REACHING OUT

FALL 2019

GENETIC TESTING 101

Coping Corner

Fighting for Insurance, Fighting for Understanding

Embrace the Adventure in Tulsa

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

Director's Message

Have you ever watched a great performer and noticed the way they get a song out? How they reach for something that is deep inside themselves to share with us? I've always admired those who leave nothing on the table and give it their all. The same is true with sports. Consider Tom Brady of the New England Patriots or Kawhi Leonard of the Los Angeles Clippers – these are two of many athletes who give 100% all the time.

As parents we are performers too. One of the greatest performances of our lifetime is raising children. Each child is unique and they don't come with a how-to manual. Most of the time, we figure it out as we go along. In the end, whoever they turn out to be, our mark as their parent/caregiver will be indelibly left on them. P. Edmund proclaimed that "people do not decide to become extraordinary, they decide to accomplish extraordinary things".

In this issue, we explore the extraordinary choices of parents, who despite their child's diagnosis went on to transform the circumstances in to something beautiful. You will learn about Ethan, Breeze, Sebastian and Caleb. Their families stood tall through heartache and pain, and through it all found the inspiration and strength they needed to be #cdlsstrong.

We are inspired by you each day. Our hope is that you are inspired by the stories of your peers in this issue of *Reaching Out*.

Warmly,

Bonnie Royster
Executive Director

Bonnie Royster, CdLS Foundation Executive Director

Please note, **Gifts that Count** can be found in our electronic version of *Reaching Out*; which is located on our website: www.cdlsusa.org/resources.

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The Importance of Allied Health Professionals

Many areas of medicine impact any child with CdLS. Medical professionals include doctors (physicians and PhDs), dentists, nurses and pharmacists. Although critical for health care, these impact only a small portion of medicine. One of the most important aspects of medicine is the field of allied health. Allied health professionals include the "hands-on" care the child encounters regularly, plus others "behind the scenes".

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

The lifeline at the CdLS Foundation for families are three Family Service Coordinators, all in social work. Social workers have many roles; coordinating day-to-day needs of children and families, counseling and giving support. Another prominent group of allied health professionals are genetic counselors. Whether in a prenatal setting, discussing an ultrasound with unusual findings or working with the geneticist; genetic counselors connect with families on their journey to a diagnosis, explain complicated genetic testing results, make them easily understood and prioritize discussions. We are fortunate to have three genetic counselors on our Clinical Advisory Board (CAB), who are always willing to meet with families during our national conference.

More direct care allied health professionals include therapists, such as physical (PT) and occupational therapy (OT), and speech pathology, that help children with developmental and educational needs. PT addresses gross motor skills, focusing on legs and mobility. OT addresses fine motor skills, focusing on the arms, oral-motor skills and sensory integration issues. Speech-Language Pathologists work with speech, feeding and oral-motor skills. These therapists are well represented on our CAB and Professional Development Committee (PDC) and do consultations at Conference. Other allied heath professionals include Dental assistants and hygienists who work with children and play a strong role in good oral hygiene. Audiologists perform hearing tests. Ophthalmology technicians perform vision tests. Hospital-based allied health professionals have less direct contact but are equally important: radiology technicians, phlebotomists, lab technicians and medical record specialists. Overall, while gratitude for the medical community should certainly exist, please never forget the work by the tireless, valuable and critical allied health community.

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Genetic Testing 101

By Amy Kimball, M.S., Certified Genetic Counselor, Greater Baltimore Medical Center, Harvey Institute of Human Genetics

The Foundation has received many inquiries to help interpret genetic testing results. Common questions include 'does this result mean my child has CdLS?', 'does this mean my child does not have CdLS?'. Unfortunately, answers to these are not always clear and straightforward.

As a genetic counselor, I often discuss test results. I like to explain that genes form the instruction manual our body uses for growth, development and functioning. Each gene is a separate and important contributing chapter in that huge instruction manual. **A significant change to the letter sequence or code of a specific gene or 'chapter' causes CdLS.**

Genetic testing usually involves sequencing, or examining the code of the gene letter-by-letter. Testing for CdLS takes into consideration many factors. It can include only genes known to cause CdLS, if the diagnosis of CdLS is likely. The testing could be for a panel of genes causing conditions similar to CdLS if the diagnosis is less certain, or a test targeting more conditions, or all known genes (exome sequencing), if CdLS has not yet been considered.

If a result is negative, the lab found no sequence changes in any genes being tested. When a test result is negative, that individual may not have CdLS. Or, that person may have CdLS from a gene variant undetectable by the test performed. Many individuals (20-25%) with CdLS have negative testing.

A positive test means the laboratory detected a variation in the typical sequence of the gene. These variants are reported as *benign, variant of unclear significance, or pathogenic*.

A *benign* variant in the gene sequence is harmless. If a gene is a chapter in our body's instructions, a benign variant is like an extra period at the end of a sentence. It does not change the meaning of that sentence or chapter.

A *pathogenic* variant is a true change disrupting that gene's information, like a deleted paragraph in the chapter, or the chapter ending prematurely. This prevents the gene from providing correct information the body needs. The gene does not function properly, causing CdLS.

