

Reaching Out

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



Spring 2013

Highlights

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2013 de Lange Society
Honored

Spotlights:

The Importance of GI
Testing

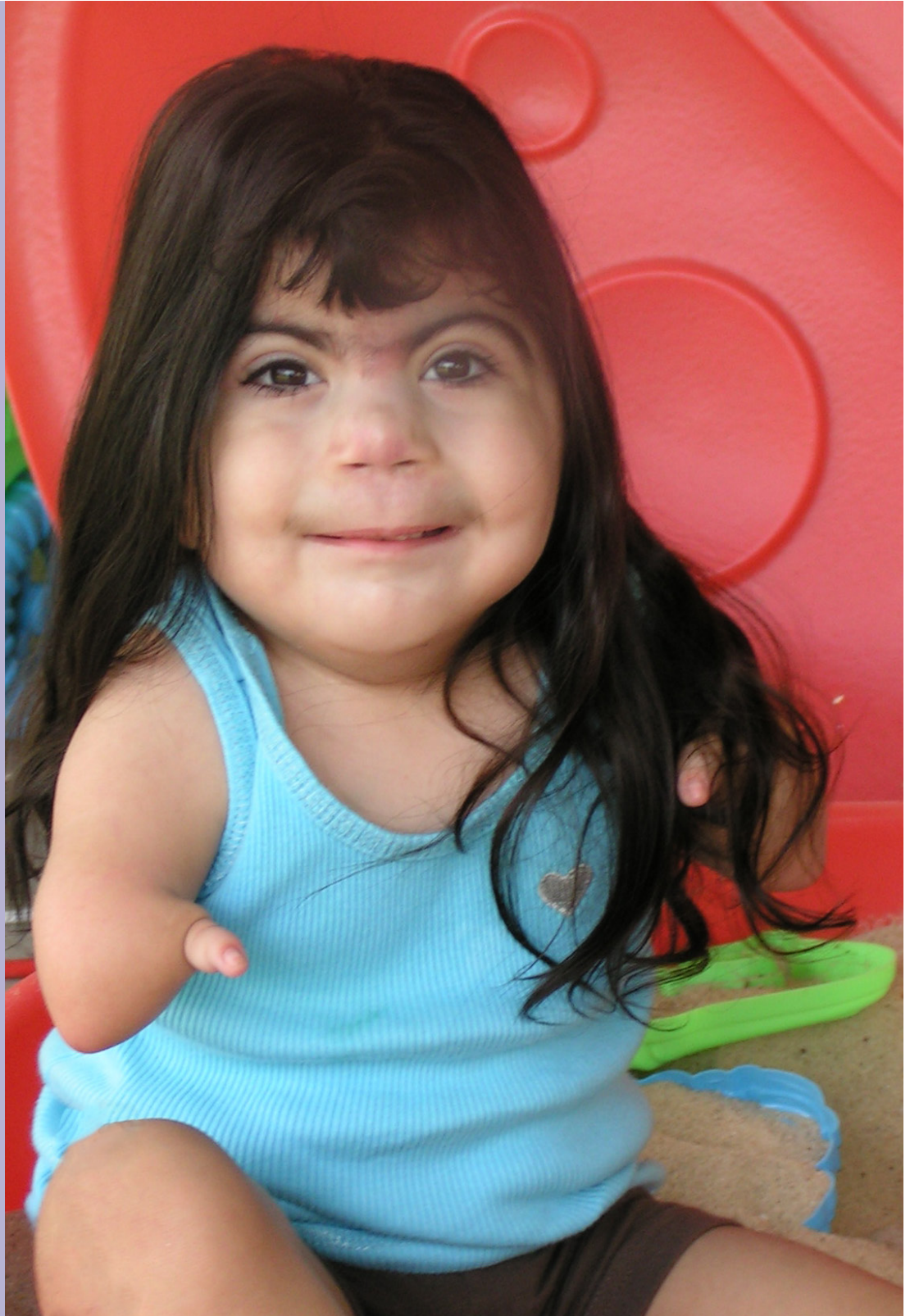
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Difference





Director's Message



2013 de Lange Society Honored

I recently had the pleasure of meeting a group of very special people whose work has made the CdLS Foundation what it is today — a thriving, professional organization still doing what it was created to do more than three decades ago: helping families and professionals caring for someone with CdLS.

