Director’s Message

Dear Families and Friends,

Twenty seventeen was a year of both leading and learning!

I met so very many of you and learned what the staff here at the Foundation has meant to you and your families as you navigated the landscape of CdLS. You shared with me the sleepless nights, the many visits to the doctors, the challenges with family members and school systems. But most importantly, you shared those remarkable milestones arrived at by your children – taking a step, finding something that brings them immense comfort and joy, attending the prom, going out for Halloween and inspiring those around them. Let us hold onto those triumphs.

One of my most meaningful lessons is learning that in spite of challenges we face – the CdLS Foundation is a community. We have a proud history of pulling together in times of joy and sadness. The Foundation provides many opportunities for support and awareness. In 2017, we touched the lives of so many individuals with CdLS and their families, by:

• Providing over 1,550 different support services, including addressing educational and medical questions
• Hosting 9 family gatherings across the United States (almost 600 attendees)
• Supplying travel support to attend CdLS Specialty Clinics across the country; and
• Awarding $48,895 in Research Grants to better understand CdLS symptoms and improve treatment.

None of this would be possible without the consistent support of our donors and corporate sponsors. We are incredibly grateful and hope we can continue to count on you to help us create life-changing experiences for our families.

Thank you for allowing me the privilege of leading this great organization.

Bonnie Royster
Executive Director
2017 Statistics

**INCORPORATED**

**INCOME**
- Clubs and Organizations: 3,834
- Conference/Program Service Fees: 3,373
- Corporate Donations: 19,400
- Direct Mail: 35,450
- Federated Campaign: 41,948
- Foundations: 14,668
- Gifts that Count: 46,032
- Individual Donations: 19,241
- Legacies/Bequests: 41
- Major Donors: 220,312
- Research: 8,484
- Special Events: 483,410
- Symposium: -
- Investment Income: 7,099
- Total Revenue: 903,292

**EXPENSES**
- Program Services
  - Family Support: $329,663
  - Public Information: $228,867
  - Professional Development: $213,672
  - Research: $57,479
  - Total Program Services: $829,681
- Support Services
  - Management & General: $79,945
  - Fundraising: $88,088
  - Total Support Service: $168,033
- Total Expenses: $997,714
- Surplus/Deficit from Operations: $(94,422)
- Unrealized Gains/Losses: $90,736
- Total Surplus/Deficit: $(4,686)
- Total Net Assets: 1,203,182

2017 Audited Expenses

- Family service coordinators welcomed new families: 106
- The Foundation sponsored Family Gathering’s
- Ask the Expert questions were submitted: 321
- Team CdLS was accepted as a charity partner at the TD Five Boro Bike Tour

- 68 families attended a CdLS Clinic
- Foundation sponsored Family Gathering’s
- 106
- 321
- 1,203,182
- 33%
- 23%
- 21%
- 8%
- 6%
- 9%
- 8%
April and Jesse are the parents are 21-month old Chase. They have been married for 7 years and tried from the beginning of their marriage to start a family; they finally got their wish through IVF treatments. April remembers her pregnancy as being a typical one, however Chase stopped growing in utero so they had to meet with a neonatal neurologist. When Chase was born, he was immediately rushed away because he stopped breathing and the neonatal neurologist suspected “syndrome”. It wasn’t until April insisted on knowing what was going on with their son that a geneticist finally confirmed to the family that Chase had CdLS.

Jesse jumped into action and got onto the internet to learn more – and what he initially learned terrified him. He reached out to the Foundation and was comforted by the voice of Lynn Audette. She not only helped him put his mind at ease, she welcomed the family into the community. They had a conversation that lasted hours and she helped Jesse understand more about CdLS. April on the other hand was having a hard time coming to terms with the diagnosis. She would be at the hospital all day and come home to sleep – she wasn’t ready to look at the information on CdLS and wanted to do genetic testing. One day Jesse came to the hospital with Lynn on the phone and said to April “I have someone I want you to talk to…” That was what helped April finally accept Chase’s syndrome, “it was scary but comforting to talk to Lynn.” Once April looked at the pictures Jesse had, she knew in her heart that Chase had CdLS.

