq., TX
- Marc Needlman, IL
- Julia O'Connor, Ph.D., MD
- Mary Opitz, TX
- Fran Rissland, GA
- Beth Smisloff, NY

PROFESSIONAL STAFF

EXECUTIVE DIRECTOR
Marie Concklin-Malloy

MEDICAL DIRECTOR
Antonie Kline, M.D.

ASSISTANT EXECUTIVE DIRECTOR
Kelly Brown

DEVELOPMENT MANAGER
Gail Speers

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

FAMILY SERVICE COORDINATORS
Lynn Audette, M.S.W.
Michelle Kiely, M.S.W.

COMMUNICATIONS COORDINATOR
Brenda Shepard

GRAPHIC DESIGNER/WEB MASTER
Francesca Scognamiglio

DONOR RELATIONS ASSISTANT
Ciara Duggan

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*.



Understanding Seizures/ Epilepsy



By Lynne M. Kerr, M.D., Pediatric Neurologist, Division of Pediatric Neurology, University of Utah Medical Center

A seizure is the change in attention or behavior that occurs due to an episode of abnormal electrical activity in the brain. Epilepsy is a brain disorder in which a person has repeated seizures over time.

For unknown reasons, individuals with Cornelia de Lange Syndrome (CdLS) have an increased incidence of epilepsy compared to the general population; seizures are observed in approximately 15 percent of children with the syndrome.

The first question a medical professional considers is whether the episode was in fact a seizure or some other kind of spell. In children with CdLS, reflux episodes causing arching may look like seizures. Other spells that mimic seizures include breath-holding, daydreaming, tics, and stereotypies, which are repetitive, apparently non-functional, possibly self-stimulatory behaviors. It is important to definitively diagnosis the episode as being due to a seizure, since other types of behavioral events will not respond to anti-epileptic medication and these medications can have unwanted, sometimes dangerous, side effects.

Conversely, sometimes children with no behavioral or consciousness changes will have an electroencephalogram (EEG) that is abnormal. An EEG is a non-painful test involving the application of electrodes to the head to record brain waves. Although there are exceptions, in general we do not treat children for abnormal EEGs without behavioral events that may be seizures. For example, abnormal EEGs without behavioral correlates are fairly common in children with autism, and this may be true in children with CdLS as well.



If the physician feels that the episodes that the child is having are likely to be seizures, she or he may perform an EEG. EEGs are not performed unless there is a strong clinical suspicion of a seizure disorder.



Since it's often difficult to tell if a particular episode is a seizure, family members may be asked to keep a record of the events for a few weeks, and even videotape the activity they are concerned about. A follow-up is then scheduled with the neurologist. If events that are concerning seizures are occurring fairly often, and

a standard EEG, which is only 20-30 minutes long, does not show epileptiform activity, the neurologist may perform a longer EEG.

These can be done in two ways. The first is an ambulatory EEG where the EEG wires are worn while the individual performs their normal activities at home for several days. While the ambulatory EEG is in place, the individual or family is able to push a button to demonstrate when the activity of concern occurs which then can be correlated with the electrical activity in the brain. The second is an in-hospital video EEG where the behavior/consciousness changes that may be seizures are monitored with video while the individual wears an EEG montage.

As a member of the Clinical Advisory Board for the CdLS Foundation, and as a practicing pediatric neurologist, I have been asked many times about behavioral episodes and whether or not these are seizures. One example is an individual with episodes of aggression. It is not likely that behavior changes such as rage or aggression are due to seizure activity. However, if the questions persist it is possible to determine if events of concern are seizures or not by recording the brain's electrical activity during the event by either ambulatory EEG recording or a prolonged in-hospital EEG. The second example is a child whose heart rate went up to a high level and stayed that way for a while after being given food by his gastrostomy tube. The certainty that this testing can offer can allow the event of concern to be addressed directly and lays the question of underlying seizure to rest.



If seizures are diagnosed, the physician often orders further testing, including a brain MRI to determine if there are any structural changes in the brain such as scarring that may be causing the seizures. MRI is preferred to a CT scan because although individuals may need to be sedated for MRIs, the images obtained of the brain are much clearer and there is no exposure to radiation.

While the type of seizures and age of the child guides the choice of medication, convenience and expense for the family should be considered. All seizure medications have potential side effects, including changes in mood, activity level and learning. Every family should ensure that they have been thoroughly informed regarding the benefits and risks of the medicine being prescribed for their child.

Families should also be aware that if the first medication chosen causes unwanted behavior changes such as difficulty sleeping or difficulty focusing, they should contact their neurologist regarding changing medications. There are alternate therapies as well that are sometimes used when children have difficulty controlling seizures. These include a special diet (the ketogenic diet), epilepsy surgery, and implantation of a vagus nerve stimulator. Your neurologist may refer you to a specialized epilepsy center if seizures are difficult to control.

Questions regarding events that may be seizures, which medication is the right medication, and possible side effects should be discussed with the child's neurologist. It's helpful to find a neurologist familiar with children with CdLS when possible.

