

Reaching Out

The Newsletter of the Cornelia de Lange Syndrome (CdLS) USA Foundation, Inc.  Fourth Quarter 2013

Highlights

Director's Message:

Thankful

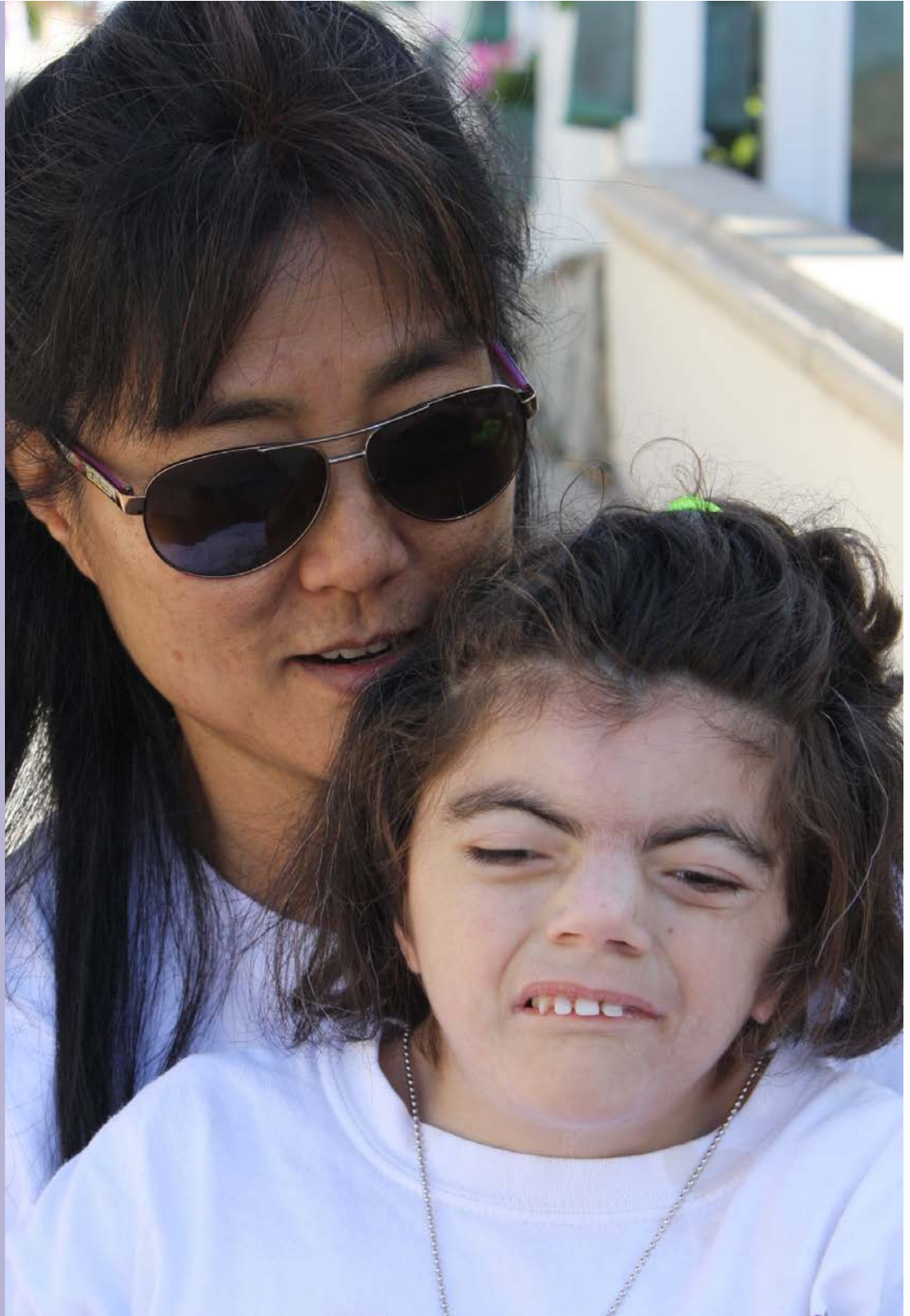
Spotlights:

Adapted Physical
Education

Orthopaedic Issues in
CdLS

Super Siblings:

Julie and Robin





Director's Message



Thankful

November is a time to be thankful for what we have, to reflect on the past and to plan for the future. So, here I go:

I am thankful for a great staff of hard-working, caring people who truly enjoy spending their days together (and bring leftover Halloween candy to the office). I am thankful for completing my first 10K at age 43, and being able to do it with people who care about CdLS. And I am thankful for a spouse who steps in when I'm traveling for work or staying late at the office.

