

# 2019 ANNUAL REPORT



# MESSAGE FROM THE EXECUTIVE DIRECTOR

Looking back over 2019, I feel a great sense of gratitude and pride for the many ways—with your help—we were able to impact so many lives.

We were privileged to welcome 135 new families to the Foundation, expanding our CdLS community to over 3,600 families across the United States. Together with our dedicated team of experts in CdLS, we hosted a series of webinars that delivered timely information, insights and support to hundreds of people in our community, directly into their homes. We reached over 600 people through these webinars that received rave reviews from those who tuned in. In 2019, the impact momentum continued with:

- 513 people attending Family Gatherings across the country
- 18 individuals and their families received support to travel to a life-changing Multidisciplinary Clinic,
- Over 925 individuals with CdLS and their families received direct services and emotional support from our ever-ready Family Service team.

On the research front, the Foundation awarded \$24,447 to three researchers in various fields, who are engaged in passionate pursuit of furthering the understanding of CdLS and how to improve treatment and lives.

This is just a glimpse of a landmark year of progress. In the following pages, you can read more about how our efforts and your financial support made 2019 a year to remember. We look forward to your continued support as we face the challenges and opportunities ahead for all of us in 2020 and beyond.

Sincerely,



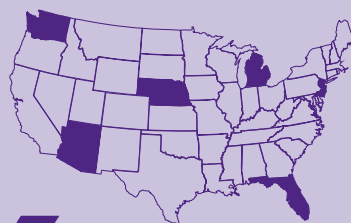
Executive Director

CdLS Foundation

# 2019 HIGHLIGHTS



The CdLS Foundation welcomed **135** new families



**6**

Foundation sponsored Family Gatherings were held with 513 families in attendance, where friendships were formed

**359**

Ask the Expert questions were answered



Research grants were awarded totaling **\$24,447**

**\$116,737**

raised at 2 golf tournaments



**\$154,110**

was raised by Team CdLS for individuals with CdLS and their families



Our PBS Segment was launched on February 28th (Rare Disease Day)



With over **7,000** views on our YouTube Channel



The CdLS Foundation hosted **6** webinars that discussed important topics including: behavior, genetics and GI





# Jupiter Gunter

## LIVING MOMENT TO MOMENT

When a child is born with CdLS, parents often reach out to the CdLS Foundation seeking advice. In 2019, when Jupiter Gunter was born, he was immediately diagnosed with CdLS. His parents, Shaunita and Christian laid in the hospital bed and knew they could not just be defeated. Shaunita looked up “help me understand Cornelia de Lange Syndrome” and the Foundation was the first thing that came up. They reached out to the CdLS Foundation the day their son was born. That very night, both parents made a commitment to educate themselves and their family about what the road ahead might be for their precious son.

The young parents had to quickly adapt to a “new normal” that included the challenges of not being able to hold their baby for weeks at a time and enduring the pain of watching him go through multiple surgeries. The CdLS Foundation offered emotional support to Shaunita when Jupiter was going through those surgeries. Family Service was there for her when she needed guidance and help navigating the intense medical arena.

Shaunita and Christian also came to rely on support from the CdLS Foundation and believe it significantly helped lighten their load and

hearts. For instance, when their doctors insisted that Jupiter needed a tracheotomy, they turned to the Foundation for advice. Whitney from the Foundation’s Family Service provided a referral to a CdLS specialist that helped Shaunita and Christian make a more informed decision with which they were all comfortable.

Shaunita confirms that, “Without the Foundation so many families would be lost. Having someone with more in-depth insight on what Jupiter needs helps us pass along information [to our son’s doctors] so that our son receives the best care.” Above all, Shaunita and Christian believe, “The CdLS Foundation has shown us the definition of unconditional love and that our babies are simply perfect.”