For more information on seizure disorders, visit www.medhomeportal.org, www.epilepsyfoundation.org or www.epilepsy.com.

A Closer Look at Seizures: Ryan's Story

We want to thank Ryan's mother, Shari, for sharing Ryan's story with us.



In April 2012, Ryan was at the group home getting ready for a day program [when he had his first seizure]. This seizure lasted 90 seconds and this is what the nurse noted as his symptoms: "Pale, eyes turning to left, slumped forward, both arms trembling." He returned to baseline after the seizure. The home called it a Petit Mal seizure.

A few months later, he had another seizure. This seizure lasted 50 seconds and this is what the nurse noted: "Staring spell, slow, flushed, upper extremities became cool to the touch. Twitching of the lips. Return to activity engaged in prior to seizure."

The next one was a few months later, and lasted about 120 seconds. The nurse noted: "Violent shaking of entire body, staring spell, shallow, ashen, cyanotic, unresponsive drowsiness, return to activity engaged in prior to seizure, complaints of headache, maintained safe environment, contacted nurse, contacted Emergency Services, sent to emergency room via ambulance. Precipitating Factors: None to note. Had just finished programs and shower. The home called this one a Grand Mal seizure."

The doctor was wonderful with Ryan. She understood that he wasn't a candidate for a monthly heart monitor to "try" to catch a spell, because he would never have kept it on. She compared his EKG from one at birth and it showed a pattern where it slows down. She determined that it was the vesovegal reaction causing Ryan's blood pressure to drop and hence less oxygen was going to the brain, causing seizures.

continued on page 13



Behavior Scale Can Help Clinicians



Dave Richman, Ph.D., Professor of Educational Psychology and Leadership, Texas Tech University

First and foremost, I thank all of the CdLS families who participated in this study. If you were one of the 179 families that filled out a series of “endless” questionnaires about your child’s behavior problems at one of the

CdLS conferences or online, the fruits of your labor resulted in the publication of *Behavior Problems in Individuals with Cornelia de Lange Syndrome: Population Specific Validation of the Behavior Problem Inventory*.*

The Behavior Problem Inventory** is a rating scale that assesses the frequency and severity stereotypic (stereotypy, repetitive behavior, etc.), aggressive, and self-injurious behaviors. It contains a list of 51 challenging behaviors often observed in individuals with intellectual disabilities.

Clinicians and researchers use rating scales as a way to quantify how big of a problem challenging behaviors are from the perspective of a parent, teacher, or other caregiver. This is a very important task because many individuals with CdLS have difficulties self-reporting these behaviors, and caregivers spend the most time with the individual and know the most about his/her challenging behaviors. Therefore, it is very important that we use a rating scale that we are quite confident will produce an accurate representation of the challenging behaviors from the caregivers’ perspective.

The BPI-01 has been shown to provide an accurate representation of challenging behavior for individuals with intellectual disabilities, but it has not been shown to produce accurate results for individuals with CdLS (along with all the other rating scales available to clinicians and researchers). The purpose of the study is to validate the BPI-01 specifically for individuals with CdLS. Without going into the technical aspects of the statistical analyses, I am happy to report that the BPI-01 is appropriate to use for measuring challenging behaviors exhibited by individuals with CdLS.

One critique of the BPI-01 is that it may not list all challenging behaviors exhibited by an individual with CdLS but this limitation is handled via an “other” line that allows the reporter to write down any challenging behavior and rate it on frequency and severity.

The big advantage of knowing that the BPI-01 is appropriate to use with CdLS is that it provides assurance to clinicians that the BPI-01 is a good measure of challenging behavior to use with individuals with CdLS. It also sets the occasion for CdLS researchers to incorporate the BPI-01 into their studies so they can more easily compare and aggregate results on challenging behavior in CdLS across studies. These two advantages may result in better clinical assessment and monitoring of treatments for challenging behavior, and help us more clearly study how these challenging behaviors develop and maybe, eventually prevent some forms of these behaviors in CdLS.

In closing, we thank the CdLS Foundation for providing a grant to partially support the cost of completing this study. I am truly amazed each and every time I interact with the CdLS Foundation staff. I wish each genetic disorder foundation was equally well organized and careful with how they continually attempt to help those with CdLS in need, along with funding research that will hopefully help individuals with CdLS live a better life.

**Rojahn, Barnard-Brak, Richman, Dotson, Medioros, Wei, & Abby, 2013.*

***The Behavior Problem Inventory-01 (BPI-01; Rojahn et al. 2001)*



A Closer Look at Behavior: Aiden's Story

We want to thank Aiden's mom, Jackie, for sharing Aiden's story with us.