As I look back on 2013, there are plenty of data and graphs that summarize what we've accomplished, but each number represents someone we've helped. Family service coordinators welcomed nearly 100 new families, facilitated close to 300 *Ask the Expert* questions and visited with more than 450 people at family gatherings nationwide.

Staff completed year one of a three-year strategic plan, meeting many goals along the way, including creating online learning opportunities for parents and professionals, preparing to set up an annual west coast clinic and creating two new publications—the *Educational Handbook* and *Guide to Transition*.

As we plan for the future, 2014 brings the National Family Conference in Costa Mesa, CA, and two new publications—one for parents who have lost a child with CdLS and the other summarizing best practices for transitioning children with CdLS from pediatric to adult medical providers. We're also working on a new logo, preparing to "retire" the cube that's been our trademark since 1997.

The development staff will work to expand Team CdLS—our biggest fundraiser—into new races, as well as promote the new 5K Club, aimed at shorter distance runners.

What are you thankful for, reflecting on and planning for? I'd love to hear. Email me at director@CdLSusa.org.

Marie Concklin-Malloy
Executive Director



P.S. I don't like to sit behind my desk. Put together a Team CdLS team in a 5K or 10K race next year and perhaps I'll join you at the starting line. Here I am at the finish line of the Team CdLS Manhattan Beach 10K in October.

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Never Give Up



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

Determination and perseverance are two traits that help an individual succeed in life. Both are crucial for many tasks, like learning to ride a bike without training wheels, doing well in school, getting a job, having a successful career, and raising a healthy family. In the middle of an athletic event, such as a long run, all it takes is determination to complete the miles and get

up the next hill, and perseverance in continuing to run even when faced with fatigue or daunted by the height of the hill.

While training for my relay marathon this fall, as part of my Team CdLS race in Baltimore, it occurred to me that this is true for CdLS itself. Children and adults with CdLS have both determination and perseverance in spades. This not only helps them be successful, each in their own way, but also sets CdLS apart from most other conditions.

Think about the baby with CdLS missing some fingers. He or she will work over and over until he or she can pick up a toy with the digits he or she has, exhibiting determination to master the task and perseverance until the toy is obtained. Consider the school-aged child with CdLS learning to use an iPhone or iPad. Think of the teenager who lets go and walks with no walker at the age of 16 years, or the young adult who finally uses the telephone correctly to make a call for the first time. These are all true scenarios for individuals with CdLS, each having determination to succeed and persevere until it happens. I can't think of more important qualities for success than these, and they should be kept in mind for caretakers as children with CdLS age.

Individuals with CdLS never give up. They continue to learn throughout life and achieve their goals. Both determination and perseverance provide a huge advantage for anyone with CdLS, compared to other genetic disorders, and allow the rest of us to maintain hope for them to master just about anything.

On the Cover

Nicole is a beautiful 20 year old "girl", well young lady is actually more correct. She is non-stop in perpetual motion. Nicole is constantly on the move, climbing all over the house (really, she can scale anything). She can often be found at the piano or other various keyboards scattered around the house. She has been banging out "tunes" that give her a tremendous amount of satisfaction. In any one day, Nicole will provide a great deal of entertainment for herself and family, laugh a bunch, and cry too. It is all part of "our day" that begins very early... like 4 to 5 a.m. – Karen, Nicole's mom (pictured on cover)

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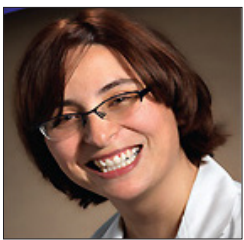
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Orthopaedic Issues in CdLS



*Carmen P. Pichard-Encina, M.D.,
Assistant Professor, Johns Hopkins
Orthopaedic and Spine Surgery,
Good Samaritan Hospital Baltimore,
MD, and member of the Foundation's
Clinical Advisory Board*

Musculoskeletal problems are common in people with CdLS.