The biggest obstacles Chase and April face are uneducated doctors and isolation. It is frightening to them to put their son’s life into medical professionals’ hands who are unsure on how to care for him. They are constantly advocating for Chase and at times feel like no one truly understand what they go through. In April’s opinion the CdLS Facebook Discussion Board helps to combats their obstacles, she recalls when Chase was first born she didn’t post pictures on social media. However, when she joined the discussion board she was made to feel welcome. She finally felt comfortable to show him off. The discussion board gives April trusted advice. She knows she can reach out at any time and receive the encouragement she needs.

When Chase was first diagnosed, the future looked very scary for the family. However, with constant encouragement from the Foundation and families all around the country, their future holds anything they want it to hold. They don’t fear the diagnosis and see peace in their future. “Chase has a community of brothers and sister. We gained a huge extended family. I would never have that if Chase was a typical child.”
Family Gatherings -
Connecting and Inspiring

Family Gatherings are one of the most impactful programs at the CdLS. The first family gathering was hosted in 1980 and for four years, they increased in numbers of families and professionals, until the size called for a larger venue. In 1985, the Foundation hosted its first conference in Indianapolis, which led to the biennial format that exists today. Family Gatherings continue on an annual basis and expanded regionally to meet the needs of families throughout the years. They specifically address the need for communal support through a unique and innovative approach.

In 2017, the Foundation and its volunteers sponsored nine family gatherings throughout the country – one in each region. Almost 600 people attended gatherings across the United States. A family gathering is a wonderful opportunity to meet with other members of our community to share stories, resources and information. When possible, a member of the Foundation’s Clinical Advisory Board or Professional Development Committee attend to consult with families.

Having the chance to meet with others who share similar challenges and triumphs as parents of children with a rare syndrome can be life-changing. Children with limb differences have met at family gatherings, and were able to hold hands with another who has limited digits; for the first time, a sibling did not have to explain their brother or sister’s condition to unaccepting strangers. Many attendees also meet others who live very close that they were unaware of, and plan future play dates for their children. The Foundation prides itself on making these gatherings based in quality, accountability and respect for individuals with CdLS.

Gatherings allow not only families, but relatives, caregivers and medical professionals, to come together and meet another individual with CdLS, sometimes for the first time. This contact can often lead to lifetime friendships and the creation of a local support system. Specialists from the various fields of speech pathology to nurses have the rare opportunity to make connections and share the progress and setbacks of their individual CdLS treatment regimens. Family Gatherings are an important and invaluable event for every individual with CdLS and family member we are able to touch.
Award Winning Medical Director - Antonie Kline, M.D.

Dr. Kline is the Medical Director for the CdLS Foundation and the Director of Pediatric Genetics at the Harvey Institute for Human Genetics at Greater Baltimore Medical Center. For more than twenty years, Dr. Kline has given her time and expertise to listen, help and guide countless individuals with CdLS and their families. Dr. Kline is continually inspired to conduct research, advance knowledge about the syndrome and make a difference in thousands of lives.

Since 2001, Dr. Kline has coordinated a free biannual Multidisciplinary CdLS Clinics for Adolescents and Adults at Greater Baltimore Medical Center (GMBC). During these clinics, patients meet with Dr. Kline and a group of specialists. Patients leave with behavioral plans, treatment protocols and valuable information on expectations for the individual’s current stage of life. This assists families in knowing what to expect as their loved one with CdLS ages. Dr. Kline has been instrumental in many other successful initiatives at the Foundation. As a fellow of Dr. Jackson, Dr. Kline created the first CdLS growth chart, which indicated an average range of height and weight for newborns with the syndrome.

Dr. Kline is responsible for recruiting a team of 60+ experts and specialists in varying fields to provide answers to the Foundation’s Ask the Expert program, where parents can submit their concerns about all aspects of health within the CdLS spectrum.

As the Medical Director, she provides diagnostic consultations by reviewing medical records and photographs free of charge. Dr. Kline also provides free, one-on-one consultations to hundreds of families at the CdLS Foundation National Family Conference and offers medical recommendations so families can share it with their own practitioner. She has seen over 250 individuals with CdLS as Medical Director. For many families, she is the first doctor they meet who has a full understanding of the syndrome. In 2017, Dr. Kline received an award from Global Genes as the RARE Champion of Hope in Medical Care and Treatment.