Aiden's aggression and defiant behavior increased after starting Public school (pre-K), and seemed to worsen as he got older. Some behaviors that Aiden has exhibited are: hitting, running away from adults, throwing himself on the floor, laughing uncontrollably at inappropriate times, slamming doors, running around the room/classroom, taking off his clothes, throwing toys, shoes and food, locking and unlocking doors, putting non edible objects in mouth, jumping off furniture, grinding teeth, and becoming frustrated quickly and easily with puzzles, dressing, etc.

Aiden started Occupational Therapy (OT) at age one, and sensory overload was mentioned at that time. The therapist performed and taught activities to help decrease sensitivity to outside stimuli, but that was as far as the teaching went. I did not relate his abnormal behaviors as he got older with Sensory Perception Disorder (SPD). He started private outpatient OT again this summer and they again mentioned how he craves sensory input, such as putting feet in a bowl of dried corn, swinging and playing with play dough.

As Aiden grew, his behavior at school and home worsened. I searched aggressive behavior, autistic behaviors, etc. and started reading about SPD. "Sensory Processing Disorder (SPD) is a condition that exists when sensory signals don't get organized into appropriate responses." (SPDfoundation.net)

My knowledge prior to this was that Aiden liked to swing and run around barefoot because it felt good. In reality, almost every behavior was sensory input seeking, such as: sucking his thumb (or fingers), stomping his feet when he walked/ran, giving bear hugs, liking tight clothing, falling on the floor intentionally, enjoying jumping (especially

on a trampoline), hitting other children and not crying when being hit or falling down and having an injury (high pain tolerance).

One thing that really got my attention was Aiden's hitting. I was thinking he was hitting to be mean or mischievous, when in actuality he hits because it gives him strong sensory input to his hands. When he is hit by another child it most likely feels like craved input, therefore hitting feels good to do and, in his mind, must feel good to others. When he goes on a hitting spree, I know it's time for "Heavy Work."

Some "Heavy Work Activities" that work for Aiden are joint compression, spine compression (pushing on shoulders while seated), squeezing in between two pillows, deep massage to feet, tight bear hugs, tight clothing (like compression shirts or footy pajamas), weighted vest (especially at school), pulling rubber band, rolling on large ball or rolling large ball over body.

I was told by his Occupational Therapist that sensory input activities should calm the nervous system for approximately two hours at a time, and Aiden would benefit most from a "Sensory Diet." We have not mastered this, but it has significantly changed the way we react to Aiden's behavior, and lets us inside his head just a little bit. We are learning every day and are open to any input from parents that have older children and interventions that have worked for them.

Learning about SPD honestly changed our lives. I have tools that I can use to help calm Aiden's nervous system and help him function more easily in life. This is not a magic tool, and it does not work exactly the same every time. We have good days and bad, but the good definitely outweighs the bad.



Super Siblings: Jillian and Delaney

Delaney is the third of four children. Her older sisters, Jillian and Emily were both competitive swimmers from a young age and they decided that Delaney would follow in their footsteps. Jillian taught Delaney water safety in the backyard pool and as a result had Delaney swimming before she was talking. It seemed that Delaney, diagnosed with a mild form of CdLS, was a natural. She was at home in the water and loved the personal attention from her idolized big sister.



When Delaney turned five, Jillian was the head coach of the neighborhood summer recreational swim team - The Mallards. Jillian, who was 18 at the time, signed Delaney up despite parents, Lori and John's, concerns. She kept a close eye on Delaney at practice where for the first time Delaney seemed completely normal when with age-group peers. Swimming was non-verbal so speech and language challenges were not an impediment. "Coach Jill," as Delaney called her at practice, was purposeful in giving physical demonstrations of all instructions that benefitted all of the five & six year olds in the pool. As a result, Delaney made new friends on the team and became a swimmer.

Every summer since then, Delaney has been a Mallard under big sister/Coach Jill with full participation and a close set of summer swim friends. She developed the muscle memory to swim freestyle, breaststroke, backstroke and even the difficult butterfly. As her swim group got older, others became quite fast and serious about competition. Delaney, however, remained the slow and steady swimmer. What she didn't have for speed she made up for with charm. She quickly became a crowd favorite and was known as "the girl who smiles when she races." Every time Delaney came up for a breath, she was smiling ear to ear. On occasion she would pause mid-flip turn to pose for a photo or wave at the crowd that was cheering her on. She modeled sportsmanship and love of

swimming for her teammates. Coach Jill was with her at the starting block at the beginning of each race, cheering her on up and down the pool.

Jillian was proud of Delaney's accomplishments and felt that she deserved more than rainbow ribbons for her efforts. In the spring of 2013, Jillian persuaded parents Lori and John to sign Delaney up for a Special Olympics swim team. They were hesitant to do so because the nearest Special Olympics aquatics team that included children and young adults was in a community 40 miles away. "I will drive her," replied Jillian, leaving her parents no way to opt out. Delaney joined the "Elk Grove Stars" - a move that resulted in life changing experiences for the whole family. Every Sunday the family made the trek to Elk Grove for swim practice in preparation for the regional meet.

