Reaching Out

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

Photo by Rich Guidotti, Positive Exposure

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Director's Message

2009 in Review

When I came on board in early 2008, my initial plan was to "maintain momentum" for a brief period as I grew into my leadership role at the Foundation. However, the economic downturn that began in 2008—and continues today—made that a staying theme. My big plans for new programs and services were put on hold as we focused on maintaining current offerings.

Although we closed 2008 with a deficit, and expect 2009 to end similarly, the Foundation maintained all services, funded its first research fellow (read about her findings on page 5), and continued to be available personally to answer questions from families and professionals.

Here are a few highlights from 2009:

To date, family service coordinators welcomed nearly 70 new families, held five family gatherings across the country, and facilitated more than 900 *Ask the Doctor* questions.

A long-time volunteer translated nine CdLS-related articles into Spanish; and we provided information to more than 300 health and education professionals working with people with the syndrome.

In the arena of public affairs, the Foundation scored big by securing Centers for Disease Control (CDC) funding of $238,000—money we will be able to tap into this fall and some of which will support the national family conference next summer.

Fundraisers *Leaps of Love*, *Yard Sale Across America* and *Vines, Wines & Meaningful Times* were launched with great success. Our biggest fundraising machine—*Team CdLS*—made its debut in marathons in San Diego and Manhattan Beach, CA. The cumulative effect of these new events was more than $60,000 that was not in the original 2009 budget.

The money from the CDC and new fundraisers helps partially offset the decline in donations occurring with the economic crisis. But, even with the significant efforts by staff and volunteers, 2009 will see a financial shortfall.

However, we are hopeful 2010 finds us on less shaky ground, allowing us to continue "maintaining momentum" and, with some luck, building on this momentum for the future of all people affected by CdLS.

Sincerely,

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

director@**CdLS**usa.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.

This Issue

CdLS Research Moving Forward in Great Strides

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

A number of Cornelia de Lange Syndrome (CdLS) research efforts have been underway for quite a few years, funded in part by the CdLS Foundation. Several have been in the basic sciences—what most people consider purely laboratory research, but which, in actuality, are extremely relevant to human disease and disorders and are very exciting.

Once patterns, or results, are found in animal models (such as mice, fruit flies or zebrafish), conclusions can be drawn for humans, and treatment trials can be initiated. This has come to fruition for Dale Dorsett, Ph.D., who recently received federal funding to test drugs approved for human use on fruit flies and yeast (see article on page 4).

Also recently, the developmental biology group at the University of California, Irvine—led by the husband and wife team of Arthur Lander, M.D., Ph.D. and Anne Calof, Ph.D.—has just published an article* on their mouse model of CdLS. Their research is another step forward in understanding how and why CdLS happens.

We know that a majority of the cases of CdLS are due to a mutation of one copy of the *NIPBL* gene, the product of which regulates a complex of chromosomal proteins called cohesin. What we don't know is how reduction of *NIPBL* gene function gives rise to developmental issues in CdLS.

A mouse model of CdLS was developed to help find this answer. (The model was created by generating mice with a deficiency of the *NIPBL* product.) These mice showed a remarkable similarity to in individuals with CdLS, including small stature, craniofacial abnormalities, hearing loss, reduced body fat, behavioral disturbances, and high prenatal mortality.

Molecular analysis of tissues and cells from the mouse models provided the first evidence that the major role of *NIPBL* in the cause of CdLS is to exert modest, but significant, effects on the expression of certain genes. Among these genes is a set involved in the development cells that make and accumulate body fat—potentially explaining reductions in body fat accumulation often seen in individuals with CdLS. Further work with the mouse model is ongoing to assess this and other aspects of the *NIPBL* changes.

In my own research on aging, we continue to evaluate older individuals with CdLS through the CdLS Multi-disciplinary Clinic for Adolescents and Adults, held each spring and fall at Greater Baltimore (MD) Medical Center.

We found there is increased incidence of snoring, and a risk for obstructive sleep apnea. This is being further assessed in a sleep study based at Johns Hopkins Hospital, under the direction of Stacey Ishman, M.D., a pediatric ear-nose-throat specialist.

Additionally, in testing smell ability, 93 percent of individuals were found to have an intact sense of smell. The more severely involved an individual is, the more likely their sensory system is affected.

