Reaching Out

The Newsletter of the Cornelia de Lange Syndrome (CdLS) USA Foundation, Inc. Winter 2009

Photo by Rick Guidotti, Positive Exposure

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New Year, New Initiatives

This issue marks the start of a new year and the completion of my first year as the Foundation's executive director. I look back proudly at the great work of this Foundation, and look forward with hope and excitement.

This year, the Foundation is implementing some new initiatives I'm excited about. These include new features in the newsletter, a national fundraising event and new volunteer opportunities.

In the fall, we asked caregivers what topics they'd like to see in *Reaching Out.* Respondents overwhelmingly said they want more medical information; therefore, we're adding a second medical article in each issue. Additionally, we're recognizing some very important people—siblings. On page 8 we debut *Super Siblings* with the story of a young woman whose college essay focused on lessons learned from her sister with CdLS.

If you have an attic, basement or garage full of things you no longer need, sign up for our *Yard Sale Across America*, Saturday, June 6. Clean out the clutter and help us "Tag the Day" for CdLS by hosting a yard sale. Learn how you can be part of it on page 10.

If one of your New Year's resolutions is to be more involved with the Foundation, now is your chance. We're recruiting volunteers for the following committees: finance, audit, family services, professional development, planned giving, special events, and major gifts.

We're also recruiting volunteers for our national conferences through the year 2016. Under a new model, conferences will be hosted by regional committees and fundraising will be conducted by the Foundation and regional volunteers. We want all regional conference planning committees in place soon. The conference locations and schedule are as follows: Dallas, TX, 2010; Lincolnshire, IL, 2012; Irvine, CA, 2014; and Atlanta, GA, 2016.

Volunteers are also needed for the 2010 National Conference Planning Committee. This committee identifies conference goals and objectives, topics for sessions and workshops, and theme. You don't need to live in Texas to serve. If you'd like more information about any of the opportunities mentioned above, contact Barbara at 800-753-2357 or info@CdLSusa.org.

It's no surprise that the struggling economy in 2008 made for a difficult year. Many non-profit organizations are tightening their belts and lowering expenses. We need to do so as well, but we aren't cutting services to the families and children we serve.

Since printing and postage are increasingly expensive, we've developed a three-part plan to cut costs and continue to provide *Reaching Out* at no charge:

Reaching Out is available online. Help us "go green" by reading an electronic version. You'll receive email alerts when the newsletter is available online. To go green, call 800-753-2357 or email info@CdLSusa.org. (We do not share your email address with other organizations.)

While we'll always send *Reaching Out* to parents, grandparents and others providing care to people with CdLS, we'll no longer mail it to friends and supporters unless they explicitly ask.

Effective this issue, *Reaching Out* goes quarterly. Even though you'll receive the newsletter less frequently, you'll find more information in each issue due to format changes.

These changes are dramatic but reflect an organization that's evolving in order to continue to meet your needs. With volunteer participation, flexibility and commitment to our mission, I'm optimistic we can work together to support one another more than ever before.

My best to you and your family in 2009.

Liana Garcia-Fresher, R.D., M.S.
Executive Director

THIS ISSUE

Medical View: Updates from the World of Genetics

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

Every year, the American Society for Human Genetics (ASHG) meeting brings together clinicians, scientists, counselors, and other professionals from all areas of genetics to hear the latest news, meet with colleagues and share ideas. In November, the meeting was held in Philadelphia, and there was an excellent representation of CdLS, both in the poster presentations and the sharing of ideas.

A number of CdLS Foundation Clinical Advisory Board (CAB) members attended the meeting, including myself, Drs. Ian Krantz, Matt Deardorff, Annemarie Sommer, Yves Lacassie, and Laird Jackson, along with the CdLS Foundation Fellow Dr. Jinglan Liu, and Genetic Counselor Dinah Clark.

In addition to the poster presentations, there was an informal meeting of professionals from around the world working with CdLS or the genes and/or proteins related to it.

We discussed recent advances in *cohesin*, the molecule most affected when there are changes in the genes causing CdLS, as well as an upcoming meeting in Italy, at which investigators will share their most recent work on cohesin.

The posters presented were both clinical and research oriented, and included the following:

- A review of social and family aspects of the multidisciplinary aging clinic held in Baltimore for the past seven years. The information showed that the greatest concerns on the part of parents were behavioral, gastrointestinal, nutrition, weight, dental, orthopedic, and reproductive.
- A discussion of prenatal findings of three cases of CdLS, demonstrating that a low second trimester maternal serum marker (PAPP-A) and an increased size of the fold on the back of the neck (increased nuchal or, in the most severe state, cystic hygroma) both can indicate CdLS (among many other things).
- An explanation of a new method of analysis being used to find underlying causes of CdLS in the 35 percent of individuals without a detectable gene change. Several candidate genes have been identified.
- An update on the molecular testing results from the University of Chicago, which present a number of interesting findings. Attempts to correlate these with clinical findings are underway.
- Research demonstrating that cells from individuals with CdLS who have a change in the gene *SMC1A* have been found to be sensitive to agents potentially toxic to our genetic material, supporting the idea that *SMC1A* plays a role in genome maintenance.
- Information on further studies on the *SMC1A* gene, showing differences between males and females due to the fact that the gene is on the X chromosome. Although many individuals have their own specific mutation, there may be one area considered a "hot spot" for mutations. A "hot spot" is an area that more commonly undergoes a change or mutation than the rest of the gene.