Dr. Kline remains the sole researcher of aging in CdLS cases in the United States, which provides challenges in its lack of previous findings and funding for such investigations. However, with every challenge, Dr. Kline has consistently searched for a solution without fail that not only benefits the families she assists, but the Foundation she cares about so deeply.
CdLS Centers For Excellence

The CdLS Centers for Excellence program recognizes demonstrated excellence and outstanding achievement in research and/or clinical work related to CdLS. The honorary distinction is given to institutions that continue to meet the established criteria of the CdLS Foundation.

Clinical Centers must offer a specialized multidisciplinary approach to patients in a specific CdLS clinic in a medical facility, demonstrate a strong knowledge base, commit to furthering clinical knowledge of CdLS through collaboration with others and distribute new clinical findings. Research Centers must pursue research that provides a significant contribution towards understanding the science behind CdLS, demonstrate a strong knowledge base of CdLS, commit to furthering scientific knowledge of CdLS through collaboration with others and discuss new scientific findings related to CdLS.

Greater Baltimore Medical Center

The Greater Baltimore Medical Center (GBMC) has been recognized by the Foundation for its premiere research and clinical work related to Cornelia de Lange Syndrome (CdLS). Pediatric geneticist Dr. Antonie Kline saw the need for a consistent CdLS clinic when she started working at GBMC in 2000. Kline says that projects that have stemmed from evaluating aging in CdLS are GBMC’s most promising endeavor in CdLS research. In the future, they envision continuing to move forward, both clinically and in basic science.

Children’s Hospital of Philadelphia

The Children’s Hospital of Philadelphia (CHOP) is one of the five Centers of Excellence for its research and clinical work related to CdLS. Drs. Laird Jackson, Ian Krantz, and Matt Deardorff have made the research and treatment of Cornelia de Lange syndrome (CdLS) and related disorders central to their body of work. The Center for Cornelia de Lange Syndrome and Related Diagnoses at CHOP provides managed medical and clinical care, and scientific research. The Center aims to improve the scientific understanding and medical management of CdLS. Research at CHOP led to the discovery of the first genes known to be associated with CdLS: NIPBL, SMC1A, and SMC3.
Saint Louis University School of Medicine

Saint Louis University School of Medicine has been recognized for its research and clinical work related to CdLS. Their involvement with the CdLS Foundation has been primarily through Dale Dorsett, Ph.D., and his laboratory staff. The staff conduct research into the causes and possible treatments for CdLS. They also have identified an FDA-approved drug that has shown positive results in the treatment of a fruit fly model of CdLS. Dorsett has hosted CdLS Foundation events, which included members of the CdLS Foundation Board of Directors and donors from the Saint Louis area. Dorsett has contributed at every CdLS Scientific and Educational Symposium.

Stowers Institute for Medical Research, Kansas City

Stowers Institute for Medical Research, is recognized for its research related to CdLS. The research taking place in the lab has helped changed the landscape of what is known about CdLS. The teams’ work also brings new hope to families of individuals who are affected by this syndrome. Stowers Investigator Jennifer Gerton, Ph.D., believes that understanding the basic biological function of the genes mutated in CdLS will reveal insights into potential therapeutic approaches. Gerton’s research contributes to a better understanding of the molecular basis for CdLS.

University of California, Irvine

University of California, Irvine, is recognized for the continued excellence and outstanding achievement in research work related to CdLS by five UC Irvine faculty members. Working with researchers at CHOP they helped identify the gene whose mutation is responsible for the majority of cases of CdLS, \textit{NIPBL}. The discovery of the \textit{NIPBL} gene led to the development of tools for molecular diagnosis of CdLS and has spawned a large body of biomedical research on CdLS and related syndromes. As part of this effort, the team has developed mouse and zebrafish models of CdLS that are being used to test methods for treating and/or helping to prevent changes such as cardiovascular disorders, changes in limb and hand structures and neurological problems that affect individuals with CdLS.
Lynn is a veteran of our mission and her work at the heart of what makes the Foundation come to life. Her experience, empathy and passion for her work as well as each individual with CdLS and their families makes her an invaluable member of the Family Services team.

Lynn began her career with the Foundation in 1996 as a Family Service Coordinator. She earned her Master’s Degree in Social Work from Boston University with a focus on working with families of children ages birth to three with special needs. Since joining the Foundation, Lynn has been an active member in the CdLS Foundation community by attending events, including Family Gatherings and National Family Conferences. She makes herself readily available to returning and new families to make sure they feel comfortable and have their questions answered.