Delaney swam her first race at the Special Olympics Regional swim meet in Roseville, CA and blew her coaches and family away. She swam the Individual Medley (IM) and her slow, steady, perfect stroke won her the gold medal. Delaney stepped down from the winner's podium and asked, "Do I keep it?" When her big sister said yes she replied with, "Can I get another one?" To which the answer was also yes. Three golds later, thanks to her big sister's support and instruction, Delaney qualified for the Special Olympics State meet in July.

The State Meet invitation was an honor that mom, Lori, was quick to decline. The meet required participating athletes to spend the weekend in the college dorms with their Special Olympics coaches only - no parents. Delaney had never slept away from home and Lori was concerned that speech and language limitations would prevent Delaney for advocating for herself when away from the family members who understand her. In stepped big sister Jillian with an assertive, "She's going." Jillian's advocacy paid off. Delaney spent an entire weekend at the event and grew notably more confident as a result. She won two gold medals as her family was in the stands cheering her on. Delaney made friends, ate in a dorm and attended her first dance party. The Special Olympics coaches told the story of Delaney demanding more whipped cream on her dessert in the cafeteria line, teaching the family that Delaney can advocate for herself when she really cares. Delaney's confidence grew from the experience and she is already looking forward to next year - all due to big sister, best friend, advocate and "coach" Jillian.



Mailbags

- Reagan -

On July 22, 2013 my life changed forever. My sweet Reagan was born weighing a tiny 4 pounds 15 ounces had a head full of jet black hair and was also born with no hands, which came as a complete surprise because it was never detected in any of my ultrasounds. From there my baby and I were transported to Winnie Palmer Hospital in Orlando, FL, where a team of doctors and specialist were waiting for us. There they diagnosed Reagan with "CdLS" Cornelia de Lange Syndrome.



My husband and I had no idea what it was or what it meant so the doctors explained. Reagan would have to have surgery right away because he could not protect his airway and fluid was leaking into his lungs so therefore a g-tube would be placed into his stomach and he would be fed through it.

After hearing all of this I sat there quietly, scared to death. I looked at my two-day-old baby, smiled and said, "he can see, he can hear, his heart is healthy and God put him on this earth for a very special purpose." I am so blessed to call this awesome little boy my son and I cannot wait to help and guide him as much as I can. All mothers are strong, but mothers of children with CdLS are the strongest. And God sure did pick the right mommy for the job.

Baby Reagan is now almost two months old and is doing amazing; he's surrounded with nothing but love and support. Although there is no cure for CdLS, it's my duty to inform everyone about CdLS.

-Reagan's mom, Julicia, FL

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

- Jasmine -

Jasmine is 22 years old and loves to dance and sing, even though she doesn't speak. She can sing gospel and anything else she wants to in her own way, but you don't always know what she's singing. She also likes watching videos, going for a ride and being outside. She likes to drop to the floor and sit there laughing while you try to talk her up. She also likes when her sister and brother, mother and uncle come over. I wouldn't trade a second for any and all of my time that she has been in my life. I'm so thankful God chose me.

Jasmine's grandmother, Patricia, IL



WELCOME NEW FAMILIES

Arizona

Christina and Aaron and daughter Anissa, born March 16, 2013

Pennsylvania

Maryann and Manny and daughter Tia, born November 21, 2013

Florida

Yolanda and daughter Aleah, born August 12, 2004

New Hampshire

Addie and Joy and son Carter, born April 22, 2010

Georgia

Monique and Rafael and daughter Angelika, born November 19, 2013

New Jersey

Lisa and Mike and daughter Lauren, born March 27, 2013

Kansas

Kayla and Phillip and son Jaystin, born June 10, 2010

Texas

Teresa and Dedrick and son Aiden, born October 7, 2009

Missouri

Haylee and Josh and daughter Charlee, born May 27, 2011

Washington

Jerry and daughter Isabelle, born September 13, 2012



Annual One Love, One Heart

“One Love refers to the universal love and respect expressed by all people for all people.” This is the theme for the “One Love, One Heart” 5K in Decatur, GA, that took place on December 7. The cold winter temperatures (it was 30 degrees warmer the day prior!) did not discourage more than 200 runners and walkers from enjoying a morning run. Seven CdLS families joined the activities helping raise awareness and funds for the Foundation.



A 5K Walk/Run
benefitting the
CdLS Foundation



Guys Playing Games Support Team CdLS

Written by Pat Lyons

For the past 14 years, a great group of guys have been getting together for what many of us classify as one of the best days of the year. We wake up early and make our way to Hingham, MA, for “The Pentathlon,” where 32 grown men, split into two-man teams, play wiffle ball, horseshoes, darts, ping pong, and eight-ball. We have a blast and we have plenty of hot dogs, burgers, beer, and laughs. This year was a little bit more special.



My good friend and host of the event, Bryan, sent an email to the crew, saying that one thing has been missing from the Pentathlon all these years, and that was “a cause.” He had gone out and made custom baseball hats for anyone who donated money to the CdLS Foundation in honor of my wife, Karen, running the ING NYC Marathon on November 3.