Overall, we should be proud of, and excited by, the new advances and increase interest related to CdLS researach.

Reaching Out
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Diet and Nutrition Issues with CdLS

By Joni Rampolla, L.D., CdLS Foundation Clinical Advisory Board Member

Diet and nutrition play an important role in the growth and development of children. For children with CdLS, balancing good nutrition, diet and willingness to eat can be a challenge. This article addresses some common dietary challenges associated with CdLS, and responds to questions frequently asked by parents.

Proper growth

The growth of a child with CdLS occurs at a slower rate than that of his peers without CdLS. It's important that your physician is plotting growth on the specific CdLS growth charts (available through the CdLS Foundation). If a child's individual growth curve levels off or begins to decline, he may need additional calories to support growth. Prior to adding extra calories, it's important to evaluate other issues that could cause weight loss, such as recent illness or diarrhea, which can indicate malabsorption. Sometimes a change in tube feeding formula can resolve a malabsorption issue (such as a hypoallergenic, or semi-elemental, or fiber containing) rather than increasing calories.

Food allergies

Many children with CdLS have a lactose (milk sugar) intolerance. Consuming milk products may lead to bloating, gas, cramps, or diarrhea. The amount of lactose that each person can handle is different. You can choose to include a lactase enzyme such as Lactaid™ to help with milk digestion or switch to rice milk, soy milk, yogurts, or cheese. Make sure your child gets enough calcium by including a supplement with vitamin D for enhanced absorption, Tums™, or high calcium foods, such as salmon, tofu, almonds, beans, or calcium-fortified foods and liquids (orange juice, breakfast bar, etc.).

Hypoallergenic supplements are available for children that need to supplement an oral diet. Two common products are Elecare by Abbott Nutrition and Neocate Jr. by Nutricia. For wheat allergies, wheat-free grains should be included in the diet. These include amaranth, buckwheat, corn miller, quinoa, rice, and tapioca. A great resource to learn about food allergies is the Food Allergy and Anaphylaxis Network, www.foodallergy.org.

Gastrointestinal concerns

Diarrhea is a common issue in individuals with CdLS. It's important to keep your child hydrated and replace lost electrolytes by including plenty of fluids. You may consider an electrolyte replacement such as Gatorade™, Pedialyte™, or Crystal Lite Hydration™. You can also include salty foods such as pretzels, bouillon or pickles, and a high potassium food or drink, such as orange juice, broccoli, spinach, tomato, summer squash, or cantaloupe. Diarrhea causes a loss of sodium and potassium, so including any of these foods will help.

Other suggestions include an over-the-counter, anti-diarrheal medication or a natural remedy of banana flakes, which controls diarrhea and can add flavor and texture to foods or tube feedings. Nana Flakes is one brand you can look for.

Some foods or beverages make diarrhea symptoms worse. These include caffeinated beverage and foods that are fried, spicy or artificially sweetened (sorbitol, maltitol, mannitol, etc.). If you notice symptoms get worse after your child eats a certain food product, eliminate that item for a few days and reintroduce after symptoms stop.

To relieve symptoms of constipation, include plenty of fluids, high-fiber foods and exercise. For added fiber, choose a high-fiber breakfast cereal or bar (five grams of fiber or more per serving), fresh fruit with the skin on, and vegetables like spinach, broccoli, collard greens, okra, or beans. Add fiber gradually to prevent gastrointestinal upset. The formula to determine the amount of daily fiber needed is the age of the child plus five. So, for a 10 year old, aim for 15 grams.

In addition to over-the-counter fiber supplements, such as Metamucil, Benefiber or Citrucel, a natural fiber source is ground or whole flaxseed, which can be added to foods for a nutty taste and slight crunchy texture. For a smooth-textured, natural remedy, try Fruit-Eze™, a prune-based fruit puree.

For symptoms of gastroesophageal reflux, certain foods may need to be eliminated from the diet, including acidic foods, such as oranges and grapefruits, fried or fatty foods, and spicy foods. A food diary can be kept to monitor foods that irritate your child. Try excluding the suspected irritant and see if the symptoms resolve. To ease symptoms, avoid dressing your child in tight fitting clothing, have him remain upright after eating, and serve smaller, more frequent meals.

A Closer Look: Coping with Food Allergies

By Mary Levis, N.C.S.P., CdLS Foundation Clinical Advisory Board Member and mother of Josh

Josh was diagnosed with CdLS when he was four years old. Once he was diagnosed, we contacted the CdLS Foundation, and one of the first newsletters we received highlighted silent reflux. So many of the symptoms described were similar to what Josh was experiencing, so we met with a pediatric gastroenterologist at Johns Hopkins Hospital. Josh was diagnosed with reflux, treated with medication, and had regular follow-ups. However, he continued to have changes and irritation in his esophagus and stomach, so he underwent a Nissen Fundoplication when he was six.

Despite this procedure, Josh continued to suffer many symptoms of gastritis and did not progress nutritionally as hoped. He was found to be lactose intolerant. He followed a lactose-free diet, but he was still nutritionally at great risk.

By the time he was 10 years old, Josh was seriously undernourished and had very poor energy. At this point, his doctors recommended that a feeding tube be placed. Since he had so many eosinophiles (white blood corpuscles) in his esophagus, food allergies were suspected. We began a course of an amino acid-based formula (Vivonex T.E.N.) through the feeding tube.