They came together at the first ever de Lange Society Luncheon, held in Avon, CT, on Saturday, April 6. This new program was formed to pay homage to people who have dedicated the past 20 years or more moving the Foundation forward by raising awareness, developing new events and helping families.

Of the 40 inductees, 27 were on hand at the event. They included doctors, teachers and moms who wear, or have worn, hats as board members, event organizers and volunteers of all kinds. Although the inductees may have different backgrounds and gifts, they share one wish: a better life for people with CdLS.

The members of the de Lange Society Class of 2013 have given their time, talent and treasures to advance the way for others. Please join me in honoring this very special group of people.

Eileen Ahearn, M.D., Ph.D.
Molly Black
Shelly Champion
Douglas Clemens, D.M.D.
Shari Drake
Susan Drexler
David Fowler
Julie Gonella
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Ken Vacovec, Esq.
Gene Van Buren
Kathy Van Buren
Kathy Wagner
Norm Winnerman

Marie Concklin-Malloy

Executive Director

See photos from the event on page 10

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The Importance of GI Testing



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

Most individuals with CdLS have some type of gastrointestinal (GI) involvement. Most commonly, this includes gastroesophageal reflux disease (GERD), in which the valve between the stomach and esophagus, the tube which connects the mouth with the stomach, does not function properly and allows the acidic stomach contents to get pushed up into the esophagus. Symptoms include frequent vomiting, bad breath, destruction of enamel on

the back of the teeth and abdominal pain (“heartburn”). With persistent reflux, there can be damage from the acid on the wall of the esophagus, including ulcers, hiatal hernia or Barrett’s esophagus, in which the lining of the esophagus changes to be more like the stomach. This latter complication can bear a risk for developing a type of cancer (adenocarcinoma) of the colon. GERD is treatable, either by medications or surgery, but can present at any age and recur, so it is important for all families to be aware of it.

In some patients, malrotation, or abnormal twisting and tethering of the small intestine, can be present at birth. If present, malrotation should be surgically repaired. If not repaired, it can lead to volvulus, an acute occurrence in which the bowel twists on its stalk, resulting in cutting off of blood supply to the intestine. This can be intermittent and resolved, or it can persist and be a life-threatening emergency, and a not infrequent cause of mortality in CdLS. This more commonly occurs in the duodenum of the small intestine, but can occur in the cecum, or the start of the large intestine. Symptoms consistent with possible intermittent obstruction include acute and significant abdominal pain, rigid (board-like) abdomen, and/or bright yellow vomiting. If these occur, volvulus should be suspected and the child should be brought to the emergency room and/or a surgeon should be contacted immediately. Possibly X-rays and/or a definitive upper GI testing series should be done. These studies are recommended even if the child is asymptomatic upon arrival but with a history of this presentation. Another significant complication in CdLS is the risk for bowel obstruction, either by volvulus, or impacted fecal material, or other unknown causes. Presentation is similar to volvulus.

There are other rarer GI complications and the most important aspect is for all parents to be aware that these can occur so signs and symptoms are not missed. The Foundation has GI experts on the Clinical Advisory Board, who can answer questions related to GI complaints.

Reaching Out
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Cornelia de Lange Syndrome
Foundation, Inc.

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The Importance of GI Testing



*By Kathleen M. Loomes, MD,
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CdLS Foundation Clinical
Advisory Board*

Gastrointestinal (GI) manifestations are very common in Cornelia de Lange Syndrome (CdLS), and include anatomic abnormalities of the GI tract, gastroesophageal reflux, constipation and feeding difficulties, as well as other diagnoses. Most children with CdLS will have some type of GI diagnostic study performed, each of which has specific indications.