She facilitated the creation of When Angels Take Flight, among other publications, and has been integral in expanding services for families throughout the years. Lynn has visited families in almost every state, welcoming them at to the community for the first time. She was also a champion in getting an individual with CdLS onto different Foundation committees. In 2016, Lynn was inducted into the de Lange Society; which represents those who have served the Foundation continuously for 20 years.

Her dedication to her work far extends beyond her job, but is evident in the compassion she shows to individuals with CdLS and their families nationwide year after year. When a new family calls, sometimes they are frantic or just needing someone to talk with. Lynn gracefully demonstrates how in that moment; they are our number one priority. With her expert knowledge of CdLS and support services, she is able to share what is needed. To ensure that families feel supported, she often follows up after that initial call to quell any lingering anxieties.

Lynn is passionate about her work because she can watch a family move from being scared, overwhelmed and frustrated to being an advocate for their loved one. Her passion comes from growing up with a brother with intellectual disabilities and knowing how important it is to see the person now the syndrome.
One Day that Changes Lives

The CdLS Foundation has been committed to expanding our ever-growing network of multidisciplinary clinics which are located in major medical centers throughout the country. The reason is simple – a one-stop location to interact with experts who have specialized knowledge on how to treat individuals with CdLS. In just one day, a family can be seen by a full complement of experts who then work together to provide families and caregivers a comprehensive treatment plan to take back home and share with their local medical and therapeutic professionals.

These very special clinics also create an opportunity for families to interact with other CdLS families and connect with the Foundation so we can help address their needs. This unique combination of medical treatment and social support creates a truly holistic experience.

In November 2017, with the generous support from donors, active family members and volunteer support, the Foundation established a clinic in the Midwest at St. Louis Children’s Hospital in St. Louis, Missouri.

Headed by CdLS geneticist, Dr. Kathy Grange, the two-day clinic gave families easy access to a wide range of specialists in gastroenterology, ophthalmology, genetics, neurology, audiology/ear, nose and throat and psychology. Families received a “head to toe” expert consultations and many attended a luncheon and a presentation about CdLS presented by Foundation Medical Director, Dr. Tonie Kline.

Jim Kesting, a CdLS Board member, had a strong desire to see a clinic opened in his home state of Missouri out of compassion for his good friend and business partner, Doug Gaines, whose daughter Amber has CdLS. Last year, Jim’s dream of creating a multidisciplinary clinic in his home state came true. “I understand how difficult it is for families to travel great distances to obtain critical information concerning their child’s health.”

We’re always working on opening new clinics around the country and look forward to creating more opportunities for families to receive this extraordinary level of care and the chance to connect with other families and the Foundation. To learn more about how you can support this life-changing initiative, please contact Bonnie Royster at director@CdLSusa.org.
In 2017, Walgreens stores across MA and RI partnered with the CdLS Foundation to help raise awareness and funds through a Scannable Event. Mike Christie, father to Taryn, CdLS Board Member and Director of Pharmacy and Retail Operations at Walgreens, commented, “Most people have not heard of CdLS, so we knew it was important to not only raise money but awareness of the syndrome that affects thousands of children and families. The folks at our stores were so proud that because of their efforts, families could get services they need, such as going to medical clinics that specialize in treating children with CdLS. The health and happiness of our community is what Walgreens is all about and we are so glad to make a difference.”

During the first two weeks of October of 2017, 95 Walgreens stores throughout MA and RI encouraged customers to purchase a scannable icon in donation amounts of $1, $3 or $5. After two weeks of fundraising, over $37,000 was raised. The funds supported programs and services provided by the CdLS Foundation, including multidisciplinary clinics, family support phone lines and awareness activities nationwide.

Caring individuals like Mike and you have the power to educate more medical, therapeutic and educational professionals about CdLS, provide additional research opportunities to further enhance the knowledge about the syndrome, as well as keep support lines open for families in need. It is simple, all you need to do is share your commitment to helping those affected by CdLS with someone else and the possibilities are endless.

You can partner with the CdLS Foundation in a variety of different ways including:

- Workplace Giving
- Monthly Giving
- Sponsorships
- Hosting Your Own Fundraiser
- Corporate Giving
- Planned Giving

“Most people have not heard of CdLS, so we knew it was important to not only raise money but awareness of the syndrome...”
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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