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Reaching Out
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All information contained herein is for the reader's personal interest. Articles on treatments, medications, or procedures, etc. are not guides for self-treatment. Questions should be discussed with your doctor or other appropriate professionals.

Researchers to Screen Drugs for Treating CdLS

Dale Dorsett, Ph.D., a member of the Foundation's Board of Directors and head of its Research Council, received federal funding to search for drugs that could be used to treat CdLS in the future.

The research is a collaborative effort between Dr. Dorsett's laboratory at Saint Louis University School of Medicine and Dr. Justin Fay's laboratory at Washington University School of Medicine, also in Saint Louis. Researchers will use yeast and fruit flies to test nearly 1,500 drugs that are already approved for human use.

"Since individuals with CdLS have only small reductions in the activities of the *NIPBL* or *SMC1A* genes, the hope is that some existing drugs could boost the remaining activities, alleviating growth and health deficits often seen in people with the syndrome," says Dr. Dorsett.

Yeast and fruit flies with gene changes similar to those that cause CdLS will be treated with various doses of the drugs. Researchers will then look for corrections of deficits in growth, development and/or gene expression. If these tests identify drugs of potential benefit, the next step would be testing the drug(s) on mouse models of CdLS.

"If these drugs also prove beneficial in mice, then clinicians can begin to carefully consider how they might be used to treat individuals with CdLS," says Dr. Dorsett.

The funding is a two-year supplement of $335,925 and is part of the American Recovery and Reinvestment Act of 2009. It supplements an existing National Institutes of Health research grant that supports research into the molecular mechanisms that underlie CdLS.

Why study fruit flies?

Genetically speaking, people and fruit flies (also known as Drosophila melanogaster) are a lot alike. About 75 percent of known human disease genes have a recognizable match in the genetic code of fruit flies, making them good substitutes for people. Additionally, the entire genome of the fruit fly has been mapped.

Fruit flies reproduce quickly and their lifespans are relatively short, so many generations of a trait can

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Study Shows Parents of Children With CdLS Experience High Levels of Stress

By David Richman, Ph.D., CdLS Foundation Clinical Advisory Board, University of Illinois, Champaign

Dr. Richman

My research team recently completed a study of parenting stress made possible by a grant from the CdLS Foundation and numerous parents of individuals with CdLS and Down Syndrome (DS).

Results from our study showed that parents of children with CdLS experienced significantly higher levels of parenting stress than parents of individuals with DS. We found that individuals with CdLS exhibit greater levels of the following problematic behaviors compared to individuals with DS: (a) self-injurious behavior, (b) challenging behavior, such as noncompliance and withdrawal, and (c) autism-like characteristics.

In fact, we found that 52% of individuals with CdLS scored in the "autism very likely" category, compared to 17% of individuals with DS. Of equal importance, 61% of individuals with DS scored in the "autism unlikely" range, compared with 28% of those with CdLS. Additional research is needed to identify whether individuals with CdLS frequently exhibit behaviors that are similar to characteristics of autism, but may not be sufficient to result in an additional diagnosis of autism.

Parents of children with CdLS said that managing their child's problematic behaviors was the most common source of stress. Forty percent of the parents of children with CdLS in our sample (vs. 22% for DS) were above the 95th percentile for Total Stress—meaning they were off the measurement scale in regards to parenting stress.

Future studies need to replicate these findings because different sources of parenting stress may be suggestive of different approaches to preventing or intervening early to decrease the stress. If the finding is replicated, one strategy may be to implement intense early intervention for the challenging behaviors frequently seen in CdLS.

Common Research Terms

Cell: The basic unit of any living organism. It is a small, watery compartment filled with chemicals and a complete copy of the organism's genome.

Chromosome: One of the threadlike "packages" of genes and other DNA in the nucleus of a cell. Humans have 23 pairs of chromosomes. Each parent contributes one chromosome to each pair, so children get half of their chromosomes from their mother and half from their father.

Cohesin: A protein essential for normal development of an embryo.

Deoxyribonucleic acid (DNA): The chemical inside the nucleus of a cell that carries the genetic instructions for making living organisms.

Gene: The functional and physical unit of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein. A mutation in the *NIPBL*, *SMC1A* or *SMC3* gene results in CdLS.

Genotype: The genetic identity of an individual.

Mutation: A permanent structural alteration in DNA. These changes can either have no effect or cause harm.