Often the first diagnostic test to be performed is an **upper GI study**, in which barium is taken by mouth or instilled into the stomach, and X-rays are taken to define the anatomy of the upper GI tract. The main purpose of an upper GI study is to assess for anatomic abnormalities of the GI tract, especially intestinal malrotation. **Malrotation** is an abnormality occurring before birth, when the intestines do not turn normally during prenatal development. The upper GI study will reveal abnormal placement of the small intestine in the abdomen, which puts it at risk of twisting on itself and causing a bowel obstruction or a life-threatening condition called midgut volvulus. If this were to occur, it would usually be associated with acute abdominal pain, distension and vomiting of bilious (green/yellow) material.

Malrotation is still relatively uncommon in CdLS, but in our experience we have found that it is more common in CdLS than in the general population, so we recommend that children with CdLS have an upper GI study done to confirm normal anatomy. Other anatomic abnormalities that might be seen on an upper GI study and have been rarely reported in CdLS would be duodenal atresia (blockage) or jejunal atresia. Often an episode of gastroesophageal reflux will be visualized on an upper GI study, but since it is just one point in time it is not the best

test to determine the frequency or severity of GE reflux.

Many children with CdLS are affected with gastroesophageal reflux (GE reflux), which may be severe at times. The term GE reflux simply refers to stomach contents coming up into the esophagus, and this condition is very common in infants. However, GE reflux becomes pathologic if it results in severe irritability, food refusal, poor weight gain due to excessive vomiting or respiratory complications due to aspiration of stomach contents. Children with CdLS may present with typical manifestations of GE reflux, such as spitting up, vomiting through the mouth or nose, crying with feeds or back arching. Children with CdLS may also have atypical manifestations of reflux, such as hyperactivity or behavioral issues.

Many times the diagnosis of GE reflux may be made clinically based on the history or physical examination, and treated accordingly. However, depending on the clinical presentation and response to therapy, additional testing may be necessary. For example, if a child is irritable and has feeding difficulty but is not actually vomiting, it may not be clear that the symptoms are due to reflux. In this case, a **pH probe study** (or pH/impedance study) may be useful. In this test, a small probe is placed down the nose into the esophagus. The sensors on the probe measure acid reflux (or both acid and non-acid in the case of a pH/impedance probe) for 24 hours and can determine how much of the time the child is refluxing and also correlate symptoms with reflux episodes. If respiratory symptoms are prominent, a nuclear medicine milk scan may be useful. In this study, labeled milk or formula is taken by mouth or instilled into the stomach, and then imaging is performed. Reflux episodes can be visualized on this scan, and it can also be used to determine whether the stomach empties normally and whether any of the labeled milk reaches the lungs, indicating aspiration.

Aspiration of oral feeds is fairly common in infants with CdLS, and may also be diagnosed by a **modified barium swallow**, in which different thicknesses and textures of food and drink are given and detailed images are taken to analyze the phases of the swallow. For children who have moderate to severe reflux, or who do not respond as expected to medical therapy, an upper endoscopy may be indicated. In this study, which is done under anesthesia, a small scope is inserted into the esophagus, stomach and the first part of the small intestine to take pictures and

continued on page 13



A Closer Look: GI and Volvulus

Nirel's Story

We want to thank Nirel's mother, Gayle, for sharing Nirel's story and experiences with us.

Since Nirel was born, she has had problems with reflux and constipation. Her pediatrician prescribed medications to help with the reflux, but Nirel was not a happy baby for the most part because she was so often in pain or discomfort. Taking what we learned from information provided by the CdLS Foundation, we decided to start seeing a pediatric gastroenterologist when Nirel was a few months old. We thought perhaps a specialist would be able to monitor Nirel's GI issues and help us find the right solutions.

When Nirel was three years old, we read about the risk of malrotation in *Reaching Out*. We expressed our concerns to the GI specialist about doing diagnostic testing. However, his reaction was not supportive. He was quite dismissive about the need to rule out malrotation because he felt it was so rare. In addition, he felt that we could go her whole life without knowing she had malrotation, and no problems would arise.

Coincidentally, the Foundation sent families an information letter written by Dr. Tonie Kline about the typical GI issues in individuals with CdLS. The letter was intended to provide information for health care providers (particularly primary care and GI specialists). The letter explained about the increased incidence of malrotation in CdLS and the dangers of volvulus if left untreated. We showed the letter to Nirel's GI specialist and we had to push hard to get him to comply. He finally agreed to a barium swallow test, a simple non-invasive test that only took a few minutes to do. But he made it clear that he was not expecting any abnormal results. He was patronizing, and made us feel like we were just being pushy parents.