# WEBINARS

## A Look into the Future

Over the past year, the CdLS Foundation hosted six webinars for the CdLS community. The goal of these workshops was to expand service lines to individuals with CdLS and their families to gain knowledge and support, especially to those in isolating conditions. The Foundation also wanted to educate and inspire individuals affected by the disorder, their families and the professionals that work with them. Experts in CdLS shared their knowledge in subjects ranging from genetic testing, addressing challenging behavior, neurodevelopmental behavior and behavioral management, GI challenges, Conference fundraising tips, and how to prepare for retirement.

Over 400 individuals registered and over 200 individuals attended the webinars live. Each webinar offered the chance for the audience to interact with the presenter and ask questions and participate in polls or discussions that enhanced the viewing experience.

Webinars were received by the community with positivity and praise. One attendee gave feedback stating, "THANK YOU!! I'm so happy there are resources available for us and for other CdLS families, between the Foundation and the FB discussion board, we are feeling supported beyond the medical folks involved in [our daughter's] care. We were worried that given the rarity of the condition, doctors would not address issues specific to CdLS sufficiently (given lack of their own experience). Having the Foundation certainly provides us a reference point and a source of valuable information."



## Gastrointestinal (GI) Challenges: Critical Care and Treatment Approaches

This webinar provided an overview of malrotation, volvulus, bowel obstruction and pancreatitis. It also reviewed treatment and care approaches to these challenges.

Presenter: Carol Potter, M.D..

## Neurodevelopmental Disorders and Behavioral Management in Cornelia de Lange Syndrome (CdLS)

This webinar provided an overview of neurodevelopmental challenges, including approaches to treatment, in Cornelia de Lange syndrome (CdLS). Individuals with CdLS can be affected by a number of developmental disorders, including intellectual disability, autism spectrum disorder, language disorder, ADHD, aggression, repetitive self-injurious behaviors, anxiety, and depression. This webinar will discuss each of these domains as it relates to CdLS, as well as strategies for pharmacological intervention.

Presenter: Siddharth Srivastava, M.D.



## How to Address Challenging Behaviors Webinar

This webinar will discuss:

- Self-injury
- Impulse control
- Destruction of property
- Determining the appropriate consequences
- Treatment options (ABA, etc.) or professional resources for families

Presenter: Julia O'Connor, Ph.D.





# Research to Expand Collaboration and Tackle Difficult Challenges

In 2019, the CdLS Foundation's Research Committee awarded \$24,447 to three researchers in various fields all in the pursuit of furthering the understanding of CdLS.

The continuing goal of the Research Program at the Foundation is to allow researchers to learn insights 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 upcoming CdLS Scientific and Educational Symposium. The recipients of the grants are:

## John Michael Falligant



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.



## Stephenson Chea

---



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. Investigating this will not only increase our understanding of when and how the first developmental changes occur in CdLS; it will also uncover new stem and progenitor cell-specific genes that could be the focus of future therapies.

## David F. Smith

---



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 also 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.

# Innovator and Pioneer of Research into CdLS

Dale Dorsett, Ph.D.

When someone mentions the name Dr. Dorsett the CdLS community collectively smiles. Dr. Dorsett has been part of the research community since 1980 when he received his Ph.D. in Biochemistry from the University of Tennessee-Oak Ridge National Laboratory and conducted postdoctoral work in molecular biology and genetics at the Weizmann Institute of Science in Rehovot Israel from 1981-1985 and Harvard University from 1985 to 1987. He joined the faculty at Memorial Sloan-Kettering Cancer Center in New York City in 1987 and moved to Saint Louis University of School of Medicine in 2000. He retired at the end of 2019 and is now Professor Emeritus of Biochemistry at Saint Louis University and Visiting Professor of Biology at the University of Rochester in Rochester, New York.



Dr. Dorsett's involvement in research into the biology of Cornelia de Lange Syndrome began because he discovered the Nipped-B gene in *Drosophila* (fruit fly) based on its diverse roles in development. Nipped-B is the *Drosophila* equivalent of human *NIPBL*, and he began a

collaboration with Dr. Ian Krantz, after the Krantz laboratory discovered that most cases of CdLS are caused by changes in the *NIPBL* gene.