Neurotransmitter: Chemicals that cells of the nervous system use to communicate with each other and with other cells of the body.

Phenotype: The observable traits or characteristics of an organism, for example hair or eye color, weight, or the presence or absence of a disease.

Psychotropics: Medications that act primarily upon the central nervous system where they alter brain function, resulting in temporary changes in perception, mood, consciousness and behavior. They include anti-depressants and anti-anxiety drugs.

Translocation: Breakage and removal of a large segment of DNA from one chromosome, followed by the segment's attachment to a different chromosome.

Fellowship Bears Fruit


Dr. Liu

The first-ever CdLS Foundation Research Fellowship ended its first year with great success, with fellow Jinglan Liu, Ph.D., fulfilling her research goals and publishing her findings in the highly respected professional journals, *PLoS Biology* and *Human Mutation.*

While at the Children's Hospital of Philadelphia, Dr. Liu performed studies to identify how changes in two genes responsible for CdLS (*NIPBL* and *SMC1A*) affect the function of other genes. *NIPBL* and *SMC1A* make proteins that are components of cohesin, a protein complex that is important for the structure and function of chromosomes. The current idea is that cohesin controls many genes needed for proper development of the human body.

With members of Dr. Ian Krantz's lab, Dr. Liu discovered that cohesin does indeed control many other genes, and that when cohesin is altered in CdLS, many genes behave differently. Dr. Liu's study supports the idea that such changes in gene regulation may underlie the various developmental changes in CdLS, including missing upper limbs, impaired mental development, cleft palate, and heart defects.

What does all this mean for people with CdLS? Better knowledge of how these genes disturb normal development may enable researchers to design better diagnostic tests for CdLS and also provide targets for eventual treatments.

Dr. Liu was unable to continue the two-year fellowship because she recently moved to a faculty position at Washington University at Saint Louis (MO), where she also serves as associate director of its CytoGenomic Laboratory. She plans to continue her work on developing diagnostic methods for CdLS.

"Dr. Liu's hard work and dedication to her research project in Dr. Krantz's lab paved the way for increased understanding of CdLS," says Liana Fresher, executive director of the Foundation. "We look forward to watching her career, and continued study of CdLS, flourish."

Behavioral Research Update

By Dr. Marco Grados, M.D., M.P.H., CdLS Foundation Clinical Advisory Board, Johns Hopkins University School of Medicine


Dr. Grados

Behavioral difficulties in children and adults with CdLS remain an area of much-needed research and service. Systematic data on the type of behaviors and effective interventions are sorely needed.

Research on the behavioral phenotype of CdLS—much of which has been funded by the Foundation's Small Grants Program—has progressed in the last two years. My research group has submitted for publication its first study on an initial group of 41 children with CdLS (there are now a total of 80 children enrolled in our studies). We report on the relation between physical features (namely limb development) in CdLS and autistic features (lack of language, repetitive behaviors). Our observations have led to the development of research questions in relation to developmental genes and their relation to autistic features in children with CdLS.

Other areas of study are repetitive behaviors in CdLS and the effect of aging on behaviors. We've observed that some individuals with CdLS experience a decline in function in teenage years, with severe behavioral and mood disturbances. We aim to better understand predictors and possible interventions to improve outcome in these children.

Additionally, we are now offering a survey on the use of psychotropic medications for individuals with CdLS. This survey will provide a much-needed database and comprehensive understanding of how medication helps manage behavior. Investigating the types of psychotropic medications that are useful may also provide clues as to which preferential neurotransmitter pathways need to be targeted in CdLS.

To inquire about any aspect of the research, including participation in studies, contact Dr. Grados at 443-287-2291 or mjgrados@jhmi.edu.

Dental Procedures and Anesthesia in Patients with CdLS

By Ellen Alpano, D.D.S., University of Maryland Dental School

To date, there has been limited research on the dental attributes and treatment common in CdLS. Oral manifestations, such as cleft palate and widely spaced teeth, and complications from anesthesia have been reported, but very little research has been done.

Through provider surveys, dental and hospital records, the goal of this study—funded by the Foundation's Small Grants Program—was to further examine dental conditions and treatment, as well as dentist perception of treating individuals with CdLS.

Study participants were recruited at the CdLS Foundation 2008 conference, through the Foundation's email list, and during the Multi-disciplinary Clinic for Adolescents and Adults at Greater Baltimore Medical Center.