The day after the test, we got a call from the GI specialist. The tone of his voice was very different. He gave us the news that Nirel did indeed have malrotation. It was bad news, but also good news that we had an early diagnosis and could do surgery to prevent volvulus. The surgery was called the "Ladd procedure." The GI specialist told us



that surgery was not urgent, but we pushed again and told him that we wanted surgery as soon as possible.

The first surgeon we met with was an experienced general surgeon who had recently moved from a high-profile institution to the children's hospital where we were taking Nirel. He had done many Ladd procedures in the past, and he explained what was involved with the Ladd procedure as an open surgery.

A few days later, the Foundation put us in touch with another family whose son had a Ladd procedure. The father said that the surgery went fine, but he wished he had opted to do it by laparoscopy because the recovery of open surgery was rough on his son.

So, we called the surgeon the next day to ask if we could do the Ladd procedure via laparoscopy. He admitted that he did not know how to do the laparoscopic surgery, and that he only could perform it by open surgery. We told him we wanted to speak with another surgeon in the department who could do the laparoscopic surgery.

The second surgeon was no-nonsense about the benefits of laparoscopic versus open surgery. He made us feel confident in him and his surgical ability, but most importantly, he listened to our concerns.

The surgery went well. Nirel only spent one night in the hospital, and her recovery was smooth. It is reassuring to know that having Nirel undergo the Ladd procedure prevents the complications associated with malrotation. In addition, it is important to us because Nirel still has abdominal discomfort and pain often, and we are able rule out that it is not due to a complication of the malrotation.



GI Medications



By Carol Potter, M.D., associate Professor of Clinical Pediatrics at the Ohio State University and Nationwide Children's Hospital and member of the CdLS Foundation Clinical Advisory Board

Gastrointestinal problems are common in Cornelia de Lange Syndrome (CdLS) and are often treated with a confusing array

of medications. I have broken some of the commonly used medications down by classification to discuss their indications and special considerations.

Medications that block stomach acid production: These medications are used when stomach acid is irritating the esophagus during gastroesophageal reflux. They can also be used when there is acid irritation to the stomach or when stomach acid is leaking out a G-tube site. These medications fall into two categories, H2 blockers and Proton Pump Inhibitors. They produce the same result by different mechanisms

H2 blockers: This class of medications include Famotidine, Cimetidine, Ranitidine, and Nizatidine. They are usually given in two or more doses a day and come in liquid and pill formulations. They are user friendly in that they work well when given with or without food. They may be given in combination with Proton Pump Inhibitors, especially at night.

Proton Pump Inhibitors: This group of medications is more potent than the H2 blockers and include Omeprazole, Lansoprazole, Zegrid, Rabeprazole, Pantoprazole, and Esomeprazole. These medications are not as user friendly in that they don't all come in liquid preparations or preparations meant to be opened and mixed in food. They are more effective when given without food. There is concern that long term use may lead to osteoporosis, making fractures more likely.

Medications for constipation: Constipation is a common problem in CdLS. Commonly used and effective medications include osmotic medications, stimulants and rectally administered medications.

Osmotic medications hold water in the stool making it easier to pass. This prevents hard stools that are painful to pass. There are several formulations which can be mixed with any fluid. They may be taken by mouth or administered down a feeding tube. They do not increase gas production. Examples include Miralax, Colyte, Glycolax, GoLytely, NuLytely, and Trilyte.

Milk of Magnesia is also an osmotic agent for constipation. Care should be given to not administer it with other medications because it may bind them and change drug levels.

Stimulant laxatives: These laxatives increase the muscle contractions in the colon and may increase the urge to have a bowel movement. They are often used in combination with osmotic agents in special needs patients. Senna is a mild stimulant and comes in many forms including liquid, chocolate chews and pills. It can safely be used for long periods of time. Bisacodyl is a more potent stimulant laxative. It comes in the form of small pills which can not be crushed. It also comes as a suppository which can be used every few days to stimulate a bowel movement. It can also be used as a chronic medication.