A *variant of unclear significance* (VUS) is just that; a change in the sequence in the gene that could be benign but also could be disruptive. In this case, there is not enough information about the VUS to determine if it is disruptive enough to cause CdLS or simply benign. To interpret a VUS result, many factors are considered to help interpret it: does the individual have features of CdLS, is the variant inherited from a parent, etc. In time, a lab may have updated information about a VUS and reclassify it as pathogenic or benign.

The most valuable resource for a family when trying to understand genetic testing results is to meet with a geneticist or genetic counselor to help explain what results mean or don't mean. Contact Family Service at 800.753.2357 to get more information on a geneticist or genetic counselor near you.

Today, Breeze is doing amazing. She is in 10th grade and a cheerleader. She is non-verbal; however, is doing well academically and looks like a child with CdLS.

Inside Look: Breeze

Margaret didn't have an easy pregnancy with Breeze. She was sick and on bed rest for 5 weeks. Breeze had restricted growth in utero, which concerned Margaret but the amniocentesis test showed no issues. Breeze was born 8 weeks early and weighed only 2.5 pounds – she had her second genetic testing which again came back normal. Breeze left the NICU with a Cerebral Palsy (CP) diagnosis.

Margaret knew there was something different about her daughter. She didn't want to brush aside the negative results, and it seemed that Breeze had different issues than other children with CP. In the meantime, the family moved and met with a neurologist who told Margaret that he wasn't convinced that Breeze's diagnosis was CP.

The doctor told Margaret to keep the diagnosis even if it was incorrect because Breeze had some rare disorder. **The doctor told Margaret that it was something that doctors won't catch because they see so many patients and they didn't have time to learn about all the rare syndromes that are out there.**

This gave Margaret a new purpose. She was a full time caregiver for Breeze and now she was determined to research what syndrome Breeze had. She pinpointed two different syndromes, Angelman and CdLS. She went to the DC National Children's Hospital and requested a third genetic test be done that would specifically test for them.

And she waited…the results came back…negative. When Margaret questioned the doctor about the results, the doctor responded, it's too rare and expensive. She knew these tests were being run at Children's Hospital of Philadelphia (CHOP) and at the University of Chicago, and was beside herself that a doctor would tell her that she couldn't spend her money on trying to help her child. Margaret couldn't believe that a medical professional just blew her off.

Breeze's blood work was sent to the University of Chicago and 8 weeks later they **FINALLY** had an answer. Breeze was diagnosed with CdLS. The doctor in Chicago told Margaret they never saw a mutation like this. They considered Breeze's mutation an "evolutionary change". The geneticist confirmed that Breeze had CdLS but couldn't tell what she would look like or if she would be high functioning.

While Margaret was waiting to hear about the results, she found the CdLS Foundation website and saw the pictures of other children with CdLS. She knew that this was Breeze and everything just clicked. When she went to her first CdLS Conference she walked in and started crying. She had never seen another child that looked like Breeze. If the CdLS Foundation didn't exist, they wouldn't have a diagnosis and she wouldn't have pushed for the testing.

"Imagine having a beautiful Christmas present that you couldn't open. You had to care for it as best you could. However, when you finally are able to open it, it is such a relief because you finally know what you are supposed to do with it…that is what it was like when I found out about Breeze's diagnosis."

The first few things we heard when he was born was "He's here, he's breathing, and he has a TON of hair!" We were already so in love with our little peanut.

Fighting for Insurance, Fighting for Understanding

Our sweet Sebastian was born 5 weeks early via emergency C-section. The first few things we heard when he was born was "He's here, he's breathing and he has a TON of hair!" He was under the 20th percentile for weight and height at 3 pounds, 4 ounces and 15 inches long.

Prior to his delivery my pregnancy became very high risk very suddenly. At 30 weeks, Sebastian had stopped moving and we found out he had congenital heart disease. There was concern that he was in heart failure but after seeing a pediatric cardiologist we learned he had four heart defects, but was not in heart failure. To compare the two felt wrong, but trading heart defects over heart failure was our only option and we decided to be thankful for the outcome. We knew when he arrived there would be a long road ahead of us and we were as ready as we could be.

At two days old, a Genetic Specialist gave us a suspected diagnosis of CdLS. This news wasn't a shock to us as we were prepared for a syndrome before he was born due to high amounts of amniotic fluid surrounding him. I think I did what any new parent would do...I googled his syndrome. Before they even gave us a confirmed diagnosis a week later, we already knew. Between Sebastian's beautiful eyebrows, little upturned nose and luscious eyelashes - we knew. Our baby had Cornelia de Lange Syndrome and it terrified me. I was angry at first, 1 in 10,000 births, *why him? What did I do wrong?* I didn't understand the syndrome or what to expect,

but I was sad. I was grieving over the child we expected while still learning to love and being thankful for the one who had just arrived. Giovanni never showed anger, only concern. It didn't matter to him 'what' Sebastian was diagnosed with, he just wanted him to get healthy.