Some problems are obvious at birth such as missing hands or fingers. The upper extremities are usually more involved than the lower extremities. Upper extremities are involved in almost one third of cases. Other problems develop with age. One study looked at twenty individuals with CdLS ages 13-50 and found that 90 percent had some degree of decreased bone density, which is commonly associated with older aged individuals. Many individuals also appeared older than their actual age. Premature aging is a topic that is being studied at this time.

The most common orthopaedic findings in CdLS are as follows:

Hand abnormalities: The differences include small hands (in 90 percent of cases), incurved and short fifth fingers (in 74 percent of cases), and short thumb placed low in the hand. More severe findings include missing fingers and hands. Surgery is rarely helpful when digits or hands are missing. Prostheses are also rarely useful. In general individuals with CdLS will find ways to use their residual limbs efficiently. Occupational therapy might be useful to help kids find tools or adaptations to make daily activities easier.

Forearms: Some individuals with CdLS have radio-ulna synostosis which is the fusion of the radius and ulna bones. The use of physical therapy on somebody with a synostosis could cause injury. Therefore it is recommended that a person diagnosed with CdLS have an x-ray of his or her forearms.

Stiff elbows: A common finding in CdLS is congenitally dislocated radial heads, simply said, the two bones that make the elbow hinge do not fit perfectly. This is found in 64 percent of patients. This can cause the elbow to be stiff and in most cases it will not extend all the way. This does not require surgery and stretching by a physical therapist

will not help. Recent studies of activities of daily living such as using a fork, knife and cell phone have shown that you do not need the entire arc of elbow motion; you can lack 30 degrees of extension and still do all of your functional activities. In other words, a stiff elbow should not stop individuals with CdLS from using their elbows fully. It is possible to detect a dislocated radial head on a physical exam but a radiograph is the gold standard for a definitive diagnosis.

Hip problems: Congenital dysplasia of the hip, or an abnormal hip ball and socket relationship at birth has been documented as well in up to 10 percent of individuals with CdLS. Treatment will depend on whether one or two hips are involved, whether the hip is subluxed (slightly out of socket) or dislocated (completely out of socket) and whether the hip can be manipulated back into the socket and maintained there with a cast or not. Depending on the various presentations, different surgical reconstructions are available.

Feet abnormalities: Individuals with CdLS might have small feet (90 percent of cases), webbing between the 2nd and 3rd toes (80 percent of cases) or club feet at birth. Small feet do not interfere with walking and need no intervention. Children born with club feet should be initially treated with the Ponseti method. This is a method of casting where children get casted weekly and the cast rearranges the position of the bones in their feet to realign their feet. As in individuals without CdLS, this is the gold standard, followed by surgery only if casting fails. With aging, patients may develop bunions and tight Achilles tendons.

At the CdLS Multidisciplinary Clinic, I've seen a very high proportion of bunions. Just as with bunions in the general population, they should be initially treated with wide toe shoes for comfort. Surgery should only be considered if they become painful or interfere with walking. We have also seen tight Achilles tendons, or tight heel cords, very frequently. The initial treatment once again should be stretching exercises which parents can ideally do daily. Surgery for release of the tendon is again reserved for failure of stretching exercises. You can tell a heel cord is tight if you cannot bend the foot to get it to a flat position, sometimes children with tight heel cords will walk on their toes and their feet are "stuck" pointing down. Despite all these possible findings, the majority of individuals with CdLS will walk independently. Often later than their



peers but they will be ambulatory by adulthood.

Treatment for these orthopaedic manifestations of CdLS focuses mainly on maximizing function and preventing deterioration of such function. Physical therapy and occupational therapy can help patients achieve good functional abilities whether they have short, missing or malformed limbs. Stretching and ambulation can help maintain good muscle tone, good flexibility and range of motion. Weight bearing exercises also prevent premature bone loss in the general population and should be recommended for patients with CdLS until we get a better understanding of the pathology of their early bone loss.

Ask the Expert Q&A

Q: My son, who is age 11, often sits on the floor cross legged. His physical therapists want to work on discouraging him from doing this as they believe it may cause genu valgus. He sits like this all the time and I am wondering about this condition and if it is related to CdLS.