Rectal medications: A variety of medications for constipation can be given into the rectum.

Minerol oil can be used as an enema to soften hard stools. In general it should not be given by mouth or feeding tube in CdLS because of the chance of aspiration pneumonia.

Saline can also be used as an enema to help pass stools. It can be mixed with one teaspoon of salt for four cups of tap water.

Phosphate enemas should be avoided in CdLS because it may cause severe problems with blood salts.

Glycerin suppositories can provide mild stimulation to have a bowel movement. Bisacodyl suppositories are more potent. Both can be used on a regular basis.



A Closer Look at GI Medication



Jared's Story

We want to thank Jared's mother, Linda, for sharing Jared's story and experiences with us.

Jared was diagnosed with severe GERD when he was a baby. He cried a lot while on the normal formula, and it would come out of his nose on a regular basis. We switched to Augmentin formula, and that seemed to help with the pain a little bit. For many years, Jared

has had an annual upper GI endoscopy. This helped the doctor refine the dosage, and type of medications that seemed to have the best result. Most of the time, the initial look at his stomach was showing no inflammation, but then the biopsies would show some inflammation. At this time, the endoscopy has been eliminated unless symptoms recur.

Since birth Jared has been on various types of medication for GERD. All this time, Jared has always been able to eat by mouth and have normal bowel movements.

The doctor's initially had him on Zantac and Reglan. As he grew, and we changed doctors, Reglan was discontinued since it has some serious long-term side effects, but he remained on GERD medicine. Jared is currently on Nexium once a day and it seems to keep the reflux in check.

We usually knew within days if the medication was working or not. We sometimes give Jared Mylanta if he seems uncomfortable, and although the doctor didn't think that really had much of an effect, it seem to help Jared.

We have had a few bouts of instability throughout the years with inexplicable pain. One such time was when Jared was approximately seven years old. The doctor

did some research and decided to try Jared on Carafate, thinking that it might actually be pain from bile vs. acid. It really worked. We used that for about a year and a half, until we slowly discontinued using it altogether and haven't had any painful periods since.

There were some positive effects that come with these medications. Even though Jared's food must be pureed for him to eat, he pretty much can eat anything. We avoid spicy or tomato-based foods as a precaution but he seems healthy and happy. He loves to eat and drink (don't we all!).

We have been lucky that he seemed to respond over the years with minor adjustments. The early years were the hardest with formula.

The CdLS Foundation Goes Social

Find us online on these social media outlets! Connect with friends around the country, as well as the Foundation and its staff.



Twitter - [Twitter.com/CdLSFoundation](https://twitter.com/CdLSFoundation)



Facebook – [Facebook.com/CdLSFoundation](https://facebook.com/CdLSFoundation)



LinkedIn - www.linkedin.com/pub/cornelia-de-lange/64/764/604/



Pinterest – [Pinterest.com/CdLSFoundation](https://pinterest.com/CdLSFoundation)



Youtube–www.youtube.com/user/CdLSFoundation



Instagram – [Instagram.com/CdLSfoundation](https://instagram.com/CdLSfoundation)



Super Siblings: Sisters Making a Difference

Madeline

Madeline, (pictured right with brother, Aiden) who is 12 years old and lives in Connecticut, has been selling handmade rock magnets at craft fairs and flea markets this year to donate a portion of proceeds to the CdLS Foundation. Her brother, Aiden, has CdLS.



“During a trip to Maine, Maddy began painting on rocks and creating magnets out of them,” said Carol, Maddy’s mom. “We joked around calling them ‘Maddy’s Magnets’ and we all thought, ‘that’s actually a really neat idea.’”

Maddy had bookmarks and information on CdLS on the table where she was selling the magnets as well, in order to raise awareness for CdLS. Her magnets are currently sold out, as they were such a hit.

Maddy raised \$80 for the Foundation.