Sebastian spent the next 101 days in the hospital. He underwent three heart surgeries and numerous small procedures including lumbar punctures, CT's and MRI's. He was intubated for two months straight at one point with no signs of life. In addition to his diagnosis of CdLS and CHD, Sebastian was also diagnosed with encephalopathy which is a really big word for an incurable brain disease. He is deaf in his right ear and has severe hearing loss in his left ear. At this time, he still has no useable vision. **But none of that stops him.**

The day before Sebastian's third and biggest heart surgery we found out his insurance had changed and he was no longer on state care. I remember the hospital's social worker coming in to tell us, but reassuring us that it must have been a mistake and they were already on the case. However, it was hard not to stress about. We are comfortable financially but by no means could we afford a 10-hour open heart surgery and lengthy recovery. Within a week his insurance was back, it turns out they did make a mistake. The stress seemed to fade as quickly as it was brought on and soon Sebastian was finally released to come home after three and a half long months.

Over the next year we didn't have any issues with his insurance or appointments. Sebastian was thriving at home and in all of his therapies. He was gaining weight and interacting with us more and more each week.

When Sebastian turned 18 months old the state re-evaluated his case and decided he no longer qualified for state care because our household income was too high. Sebastian's father, Giovanni, and I are not legally married yet and I stayed at home to care for Sebastian. Giovanni was the only one working and paying for everything. They didn't even give us a warning, or even try to look into where the finances were going. They sent us a letter letting us know he had been taken off state care.

Our financial state was a typical one, we had everything we needed, but by no means could we provide everything Sebastian needed on a daily basis. Not to mention the amount of medical follow-ups and tests he needed monthly. I was devastated. We attempted to put Sebastian on his father's insurance and the first representative told us it wasn't open enrollment, even though this should've been considered a life changing event. The second person said we could find a plan for him, but it would be pricey considering his needs. The second representative also told us that given Sebastian's medical needs, he **SHOULD** get care through the state. They told us to fight for it, and so we did just that.

It was never about the money, we could always find a way to make it work. It was about principle. Sebastian is disabled and should qualify for assistance. I felt it was my responsibility to fight for inclusion for him. We fought for months and finally won his case. His insurance has been reinstated and we are waiting for an approval from social security to further benefits for Sebastian's future, because that's what he deserves. **Security.**

Sebastian losing his insurance for a second time really took a toll on our family in different ways. Giovanni felt he had to push harder at work because there was no guarantee this would end in Sebastian's favor. I went back to work and found a nurse who worked for a lot less than she deserved. All while I tried so hard to keep my sanity. It was hard, I was stressed and I was upset. I tried so hard not to take it personal. The state doesn't look at Sebastian and who he is, they look at facts. To them, they saw a family who didn't need help. My anxiety was higher than it had ever been and I almost became unable to function. Missing appointments and not returning important phone calls just because I couldn't handle things.

Through all of the stress I fought for his right for assistance and when it paid off, it felt like the heaviest weight was taken off my body. We were able to move forward in the process of getting Sebastian his glasses and hearing implants, things we could not afford even if he were on his dad's insurance. He was able to see his cardiologist and for the first time in 22 months, he was cleared to come off oxygen! The light at the end of the tunnel had finally reappeared and everything we fought for paid off.

The only thing I want other parents to take from this is that if you think your child deserves something, fight for it. I can't attest to what other family units and their income looks like, or what their situation may be but I do know that there are resources out there for a reason. Sometimes you just have to fight for them and that is the duty we have been given as special parents. It's hard at times, but fighting for our children is a pride and responsibility I will never let anyone take away from us. No one should have to feel like they have to go without, and I'm here to remind you that you are capable of being the advocate your family needs. **We are CdLS strong.**

HIGHLIGHTED Events

SPARE SOME LOVE FOR CdLS

Kristin, Joe, Kyla and Connor Willey hosted their very first, wildly successful "Spare Some Love for CdLS" Bowling Fundraiser on May 18th. Their event included a silent auction and raffle. They raised over $10,000, which smashed through their original goal of $2,000. The event proceeds went to support programs and services provided by the Foundation. Proving that it really does take a community…Kristin shared, "A special thanks to our family and all of our volunteers for all you did! We couldn't have done it without each of you!! Yesterday was perfect! After nine months of planning, it was incredible to watch a vision become a reality."

SECOND ANNUAL ROLLING FOR KENSLEY

A fun filled weekend was held June 29th and 30th at Crickets Bar & Grille in Youngstown, Ohio. The Lupo & Holovatik families hosted a fundraiser which featured a bocce tournament, Chinese auction, raffle and dinner. A portion of the proceeds went to support the CdLS Foundation. The weekend event climaxed with an exciting special guest performance by country rock duo, Adair's Run, Pat Pollifrone and Todd Cameron, who donated their time and talents, arranging their tour schedule to be able to support this event.

STUDENT LED FUNDRAISER FEATURING HISTORICAL FIGURES

The Most Holy Redeemer School, in Illinois, raised funds during a student led "Historical Wax Museum". Students dressed up as historical figures and visitors made a monetary donation to hear the student share a two-minute personal oral history as told by the figure they represented. A portion of the proceeds gathered was gifted to the Foundation in honor of Maddie Busk's twin sister, Abby.