The plan was to reintroduce foods gradually to discover what he was allergic to. The feeding tube was supposed to be temporary until the allergens could be identified. We discovered Josh is allergic to casein (milk protein) as well as lactose (milk sugar). Additionally, he is highly allergic to certain shellfish, in particular mussels, oysters and scallops.

Today, Josh is 26 years old and still using the Vivonex T.E.N. for supplemental nutrition. Each time we have tried to wean him off these feedings, his weight and health have declined dramatically. Josh carries a list of the foods he needs to avoid so that when he goes grocery shopping or is at a friend's house, he can inspect the labels of the food before he eats it. He manages this quite well independently and continues to progress and be healthy.

Josh with family members

Did You Know…?

The CdLS Foundation Web site is a valuable resource for previously published information about CdLS. You can search an index of archived *Reaching Out* issues, Ask the Doctor questions and answers, genetic information, and much more. Visit www.CdLSusa.org and click on the Be Informed link to take advantage of the Foundation's information bank.

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Feeding Issues in Individuals with CdLS

By Cheri S. Carrico, Ph.D., CCC-SLP, CdLS Foundation Clinical Advisory Board Member

Many individuals with CdLS experience feeding concerns at some point in their lives. Challenges can include food aversions; being a messy, slow, or picky eater; eating limited varieties and small amounts of food; taking very small bites; spitting food out; and refusing to eat. On occasion, however, some individuals "stuff" food in their mouths.

There are a variety of factors that impact feeding, including gastroesophageal reflux disease (GERD), low muscle tone in and around the mouth, a small jaw (micrognathia), oral defensiveness, and aspiration of food or liquids. Symptoms of oral feeding difficulties include choking, coughing, gagging, vomiting, a "gargly" sounding voice after feeding, aspiration, and food aversions.

Infants with CdLS may have difficulty sucking or coordinating sucking, swallowing and breathing. Some difficulties may not be observed until infants are a few months old, when anatomical changes in the mouth and neck may make it difficult to control food to swallow it safely. Other challenges become present as children move from formula to solid foods.

Approximately 90 percent of individuals with CdLS experience GERD. These symptoms include spitting up or vomiting during or following meals, as well as pain or discomfort associated with feeding. Consequently, individuals affected by GERD may develop feeding aversions. Food aversions also are frequent among individuals with a history of tube feeding. When children are tube fed, the tube feedings bypass the need for oral feeding and can result in a lack of exposure to a variety of tastes, textures, temperatures, smells, and food presentations that provide oral stimulation and make meal times a pleasant, social experience.

In addition, individuals who are tube fed may be fed in places or at times that are not typically associated with feeding experiences (such as bedrooms or during the night). Thus, it's essential to provide individuals who are tube fed with opportunities to experience both the oral and social stimulation associated with meals.

Individuals who exhibit feeding challenges should be referred to a speech-language pathologist, and medical clearance should be obtained prior to initiating oral feeding. Individuals with swallowing difficulties, such as coughing, choking, or gagging during meals, typically are recommended for a swallow evaluation. The evaluation will determine whether aspiration, the accidental sucking in of food particles or fluids into the lungs, occurs. If no aspiration is evident, it's probably safe to feed orally. If there is aspiration, modifications such as thickening of liquids or pureeing of solids may be recommended. Liquids may be thickened with baby cereals, baby foods, oatmeal, flour, corn starch, yogurt, applesauce, and commercially available thickeners. Soft pieces of pasta, rice, potatoes, cooked vegetables, and canned fruit may gradually be added to provide textures that are relatively easy to manage. Gradually increase the number of pieces and the firmness of the pieces before moving to firmer foods, such as bread, soft breadsticks, medium cooked vegetables, lunch meats, and fish sticks. Eventually, transition to firm solids, including apples (they may initially need to be peeled or cut into slices), carrots, meats, teething biscuits, crackers, pretzels, granola bars, and so forth. Remember to proceed in small, gradual steps as broader food preferences are encouraged.

No oral feeding should occur unless medical clearance is obtained for individuals with a history of feeding difficulties. If oral feeding is contraindicated, oral stimulation should still be provided, using a variety of tastes (sweet, salty, spicy, sour, bland), textures (smooth, bumpy, soft, firm, "squishy"), and temperatures (warm, room, cool, cold, and frozen).

Taste stimulation can be provided by adding objects to a child's hands, such as traces of food or liquid, toys, pacifier, toothbrush, cloth baby book, teething ring, and wash cloth, or by putting solid foods in a safety feeder (a small device with a mesh bag attached to a solid plastic ring, available where baby feeding items are sold). To provide temperature stimulation, appropriate items can be placed in the freezer or refrigerator, warmed in the microwave oven or in warm water, and so forth. Texture stimulation should be provided by utilizing items of various textures, such as those described, as well commercially available "chewy tubes" and refrigerator tubing.

Initially introduce only one stimulus (taste, texture or temperature) at a time, if a child has a history of feeding aversions. Once each is reasonably well tolerated, stimuli can be combined, for example, by trying cold applesauce

that previously was tolerated at room temperature. Also stimulate the oral area using deep touch. The lips, cheeks, and chin can be strengthened by applying firm pressure and massage. Strengthen the tongue by applying pressure with a finger down the center of the tongue and by gently pulling on the tongue and shaking it, while holding it with a wash cloth. These techniques may also reduce oral defensiveness and lead to better oral feeding skills. Be sure to engage your child in a very playful manner.