A: Children (and adults, too) will sit in the most comfortable position when on the floor for long periods of time. Comfort is dictated by the anatomy of their lower extremities, especially hip range of motion and the lower extremity rotational profile. In the past there has been concern that sitting postures, especially “W sitting” caused or exacerbated intoeing. Now pediatric orthopaedic surgeons believe that sitting positions do not affect changes in anatomy, but just result from them. I do not think your son will develop any problems because of his sitting position. It is not possible to be absolutely 100 percent certain without seeing him, so you may wish to have him evaluated by a pediatric orthopaedic surgeon.

A Closer Look at Orthopaedic Issues

Shelby was diagnosed with CdLS in 1997 at the age of five. She is now 20 years old and has grown into a fine young lady. Along the way we have had many obstacles to overcome. Thank goodness they have not happened all at once. One issue in particular is Shelby’s orthopaedic needs.



Shelby did have issues, once she began to walk, at 18 months with tiptoeing. This continued for several years until we took her to an orthopaedic specialist in Evansville, IN. He fit her for some orthotics that she was to wear at all times during the day whenever possible. She was in these several months when he approached us and said that he thought she probably needed surgery to fully correct the problem. He then recommended we see some orthopaedic specialists in St. Louis, MO, at the Shriner’s Hospital.

Within a couple of months we had the paperwork in and a surgery date set. Shelby did very well throughout the surgery. Post surgery she learned to walk in full walking casts all the way up to her mid-thighs. She didn’t want anything to do with the crutches or walker that was available to her. Within about six weeks time she had gone from full casts to half casts. She did excellent. There were some exercises involved but those were all shown to us by professional staff at the hospital.

As of today many years later, we have had no other orthopaedic issues. Shelby is 100 percent mobile and walks, runs, bikes, dances, and even swims.



Adapted Physical Education



Geir Rosvik, Adapted Physical Education Specialist, Seattle, WA, Public Schools and dad to Iselen

Students with special needs are required to receive at least the same amount of Physical Education (PE) as students who are typically developing. Federal law defines PE as the development of physical and

motor fitness, fundamental motor skills and patterns, and skills in aquatics, dance and individual and group sports (including intramural and lifetime sports).

Some students with disabilities are able to participate in regular PE activities. Adapted Physical Education (APE) is PE that is customized and modified to address the needs of the individual.

In January 2013, the U.S. Department of Education Office for Civil Rights determined that schools need to offer equal access to extracurricular activities for students with disabilities. Extracurricular activities include club, varsity, and intramural sports programs.

The following are core principles that should be applied when providing APE to students with disabilities.



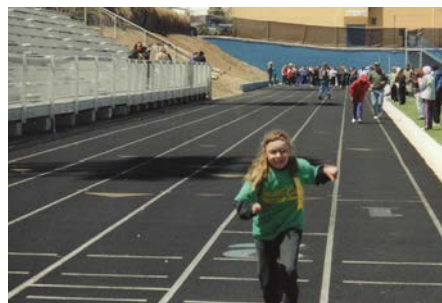
Safety

When developing a plan, teachers and other staff need to determine students' abilities and consideration should be given to things such as range of motion, weight bearing, and sensory issues. An example for a student with limb differences might be lowering an activity to the floor, using assistive devices for the affected limbs or designing

an activity that uses the unaffected limbs. Teachers should adapt activities to meet the needs of the student, rather than expect the student to adapt to the activity.

Placement

Adapted PE services should be provided in the least restrictive environment possible. According to Dr. Ronald W. Davis from Texas Women's University, "Laws requiring integration of students with disabilities could not be clearer; students with disabilities should have meaningful participation, equal opportunities and inclusion with students without disabilities to the maximum extent possible." I work to develop peer partners in the classes I teach. Peer partners are students without disabilities who work with the student with a disability to facilitate that student's participation and inclusion.



IEP

APE can be included in a student's Individual Education Plan (IEP). Sometimes parents/guardians need to advocate for their sons or daughters to have APE on their IEPs.

Classroom structure

Universal design of the classroom meets the needs of a broad spectrum of learners. Examples of universal design are a consistent structure, visual schedules, attention to transition, grab bars on the walls, three dimensional cues, class wide peer partners, the use of sign language and using auditory, visual, gestural and physical prompts. I try to provide a variety of activities using multiple strategies when I work with students.