When you have a child with any type of special needs you always do so much to help them. I find it so comforting when someone does something for you, especially when it just totally catches you off guard. It was so nice to have my brother as my teammate this year, as he knows how hard things have been at times. They raised more than \$600 at the 14th Annual Pentathlon, putting Karen above her fundraising goal. We are so lucky to have such a great group of friends and family to support our family and the CdLS Foundation.

Where will your sneakers land in 2014?

Contact the Foundation to find your next race at [800.753.2357](tel:800.753.2357) or

TeamCdLS@CdLSusa.org



CdLS Champions Spotlight

The CdLS Foundation recognizes the importance of acknowledging CdLS Champions and provided an opportunity to honor them publicly through the “Celebrate Your Champions Yearbook.”

We thank everyone who responded to the CdLS Champions program, but would like to especially recognize Richard and Dianne Haaland, who highlighted five of their personal CdLS Champions:



Margaret & Richard Bergstrom – During Joey’s brief life, Richard and Margaret (Grandpa & Grandma) were always so supportive and loving each and every day! Thank you so much!

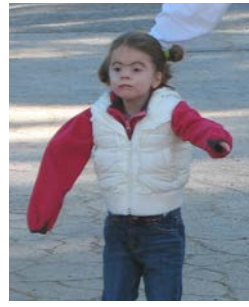


Amy Haaland – What a fantastic younger sister to Joey! Amy has always been the most tender and loving support possible!



Rich Haaland – Rich Haaland is certainly the best big brother anyone could imagine. Now many years later, he continues Joey’s legacy with his active participation with the CdLS Foundation.

Monica Haaland – Monica has been a support for Rich in his activities with the Foundation. Also, Monica coordinates a team of student volunteers from Our Lady of Mercy High School for the “One Love One Heart” 5K walk/run in Decatur, GA.



Maya Pomfret – We have followed Maya’s progress throughout our son’s encounters with her and her family. Last December, we finally had the opportunity to meet her and her family at the “One Love One Heart” race. We thought that was special, but not as much as when Maya ran up to Rich, motioned to be picked up, and then snuggled against his chest! This was a warm memory we will always cherish!

Recognize your CdLS Champion in 2014 by visiting www.CdLSusa.org/champions, or call Gail to share your story, at 800.753.2357.



2013-14 CALENDAR

April 4
Thank You Reception

April 4-5
Board of Directors Meeting

April 12
Multidisciplinary Clinic for Adolescents and Adults
Greater Baltimore Medical Center
Baltimore, MD

May 10
National Awareness Day

May 10
Northeast Foundation Sponsored Gathering
Katonah, NY

May 19
22nd Annual New England Golf Tournament
Ipswich, MA

June 25-26
Scientific & Educational Symposia
Costa Mesa, CA

June 26-29
CdLS Foundation National Family Conference
Costa Mesa, CA

June 29
de Lange Society Induction
Costa Mesa, CA



Save the date: CdLS Foundation National Family Conference: June 26-29, 2014

**CdLS Foundation National Family Conference registration opens
February 1, 2014.**

Don't miss your chance to attend the CdLS Foundation's hallmark event, taking place in Costa Mesa, CA.

Adult registration is \$320 before March 26 and \$350 after. Children under 18 are \$220 and \$235, respectively. Registration for children two and under is free, but childcare costs still apply.

The registration packet includes information on the hotel, child care and sibling programs, the event schedule and consultations. If you need a copy mailed to you, call the Foundation at 800.753.2357. You can also download it from our web site.



A limited number of **scholarships** are available for first-time families attending conference. The scholarship will cover up to two registrations.

Go green by downloading your registration packet from the CdLS Foundation's web site, www.CdLSusa.org.



Workshop topics include behavior, puberty and communication.



Meet the Staff: Brenda



Brenda Shepard joined the CdLS Foundation as Communications Coordinator in July of 2012. She received her bachelor's degree in communication at George Mason University.

“There is always a new project to work on, new people to meet and great opportunities to engage others. I enjoy putting publications together – I feel it’s like a puzzle and when all the pieces come together, it makes me very proud.”

Prior to joining the Foundation, Brenda worked as a Marketing Associate at the Connecticut Science Center, Admissions Representative for George Mason University, and held two internship positions at CNN and NBC in Washington, DC. She grew up in coastal New Hampshire.

“From a young age, I always envisioned finding a career where I could make a difference in the world. I had the opportunity to introduce Hillary Clinton in high school. I decided to pursue a degree in journalism and gained an interest in nonprofit work. I also volunteer with a nature project documenting the surroundings where I live. I’m a music junkie and having lived in the south during college, I even developed a love for country music. Sunday afternoons, you can find me jogging, watching sports or reading.”

A Closer Look at Seizures cont.

Ryan began Keppra in October 2012. We then worked with the dosage until we found the right fit for Ryan. It was given twice a day and he had it rechecked with a neurologist a year later. It was determined to keep him at the same dosage, which seems to be working (knock on wood).