“Without his work we would not have an in depth understanding of how the genes that cause CdLS function and he has led the charge to find ways to try and correct these changes and to begin the process of developing targeted therapeutics for CdLS. Dale as a basic scientist, is also unique in his drive to try to understand the impact that CdLS had on the lives of the affected individuals and their families – I remember that first year when we identified the gene Dale came to the Foundation meeting and presented his work to the families. Trying to describe *Drosophila* (fly) genetics to non-scientists was no easy task – but after his talk there was a standing ovation and at least one family came up to him afterwards to say they were throwing out their fly swatters now that they realized the importance of fly research!! Dale went on to work for the foundation and support their research efforts and regularly attends the Foundations conferences. Dale’s work led to many National institutes of Health (NIH) and March of Dimes (MOD) grants to study cohesin and CdLS and he has transformed our understanding of the causative genes and has made a lasting impact on the families and on the direction this research has taken and will continue to take in the future,” reminisced Dr. Ian Krantz.

He served on the CdLS Foundation Board of Directors for six years, as well as the Clinical and Research Advisory Committees. Dr. Dorsett received the Laird Jackson award for his contributions to CdLS Research in 2010 and his laboratory at Saint Louis University was designated a CdLS Research Center of Excellence from 2008 until his retirement at the end of 2019. Dr. Dorsett currently serves as a Deputy Editor of Biology for the Science Advances journal published by the American Association for the Advancement of Science.



# Whitney Rinaldi

## A Compassionate Friend to Walk the Journey With

There is a moment in a person's life that defines who they have become in their career. Whitney's defining moment was when she attended her first Family Gathering. When she arrived in Arizona she was overwhelmed with excitement. Up to this point, she had been communicating with families mostly through email or phone calls, now she was meeting them in person and this completed the circle for her. It was an incredible moment to see

individuals with CdLS and their families connecting – for Whitney it made her feel like she was where she was meant to be.

Whitney is a part of the dynamic and caring Family Service Team and has been part of the CdLS Foundation for over three years. On any given day she is tasked with helping a family cover the entire CdLS journey. She can help a mom with a newly diagnosed child to a veteran family that has questions about a new behavior issue. There are sessions of joy, when a child learns to use an AAC device and there are discussions of sorrow, when a loved one passes away. Whitney believes that there is a such a broad spectrum of care and she wants to be able to provide any service a family may need.

The families and individuals with CdLS are what inspire Whitney. With the nature of her job she never knows what the day may hold. She really loves being the person they call when they feel they need help, connections, to vent or share a great achievement. Being able to talk to someone that understands is something that Whitney finds value in. Many times,





families are able to just talk to her and know that she understands. Whitney believes that the Foundation provides a safe haven for families to call, receive assistance with confidentiality and not feel judged.



“There is always a need to enhance and update services. The more we are in contact with families, the more we are aware of the needs of our community. Identifying needs and being creative in the implementation of services is a way to address the demand,” Whitney shared. “Working with individuals with CdLS allows me the opportunity to make a difference – however large or small, in the lives of families. This community shows me how important relationships are in any given situation. As a new mom, it opened my eyes to how important it is to connect with other people for your physical and emotional health. You are not alone.”

The support Whitney provides to individuals with CdLS and their families is life-changing and allows someone to get assistance no matter where they are in their journey. Family Service wants families to know they are available even when there isn't a need.

*“Working with individuals with CdLS allows me the opportunity to make a difference – however large or small, in the lives of families.”*

# Planned Giving:

## WHY START NOW

For a moment, think about all the ways the CdLS Foundation has been here to support your child and family over the years. Now consider how important it is that the Foundation continues to be here when a child is first diagnosed; when a family has questions; when a medical professional needs to consult with a CdLS expert to create a better treatment plan; when a researcher needs the funds to pursue a new lead in understanding the syndrome that will result in better care for all.