Adaptations and modifications

The environment may need to be changed to meet the needs of the individual student or students. It is important to determine what activities the student likes or desires to do. Activities should be broken down into tasks and



skills. The teacher needs to determine what the student is able to do, if the activity needs to be changed, what equipment is required and what supports are necessary. Some examples of adaptations are short, simplified rules, shorter or longer playing time, rule changes, increased rest time and slowing down the game, lowering the target, increasing the goal size, making the field smaller and removing obstacles. Games may be played sitting or lying down. The equipment may be smaller or larger, a different color, varying textures, lighter or softer. Balls may be suspended from stationary objects or partially deflated.

Activities

A variety of organizations provide recreational opportunities for people with special needs. Outdoorsforall is an organization in Seattle. There are others throughout the country. A good source for adapted PE information is: www.pecentral.org/adapted/adaptedsites.html.



Lifetime activities

Our family enjoys recreational activities with Iselen, our daughter who has CdLS. These activities include, biking, swimming, hiking, dancing and walking the dog. Our family focus is on improving her lifetime fitness skills that will improve her overall health.

As an Adapted PE Specialist, I believe we are limited mostly by our lack of imagination and creativity regarding participation by students with disabilities in PE and extracurricular activities. We as parents and caregivers can make a difference in our sons' and daughters' lives by offering them opportunities to participate in recreational activities both in school and in the community.

A Closer Look at Recreation

Peter is now 18 years old and in his senior year of high school. Peter has played many sports over the years through youth sports, school sports and Special Olympics but golf is the sport that has come to the top for him. Both my husband and I golf, so we began taking Peter with us to the course and put him in some lessons. He enjoyed golf, especially seeing how far he could hit balls, but struggled staying focused for more than a few holes at a time on the course. At age 15 he made the decision that he wanted to go out for the high school golf team. He practiced, even in the snow, and he began watching golf on television. He is a very visual learner and seems to be able to emulate what he sees. His freshman year he made the JV team at school and played in several tournaments.



The past two years he has played on the varsity team and expects this coming season to play again. During the golf season the team travels all over eastern Washington to tournaments, even some overnights. The experience has been tremendous for Peter in many ways. His confidence has grown in leaps and bounds. Peter tested out of speech therapy services at school after the second golf season which we attribute to the experience of participating in golf. His focus and attention are greatly improved. It is the perfect sport for Peter because it is an individual sport physically, yet very social, which he is. He does like people watching him and he is very proud of his plaques, his varsity letter and pins. While a great confidence builder and learning experience for Pete, it has been a "letting go" experience for us as parents. We have had to let him make his way out there.

Peter is well known at our local golf course and has applied to work there next season. On his own he calls friends, fellow team members, coaches and neighbors to make golf dates. He makes his own tee times at the courses and gets himself out of bed at 5 a.m. to play during the summer. Peter joined the Men's club at a local course and has played a few tournaments with his Dad. He is now routinely scoring better (low to mid 80's) than his Dad and has a lower handicap which is a source of lively discussion and laughter. We truly enjoy playing together.



Mailbags

– Jason –

Jason is our only child and is 24 years old. We found out Jason had CdLS when he was around age six. At age nine, he had surgery for malrotation of the intestines. He has several other health



issues that we are not sure are common. Jason gets bad migraine headaches; he has an ulcer in the lining of his intestines, malabsorption and has osteoporosis. He takes medication and high doses of vitamins for this.

Jason goes to a day program four days a week and will be changing to two new day programs. At one he will learn life skills and go on trips, the other he will volunteer in the community. Jason and his friend Ethan belong to the Rotary Club in Colorado. It is a Rotary Club to be made up of adults with disabilities. They meet once a month at a restaurant with a guest speaker, and go out in the community several times a year to volunteer.

In June, Jason went on a ten-day trip, without his parents, to Israel, called “Israel Free Spirit Birthright.” The group was made up of all young adults with disabilities. They traveled all over the country and Jason had his Bar Mitzvah. It was a wonderful experience for him and he made many friends on the trip and are keeping in touch through Facebook.

Having a child with a disability has been a challenge and a joy. Jason hopes to live on his own someday, with peers his own age and an adult to help him with things he is not able to do, such as cooking, medical appointments, driving, etc. We hope Jason will live on his own and grow to be a productive, happy and healthy adult. He is the love of our lives. We are very proud of him and the young man he has become.