MUD RUN

On June 8th, Mike Feehan completed his tenth annual mud run event, on behalf of Team CdLS and in honor of his 11-year-old son, Connor. This one-of-a-kind 18+ obstacle course was surrounded by mud-filled pits, trenches and the new and improved Slippy Sloppy slide. At the end of the event, Mike and his family celebrated his athletic accomplishments, as well as the dedication and commitment of their friends, family, co-workers and community who helped them raise over $30,000 this year, bringing their lifetime fundraising total to $200,000!

Mike provides his professional knowledge to the CdLS Foundation as the Treasurer of the Board of Directors. He generously commits his time to the Board of Directors to help establish, oversee and direct the Foundation's mission and vision, all while ensuring its financial health.

CARR FAMILY SPARTAN RUN

Led by Shawn and Kim Carr, the team of 12 took on their toughest and most challenging Team CdLS event yet – the Palmerton Super Spartan course. The elite event challenged the team to maneuver through intimidating steep slopes, long climbs and navigating in and out of dense forest. The team was rewarded at the end of the race with breath-taking views from the top of Blue Mountain.

"It has been over five years now since the birth and passing of our daughter, Layla, who was born with a rare genetic condition: Cornelia de Lange Syndrome…We have had a lot of positives over the past five years and have an amazing group of family and friends who are always there to step up for us," says Shawn. This year the team raised nearly $3,500 – bringing their lifetime fundraising Team CdLS total to $29,000.

CALENDAR

December 3, 2019
Giving Tuesday
National Day of Giving

February 29, 2020
Rare Disease Day

March 11, 2020
CdLS Clinic at
Shriner's Hospital
Salt Lake City, UT

March 15, 2020
United Airlines NYC Half
New York, NY

May 3, 2020
TD Five Boro Bike Tour
New York, NY

May 9, 2020
National CdLS
Awareness Day

May 19, 2020
New England Charity
Golf Tournament
Ipswich, MA

June 24 - 25, 2020
Ninth Biennial CdLS
Scientific & Educational
Symposium
Tulsa, OK

June 25 - 28, 2020
CdLS National
Family Conference
Tulsa, OK

October 11, 2020
Bank of America
Chicago Marathon
Chicago, IL

Matt & Julie Guidry

There's never any deception with Caleb. Whatever he is expressing, however he is trying to connect, is always the absolute truth. That and the fact that he has unbelievable comedic timing, and a mischievous side to him that we much admire. These traits are what make him so unique.

Caleb was diagnosed with CdLS two days after he was born. I remember the first five weeks spent in the NICU, and then many weeks in the hospital those first two years, as a blur of memories filled with so many emotions. But from the beginning, Caleb's presence always brought people together. Inspired by the way Caleb expressed himself within his community, and how they responded – my wife Julie (Caleb's stepmom) and I founded Upstream Arts 13 years ago.

Upstream Arts is a team of committed individuals who come together to learn and practice social skills through creativity and play. Creativity is defined as "the ability to transcend traditional ideas to create meaningful new ideas." I'm sure everyone in the CdLS community can understand the need to move beyond traditional ideas.

The approach of our work is modeled after the community support we experienced with Caleb. We use the power of creative arts to develop skills and a practice for everyday communication for individuals with disabilities. It's about play and being in a community with one another, and finding the many avenues that the arts can provide for self-expression, self-advocacy and social interaction. **We wanted to give individuals with disabilities a space to tell their story and a place to be heard.** Caleb previously had few tools to communicate; but he began to use the physical movements, body language and facial expressions, he learned through the arts to communicate and engage with those around him. Out of this spirit and determination, Upstream Arts was born.

I strongly believe that exposure to the arts has provided ways that Caleb uses to connect with those around him. We see the real value of arts as creativity, not simply the end result of making art. Caleb always connected to and is fascinated by dance and movement. And he loves to mirror what he sees. Exposing him to these things gave him a physical vocabulary, and a way to have non-verbal conversations with those around him. It's his way of socializing. It moved our interactions from a verbal practice, to a give and take, exchange using dance, rhythm, and play. Always taking turns, the basis of conversation. It has allowed us to better understand his wants and desires, and more importantly, who he is as a person.

All of these experiences we have had over the years with Caleb have gone into giving individuals with disabilities the tools to better engage with their friends, families, teachers and community. Now, we run over 35 classes a week in schools, adult day programs and other community organizations, led by professional artists. We have also become advocates with the goal of changing mindsets about ability and disability.

To learn more or to find out how you can get involved, visit **www.upstreamarts.org**.

WELCOME
New Families

THE PETERSON FAMILY

Ryan and I welcomed our valentine, Aisley, on February 14, 2019. At twelve days old she was given a clinical diagnosis of CdLS and then later genetics discovered she has a truncated *NIPBL* mutated gene. It has been quite the journey; we are lucky to have our CdLS family. We live in Healdsburg in Northern California. For our first CdLS awareness day our friends and family helped us raise over $2,000!

THE SALLEY FAMILY

Hi, I'm Gentry Brinkley Salley. I live in Greers Ferry, Arkansas. I am nine months old and I enjoy boat rides and playing with my older brother and sister. I am a girl on the go and I'm excited about spending my first birthday with Minnie Mouse at Disney World!