Ryan can feel a seizure coming on. He begins by taking his hands and putting his fingers together, and stares at them. His skin gets pale and mottled. He then becomes unresponsive. The trigger that I’ve noticed before Ryan has a seizure is that it’s usually after a feeding and he has been moved either from his wheelchair to bed or to shower. The staff at the group home now feeds him after his shower instead of prior.

We learned things to do that help Ryan when a seizure starts to come on. The best thing I’ve found to do is to put him in bed and elevate his legs to bring his blood pressure back up (per his cardiologist). I have tried preventing the seizures a few times while visiting him. He will be playing with his blocks and then starts to do the finger thing. I try to stimulate him by taking his hands, kissing them, his arms and cheeks to try to get his attention and bring his blood pressure up. I’ve done this a few times and it has worked.

The treatment at the group home where Ryan lives and at our family home is the same. The staff monitors that he won’t choke on saliva if he is frothing at the mouth and now they elevate his legs. They also talk to him and tell him that they are there with him during the seizure. I think that really helps him because I think he would be scared not knowing what is going on.

Spread Kindness

February is kindness month, and at the CdLS Foundation, we know you all have incredibly kind hearts—we see it daily through your actions and generosity. To thank you for your kindness, during the month of February, every person who gives a donation to the Foundation receives a CdLS Kindness Coin. It’s a small way to show you how much you mean to us.





Donations from 10/1/2013-11/30/2013

Gifts that Count - In Honor/Celebration

Aiden Meyer
Mary Jenkins

Alexander DeCillis
Helene Kriegstein

Alexandra Boteler
Cristine Ponthier
Duhon & Company, LLC
Edward O'Leary
Joyce and Bernard Seary
Kenneth Charpentier
Peggy and Thomas Williams
Pistol Power, LLC
Skin Saver

Andrew Pacheco
Dorothea Barreto

Andrew Patitucci
Judith and Theodore Felix

Anya Janoski
Beth Stern

Audrey Barton
Donna and David Viland

Benny Fromhartz
Carol Galin
David Podell
Jill and Scott Gelber
Joanne Geller
Pamela Raab
Paul Granett
Rose Gelber
Zeporah and Joseph Geller

Caleb Wherry
Kathy and Andy Wood

Colm Toman
Mary Metzger

Delaney DeMaria
Vincent Berardini

Devin Miller
Melissa and Jonathan
Widdicombe

Diane Friedman
Lois Braverman
Ellen Braverman and
Robert Fishal

Elijah McGaw
Jeryl McGaw

Emma Pietrafesa
Lisa Whetstone

Ethan Walters
Penelope Keating

Francie Barber and
Craig Cheney
Mary and Jim Micallef

Gretchen Heinrich
Ellen and Daniel Smock

Harley Butler
Mary and James Fenske

Hayley Turbyfill
Carol and Glenn Zeliff

Jaclyn Fielder
Billy Williams
Janet Eastburn

Jakayla Williams
R.P. Gupta & Associates.,
M.D., P.A.

Jonathan Swanson
Judith and Ross Soltess

Kelly Noyes
Carolyn Kingman
Linda Zeller

Logan Fowler
Edward McCrossin
Sarah and Douglas Fleming

Lucas Hoen
Marjorie and Carl Paffendorf
Wendy Shuford

Mason Linehan
Chris Lienhard
Valerie and Jay Lienhard

Maya Pomfret
Ninetta Violante

Nathan Frischmeyer
Allen Stickfort
Michael Shelton

Ollie Wilson
Rosie and Arthur Jefferson

Paula Hauffe
Jacquelyn Snyder

Peyton Howland
Evangelyn Davis
Jennifer and Jerry Lindler
Marsha Corley

Riley Rissland
Anne and Wesley Johnson

Robert Brough
Barbara Brough

Ryan Elphingstone
Susan and William Elphingstone

Sam Miller
Susan and Michael Brown

Sandra Kaufman
Diane and Michael Friedman
Lois Braverman

Siena Renteria
Mary and Jim Micallef

Sydney Fasnacht
Jodi and David Fasnacht

Thomas Scruggs
Lisa Ratliff

Will Smisloff
Mark Whalen

In Memory

Allison Tryba
Jane Fischer Shebeneck and
Paul Shebeneck
Veronica and Ernest Tryba

Bill Dambrose
Angela and Wayne Wrolstad

Charles Krattenmaker
Myrna and Alan Kaplan

Claudia Tiogson
Barbara and Regis Koontz
Carolyn and Steve Hofmann
CdLS Foundation Board
and Staff