-Joyce and Robert K., Aurora, CO

– Levi –

Our son and daughter-in-law were blessed with three boys, Adam, Tyler and Levi. Soon after Levi’s birth, he was diagnosed with CdLS. Not many, including ourselves, knew anything about it. Doctors suggested his life could reach only 12 or 15 years. He was very small boned, one arm is slightly shorter, and doctors said his little muscles were too tight for their length. He had, and



still has some trouble making his words distinct, however, most of the time we can understand him.

Levi became one of the most popular kids in the community, and in school. When he attended his first day of middle school, it is reported that he went to a young lady in his class and took her hand. This started a caring, sharing relationship that lasted through high school. She became his “bud.” This young lady and many other students, teachers, and parents consider Levi their friend.

We want to recognize the love and consideration of his “bud,” teachers and community. Central High School has a “promenade” where they recognize the seniors each year. The ladies wear their beautiful gowns and the guys wear their tuxedos. The young lady insisted that she would escort Levi (or vice-versa). For one person with CdLS, it was CSDL (Caring Students Displaying Love).

Among all the outstanding academic, sports players, and most likely; Levi was voted “king” of the prom. His dad practiced dancing with him. Levi was so proud of his crown, sash and medallion. He insisted on wearing it the next day at a restaurant. The young lady and many others say Levi is still their bud.

When I hear how some young people are “self centered,” I say, not here. When Levi was announced to go on stage to receive his diploma, he received the loudest and longest applause.

Written for his family,
- G-pa and G-ma, Fowler, IL



Super Siblings: Julie and Robin

My wife, Glenda, and I are the proud parents of two daughters, Robin and Julie, both born with CdLS. Robin, now 37, is non-verbal, and legally deaf/blind. She teams up with me to create Wind Dancers table tops and canvas paints for people's homes. Julie just turned 39.

It all started with Robin scribbling on an empty pizza box. Robin draws the lines and circles however she wants, then I paint between the lines apply three coats of polyurethane, attach a hook and chain to the "dancer", hang it on the deck or tree limb and let the wind do the rest.



Robin and I took two to our neighbor and her sister. This has lead to the sale of six, and others are for sale in a local store. Robin's crowning work has been the three, 36x24 canvases that we have been commissioned to paint for a couple's home in Dallas.

Julie likes to sit and strum the guitar with me. In addition, she does a great deal of humming her own songs. The humming sounds repetitious to us, but then again, most songs are also very repetitious.

She and Robin both go into the community with their friend and caregiver, Karen, Monday through Friday from 9 a.m. to 7 p.m. They work at two food banks, filling orders and stocking shelves and boxes, as well as interacting with the other workers and clients. Karen thinks out of the box with them, and they go and do all kinds of things.

One day they went into a shoe store for a change of scene. Once inside, a clerk was opening boxes of shoes and putting them out. Karen asked the manager if she and the girls could do the same. After the manager was assured Karen and the girls were not asking for a paid job, rather just an activity, he agreed. Julie had a ball taking shoes out of boxes and putting others here and there, she was laughing most of the time.

Submit your Mailbag or Super Siblings Story!

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

WELCOME NEW FAMILIES

Arizona

Tiffany and Kevin and son Troy,
born September 25, 2011

Hawaii

Laurie and son Bucky,
born May 5, 1999

Connecticut

Ashley and Michael and
daughter Lindsey,
born November 20, 2011

Kentucky

Jing and Jack and son Caleb,
born June 26, 2009

Florida

Julicia and son Reagan,
born July 22, 2013

Maine

Shannon and Zach and daughter
Luna, born August 11, 2013

Latasha and son Jason,
born June 5, 2013

Missouri

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

Cari and son Koby,
born June 28, 2013

South Carolina

Mona and Dave and son Tommy,
born 2007

Georgia

Jennifer and daughter Ellie,
born July 27, 2010

Texas

Ginny and Sean and daughter
Bella, born July 8, 2013



2013 Walk for Will and Hope

The Kurth family hosted their annual Walk for Will and Hope, a fun two mile walk at Stony Creek Metropark in Shelby Township, MI, October 13, in honor of their children with CdLS. Joel and Maureen have raised more than \$17,000 in the past few years. This year, the event brought in 126 walkers including six CdLS families, their largest turnout ever. Congratulations to the Kurth family. Joel ran the ING NYC Marathon with six other Team CdLS runners that day to cross the finish line for people with CdLS.