Arkansas
Amber and James and daughter Everly, born July 17, 2016

Arkansas
Lindee and daughter Gentry, born October 29, 2018

California
Emily and Ryan and daughter Aisley born February 14, 2019

California
Wanda and Kevin and daughter Ariel born October 21, 2016

California
Jeannette and Kyle and son Greyson, born October 20, 2017

California
Brooke and Matt and son Grayson, born May 5, 2019

Illinois
Elizabeth and Tyrone and daughter Michelle born May 20, 2019

Missouri
Josie and Jordad and daughter Payton born September 11, 2017

Missouri
Jessica and son Doniven, born July 25, 2008

North Carolina
Coretta and Jeffery and son Cameron born April 23, 2019

North Carolina
Yulissa and Jesus and sons, Jhon and Wilian born April 9, 2015

Texas
Wanda and daughter Jeysha, born April 22, 1998

Ethan's family never imagined that his smile and the sound of his laughter would be so heartwarming and contagious. "Ethan's joy and meaningfulness to life tells us more than we ever have to hear him say."

Ethan Beas

On November 18, 2015 we welcomed a baby boy into our family: Ethan Greg Beas. At birth, we were told there was a 70-80% chance that Ethan had CdLS. Ethan has many characteristics of CdLS; low birth weight, limited mobility in his arms and hands, distinctive shape of the face and head, connecting brows, breathing and feeding problems, acid reflex, is considered profoundly deaf in both ears and other cognitive delays. Ethan's diagnosis was confirmed in January 2016 by the Children's Hospital of Philadelphia (CHOP).

Ethan's suppressed immune system causes him to be confined to the house most days in the winter to avoid catching any infection. There have been many times when Ethan gets so sick that he must spend days or weeks in the hospital.

He has an older sister, Sophia. One thing that we enjoy the most is when Ethan and Sophia play together. The love they have for each other is unmatched. We are blessed with a loving daughter who looks beyond her brother's disability. Every time he sees her he laughs and they smile as if they have inside jokes. Although Ethan is not yet able to walk and chase after his sister it is something we are looking forward to.

Ethan is attending school and working on sign language, Occupational and Physical Therapy. He has improved so much in the last year and a half. What comes easy to most children Ethan has to work very hard for. He went from sitting up, to standing with assistance, to cruising in his walker within a few short weeks. He gives it his all and we couldn't be prouder.

Although Ethan was born with CdLS we try to make his life as normal as possible, we do not stop him from being a kid. He is our strength; he shows us so much determination every day along with so much self-motivation. From all the new adventures this life brings us, one of the most important lessons that Ethan has taught us is that love needs no words.

In the short but meaningful life Ethan has lived, he has faced many challenges. But through it all he's managed to find a way to overcome them. Challenges are what make life interesting and overcoming them is what makes life meaningful. His joy and laughter goes to show he's a fearless warrior. His life will be a battle and an adventure that has only just begun. Not knowing in which direction we will be pulled in next can be frightening, but with Ethan's fearless spirit, I'm sure he will overcome anything that crosses his path.

Wallet Free Fundraising

"For my 70th birthday this year, I created a Facebook fundraiser to support the CdLS Foundation. I chose the Foundation because their mission means a lot to me, and I hoped that friends and family would contribute as a way to celebrate my milestone birthday.

Our 30-year-old son, Miles, was diagnosed with CdLS a day after his birth. At that time, we were told his life expectancy, because of the severity, was five-eight years. But that was based on a bunch of old information that was published in medical journals at the time.

The CdLS Foundation contacted me at Baystate Medical the second day of Miles' life and from that day forward they have been our lifeline to all things regarding his health issues. He has been on death's door so many times and has continued to amaze and inspire me by his strength and will to LIVE."

Billy's Facebook fundraiser brought in over $2,500 in just two weeks. Since the inception of this program the Foundation has generously received over $44,500! Creating a fundraiser is a quick, and easy way of celebrating your birthday or other special occasions.

The CdLS Foundation sincerely thanks all those who have created a Facebook Fundraiser – you are not only helping to raise funds but also awareness to hundreds of thousands of people. **THANK YOU!**

How to Create a Facebook Fundraiser:

1. Click **Fundraisers** in the left menu of your News Feed.
2. Click Raise Money.
3. Select Nonprofit, Cornelia de Lange Syndrome Foundation.
4. Choose a cover photo and fill in the **fundraiser** details.
5. Click Create. *It's that simple!*

Helpful Hints

DIAPER ASSISTANCE

The National Diaper Bank Network (NDBN) provides basic necessities required to build the strong foundation all children, families and individuals need to thrive and reach their full potential. They work in partnership with a network of diaper banks, allied programs, donors, sponsors and elected officials to end diaper need and period poverty in the United States. If you or someone you know needs diapers, there may be assistance in your community, visit **nationaldiaperbanknetwork.org/get-help-now/**

SPECIAL NEEDS LAWYERS

The Special Needs Alliance is a national organization comprised of attorneys committed to helping individuals with disabilities, their families and the professionals who serve them. Many of the members have loved ones with special needs; all of them work regularly with public benefits, guardianships/conservatorships, planning for disabilities and special education issues. They collaborate with advocates throughout the special needs community to improve quality of life for individuals with disabilities. Special needs attorneys have daily experience with the issues that matter to individuals with disabilities – but which are largely unfamiliar to other counsel. They can advise you about:

- qualifying and applying for appropriate public benefits
- creating special needs trusts (SNTs) to protect eligibility for means-tested government programs
- availability of local services and supports
- special education
- guardianship and powers of attorney

To learn more, visit: **www.specialneedsalliance.org/**

ADAPTIVE CLOTHING

For many individuals with CdLS it can be a challenge to get dressed every day. New Horizons Un-Limited Inc. put together a list of resources for adaptive clothing designed for people with disabilities, who may experience difficulty dressing themselves. They provide resources for dressing accessories, aids or devices that assist the self-dresser to manipulate closures and to dress as independently as possible.