Madison

Madison’s teacher gave each student in her class \$2 and encouraged them to use the money to help make the world a better place. Maddy, (pictured below with sister,



Isabel) who lives in Michigan, decided that she wanted to raise money for the CdLS Foundation because her sister, Isabelle, has CdLS. She also felt it would be a good way to help raise awareness about the syndrome,

because there are still so many people who have never heard of it.

Maddy took her \$2 and created two prizes (a picture frame containing pictures of Isabelle, and a handmade birdhouse). She then posted on her mother’s Facebook page, asking people to help her raise money for the Foundation by sending her donations. Every person who donated had their name placed in a drawing to win one of her prizes.

In total, Maddy was able to raise \$527.50, which she donated to the Foundation in honor of her sister.

Sophia

Californian Sophia (pictured right) made a difference in her 4th grade classroom, by telling her classmates about her sister Isabel, who had CdLS.



“My sister Isabel had CdLS, and the Foundation sent us a book about it for kids. I wanted to bring it to my school for Rare Disease Day,” said Sophia. “One of the fourth grade teachers at my school, has a daughter who has a rare disease called AT (ataxia telangiectasia) and so I thought it would be a good idea to bring in the book about CdLS.”

Sophia made a poster card and brought it into school that morning. At recess she and some friends got all of the fourth graders, some of the coaches and other fourth grade teachers to sign it. They then gave it to the teacher whose daughter has AT.

“We went into her classroom and all the other kids in her class knew what was going on,” said Sophia. “When she saw the card she started to cry, and said she would show it to her daughter.”



Mailbag

- Declan -

Our little angel was born June 28, 2012, unexpectedly, by emergency C-section five weeks early due to small measurements and lack of amniotic fluid. At birth we discovered that he was missing two fingers on his left hand and on his right, two fingers were fused together. As far as I could tell he was perfect, but a very keen neonatologist in the NICU consulted with a geneticist in his group and through pictures they determined that he had CdLS. While it was a lot to take in at three days old, I am very grateful we found out right away so we could begin therapies and be aware of what to watch for.



At two months old while at the GI doctor for severe GERD, we were referred for further testing and learned that he had intestinal malrotation that required immediate surgery. During the hospital stay for that they found that he aspirated when swallowing and so is now being fed through a G-tube. We are very hopeful that this is temporary. He is due for several other surgeries in the coming months--one to correct his ptosis, one to separate the fused fingers and a third to correct his hypospadias.

While this has all been very overwhelming and sometimes almost unbearable, every time I hold him or even now as I look at him while writing this, I am amazed by this perfect, wonderful angel.

Now at the ripe old age of 40 weeks, I can already tell that he will do great things! His therapists say he is a fighter and so strong. I would encourage anyone with a new diagnosis to seek services early. I look forward to the future with a mixture of excitement and fear. Excitement, because I know he is capable of so much good, and I can't

wait to see all the goodness he will bring into this world. Fear, just of the unknown.

The times when he is made fun of, or told he can't do something will be the hardest, I think. But our family will be there to support him through it. We have been so blessed in this short 40 weeks that we have had Declan--we are lucky to have him in our lives! He is growing well, meeting milestones and amazing everyone along the way. It is impossible not to fall in love with him when you meet him.

Jenni, Declan's mom, AZ

Submit your Mailbag or Super Siblings Story!

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

WELCOME NEW FAMILIES

Arkansas

William and son Maxx, born
November 19, 2006

Oregon

Eva born May 28, 1970

California

Ramona and Jose and son Joshua,
born January 27, 2011

Pennsylvania

Amy and Mark and daughter
Olivia, born June 4, 2008

Illinois

Diana and son Iziah,
born July 30, 2011

Texas

Micheala and daughter Addison,
born November 15, 2012

Michigan

Stacy and daughter Ellie, born
December 1, 2012

Utah

Miriam and Allan and daughter
Harper, born June 22, 2011

New York

Barbara and son Khaif, born
January 14, 2013

Washington

Shana and Dominick and son
Dane, born December 27, 2012

Anna and son Logan,
born February 24, 2013

Heather and daughter Chloe,
born February 8, 2008

North Carolina

Angeles and daughter Ashley,
born November 26, 2003

West Virginia

Barbara and Stephen and grand-
son Ezekiel, born October 7, 2011

Ohio

Patricia and David and daughter
Kimberly, born April 3, 2012



Event Highlight: de Lange Society

The de Lange Society, named after the doctor who cared so much about children with the syndrome, publicly recognizes individuals and volunteer groups, who, like Dr. Cornelia de Lange, lead the way for others. Read about the first ever de Lange Society event on page 2.