A Big Thanks for Awesome Events

Lapel Village Fair

Lapel, IN

Scott and Tammy Hersberger

Maddy's Run

Salt Lake City, UT

Lynn Kerr and Hunter Jackson

4th Annual Brew & BBQ

Atlanta, GA

Fran Rissland & Suzanne Musial

Arzillo Softball Tournament

Virginia Beach, VA

Julianne Arzillo & Arzillo Industries!

Sarah's Bowl

Batesville, IN

Anne Suttman

25th Annual Missouri Golf Tournament

A full field of golfers and their families enjoyed beautiful weather at the 25th Annual Missouri Golf Tournament on September 16 at Pevly Farms Golf Club in St. Louis, MO. A total of \$49,508 was raised. The Foundation's executive director, Marie Malloy, attended the event in its milestone year. Golfers also received a 25th Annual Missouri Golf Tournament commemorative golf shirt.



The CdLS Foundation would like to thank our 2013 Tournament Sponsors: Madison County Wood Products, PLM, Crown Machinery, Gallagher Benefit Services, Inc., Weiss Attorneys at Law, UHY LLP Certified Public Accountants, The Daniel and Henry Co., Montgomery Bank, Midland Davis Corporation, Pennsylvania Lumbermens Mutual Insurance Company, Magnum Fasteners, PECO Pallet, Disaster Relief & Innovative Protection Systems LLC, Hampton Kiwanis, Cass Commercial Bank, and Barbara and Doug Gaines.

Thanks to Bryan Kesting, Jim Kesting, Jim Morton, and Steve Snodsmith!





Donor Spotlight

Every bottle counts



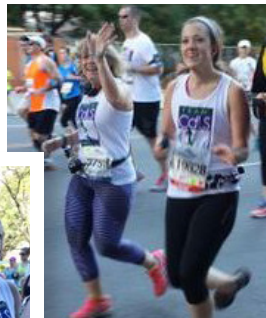
Cathy and her daughter, Jenna have been collecting recyclable items to raise money for the CdLS Foundation.

“We collect all the time, but we have a big collection after our homecoming game.”

They currently have raised \$340 dollars.

“My daughter Jenna is working with the Parent Teacher Student Association (PTSA) and the Bridges Program at the High School where Will (who has CdLS) attended,” said Cathy. “It’s become a community-wide thing.”

Both Cathy and Jenna also ran in the Chicago Marathon as part of Team CdLS.



2013-14 CALENDAR

December 7
One Love, One Heart
5K Run/Walk
Decatur, GA

April 12, 2014
Multidisciplinary Clinic for
Adolescents and Adults
Greater Baltimore
Medical Center
Baltimore, MD

May 19, 2014
22nd Annual New England Golf
Tournament
Ipswich Country Club
Ipswich, MA

April 4, 2014
Thank You Reception
Dallas, TX

April 4-5, 2014
Board of Directors Meeting
Dallas, TX

May 19, 2014
22nd Annual New England
Golf Tournament
Ipswich, MA

June 25-26 2014
Scientific & Educational
Symposia

June 26-29, 2014
CdLS Foundation National
Family Conference
Costa Mesa, CA

June 29, 2014
de Lange Society Induction
Costa Mesa, CA

¡Buscamos voluntarios!

Nos gustaría empezar a ofrecer un boletín de noticias trimestral traducido para nuestras familias hispanohablantes y buscamos voluntarios que estén dispuestos a traducir su contenido. Si le interesa ayudarnos a hacer llegar información valiosa a las familias hispanohablantes de todo el país, email Deirdre en familysupport@CdLSusa.org.

Además, muchas familias hispanohablantes se ponen en contacto con nosotros para comunicarnos que les gustaría entablar relación con otras familias. Si ser un “Coordinador hispanohablante” le parece una buena manera de ofrecerse como voluntario para nosotros, email Deirdre en familysupport@CdLSusa.org.

Al convertirse en un recurso para la Fundación CdLS y para otras familias en cualquiera de estas capacidades, usted ayudará a transmitir información valiosa, recursos y tranquilidad a las familias que buscan respuestas a sus dudas.