To learn more, visit: **www.new-horizons.org/adpclo.html**

MEDICAID WAIVERS

Medicaid Waivers help provide services to people who would otherwise be in a nursing home or hospital to receive long-term care in the community. Although there are waivers for many conditions, our focus is towards waivers for people who have intellectual disabilities, developmental disabilities, and autism.

Many people who qualify for waiver services are not even aware that they exist. Forty-four (44) states and the District of Columbia have received waivers to provide home- and community-based services to people with developmental disabilities (DD). Depending on each state's DD definition, these waivers may cover services to people with autism.

For more information about Medicaid Waivers, visit: **bit.ly/2pBHDI1**.

Our Deepest Sympathy

Angela Shontaine Latham

March 20, 1968 – March 4, 2019
Daughter of Lynda and Alex Latham
305 S. Free Street
Portia, AR 72457

Benita Hudson

February 26, 1989 – August 7, 2019
Sister to Charletta Hudson
P.O. Box 230101
Grand Rapids, MI 49523

Herbert Argueta, Jr.

August 9, 1985 - April 14, 2019
Son of Carmen Anaya and Luis Prado
1226 Simpson St. Apt. A
Bronx, NY 10459

Laurie Grace Johnson

December 14, 2017 – August 24, 2019
Daughter of Molly Warren and Scott Johnson
2941 Park Terrace Dr. Apt. 4
Rockford, IL 61114

Our Memorial page is a revered space for family and friends to share happy memories or say a few words about the life of their loved one. This page honors all of those who have passed away. You can share a memory or submit your story today. Visit: www.cdlsusa.org/in-memoriam.

"Cherish the good memories of your child and remember: hold on to hope, rest when you're weary, take time to grieve, and let your heart mend."

–Excerpt from *"When Angels Take Flight: the Loss of a Loved One with Cornelia de Lange Syndrome"*

If you would like to speak with Foundation staff or connect with other families who have lost a child with CdLS, please call 800.753.2357.

Embrace the Adventure in Tulsa

Join us for the CdLS Foundation National Family Conference, June 25 - 28, 2020, in Tulsa, OK. The CdLS Conference provides education and support to individuals with CdLS and their families. Attendees receive free head-to-toe consultations with experts from a range of medical and educational fields; attend workshops on legal concerns, educational issues and medical/behaviors challenges; and have opportunities to meet other families facing similar challenges.

Over the next several months, our committee and staff members will work together to determine workshop topics, solicit speakers for the sessions and professionals to provide consults, menu planning, layout of rooms and much more. A big piece of the conference planning puzzle, includes seeking financial support from various sources, from large national corporations to small local businesses, to help offset the costs. **Conference is not a fundraiser for the Foundation**–in fact, registration fees don't cover the true costs of this event, so finding financial support is key in being able to keep the event viable.

As we get closer to the conference, be on the lookout for more information and tips. We hope to see as many families as possible at the event. It's truly a highlight for staff to see families in person, so we'll see you on-site and look forward to handshakes and hugs in Tulsa!

FAQs

When can I register for conference?

Registration for conference is scheduled to open Tuesday, March 3, 2020. The early bird registration deadline is April 7; the final deadline is May 19. Registration is available online through the CdLS website, visit www.cdlsusa.org/conference/. Conference can accommodate up to 150 families. Once that number is met, registration is closed.

What does registration cover?

The registration fee covers all workshops and sessions, medical and educational consultations, and the following meals: Thursday night ice cream social; Friday breakfast, lunch and dinner; Saturday breakfast, lunch and dinner. It does not cover hotel or travel expenses.

Registration Fees		
	Before 4/7	After 4/7
Adult 18+	$370	$410
Person Providing Childcare	$250	$280
Children 3-17	$190	$215
Children 2 and Under	No cost	No cost
Person with CdLS	No cost	No cost
One-day Professional	$150	$150

Where are we staying, and when can I book my room?

The conference takes place at the Marriott Tulsa Hotel, Southern Hills (bit.ly/marriottsouthernhills), just 20 minutes from the Tulsa International Airport. **Access to booking hotel rooms is through online registration.** The reduced room rate is $125/night plus tax and is **only** for people registered for conference.

What are my childcare options?

Children with CdLS (any age) can attend **Jindi's Place** at no cost. Activities include arts and crafts, movies, games, toys, reading and a quiet room. Arrangements can be made for tube and bottle feedings. More details regarding child care options will be available soon on our website over the coming months.

More details on Conference, as well as registration links, amusement park discounts, and transportation information will be on the conference web page, www.cdlsusa.org/conference/.