Charles Fracisco
Cindy Redding
Danielle Bennion
Dolores and Robert Schoen

Frank Brosnan
Heather Boe
Jesada Homchand
Joe Vassar
Jon Reed
Jon Tiogson
Karen and Larry Prada
Kimberly Young
Linly Kwong
Liza and Adon De Leon
Margi Stern
Marisa and Russ Chu
Mark Pedersen
Mary Horan
Michael Mori
Michelle La Grandeur
Rennett Macintyre
Roshan Shrestha
Som Sivongxay
Stacey and Paul Turvey
Susan Tiffany
Tammy Salzman
Vacaville Christian Schools
William Bassett

Emily Johnson
Lynda and Jay Steller

Frances Ern
Martha and Robert Fisher

G. Alex Black
Pam and Vince DeGeorge
Sue Ann Houser
Susan and David Fowler

George Kesting
Bob and Teddy Garagiola
UHY Advisors Tax &
Business Consultants

Genevieve Jansen
Veronica and Ernest Tryba

Harry Whitworth
Angeline Pappas and
Catherine Brown

Jack Brigham III
Jack Brigham II

James Alevizos
Angeline Pappas and
Catherine Brown

Jarvis Winslow
Tony and Karen Spivey

Jeremiah Caudillo
Barbara and James Galyean

Jessica Murphy
Roberta Murphy

Jindi Van Buren
Lynn and Kenneth Colombo

Joshua Ackles
Barbara and James DeBastiani
Barbara and Regis Koontz
Barry Pugh
Carole and Richard Chapman
CdLS Foundation Board
and Staff
Chimes Metro, Inc.

David and Patricia Boulden
David Geiszler
E. Starkey
Janet Brown
Joan Pulley
Lawrence and Linda DuBow
Lisa Synnott
Lynnette and Dean Miller
Mary Davis

Monica and Richard Haaland
Pamela Onori
Patricia Matanis
Rhea and Blair Brekke
Susan and David Fowler

Kerry Myrthel
Willing Workers of
Paradise Valley

Leonard Hoeft
Angeline Pappas and
Catherine Brown

Maria Damonte
Alda Houchin

Liddy Horsey
Karen Manning and
Thom Horsey
Mary and Robert Geis

Michael Gonella
Susan and Raymond Molinari

Mitchell Rice
Angela and Wayne Wrolstad

Nancy Cataline
Betty and William McMahon
Carol Spina
Christine Baker
Elizabeth and Richard Brooke



- Finger Lakes Football Association
- Florence Stajura
- Gwendolyn and Aaron Dell
- Jeneane and Joseph Ciancaglioni
- Joe and Mary Carnovale
- Marie Combs
- Marie Zeoli-Merkel
- Maura and Frank Cook
- Michael Zeoli
- Nancy Palmisano
- Peter and Margaret Zeoli
- Wanda and Stephen High
- Salvatore Viola
- Cheryl and Art Eng
- Deborah Walsh
- Lisa and Vincent Viola
- Sean Brisbane
- CdLS Foundation Board and Staff
- Lois Brisbane

- Sherry Millar
- Louise Crowe
- Stephanie Smith
- Crystal and Jerry Smith
- Tara Joyce
- CdLS Foundation Board and Staff
- Irene Smith
- Tom Kurth
- Helen Casey

- William Nicklow
- Angeline Pappas and Catherine Brown

On the Cover

Tayvion is our “cover boy” this issue and according to his mom, Holly, he made great progress in 2013.



“Tayvion continues to learn best when a multi-modal approach to learning is used and, of course, when music is incorporated into his learning activities,” said Holly. “He has made some wonderful gains since we began homeschooling him, thanks to his brother Tre’, and his nurse (aunt) Keisha. The best part about homeschooling has been the decrease in his self-injurious and aggressive behaviors.

We are so proud of Tayvion and the progress he has made and continues to make each day! He has grown into a beautiful, smart, stubborn little boy, with a personality and smile that can make your day brighter... despite all his struggles medically and developmentally, if he can smile then we all can too!”

We thank Holly for sharing Tayvion’s photo with the rest of the CdLS Foundation family as our first cover of 2014.

Our Deepest Sympathy

Claudia Rose Tionson
September 7, 2001 – November 2, 2013
Daughter of Nicole and Phillip Tionson
261 Willow Green Way
Vacaville, CA 95687-4301

Tara Maureen Joyce
October 29, 1978 – October 17, 2013
Daughter of Patricia and Gerry Joyce
9214 Ridge Blvd Apt 6D
Brooklyn, NY 11209-6212

Sherry Millar
February 25, 1965 – November 8, 2013
Daughter of Janelle Millar
225 Upton Pyne Dr
Brentwood, CA 94513



CdLS Foundation

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

Select a boat to be displayed at the National Family Conference:

\$25.00
Skiff (Small)

\$50.00
Sloop (Medium)

\$100.00
Schooner (Large)

(Please indicate how you wish the name to appear on the boat)

To: _____	From: _____	<input type="checkbox"/> Skiff	<input type="checkbox"/> Sloop	<input type="checkbox"/> Schooner
To: _____	From: _____	<input type="checkbox"/> Skiff	<input type="checkbox"/> Sloop	<input type="checkbox"/> Schooner
To: _____	From: _____	<input type="checkbox"/> Skiff	<input type="checkbox"/> Sloop	<input type="checkbox"/> Schooner
To: _____	From: _____	<input type="checkbox"/> Skiff	<input type="checkbox"/> Sloop	<input type="checkbox"/> Schooner

Harbor of Hope

Enclosed is my check for \$ _____

You may also dedicate your boat online at www.CdLSusa.org or by contacting us at 800.753.2357.