Ultimately, every attempt should be made to keep feeding social, enjoyable, and fun.

A Closer Look: Families Consider Intensive Feeding Programs

By Jennifer Sangeloty, Communications Coordinator, CdLS Foundation

It's only natural for parents to want the best for their children. For parents of children with CdLS, it can be difficult to decide whether to enroll their children in intensive feeding programs if they are tube fed. Below are the stories of two families currently facing this decision.

Julie and Darian of Maryland recently completed an intensive feeding program with their two-year-old son Fletcher. "The main reason we wanted to do this program was the desire for normalcy," Julie says. "We want Fletcher to live as normal a life as possible, and eating through your mouth is a big part of that."

Fletcher

Fletcher attended the feeding program at Mt. Washington Pediatric Hospital in Baltimore. His regimen consisted of three half-hour feedings per day Monday through Friday for seven weeks. By the end of the program, Fletcher readily accepted a half ounce of pureed food per meal.

Fletcher's oral motor skills also improved. "By the end of the program, if he didn't open his mouth automatically when he saw the spoon, he would open it if you asked him," says Julie. "And not all but some of the food stayed in his mouth. This was a huge development."

Julie and Darian haven't stopped their efforts to help Fletcher eat. "We are so convinced that Fletcher needs to eat, that we are willing to do absolutely anything to get him there," Julie says. Fletcher is currently on a waiting list for a more aggressive feeding program at the Kennedy Krieger Institute in Baltimore.

Isabelle

For Jennifer and Brent, parents of Isabelle (pictured on the cover), who will be three years old in May, the decision to enroll their daughter in an intensive feeding program has been difficult. Isabelle is on the waiting list for a program that consists of five feedings per day, five days per week for eight weeks. In the meantime, the Michigan couple has gone back and forth between different forms of feeding. After gagging spells and Isabelle's frustration with bolus (all at once) tube feeds, the family is focused on drip feeding while Isabelle sleeps.

"The drip feedings while sleeping seems to fit well with Isabelle. She is healthy and happy," Jennifer says. "It seems that the less pressure I put on her to eat, the more she eats on her own. I'm worried that we may take a step in the wrong direction if we put her in the intensive program and try to force her to eat. I think she will resist even more," she says. "We are still weighing our options … and we will continue to encourage eating and weekly feeding therapy, just not in an aggressive manner."

Jennifer and Brent would like to speak to families whose children have had success in an intensive feeding program. Contact the Foundation at 800-753-2357 or familysupport@CdLSusa.org and we'll share your contact information with the family.

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 lifetimes.

Super Siblings

When Ruth from Texas applied to Texas A&M University, she was required to write an essay describing the person who has most influenced her life. "I knew I had to write about my sister Blair because she has been a huge part of my life that shaped who I am today," Ruth says.

Blair, now 24, has CdLS. While Ruth is the younger sibling, she grew up with the responsibility of helping her parents care for Blair. In her essay, Ruth explained that she didn't always appreciate how special her sister was until Blair moved away from home.

The saying goes, "you don't know what you've got until it's gone." When I was younger, I thought the saying only applied to material objects. It was not until my sister, Blair, moved away that I realized that the most important things in life are not necessarily tangible.

Ruth admits that growing up with a sibling with CdLS is full of challenges.

Although I loved my sister's spirit and silliness, I would often become aggravated and annoyed with her unceasing dependence. Instead of going out with my friends, I would have to stay home and watch her.

Despite these challenges, once Blair moved to a group home at age 18, Ruth learned that her sister influenced her life in many ways.

Our family was not the same without Blair; we could not live without her. Even though having her gone allowed me the freedom I had always dreamed of, I did not have her. I thought I was escaping the burdens of living with a mentally handicapped person; I thought life would be better, easier. However, I realized that aside from her silliness, I missed the babysitting. I found that she was what made my life happy.

It wasn't long before Ruth's family brought Blair back home to live with them. Ruth has spent time apart from Blair for the past three years while she studies Animal Science at the university. "It's been difficult for both of us with me being away from home. It's definitely a big change for Blair," Ruth explains. "But since we have minimal time together, we try to make every moment count."

Ruth says that the most valuable lesson she has learned from living with a sibling with CdLS is that of patience. "It's not easy to understand or deal with a family member with special needs. When I was younger I didn't understand that my sister was different, that she had special needs. But now that I have learned patience, we get along better and are closer because I have a better understanding of my sister."

The last lines of Ruth's essay express her outlook on life with Blair.

I know now that I have to take the bad with the good, the burdens with the happiness. I now cherish every day I share with her.

Send your "Super Siblings" story to communications@CdLSusa.org or call 800-753-2357 and your experience may be featured in an upcoming issue of *Reaching Out*.

The fifth bi-annual CdLS World Conference takes place in Brighton, England from July 22-26, 2009. This year's conference is sponsored by CdLS Foundation UK and Ireland. For more information about the 2009 CdLS World Conference, visit www.cdlsworld.org.