Planned giving is one of the most significant ways to give yourself the peace of mind that the CdLS Foundation will continue to be here to serve families like yours in the years to come.

Planned giving may sound complicated, but it's actually simple—and there are a variety of ways to do this. For instance, you can request a gift be given to the Foundation through your will or a trust; you can make the Foundation a beneficiary in your life insurance policy. And whatever you plan to do to support the future well-being of the CdLS Foundation, it will not alter your current lifestyle and can be easily changed at any time.

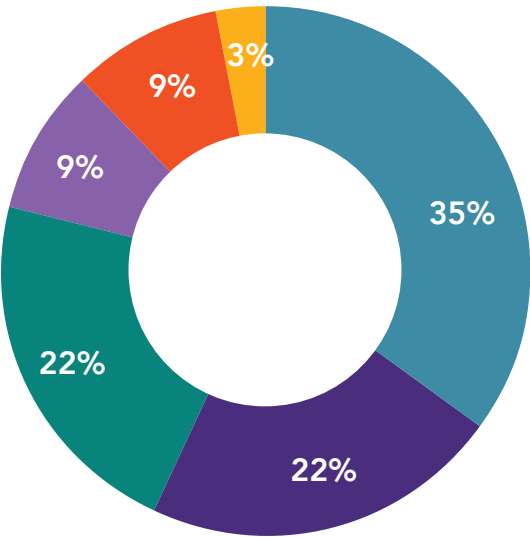
Planned giving has become a source of stability hope for the future because of people like you, families and friends of the CdLS Foundation. "The Foundation has been so important to us. Knowing how important it has been to others, we knew we wanted it to continue in perpetuity. You prepare your will to take care of your family. We feel like the Foundation is a part of our family." -Planned Giving Donor

If you would like to learn more about planned giving talk to your financial advisor and learn more about the easy ways you can make a Planned Giving gift. To learn more, contact Annette Scheidecker, Development Director at [PlannedGiving@CdLSusa.org](mailto:PlannedGiving@CdLSusa.org).

# 2019 Audited Income and Expenses

## 2019 Expense

January 1 - December 31, 2019



- Family Support Program

■ General & Administration

■ Public Information & Education

■ Fundraising

■ Professional Development Outreach & Awareness

■ Research

INCOME	
Individual Donations	21,316
Corporations/Company Donations	28,927
Foundations/Grants	8,600
Major Donors	229,681
Direct Mail/Foundation Drives	29,020
Clubs and Organizations	4,092
Gifts that Count – Memorial/Celebration	39,522
Special Events/ Third Party Fundraising	401,123
Federated Campaigns	67,110
Bequest	1,443,368
Program Services Fee/ Nat’l Conference & 21st Century Fund	16,890
SMC1A Research	16,062
Total Operating Revenue	2,305,713
Net Loss from Asset Disposal	(2,846)
Investment Net Income	134,291
Total Revenue	2,437,157

EXPENSES	
Program Services	
Professional Development, Outreach and Awareness	219,967
Research	26,265
Family Support Program	343,002
Public Information and Education	215,549
Total Program Services	804,783
Support Services	
Management and General	83,987
Fundraising	86,898
Total Support Service	170,885
Depreciation	6,158
Total Expenses	981,826
Net Assets, Beg of Year	975,826
Surplus/Deficit from Operations	1,321,041
Unrealized Gains/Losses	134,291
Total Net Assets	2,430,453