Charge my gift to: VISA MC AMEX

Card #: _____

Expiration: _____ Security Code: _____

Print name on card: _____

Signature: _____

Email address(es): _____

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

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

Return Service Requested

Harbor of Hope

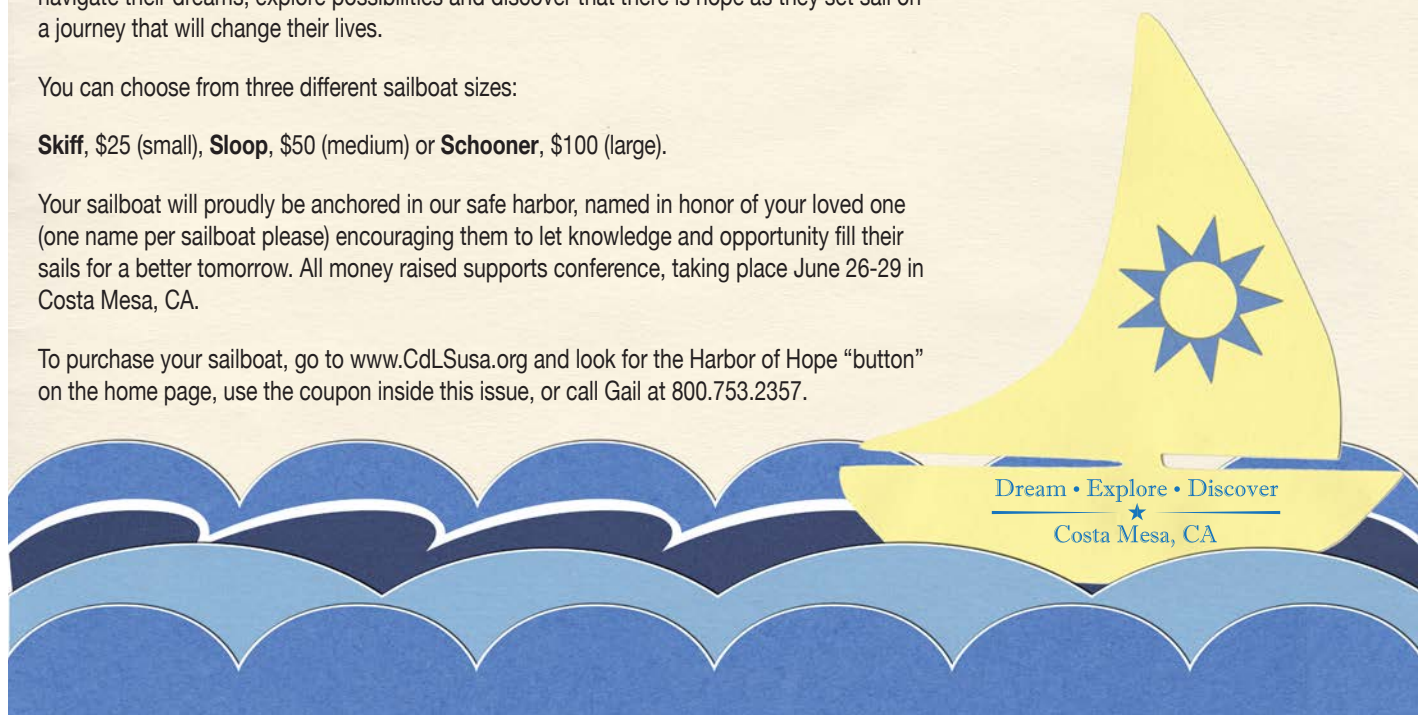
Anchor a sailboat in the CdLS "Harbor of Hope" to honor someone you love at the CdLS Foundation National Conference this summer. What better way to inspire your loved ones to navigate their dreams, explore possibilities and discover that there is hope as they set sail on a journey that will change their lives.

You can choose from three different sailboat sizes:

Skiff, \$25 (small), **Sloop**, \$50 (medium) or **Schooner**, \$100 (large).

Your sailboat will proudly be anchored in our safe harbor, named in honor of your loved one (one name per sailboat please) encouraging them to let knowledge and opportunity fill their sails for a better tomorrow. All money raised supports conference, taking place June 26-29 in Costa Mesa, CA.

To purchase your sailboat, go to www.CdLSusa.org and look for the Harbor of Hope "button" on the home page, use the coupon inside this issue, or call Gail at 800.753.2357.



Printing and distribution
supported by the American
Legion Child Welfare
Foundation Inc.