2013 de Lange Society Inductees with Executive Director Marie Malloy



Inductees Eileen Ahearn and Laird Jackson being pinned.



Inductees and guests enjoying the presentation.

2013 CALENDAR

May 20
21st Annual New England
Golf Tournament
Ipswich Country Club
Ipswich, MA

June 8
Kristian's Bowl
Madisonville, KY

Dodge Balls for Camden
Edgerton Elementary
Edgerton, KS

June 22
Concert for CdLS
Sami Center, Spirit Lake, IA

June 23
Merrell Down & Dirty National
Mud Run Series
Hartford, CT

July 13
Lapel Village Fair CdLS
Benefit 5K
Lapel, IN

July 14
Merrell Down & Dirty National
Mud Run Series
Philadelphia, PA

September 7
West Region Family Gathering
Oakley, CA

September 15
Saratoga Palio 5K and
half marathon
Saratoga Springs, NY

September 16
25th Annual Madison
County Wood Products Golf
Tournament
St Louis, MO

September 28
4th Annual Brew & BBQ
Sweetwater Brewery
Atlanta, GA

Maddy's Run 5K walk/run
Salt Lake City, UT

Southwest Region
Family Gathering
Harvey, LA

October 5
Manhattan Beach 10K
Manhattan Beach, CA

For more scheduled events visit calendar.cdlsusa.org

Kennedi's Bowl

On April 21st, Kenya Ballard hosted the Faith-Hope-Charity Bowl in honor of her seven year old daughter, Kennedy. Over 60 bowlers came out to Collins Bowling Center in Lexington, KY, to support the CdLS Foundation. Kenya said attending conference last year inspired her to hold this event. "Conference meant the world to me," she said. "Knowing what the CdLS Foundation does for families, I wanted to do something for them. I'm excited and already looking forward to next year."





Plant Seeds of Hope with Cornelia's Garden

Fill your garden with beautiful wildflowers that represent the hope the CdLS Foundation gives to people with CdLS and their families. Request your Cornelia's Garden kit, complete with the seeds and the materials that you need to ask 10 people to plant wildflowers in honor of a person with CdLS. They will be rewarded not just by the beauty of the flowers, but also by knowing that they helped someone with CdLS grow and bloom.



For more information, or to request a Cornelia's Garden kit, contact Kellie at specialevents@cdlsusa.org.

*Our children are like wildflowers
Each unique in their own way
They share with you their beauty
Giving joy to you each day...*

Reaching Out in your Community

- 1) Are you familiar with these clubs and organizations in your community?
- 2) Do you or someone you know belong to one of these clubs and organizations?
- 3) Would you be willing to speak at their meetings?

If you can answer yes to one or more of these questions, please contact Kellie at specialevents@CdLSusa.org the CdLS Foundation to see how you can raise awareness in your community through these groups and others.



Diary of a Yard Sale Enthusiast

Every year, I say this is my last one. It never is...

Gathering other people's clutter to organize a yard sale to benefit the CdLS Foundation is an obsession for me, like morning coffee, walking on my treadmill and "American Idol." Now, my friends and co-workers expect my annual requests to donate a box (or three) for the sale. My husband looks forward to buying back his "treasures" that I have donated (without his permission). My sister anticipates my ask for help.



I've made it an annual event. Why? It is the only support I can give the Foundation for the services they provide families faced with the challenges of CdLS. My efforts and time put toward organizing a yard sale seem minimal compared to the impact the funds raised have in other's lives. Amazingly, over the last four years, I have raised \$2,000. (Pretty impressive for a person hoarding other people's clutter.)

Join me, and others nationwide, by contacting the Foundation for your "Yard Sale Across America" Kit that has everything you need, including stickers (yes, you should price your items) to tag the day for CdLS. Contact specialevents@cdlsusa.org to learn more.