A survey about dental condition, treatment and provider perception when treating patients with CdLS versus other patients with special needs was mailed to 51 participants' dentists, of which almost half responded. Request for dental and hospital records were also made, of which 28 and 12 were returned, respectively. Results were as follows:

According to the dental provider survey, there was no difference in extraction of teeth due to cavities, tolerance of advanced dental procedures or access to care compared to other patients with special needs; however, patients with CdLS had more malocclusion (misalignment) and parents were present more often during dental treatment.

Dental records showed that patients with CdLS were treated under general anesthesia more frequently compared to in-office sedations and use of restraints. Also, patients with CdLS had more extractions of permanent teeth compared to primary teeth. These trends may be useful for dental providers who have not treated a patient with the syndrome.

Based on the hospital records, nearly 80% of patients who received the sedative Versed prior to a procedure had an adverse post-operative event. Of those who did not receive Versed, 40% had an adverse post-operative event. Based on this information, an alternative pre-medication may be a better choice for patients with CdLS.

The Support Group Experience: Perspectives from Parents of Children with CdLS

By Adrianne L. Baxter, M.S., University of Utah


A. Baxter

The purpose of the study was to explore parents' unique support group experiences. Seventy parents responded to an email invitation to participate in the survey; fifty surveys were completed.

About half of the reported experiences were regarding groups specific to the CdLS diagnosis. The other support groups parents participated with included groups through their child's early intervention or school program, feature-specific groups (such as feeding issues, autism or other unique aspects of their child's condition), or generalized groups for families of children with special health care needs.

There was not a significant difference in the overall experiences of groups that were specifically for CdLS and those that were not. Parents found the majority of both met their expectations and were helpful.

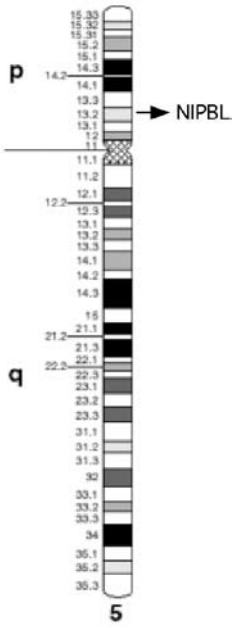
Nearly three-fourths of support group experiences reported had an Internet-based component. With the use of this technology, constraints of time and location are removed, allowing to access support groups as needed. Parents did not report a significant difference in the overall experience of online support groups as compared to in person.

These findings suggest that parents of children with more rare conditions, like CdLS, can benefit from a variety of support group experiences. Internet-based groups can be diagnosis-specific, like the parent-run **CdLS Kids** (http://groups.yahoo.com/group/cdls-kids), or focused on a particular feature or challenge. Local in-person groups often have a more general focus on children with special health care needs rather than the specific diagnosis. As one parent expressed, these groups are great because, despite differences, parents have shared goals and desires for their children.

Selected *NIPBL*-Related Findings at the University of Chicago Genetic Services Lab

By Melissa Dempsey, M.S., CdLS Foundation Clinical Advisory Board, Eden Haverfield, Ph.D., and Soma Das, Ph.D.

The University of Chicago Genetic Services Laboratory offers testing for more than 40 rare conditions, including CdLS. Our lab performs sequencing of the *NIPBL* and *SMC1A* genes, along with deletion/duplication testing for *NIPBL*. Since 2006, we've tested more than 450 patients for CdLS. Although our primary focus is on clinical testing, we've conducted several projects to improve diagnostic methods and mutation detection rates.



Small deletions (missing part of a gene) or duplications (part of a gene repeated) within the *NIPBL* gene are thought to cause CdLS, although very few have been described to date. Our laboratory has identified six deletions and one duplication in 208 individuals tested by *NIPBL* deletion/duplication analysis—a detection rate of 3.4%. Our findings highlight that small deletions/duplications in *NIPBL* may be more common than originally thought, and represent a significant cause of CdLS.

We have also found some interesting trends in prenatal testing for CdLS. Patients with CdLS have several findings that can be detected during pregnancy, including upper limb defects, growth delay, micrognathia (small jaw), and increased nuchal (skin fold behind the neck) translucency detected by ultrasound in the first trimester.