DOLLAR FOR DOLLAR

Each day, we receive generous donations from our community members. Each dollar we receive goes to a program to support individuals and families affected by CdLS. Below is a breakdown of your donation dollars hard at work:

$10 supports sending out a New Family Packet, which includes 4 publications & treatment guidelines

$20 pays for an Awareness Packet to expand our outreach and education to local communities

$25 provides 10 professionals with a packet containing Treatment Guidelines, fact brochure, personalized material for their medical field and a copy of *Reaching Out*

$40 provides 20 new grandparents with educational materials to help themselves and their loved ones in caring for individuals with CdLS

$50 allows 10 families to receive *Reaching Out* three times a year at no cost to them

$400 provides supplies for one CdLS Foundation sponsored Family Gathering

$1K supports the Foundation in providing webinars free of charge

Any amount has a BIG impact on what the Foundation can do for you.

Please consider making a donation today, visit: www.cdlsusa.org/general-donation.

Thank You, Lynn!

Lynn has been a lifeline to our families for over 20 years. Her compassion, understanding and presence is a driving force behind our mission. At the end of 2019, Lynn will be retiring from the CdLS Foundation. It is with great sadness that we must say goodbye. Lynn is an inspiration to her coworkers and her dedication to her work far extends beyond her job and is evident in the care she has shown to individuals with CdLS and their families over the years.

We thank you, applaud you, and above all we will miss you. Please take a moment to send a message to Lynn by visiting: **www.cdlsusa.org/goodbye-lynn.**

We Lost a Giant

With a heavy heart, we report the passing of **LAIRD JACKSON** – the CdLS Foundation's first Medical Director. Before the founding of our beloved Foundation there was the *Reaching Out* newsletter. The early pioneers sent out a request to the medical community asking for help to better understand the little-known genetic disorder of CdLS. One man answered the call – Dr. Laird Jackson. He not only helped countless families with the many medical complications and behavioral issues faced by children with CdLS by recruiting other specialists and forming a scientific advisory committee, but he also worked tirelessly in the search for the causal genes by seeking out important patient samples and recruiting and encouraging scientists to pursue this elusive quest successfully.

He was dedicated in his efforts to not only understand the genetic causes of CdLS, he was equally interested in pursuing what complications led to the death of those with the condition. He published many articles and unselfishly shared his research in pursuit of that understanding. Modest to an extreme, he is best known for furthering the field of Cytogenetics

Dr. Jackson's passion for teaching others about CdLS was contagious. He mentored two important figures in our community – Dr. Ian Krantz and Dr. Tonie Kline. Dr. Krantz is a leading clinician and researcher at Children's Hospital of Philadelphia and Dr. Kline among her many other accomplishments, went on to succeed Dr. Jackson as the Foundation's second Medical Director. Laird G. Jackson, M.D., Professor of Internal Medicine, Obstetrics and Gynecology and Pediatrics, contributed so greatly to the field of Medical Genetics that he was awarded the first David L. Rimoin Lifetime Achievement Award in 2017 from the American College of Medical Genetics and Genomics, for which he was a founding member.

Many recall his thoughtful and insightful presentations on Genetics to the first-year medical students, inspiring some to enter fields related to Genetics. He had a passion for model airplanes, old cars, and sharing stories. He enjoyed his time with his wife and their many dogs. He will be greatly missed by those who knew him and had the honor and privilege to work with him.

Perhaps, Dr. Jackson's greatest legacy is the love, care and hope he provided for so many in those early years. We offer the family and his many friends our deepest condolences.

2019 CdLS Grant Researchers Announced

As part of the 2019 Research Grants Program, the CdLS Foundation has awarded $24,447 to researchers studying various aspects of CdLS. The goal of our program is to have research projects thatprovide insight into the causes of CdLS, as well as evaluate potential ways to improve the lives of individuals and families affected by this syndrome. Researchers will present their findings at the 2020 CdLS Scientific and Educational Symposia. The recipients of the grants are:

Stephenson Chea, Graduate Student Researcher, University of California, Irvine, Center for Complex Biological Systems: *Investigating Cell Fate Misallocation as a Source of Developmental Defects in Cornelia de Lange Syndrome*. This study will be using single-cell RNA sequencing to investigate whether similar disruptions in the allocations and fates of embryonic stem and progenitor cells are also responsible for defects in the brain and gastrointestinal system, two critical organ systems that often show abnormalities in CdLS.

John Michael Falligant, MS, BCBA, Kennedy Krieger Institute, Maryland: *Behavioral Assessment and Treatment of Problem Behavior in Children with CdLS*. This study will improve our ability to effectively treat problem behavior in CdLS, as well as identify key variables associated with problem behavior in CdLS which may be examined in future studies and clinical practice to foster early intervention and prevention efforts.

David F. Smith, MD, Children's Hospital Medical Center, Cincinnati: *Characterization of Sleep Patterns in Patients with CdLS*. The aim of this study is to use objective measures to characterize the sleep patterns of patients with CdLS by sleep logs, validated sleep surveys, and actigraphy in order to better characterize the sleep disturbances noted to be prevalent in these patients. We aim to correlate these findings with patient phenotypes (using quality of life surveys and a review of previous medical records) as well as assess their sleep patterns against familial controls.