WELCOME NEW FAMILIES

Illinois
Vicki and Gail and granddaughter Olivia
born December 25, 2007

Kansas
Teri and JD and son Camden
born October 11, 2007

New Jersey
Theresa and son Bryan
born February 18, 1997

New Mexico
Roberta and Kevin and son Isaiah
born August 8, 2006

New York
Melissa and Ken and son Tristen
born June 26, 2007

Tennessee
Monique and son Elvis
born October 21, 2008

Texas
Dolores and son Trent
born October 4, 1988

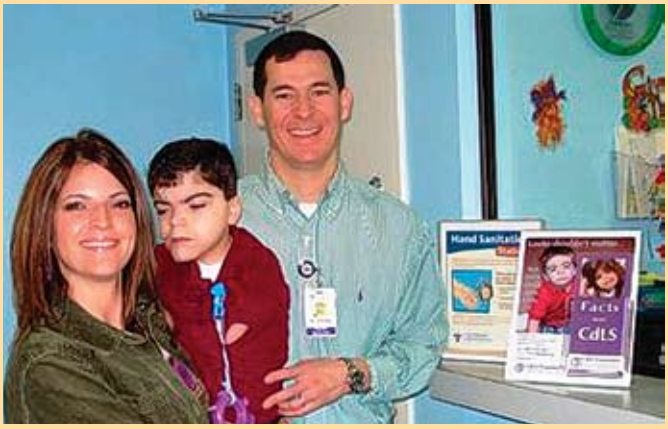
Utah
Bobbi and Todd and daughter Kinzi
born March 23, 2006

Washington
Meliza and Don and daughter Danika
born March 1, 2008

Only new families who have given us permission to print their information are listed above.

Awareness Around the Country

Candace, an Awareness Coordinator (AC) in Louisiana and mom to Ethan (pictured at right), set up a CdLS brochure display in Ethan's pediatrician's office. Candace was one of a dozen ACs who set up the displays as part of the Awareness by Month program.



CdLS Awareness Day celebrates 20 years

CdLS Awareness Day marks 20 years on Saturday, May 9. Awareness Day had its start as an international yard sale in May 1989. Later, it became an official national health observance day, occurring the second Saturday of May.

This year, you can help raise awareness of CdLS in your community by playing Awareness Day BINGO. Try some or all of the activities in the squares at right. If you get BINGO, you can even compete for a prize.

For templates and materials for any of the activities, contact Marie Malloy at outreach@CdLSusa.org or 800-753-2357. Good luck!

Got BINGO?

To be part of a drawing for a prize, submit your BINGO sheet to Marie by June 1. To be eligible, you must have completed activities **in five consecutive boxes.**

Fax: 860-676-8337
Mail: CdLS Foundation
302 West Main Street, #100
Avon, CT 06001

C	d	L	S	
Set up an awareness booth at a popular shopping area or town event. Include a poster board display.	FREE	Create a postage stamp with your child's picture (go to zazzle.com or photostamps.com to create it).	Request an official Awareness Day proclamation from your mayor or town supervisor.	Email your friends and family about Awareness Day. Include a link to the Foundation web site and an update on your child.
Hang a CdLS poster at your place of work.	Ask the bank to put an Awareness Day message on its marquee (or any other business with a marquee).	Put a CdLS brochure in your mailbox for your mailman/woman.	Call a newspaper or TV health reporter and ask if he/she would do a feature about CdLS.	FREE
Be part of the outside audience of the Today Show or other a.m. news show. Don't forget your Awareness Day sign.	Submit your child's photo and info to the Foundation's Online Photo Album (if you haven't already).		Submit a public service announcement to your local radio station.	Add an Awareness Day message to your email signature.
FREE	Send a CdLS goodie basket to your child's school or health professional's office. Include candy or bubbles and CdLS materials.	Put a CdLS flier on the community bulletin board at your post office or other busy public place.	Get together with another CdLS family (the Foundation can provide a list of families in your region).	Present info about CdLS to your child's or a sibling's classroom.
Ask your favorite salon or coffee shop to donate a percentage of the day's proceeds to the Foundation.	Put a CdLS brochure rack in your child's doctor's office.	Make Awareness Day T-shirts for your family	FREE	Send a letter to the editor to your local newspaper about Awareness Day.

Yard Sale Across America

Saturday, June 6, "Tag the Day for CdLS"

Over the years, neglected, outgrown and unused items inevitably begin to consume your closets, basement and attic. If you're ready to eliminate clutter and recycle belongings you no longer need, you can do so while supporting a great cause.

The *Yard Sale Across America* takes place on Saturday, June 6. This cross country event supports the Foundation by raising money and awareness. The concept of a national yard sale to benefit the Foundation originated in 1989 when Shelia Penedos, mom to Rachel (CdLS), devised a plan to educate hundreds of ardent bargain hunters about CdLS while raising funds, then urged others to do the same. Yard sales raised more than $8,000 that year.

This year, you can "Tag the Day for CdLS" by organizing your own yard sale to benefit people with CdLS. The Foundation's goal is to have at least one yard sale in every state. Get your neighbors, friends and relatives to donate items to your sale, or organize a neighborhood yard sale, with proceeds benefiting the Foundation. While the official event date is June 6, you can host your yard sale any time during June.

The *Yard Sale Across America* Web page provides information and tips on preparing your yard sale, collecting and pricing items, advertising, disposing of unsold items, and much more. The Web page also includes a national map showing all CdLS yard sales, so even if you can't host a yard sale, you can shop at one. Visit the *Yard Sale Across America* Web page on the Foundation Web site, www.CdLSusa.org.