Confirmation of gene changes prenatally has rarely been reported. Our lab performed *NIPBL* testing on seven fetuses and identified gene changes in six of them. Five of the *NIPBL* mutation-positive cases with clinical information available had severe upper limb defects, while four had micrognathia noted on ultrasound, and three had increased nuchal translucency measurements.

NIPBL testing revealed that four of the five had severe mutations with an obvious effect on the protein. These findings demonstrate that fetuses with significant ultrasound findings consistent with CdLS are likely to have *NIPBL* mutations.

Thrombocytopenia in CdLS

By Michele Lambert, M.D., Children's Hospital of Philadelphia


Dr. Lambert

Thrombocytopenia—or, in layman's terms, low blood platelet count—was first reported in patients with CdLS in 1993. Despite these reports, it's an under-recognized manifestation of the syndrome. Our study aims to further understand the reasons for the increased risk of thrombocytopenia in individuals with CdLS.

At the Children's Hospital of Philadelphia (CHOP), we have information on 1,740 patients with a definite or probable diagnosis of CdLS. Of these children, 43 have platelet count data available.

When we looked at the data from these 43 patients, we found that 14 (one-third) had thrombocytopenia, seven of which had presumed Immune Thrombocytopenia (ITP), a disease in which the immune system destroys platelets. The severity of the ITP varied from mild to one case of spontaneous bleeding in the patient's head.

Even if we assume that the other 1,697 individuals without platelet count data have normal counts, this is a significantly higher proportion than expected given that ITP occurs in 5 to 13 per 100,000 persons.

This study—funded by the CdLS Foundation's Small Grants Program—is ongoing. Currently, we are evaluating available bone marrow specimens and blood samples to further understand the reasons for the increased risk of ITP.

If you or your child have a low platelet count and would like to participate in the study, please contact : Dr. Michele Lambert, Children's Hospital of Philadelphia, 3400 Civic Center Blvd., Division of Hematology, Philadelphia, PA 19104 or 215-590-4667.

See page 13 for information on Thrombocytopenia

Support Service Needs of Adult Siblings of Persons with CdLS

This study evaluates the use and needs of support services for adult siblings of persons with CdLS. The purpose of the study is two-fold: 1) to understand the support needs and life experiences of these adult siblings, and 2) to assist service providers and organizations like the CdLS Foundation develop and implement services for this population.

Participants must be at least 23 years of age and have a sibling with CdLS. Multiple siblings from the same family are welcome to each complete a survey. The survey takes approximately 30 minutes to complete and is available online at http://tiny.cc/cdlssibs. Participation is voluntary, completely anonymous, and greatly appreciated. The survey is open from November 1 to December 31, 2009.

The study is conducted by Veronique Weinstein, a genetic counseling student at the University of Maryland. E-mail vwein001@umaryland.edu with questions or concerns.

Educational Research Opportunity for Parents with Children Ages Birth to 21

The University of Connecticut's Center for Developmental Disabilities (UCEDD) invites parents to participate in a survey to share their perspectives on their child's experiences. Topics covered include therapies, social interactions, and the educational system.

With information gained from this survey, researchers hope to develop educational guidelines for parents, as well as professionals working in the school arena. This project is a follow-up to a broader survey in 2007, which saw overwhelming support from parents.

The survey can be accessed online at http://www.surveymonkey.com/s.aspx?sm=556cwt76DDNZ9Tt2rbVFLg_3d_3d; for a paper copy, contact Cristina Wilson, Ph.D., at 860-679-1500 or cmwilson@uchc.edu.

Questions regarding this study may be directed to the Principal Investigator, Mary Beth Bruder, Ph.D., 860-679-1500. Questions regarding the rights of research subjects should be directed to an IRB Representative at (860) 679-8729 or (860) 679-3054.

Conference 2010: Living & Learning in Dallas

The countdown to theCdLS Foundation's National Conference is on, and planning is in full-swing. The biennial event takes place June 24 – 27, in Dallas, TX. This year's theme is *Living & Learning for a Lifetime;* workshops and sessions will focus on the issues and challenges individuals with CdLS face throughout their lives.

You can support conference now by contributing to the 2010 Conference Fund or participating in a letter-writing campaign that asks friends and relatives to support the event. Call 800-753-2357 or access materials online at www.CdLSusa.org. Go to the **Be Connected** tab, then click **Attend a Conference**.