7.1.19 - 10.1.19

In Honor/ Celebration

Zackery Arrowood
Theta Mu Sorority

Brynnlee Beekman
Bonita Boxell
Mary and Glen Dehaven

Hannah Boehman
Jasper Engines & Transmissions Inc

Philip Braverman
Howard Braverman

Liam Brundage
Element Wholesale LLC

Abigail Busk
Most Holy Redeemer School

Kaitlynn Clark
Carmen Castaneda

Halston Dorow
Caroline Dixon

Chase J. Duff
Barb Robinson

Emmalee N. Ellis
Whitney Wilson

Mary Fiori
Ted and Carolyn Williams

David B. Fowler
Charles Weisman

Logan Fowler
Sydney and Emory Wishon

Lindsay Franco
Susan and Scott Coleridge

Diane Friedman
Howard Braverman
Sandra and Stuart Kaufman

Manuel J. Garcia
Laura Garcia

Aubrey Garigen
Western New York Oldsmobile /GM Club

Charles Gladson
Margaret and Stephen Gladson

Brenden Keating
Karin Csolty

Jared Koelling
Melissa and Jerry Koelling

Aelycia Monasmith
Whitney Wilson

Jake A. Marcus
Best Lawns, Inc.

Jayme B. Naples
Whitney Wilson

Alanna Neuendorf
Whitney Wilson

Emma G. Perez
Whitney Wilson

Brianne N. Prada
Karen and Larry Prada

Isabelle R. Skelonc
Susan and Robert Burns
Marilyn and Rix Robinson
Janet and Gerald Skelonc
Jill and John Vanoeffelen
Marcella and Ronald Wesche
Bonnie and Gary Woodruff

Hayley N. Turbyfill
Carol and Glenn Zeliff

Emily Turner
Mrs. and Lt. Col. Gus Meuli

Nathan Riley
Whitney Wilson

Peyton Howland
Joyce and Wilson Brown

Sloane I. Faber
Meghan Altier
Allene Faber
Anita Fleisch
Carly Owen
Charles Radtke

Susan Murphy
Martha-Jane and Richard Moreland

William E. Smisloff
Laurie and Michael Nelson

In Memory Of

Herbert Argueta, Jr.
Board and Staff of the CdLS Foundation

Jean Ann Brennan
Susan Johnson

John Cataline
Sheila Lieder

Layla A. Carr
Margaret and James Ludgate

Rosemary Christie
Anonymous
Angela and Gregg Garguilo
Anna O'Herren
Cathleen O'Brien
Darcy Campbell Moniz
Deborah and Merle Rinehart
Dennis and Sarah Drislane
Donna DiBona
Jessica Tiegs
Kabrina Hudson
Laurence Pagnoni
Lisa and Gil Pagnoni
Marilyn Parsons
Robert Zuczek
Tiffany Bartke
Tonya and Bradley Kingston

Lisa A. Chuilli
Norma James

Michael Gonella
Patricia and Aldon Daniels

James Goodbrake
Norma James

Theodore Hanneman
Anita and Jerrell Gaulf

Benita Hudson
Board and Staff of the CdLS Foundation

Jeffrey Ivins
John Alexander
Marcie Cramer
Linda Whatley

Anya Janoski
Tau Upsilon Omega Chapter/Alpha Kappa Alpha Sorority, Inc.

Laurie Grace Johnson
Board and Staff of the CdLS Foundation

Daniel Krimpenfort
Mary and Jim Krimpenfort

Angela Shontaire Latham
Board and Staff of the CdLS Foundation

Mia M. Mannino
Norma James

Duane Manzey
Mary Schow

Lily Molett
Norma James

Jessica E. Murphy
Ms. Roberta Murphy

Roland Owen
Norma James

Angela Ray
Norma James

Justin M. Roesler
Norma James

Barbara Skelonc
Tami Bullen
Susan and Robert Burns
Karen Casey
Donna and Dennis Larko
Marilyn and Rix Robinson
Janet and Gerald Skelonc
Jill and John Vanoeffelen
Constance Wert
Marcella and Ronald Wesche
Bonnie and Gary Woodruff

Claudia R. Tiongson
Dolores and Robert Schoen

Jindi H. Van Buren
Marilyn Sheehan

Molly Wagner
Kathleen Augustini
Norma James
Seth and Ashley Wagner

Aislinn K. Walsh
Mary Stasko

Ethan Walters
Penelope Keating
Candace and Shane Kelly

Melinda Walters
Helen Miller

CdLS Foundation
Cornelia de Lange Syndrome Foundation, Inc.

30 Tower Lane, #400
Avon, Connecticut, USA 06001
800.753.2357
www.CdLSusa.org

#GIVINGTUESDAY

12.03.2019

When you give this year on #GivingTuesday you are committing to help those affected by CdLS. Your contribution allows the CdLS Foundation to continue as the only organization in the country solely dedicated to those impacted by this condition. When you give to the CdLS Foundation on Tuesday December 3rd you make a real statement about the great tradition of generosity in this country.

To learn more bit.ly/CdLSGivingTuesday2019 or contact Alex@CdLSusa.org.

Funding provided by William and Alice Mortensen Foundation.