## 2019 Sponsors

BELLON WRECKING & REHABBING	DOUG GAINES	MCCLAIN FOREST	PLM	WILSQUARE CAPITAL	NORTHERN BANK & TRUST COMPANY
AMBASSADOR ENTERPRISES	DRIPS/BIG BAGS	MCCLAIN FOREST PRODUCTS	R&E PROPERTIES	ALL AMERICAN/EVERYDAY GOURMENT	PAUL FRISOLI
AMRISC	FSK	MADISON COUNTY WOOD PRODUCTS	R.C. STORES	ARDITO, TOSCANO	QUMULO
BOTKIN LUMBER	GUARDIAN	MCWP EMPLOYEES	REED LUMBER CO	BRIAN & MARIE BRUECKS	ROGER & PATTY LINNEMAN
BURNS & WILCOX	HAMPTON KIWANIS	MENOMINEE SAW	ROBERT ABOUSSIE FAMILY	CAMP EATON	TECH CENTER/MAIRANO
CABKA	HENGES INTERIORS	MFA OIL	RUSTIC WOOD	CATRICKES	THE CHAMPION FAMILY
CAMBRIDGE AIR SOLUTIONS	HOGAN	MID-CONTINENT NAIL	SARGENT CONSTRUCTION	DANIEL HOPGOOD	TONIA KINGSTON
CASS BANK	HUB INDUSTRIAL	MO FOREST PRODUCTS	SCHNUCKS	DIANNE & VAL LESSA	WORKER'S CREDIT UNION
CHEROKEE PASS MOBIL	JIM & JACKIE DUFFER	MO POWER TRANSMISSION	SCHWARZE TRAILER REPAIR, INC	DUKE PASCUCCI	DOWN TO BASICS
CLEAN UNIFORM	JOHN HENRY FOSTER CO	MO WOOD INSURANCE	SONUS BENEFITS	FISHER ENGINEERING	EDWARD JONES INVESTMENTS
COOPER MACHINE	KAISER & JOHNSON	MONTGOMERY BANK	SWINFORD REALTY	FRED & LYDIA KEACH	FGMK, LLC
CRANE AGENCY	KAMPS	NAEGER FOREST PRODUCTS (NFP)	TAG TRUCK CENTER	JERRY DOYLE	GEOFFREY METALS, INC.
CRH TRANSPORTATION	KESTING & DIECK FAMILIES	NEW YORK LIFE INSURANCE	THE MONSCHEIN TEAM	JOHNSON FAMILY	GEROME TECHNOLOGIES
CROWN MACHINERY	MAGNUM FASTENERS	ON/OFF LOGISTICS (FALM)	TIMBERLAND FOREST	LAWRENCE & JEAN TODESCA	J.C. ANDERSON, INC.
DANIEL & HENRY	MARCK INDUSTRIES	PALLET MACHINERY GROUP	UHY	LYNN WELLS	
DAVID WIESS	MARCO FINANCIAL	PALLET REPAIR SYSTEMS	UMB ST. LOUIS	MCCARTHY INSURANCE	
DIVA DANCE COMPETITION, INC.	MARIANO FAMILY		UNITED LUMBER & REMAN	NORM & BARB WINNERMAN	
	MCBRIDE MACK SALES				

# The Current CdLS Foundation Family

## PROFESSIONAL STAFF

EXECUTIVE DIRECTOR  
Bonnie Royster

MEDICAL DIRECTOR  
Antonie Kline, M.D.

FAMILY SERVICE MANAGER  
Deirdre Summa, L.M.S.W.

FAMILY SERVICE COORDINATORS  
Whitney Rinaldi, B.S.W.  
Alicia Jackson, M.S.W.

DEVELOPMENT DIRECTOR  
Annette Scheidecker

DEVELOPMENT ASSISTANT  
Vanessa Hatten

COMMUNICATIONS MANAGER  
Gabrielle Nadeau

GRAPHIC DESIGNER/WEB MASTER  
Francesca Scognamiglio

OFFICE COORDINATOR  
Bonnie Kraft

FINANCE DIRECTOR  
Silvia Goldman

## BOARD OF DIRECTORS

PRESIDENT  
Mike Christie, Massachusetts

VICE PRESIDENT  
Eric Johnson, Connecticut  
Lynne Kerr, M.D., Utah

TREASURER  
Mike Feehan, Pennsylvania

SECRETARY  
Katherina Terhune, Kentucky

DIRECTORS  
Jennifer Gerton, Ph.D., Missouri  
Jim Kesting, Missouri  
Arthur Lander, M.D., Ph.D, California  
Patrick Lyons, New Hampshire  
Emily Turner, Alabama  
Angie Young, Florida