Coming in late January is the Shining Stars program, which lets friends and relatives buy stars for children with CdLS and write a personal message on them. The stars will be displayed at conference. Look for details on this program and the conference in early 2010.

A limited number of first-time conference attendee scholarships are available. Application information is in your registration packet, available in February.

Leap for those who can't

You can help children who can't always leap themselves by participating in the CdLS Foundation's second annual *Leaps of Love.*

This Valentine's Day-inspired educational program leaps into child care centers and elementary schools across the country in February 2010. Participants receive supplies to make heart-shaped cards for a child with CdLS and hear a story about Emma, a little girl with CdLS.

Children raise money by collecting pledges for each leap they do in a minute. Prizes are awarded to all "leapers" raising more than $25, and the school or child care center raising the most wins a $250 gift card to Barnes & Noble.

Ready to leap? Contact outreach@CdLSusa.org or call 800-753-2357 for more information or to sign up.

WELCOME NEW FAMILIES

Arkansas
Margaret and Billy
and daughter Tesla,
born May 8, 1999

California
Christina and Jacob
and daughter Eva,
born March 3, 2009

Florida
Lawan and son Antonio,
born February 16, 2007

Massachusetts
Sue and Louis
and daughter Jaclyn,
born January 7, 1985

Michigan
Stacy and Zeb
and daughter Abby,
born May 18, 2006

Minnesota
Kelly and Darin
and daughter Marrianna,
born August 4, 2009

New York
Letticia and Arthur
and son Santio,
born February 10, 2009

Kires and Leonard
and daughter Glori,
born April 19, 2006

North Carolina
Barbara and
daughter Patience,
born June 4, 2009

Raelynn and Jim
and son Alexander,
born February 21, 1999

Virginia
Marianne and Michael
and granddaughter Sophie,
born October 31, 2006

2009 CdLS Events

For the Love of it- CdLS Awareness 5k Run/Walk
Decatur, GA

11th Annual Skating Party Benefit
Warwick, RI

Leaps of Love
Nationwide

Basket Bingo,
Wind Gap, PA

CdLS Bake Sale
Decatur, TX

Pennies for Jessica
Nationwide

"You Light Up My Life" Dinner Dance
Slate Hill, NY

CdLS Awareness Night at PNC Park
Pittsburgh, PA

New England Golf Tournament
Georgetown, MA

Hoof it for Hunter- 2 Mile Fun Walk
Seabrook, NH

Yard Sale Across America
Nationwide

10th Annual Lapel Village (IN) Fair CdLS Run/Walk

11th Annual IL Golf Tournament
Wheeling, IL

Team CdLS- 32nd America's Finest City Half Marathon and 5K
San Diego, CA

21st Annual Madison County Wood Products Golf Tournament
Wood River, IL

Vines, Wines and Meaningful Times
Coventry, CT

Team CdLS- Manhattan Beach 10K
Manhattan Beach, CA

ING Hartford Half Marathon
Hartford, CT

Walk for Will & Hope
Shelby Township, MI

Team CdLS- Bank of America Chicago Marathon
Chicago, IL

5th Annual CdLS Dinner Dance
Montgomeryville, PA

Sarah's Bowl for CdLS
Batesville, IN

… and numerous other events that help
the Foundation make a difference in so many lives.

Don't Look the Other Way

By Lynn Audette, M.S.W, CdLS Foundation Family Service Coordinator

Have you ever noticed a child you think may have CdLS, but weren't sure if you should—or even how to—approach the parent?

If you feel brave enough, and the parent doesn't look too harried, try to establish eye contact, smile and open with something like: "What beautiful hair your daughter has," or, "Your child reminds me a lot of my son, who has CdLS." Then give examples of the visible similarities, such as long curly eyelashes, connecting eyebrows or small hands and feet. This is most easily done when your own child is with you.

If the parent is receptive, she may ask questions about what CdLS is. She may even say her child also has CdLS and she's thrilled that someone else is familiar with the syndrome. Best of all, your bravery may present the parent with a new idea to explore for an undiagnosed child. Having a CdLS Foundation awareness card or bookmark (available from the Foundation) with you to give out is very handy during this exchange.

If faced by a parent who appears stunned or does not want to engage any further, simply say you had not meant to intrude and be on your way. This outcome may not be what you hoped for, but you can feel good that you made the effort.

If you've been in this situation, we would love to hear from you since your first-hand knowledge is valuable to share with others. Email families@CdLSusa.org or call 800-753-2357.