Meet the Staff: Michelle



Michelle Kiely joined the CdLS Foundation as a family service coordinator in July. She received her bachelor's degree in human development and family studies, and her master's in social work, at the University of Connecticut. Her desire to help people started at a

young age when she would volunteer with her mom at the ALS (Amyotrophic lateral sclerosis) Association. She turned this desire into a career and has interned at the Department of Children and Families, West Hartford Public Schools, and Vitas Innovative Hospice Care before joining the CdLS Foundation.

"I feel so fortunate to love my job. Every day I learn something new and speak with such inspiring families. I couldn't imagine being anywhere else. My interest in the medical field and passion for helping people make the CdLS Foundation a perfect fit for me."

Michelle has attended family gatherings in Louisiana and Wisconsin, and has enjoyed the opportunity to meet with families in person. You can contact Michelle with any questions or comments at familyservice@CdLSusa.org.

Our Mission

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

New CdLS Research: Cheek swab finds gene changes that blood test does not

Currently, there are five known genes that cause Cornelia de Lange Syndrome (CdLS) when mutated: *NIPBL*, *SMC1A*, *SMC3*, *RAD21* and *HDAC8*. Changes in these genes are found in approximately 65 percent of individuals with CdLS, with the majority being caused by mutations in *NIPBL*.

A study by Dr. Raoul Hennekam in the Netherlands* found that mosaicism for *NIPBL* mutations is found in up to 30 percent of individuals with CdLS whose blood tested negative for the mutation. Mosaicism means that an individual has a change in a gene that is present in some, but not all, of the cells in the body.

If an individual is mosaic for a change in *NIPBL*, we may not be able to identify it by testing blood; instead, we need to test cells from other tissues—the easiest being from the inside of the cheek, called buccal cells. These cells are collected by swabbing the inner cheek with a Q-tip like brush.

Researchers at The Center for CdLS and Related Diagnoses at the Children's Hospital of Philadelphia (CHOP) are interested in understanding the frequency of *NIPBL* mosaicism in individuals with CdLS who have had normal testing of their blood. For individuals already enrolled in the CHOP research study who had negative testing for the CdLS genes, a buccal swab kit can be sent for free. The kit comes with instructions on how to perform the swab, as well as a stamped return envelope.

If your child has had prior normal blood testing for *NIPBL* and is not enrolled in the CHOP research study, CHOP can send a consent form to enroll in the study. Once the signed consent is received, a buccal swab kit is sent.

If you are interested in participating in this project, contact Sarah Noon, CHOP genetic counselor, at 215-590-4248 or noonse@email.chop.edu.

*Huisman et al., *Journal of Medical Genetics*, 50:339-34, 2013



Small enough to know you, big enough to help you.



Photo by Eric Leslie

The following are reviews of the CdLS Foundation from Greatnonprofits.org:

“With a tiny newborn in 1977 and no information, we groped our way through each day. When the CdLS Foundation began in 1981 by a group of parents scattered across the country, hope, support, information and advocacy began for us. Today the Foundation is an extraordinary organization of families, professionals and friends, now numbering in the thousands. The information and support that can now guide families is awe inspiring! What an accomplishment! Our Adam died in 2001 but the CdLS Foundation is still there for us.” **–Linda**

“This is the best nonprofit organization I have ever been a part of ... and I work at a nonprofit!” **–Anonymous**

“My daughter was diagnosed with CdLS 16 years ago. The Foundation was an amazing help to us during that time and since then. They hooked me up with other parents in the area and I became a part of an email group that was a huge help with issues and problems specifically related to the syndrome. I live in a small town and we don’t have any children with this here and when you tell people they are really surprised... Thank you for being here all of these years and the promise to continue to be here for us!” **– Juli**

Please remember the CdLS Foundation during the season of giving.



CdLS Foundation

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9 Pewter Ct
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The next CdLS Foundation National Family Conference is in Costa Mesa, CA, June 26-29, 2014. The event takes place at the Hilton Orange County/Costa Mesa, just a few miles from the Orange County Airport. Book your room now at www.hiltonorangecounty.com or call 714.540.7000. Use the code CDLSF to get the \$109/night room rate. This discounted room rate ends May 26 or as soon as the room block fills up.

Conference registration opens February 1, 2014. Adult registration is \$320 before March 26 and \$350 after. Children under 18 are \$220 and \$235, respectively. There is no charge for children under two or anyone with CdLS.

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

Keep an eye on the Foundation web site for more information, or call 800-753-2357.