Help us "Tag the Day for CdLS." For more information or to request a *Yard Sale Across America* starter kit, contact Gail Speers at 800-753-2357 or events@CdLSusa.org.

Achieve your dream.
Give hope to others.

Run with Team CdLS
at the Chicago Marathon

October 11, 2009

If you or anyone you know would like to cross the finish line for children with CdLS, contact Gail Speers at events@CdLSusa.org or 800-753-2357.

Calendar

March 27 – 29
CdLS Foundation Board of Director's Meeting
Hartford, CT

April 4
CdLS Multi-disciplinary Clinic for Adolescents & Adults
Greater Baltimore Medical Center
Towson, MD

May 9
National CdLS Awareness Day

June 6
Yard Sale Across America

July 22 – 26
CdLS World Conference
Brighton, England

November 6 – 8
CdLS Foundation Board of Director's Meeting
Location TBD

Check our Events Calendar at www.CdLSusa.org/calendar.shtml or call 1-800-753-2357 for the most current information.

Foundation Receives Clinic Funding

Quest Diagnostics Foundation has awarded the CdLS Foundation $5,000 for the CdLS Multi-disciplinary Clinic for Adolescents and Adults.

The grant will be used to provide additional services and diagnostic tests—such as echocardiograms—that have been cost-prohibitive to offer at the clinic.

"This money will go a long way to help provide fully comprehensive services to the families," said Dr. Tonie Kline, the CdLS Foundation medical director and director of pediatrics at the Harvey Institute for Human Genetics at Greater Baltimore Medical Center, where the clinic is held. "The more diagnostic work we can do, the more we can learn about CdLS. The benefits go far beyond one clinic day."

Held twice yearly, the clinic is free to families and includes one-on-one consultations with specialists in ophthalmology, psychiatry, gynecology, and more. The next clinic is Saturday, April 4, 2009, and is open to individuals who are at least 13 years old. It can accommodate up to seven individuals who have CdLS.

For the upcoming clinic, travel and hotel stipends are available thanks to other recent grant funding. Reimbursement of at least $250 for up to seven families will be awarded on a first-come, first-serve basis. Four of the stipends are restricted to families from Maine, New Hampshire, New York, New Jersey, Massachusetts, Connecticut, and Rhode Island.

For information on the clinic, call Janette Peracchio at 800-753-2357 or email familyservice@CdLSusa.org.

Established in 2001, the Quest Diagnostics Foundation is committed to supporting health related charitable programs. Priority is given to funding programs and projects that promote the prevention and early detection of disease or provide research for curing disease.

Foundation Welcomes New Communications Coordinator

Jennifer Sangeloty began her position as the Foundation's communications coordinator in October, 2008. You will see Jennifer's communication skills at work in *Reaching Out*, the CdLS Foundation blog, awareness materials, and special events campaigns.

"I am excited to be part of the Foundation's efforts to reach out to families and help make a difference in the lives of those affected by CdLS," Jennifer says. "I've always had a passion for working in the nonprofit field, and this is the perfect opportunity to use my abilities for the benefit of others."

Jennifer received her bachelors degree in Communication from the University of New Haven in Connecticut. Her experience in nonprofit communications began with her public relations internships for the New Haven Symphony Orchestra and Orange Economic Development Corporation. Professionally, she served as an associate public relations consultant for Bridgeport Public Schools and Aquarion Water Company.

If you have a story you would like to share with Jennifer for *Reaching Out* or other publications, please email communications@CdLSusa.org.

Our Deepest Sympathy

Tony Lee Curry
February 20, 1958 –
November 13, 2008
Brother of Kathy Hedrick
P.O. Box 401
Matheny, WV 24860

Raymond Carl Grueneich
May 20,1969 –
December 29, 2008
Son of Shannon Casamo of
Santa Cruz County and
Ray Glock-Grueneich
115 Vista Prieta Ct
Santa Cruz, CA 95062

Nicholas Toffer Schurke
March 6, 2004 –
November 20, 2008
Son of Sarah and
Christopher P. Schurke
330 Perry Ct
Zumbrota, MN 55992-1291

Gail Elizabeth Truett
June 24, 1954 –
December 13, 2008
Daughter of Ruth
and Bob Truett
404 Cheswick Pl
Rosemont, PA 19010

Going Green for the New Year

Help the Foundation save money and "go green" by receiving your issue of *Reaching Out* electronically. Each time *Reaching Out* is published, you'll receive an email with a link to the latest issue. The Foundation saves printing and mailing costs while being more environmentally friendly. Various Foundation events, blogs, and other updates are most often sent via email.

To subscribe to *Reaching Out* electronically and receive additional Foundation updates, contact Barbara at info@CdLSusa.org or 800-753-2357 and let her know you're "going green."

Spanish Translation

New informational publications are now available in Spanish on the CdLS Foundation Web site. Visit **http://www.CdLSusa.org/publications/index.shtml#ESPANOL** to view a full list of translated materials.

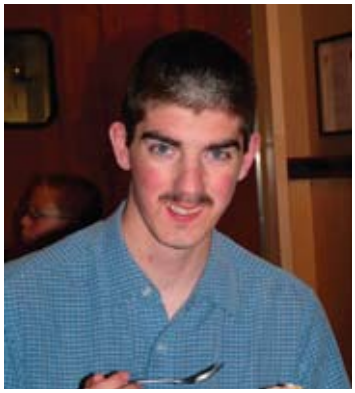
Visítenos en nuestro sitio web **http://www.CdLSusa.org/publications/index.shtml#ESPANOL** para obtener publicaciones nuevas en español sobre el síndrome Cornelia de Lange.