Texas Family Takes the Spotlight to Support Federal Legislation

Stephen

The summer of 2009 brought many exciting events for one Dallas-area family. Joanne and Stephen, parents of Stephen and longtime Foundation volunteers, were thrilled when Texas Congressman Sam Johnson contacted them to discuss the Denton State Supported Living Center School, which 30-year-old Stephen has called home for past eight years. Rep. Johnson was rallying support for a bill protecting the interests of residents living in intermediate care facilities, like Denton, when a lawsuit is filed against the facility.

Under current law, there is no federal requirement that residents or legal guardians be notified of a lawsuit, and they do not have the right to opt out. Many residents are swept into these lawsuits against their wishes. If passed, the legislation would require that residents and guardians receive notice of the lawsuit and be given an opportunity to opt-out of it.

The family shared information about Denton with the Congressman and offered their support of the bill. They were also featured in a local newspaper article about Johnson's efforts.

"We had the opportunity to promote CdLS awareness, and the reporter included information from the web site," says Joanne. "Stephen's picture made the front page, and we bought several copies!" Stephen was also featured on Rep. Johnson's web site.

Pennies for Jessica
Providing Hope Through Change

Providing Hope Through Change

Pennies for Jessica is a national coin-drive event aimed at clubs and organizations committed to community service. This new program gives civic groups a chance to support a meaningful cause while demonstrating a commitment to children with special needs.

The program was inspired by Jessica (pictured), who began collecting pennies for the CdLS Foundation more than a decade ago.

Feel inspired? Customize Pennies for Jessica to feature the child in your life. For more information, contact events@CdLSusa.org or 800-753-2357.

MAILBAG

– Kathryn –

Dear Reaching Out,

Kathryn was born on August 2, 2007. She weighed 4lbs, 10oz and was 17 ¾ inches long. When Kathryn was born, she was sent to the Neonatal Intensive Care Unit at East Tennessee Children's Hospital because she had so many problems. During her two month stay in the NICU, we found out that she had Cornelia de Lange Syndrome.

Kathryn was born with only half of her arms, two heart murmurs, a cleft palate, hearing loss, breathing problems, reflux, feeding problems, and hypothyroidism. We also later found out that Kathryn has kidney reflux. Kathryn is currently seeing specialists for all of these problems as well as other doctors that need to follow her.

We have recently gotten Kathryn some prosthetic arms so that she can progress in her development. Her heart murmurs have healed themselves. We are waiting on her to grow so that her cleft palate can be repaired. We found out that her hearing loss is due to excessive fluid in her middle ears; therefore, she has had PE Tubes placed in both ears and wears hearing aides as well. She has overcome some, but not all of her breathing problems. She had surgery before she came home from the NICU to correct her reflux and feeding problems. She now eats the majority of her food via a feeding tube, but she will tolerate some feedings by mouth. And Kathryn takes medication for her hypothyroidism.

Kathryn scoots all over the floor to get what she wants and where she wants. She babbles constantly, but the only word she can say is "mama." She loves to watch TV, indulge in self play, and be around other kids (especially her cousins). Kathryn also loves to be talked to and read to. Kathryn's favorite things are rough housing with her daddy, playing with Elmo, her play gym, and her toys that light up and play music, and being talked to by her mommy.

Kathryn has taught us so much about life and how not to complain about anything. Kathryn has many problems, but we would not change anything about her. She is our little "miracle." Kathryn is a true fighter and is very independent. Kathryn is loved by many people. We all know that Kathryn was given to us for a reason and we are very thankful for that.

David & Sarah, Kathryn's parents

Tennessee

– Christian –

Dear Reaching Out,

This is our son Christian at his school prom in June of 2009.

Christian is a wonderful 17-year-old young man with a great sense of humor. When he is feeling good he loves to do artwork and math on his computer, go bowling, bike riding and horseback riding.

Christian has had some setbacks the past few years with anxiety and increased behaviors that we think are caused from his high anxiety.

Christian is currently getting help at the Devereux residential facility in Viera, Florida. We are very lucky to have a great team of experts at Devereux that are working with Christian to help him get past some of the things that have caused his anxiety and behavior issues to increase. The entire staff has been more like an extended family to us and Christian adores all the people that are part of his staff and treatment team.

We are looking forward to Christian getting past his anxiety issues and hope he will be back home with us very soon.