My words of advice: Always have standards for the "clutter" you collect for your yard sale. Personally, I: 1) don't take clothes; 2) will pick up furniture; 3) will donate it to Goodwill if I cannot sell it.

Thank you and happy sales.

-Anonymous

Contact the Foundation for your "Yard Sale Across America" kit, which has everything you need, including tips on how to make your sale a success. To get started, contact Kellie at specialevents@cdlsusa.org.



Research Participants Needed: Association Between Brain MRI Findings and Behaviour in Clinically Diagnosed CdLS Patients.

Principal Investigator: Antonie D. Kline, M.D., Director of Paediatric Genetics, Greater Baltimore Medical Center and Medical Director of the Cornelia de Lange Syndrome Foundation

Co-Principal Investigators: Tamanna Ratti M.B., Ch.B., Paediatric Resident, Children's Hospital at Sinai Hospital of Baltimore

This study will investigate central nervous system findings on brain MRIs and compare them to behavioural aspects (such as aggression, mood disorders, self-injury, sleep disturbances) of patients with CdLS. The study will consist of parents answering a behavioural questionnaire (via phone or face to face) and signing a release for us to obtain a copy of previously obtained brain MRI scans and medical records. Medical history and physical exam findings will also be compared to findings on MRI.

Similar studies have been carried out in other developmental syndromes including Down syndrome and Fragile X syndrome. If characteristic trends can be found in CdLS, specific medical recommendations may be able to be made, preventative measures taken, and complications avoided, or, at the least, prospective information given to families.

If you or your child are interested in participating, please email cdlsmri@gmail.com for more information. Your participation would be greatly appreciated.

National CdLS Awareness Day 2013



Raising awareness of CdLS benefits so many: it engages communities, helps families around the country, and represents those with CdLS, some of whom still remain undiagnosed.

We are so thankful for your efforts and hope to highlight them in the coming months. Send your National CdLS Awareness Day photos and stories to Brenda, at bshepard@CdLSusa.org. Together, we can make a difference in the lives of those touched by CdLS.



Help us Stay in Touch

Have a new email address? The CdLS Foundation is looking to update our records and we need your help. Send us your updated email addresses so that we can stay in touch.

Please respond to info@CdLSusa.org with your current email address and any other updated information you may have.



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The Importance of GI Testing con't.

obtain biopsy specimens. Biopsies of the esophagus may reveal inflammatory changes associated with GE reflux.

In some patients with CdLS, we have found large numbers of allergic cells (eosinophils) in the esophagus, consistent with a diagnosis of eosinophilic esophagitis (EoE). This condition may require food allergy testing and treatment with dietary changes. EoE is becoming more common in the general population, so it is not clear whether it is specifically associated with CdLS. The most severe findings on upper endoscopy include Barrett's esophagus, a pre-malignant condition of the esophagus, or adenocarcinoma (esophageal cancer). With increasing awareness of GE reflux in CdLS, we hope that most patients will be treated aggressively to prevent the development of these late complications.

Monthly Giving: The Easiest Way to Support the Foundation

Monthly giving allows the CdLS Foundation to budget more efficiently to meet the needs of those we serve, as well as plan for the future. It also benefits you. How?

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For as little as \$20 per month, you can make a difference in the lives of many.

To discuss a monthly donation to the CdLS Foundation, contact Gail at 800.753.2357 or events@CdLSusa.org.

Publicaciones Nuevas En Español

Many materials are available in Spanish on the CdLS Foundation Web site.

Visit www.CdLSusa.org/what-is-cdls/cdls-en-espanol.htm to view a full list of translated materials.

Visítenos en nuestro sitio web www.CdLSusa.org/what-is-cdls/cdls-en-espanol.htm para obtener publicaciones nuevas en español sobre el síndrome Cornelia de Lange.



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Our Mission

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



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Anthony L. Ramirez

May 8, 1987 – February 7, 2013

Son of Diahann Ramirez

Grandson to Mr. and Mrs. John Ramirez

447 S San Jose Ave

Covina, CA 91723-3056

Andrew Perales

January 18, 2010 – March 16, 2013

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