Wishlist

Print and mail one issue of *Reaching Out*.....$4,500
Postage for one issue of *Reaching Out*.....$2,000
Upgrade laptop computer.....$1,500
Send a family to clinic.....$1,100
Purchase half-page ad in *Reaching Out*.....$800
Upgrade battery backup for network.....$250
Replace laser printer ink cartridge.....$150
Photos & stories of people with CdLS.....PRICELESS

MAILBAG

-Sam-



Dear *Reaching Out* Editor,

Sam spent the month of August in Antelope, Oregon at a Young Life Camp (Wildhorse Canyon) serving on the volunteer work crew. He worked up to 8 hours a day on kitchen staff. It was quite an experience. He had never been away from home that long before. For the past three years, he had gone to Wildhorse Canyon as a camper, one week a summer.

Sam was chosen to work alongside typical college students who volunteer their time for one month each year. He had a peer coach who not only worked alongside him in the kitchen, but they also spent their free times together, so he did not fall through the cracks. It was a bit rocky at first, as he was homesick in the afternoons, but with time, he was getting used to the routine and also, they tweaked it to accomodate some down time for him to do some of the things he enjoys at home. Additionally, we brought envelopes for him to open each day, the rest of his trip, so he had something to look forward to. Some contained a candy, or a funny card, some new music or a picture. We were told that made a big difference.

Two weeks into the session, we were able to see him on parent visit day for three hours. It was a 7 hour drive from home to the middle of nowhere! He gave us the royal tour and was so proud of his accomplishments. He introduced us to many new friends and we were able to swim and share a meal. Though he was glad to see us, he was happy to see us go, as he heard there was going to be a bonfire that night with s'mores!

Sam's peer coach, Jessie, was a God-send. She helped him at first and then backed off as the whole work crew (70 people) embraced Sam and made him feel welcome! By the end he was a seasoned work crew volunteer, and said he would love to do it again. When he got home, the biggest change we noticed was that his self confidence had grown so much. He had learned to do many tasks and was even more conversational.

Sam wasted no time at home. After he got back from Oregon in September, he flew to Arkansas in October to visit his former junior high teacher who has become an important part of his life and welcomes his visit each year.

Tim, Sam's dad, Washington state

-Iselen-

Dear CdLS Community,

I recently attended a CdLS Board meeting in St. Louis. Several of us who are parents and board members shared challenges our teen and young adult children are facing. I found myself thinking about how difficult it can be for our children to go through the developmental stages of becoming adults with their challenges and what we can do as parents and guardians to help our children develop positive self-esteem.

I work in mental health and serve some individuals who have developmental disabilities and mental illness. As a psychiatric nurse practitioner my work is primarily prescribing medications, but I enjoy the interpersonal aspects of my work also. I have often wondered how self-esteem impacts mental health and whether we do a good enough job supporting positive self-esteem among individuals with developmental disabilities.

In my own experience, it has been difficult for me to understand how my daughter feels about herself because of her limited verbal ability to express her feelings. One evening we were looking at pictures of former classmates and friends who have gone on to more independent lives when she pointed to her head and showed me the sign for "broken." I asked her whether she thought her head was broken and she said yes. I felt so sad but was glad she had shared that with me. Since that time, I have become more aware of the ways I interact with her that are negatively effecting her and I am trying to find ways to help her have a better sense of herself.

I would like to hear your experiences with your sons and daughters and whether issues of self-esteem have been a concern for you as parents or guardians. I would also like to hear what you have done to foster positive self-esteem for your son or daughter with CdLS. Please send your ideas and experiences to my email address at kcrosvik@comcast.net. I will review what I receive and I plan to write a follow-up article that includes ideas for fostering self-esteem among our loved ones with CdLS.

Kari, Iselen's mom, Washington state

-Phoebe-

Hello everyone,

Three years ago, at the age of 50, I thought I was finally through with the little kid part of parenting. My husband Bruce and I were the parents of three sons by birth, three daughters by adoption, and had parented about 120 other children from the child welfare system. I was tired of diapers, tired of middle of the night feedings, and tired of living in a house that was perpetually baby-proofed. I had written two books about our experiences as foster parents, *Another Place at the Table*, a story of shattered childhoods redeemed by love, and *One Small Boat*, the story of a little girl lost then found, that had done quite well. I had been on a cross country book tour and was anxious to settle down to a slower paced lifestyle.

Enter Phoebe Jean. She was seven months old and in need of a family, but her social worker was reluctant to place her with a family pre-adoptively until her special needs were assessed. I had a lot of experience with special needs babies and agreed to take her on since her stay was destined to be short lived.

By the end of her first day with us it was clear that Phoebe was indeed a baby with special needs. Her head was too small, her limbs floppy. She didn't roll over or coo or make much eye contact. I could hear her wheeze from across the room. She was hairy and her eyebrows met in the middle. Her toes were fused and she had reflux like I had never seen. Twenty, thirty, or forty times a day she spit up the small amount of formula we could get her to swallow. No wonder she didn't grow. I began to have a lot of compassion for the very young mother who had been trying to raise this needy little girl.