A BIG THANK YOU to the CdLS Foundation for all you do.

Gianni & Phyllis, Christian's parents

Florida

Committee Corner: Research Council

The Research Council oversees all research activities channeled through the Foundation. The Council is headed by Dale Dorsett, Ph.D. (pictured), of Saint Louis University School of Medicine, whose work with fruit flies helped scientist identify changes in the gene *NIPBL* as a cause of CdLS (read more about Dr. Dorsett's work on page 4). Additional council members have science or medical backgrounds and personal or professional experience with the syndrome.

In addition to reviewing research study requests, the Council oversees the CdLS Centers of Excellence designations, and reviews applications for the Foundation's Small Grants and Fellowship programs, when funding is available.

Members also create policy and procedures related to CdLS research and surveys and keep the Foundation staff and Board of Directors up to date on CdLS research news and findings.

CdLS Research Moving Forward in Great Strides

continued from page 3

This issue of Reaching Out is packed full of updates on research in the area of basic science and clinical research. I hope that when you are done reading it, you are more knowledgeable about CdLS research and excited about what the future holds for people with the syndrome.

** www.plosgenetics.org/article/info:doi/10.1371/journal.pgen.1000650*

2010 CALENDAR

March 26-27
Board of Director's Meeting
Philadelphia, PA

May 8
CdLS Awareness Day
Nationwide

June 24 -27
National Family Conference
Dallas, TX

June 25
Board of Directors Meeting
Dallas, TX

Nov 5-6
Board of Directors Meeting
Hartford, CT

February 13, 2010
One Heart, One Love
5 K Walk/Run for the
CdLS Foundation
Decatur, GA

Go to www.CdLSusa.org/calendar.shtml for the most current information.

What is Thrombocytopenia?

continued from page 8

Thrombocytopenia is a condition in which the blood has a low number of blood cell fragments called platelets. Platelets are made in bone marrow, along with other kinds of blood cells. They travel through blood vessels and stick together (clot) to stop bleeding that happens when a blood vessel is damaged.

When there are low numbers of platelets, mild to serious bleeding can occur. This bleeding can happen internally or on the skin. Bleeding on the skin is usually the first sign of a low platelet count. It may appear as:

- Small red or purple spots on the skin, often occurring on the lower legs.
- Purple, brown and red bruises that occur easily and often.
- Prolonged bleeding, even from minor cuts.
- Bleeding from the mouth or nose.

A simple blood test measures the platelet count.

Why study fruit flies? continued from page 4

be observed in a short time. Fruit flies are also cost-efficient specimens, as they require little space and less equipment than other animal models.

Since the early 1900s, fruit flies have been used by genetic researchers—some of whom received Nobel Prizes for their work. Today, fruit flies are used as genetic model for several medical conditions and disorders including Huntington's and Alzheimer's diseases. They are also being used to study aging, immunity, diabetes, and cancer.

Our Deepest Sympathy

Stephanie Smith
November 10, 1992 – July 16, 2009
Daughter of Crystal and Jerry Smith
1234 Boulder Creek Road
Richmond, VA 23225

Wishlist

- 100 memory sticks
- Coil or comb binding machine
- Printer ink cartridges
- Digital camera (minimum 10 mega pixels)
- New or refurbished computers
- Phone cards for volunteers
- Postage stamps
- Phone and gas cards for staff travel
- Pro-bono experts in grant writing, graphic design and photography

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_____ _____

StreetCity _____ State _____ Postal Code _____

❑ Use my gift to help 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):_____
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Signature:_____

To give on line: www.CdLSusa.org
To give stock or plan an estate gift, contact director@CdLSusa.org or 800-223-8355 11/09

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

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The UNIVERSITY of CHICAGO
Genetics Services

www.genes.uchicago.edu/lab.html

The University of Chicago Genetic Services Laboratories offer full gene sequencing of the *NIPBL* and *SMC1A* genes as well as deletion/duplication analysis of the *NIPBL* gene for Cornelia de Lange Syndrome.

Please contact us for more information on testing.

Melissa Dempsey
Genetic Counselor
Phone: 888-UCGENES
mdempsey@bsd.uchicago.edu

COMMITTED TO CUSTOMIZED DIAGNOSTICS, TRANSLATIONAL RESEARCH & YOUR PATIENTS' NEEDS

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