Finally, when she was ten months old, we took her to see a geneticist. He spent an hour studying this twelve-pound baby. When he was finished, he pulled out a picture of a little girl who looked just like Phoebe, a little girl with Cornelia de Lange Syndrome.

Months went by and Phoebe got everything she needed. The fundoplication put her in the hospital for three weeks, but it changed her life. She got ear tubes, seizure medication and therapy. She started to babble. She rolled over, then crawled, then walked.

Talk turned to adoption. A nurse in Boston saw Phoebe's picture, read her profile, and asked to meet her.

I can't say why it happened. Goodness knows. We've had a dozen other babies over the years. We had parented any number of bright, beautiful children with limitless futures but we were able to transition them to adoptive families without a second thought about whether it was the right decision for us or for the child. Somehow Phoebe was different. We heard about this family and we knew we could never let our Phoebe go.

"You must be crazy" is the comment we heard most, closely followed by "What are you thinking?" so here is my attempt to explain myself.

When my other kids walked and talked and did all of those things that most children do on time and as a matter of course, we were delighted. We took pictures and noted the moment, then moved on. But with Phoebe, it's different. Her accomplishments bring us, not just pleasure, but profound joy. They are hard-won steps, each of them, and we revel in every small success. Her first steps, her first words, the day she first cried for me, are etched in my mind as instants of bliss. On the day Phoebe first smiled at us, Bruce's eyes filled with tears. "I do believe God dropped an angel on our doorstep," he said.

Kathy, Phoebe's adopted mom, Massachussetts

Remember the CdLS Foundation in Your Will

One way to ensure that the CdLS Foundation will be here for children with CdLS and their families for as long as needed is to make a bequest to the CdLS Foundation in your will. In the previous issue, we provided some bequest language in order to support the CdLS Foundation with an unrestricted gift. However, if you wish to make a bequest in support of a specific purpose, you may use the following sample language.

I give to The Cornelia de Lange Syndrome Foundation, Inc., a non-profit Massachusetts corporation with its principal office in Avon, Connecticut; the sum of $_____ to be used for [state purpose].*

* Rather than leaving a certain sum, you can include language that refers to a percentage of your estate or a description of the property you wish to give. If you would like sample language to suit another type of gift or contingency, please call (800-753-2357) or email the office (giving@CdLSusa.org) to arrange for language you prefer. Thank you.

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

Enclosed is my tax-deductible gift of: ❑ other $_____ ❑ $750 ❑ $500 ❑ $100 ❑ $50 ❑ $35

Name_____ E-mail_____

Street_____ City_____ State _____ Postal Code _____

❑ Use my gift to save the lives of children with CdLS

❑ Please send me information about the Lighthouse Giving Society.

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

Charge my gift to: ❑ VISA ❑ MC ❑ AMEX

Card #_____

Expiration:_____ CCV# (3-4 digit code):____

Print name on card: _____

Signature:_____

To give on line: www.CdLSusa.org

To give stock or plan an estate gift, contact giving@CdLSusa.org or 800-223-8355

03/09

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

Return Service Requested

How Do You Spell Love? J-A-M-E-Y

"Wow, look at all that dark hair, and can you believe how tiny he is?" were my first words when I saw Jamey. With pride and amazement, I then realized that I had a huge responsibility of becoming a grandmother.

Jamey was not diagnosed with CdLS until he was six months old, and as I look back, they were difficult and challenging months. Once we had a diagnosis, we were really scared, and the doctor handed us a flyer with information about a Foundation that could offer us some comfort and information.

I'll never forget the first time I spoke with someone from the Foundation. I cried tears of relief when I hung up. We were regular callers and over the years, the staff became like family to us. Any time we had a question or concern, we called our CdLS family. As soon as they answered the phone, they immediately knew me and asked about Jamey.

Our relationship with the Foundation grew over the years. The education they provided to Jamey's doctors, teachers, dentist, and family was essential in providing him with opportunities that allowed him to grow in mind and body.

When Jamey was five, we had a chance to meet the staff at a national conference in Tennessee. I had the opportunity to attend the "Grandparents Tea." It was so nice to finally meet Julie Mairano (former executive director) and put faces to everyone we had spoken with so often.

At a regional conference, we met Alex and his family who only live about an hour and half from us. Many more gatherings provided other families and friends over the years. The best thing about attending gatherings is that you can take off the mask that you normally wear around others. Families who have children with CdLS understand exactly what you are feeling.

I have always wondered how I could give back to the CdLS Foundation in a meaningful way. When I was asked to serve on the Board, it seemed like an answer to my prayers. I am very proud to volunteer my time and serve others.

Jamey is now 19 years old and attends a school for children with autism. He is very funny and entertaining. He loves Disney's Cinderella, Snow White, Belle, and the other beautiful princesses. His all-time favorite movie is *The Wizard of Oz* and he has quite a collection of everything Oz. Jamey has an amazing memory especially with numbers. He knows everyone's birthday and how old everyone will be on their next birthday. He loves music and he likes to watch movies.

His brothers, Dave and Justin, are very loving toward him and his mother and father are very patient, kind, caring and loving. Everyone in our community knows and loves him. We have all learned so much from Jamey and I can say that I am a better person because of Jamey.

Note: *Penny serves on the CdLS Foundation Board of Directors and chairs the new Planned Giving Committee. If you would like to volunteer with her for this committee or another Foundation committee, please call (800-753-2357) or email (info@CdLSusa.org) the office.*