## PROFESSIONAL DEVELOPMENT COMMITTEE

Geir Rosvik, M.Ed.  
Julia O'Connor, Ph.D.  
Mary Pipan, M.D.  
Denise Williams, MS  
Urbi Ghosh, Ph.D.  
Rachel Hueston  
Mary Beth Bruder, Ph.D.  
Ingrid Barone, R.N.  
Christine Ackermann, M.S., O.T.  
Joann Fleckenstein, M.S., O.T.  
Amy Metrena, M.S.P.T.  
Mary Levis N.C.S.P.  
Shelly Champion, M.Ed.

Adaptive Physical Education  
Clinical Psychology  
Developmental Pediatrician  
Early Childhood Education  
Education  
Education  
Educational Psychology  
Nursing  
Occupational Therapy  
Occupational Therapy  
Physical Therapy  
School Psychology  
Special Education

## CLINICAL ADVISORY BOARD

Dale Dorsett, Ph.D.  
Jennifer Gerton, Ph.D.  
Marco Grados, M.D., M.P.H.  
Ian Krantz, M.D.  
Matthew Deardorff, M.D, Ph.D.  
Yves Lacassie, M.D.  
Antonie Kline, M.D.  
Trevor Hoffman, M.D., Ph.D.  
Arthur Lander, M.D., Ph.D.  
Joseph McCleery, Ph.D.  
Alena Egense, MGC, CGC  
Amy Kimball, M.S.  
Sarah Raible, M.S.  
Natalie Blagowidow, M.D.  
Michele Lambert, M.D.  
Soma Jyonouchi, M.D.  
Clair Francomano, M.D.  
Soma Das, Ph.D., FACMG  
Anne Calof, Ph.D.  
Kari Cunningham-Rosvik, APRN  
Ronald Berger, M.D.  
Aaron Zuckerberg, M.D.  
Jeff Rockow, M.D.  
Douglas Clemens, D.M.D.  
Richard Mungo, D.D.S.  
Rochelle Lindemeyer, D.D.S.  
Joni Rampolla, L.D.  
Loretta Gore-Harvey, R.D., L.D.N.  
Carol Potter, M.D.  
Kathleen Loomes, M.D.  
Lynette Gillis, M.D.  
Kristen Baranano  
Lynne Kerr, M.D.  
Siddharth Srivastava, M.D.  
Alex Levin, M.D.  
Christianne Schoedel, M.D.  
Susan Schloff, M.D.  
Stacey Ishman, M.D.  
Katherine Farr, D.P.T.  
Dorothy K. Grange, M.D.

Mark Kliewer, M.D.  
Richard Haaland, Ph.D.  
Cheri Carrico, Ph.D., CCC-SLP  
Patti Caudill, M.S., CCC-SLP  
Nancy Mathis, M.S., CCC-SLP  
Ming-Hsien Wang, M.D.

Biochemistry & Molecular Biology  
Biochemistry & Molecular Biology  
Child Psychiatry  
Clinical Genetics  
Clinical Genetics  
Clinical Genetics  
Clinical Genetics  
Clinical Genetics  
Developmental Biology  
Developmental Neuroscientist  
Genetic Counselor  
Genetic Counselor  
Genetic Counselor  
Gynecology, Genetics  
Hematologist  
Immunology  
Internal Medicine, Genetics  
Molecular Genetics  
Neurodevelopmental Biology  
Nurse Practitioners  
Ophthalmology  
Pediatric Anesthesia, PICU  
Pediatric Cardiology  
Pediatric Dentistry  
Pediatric Dentistry  
Pediatric Dentistry  
Pediatric Dietetics  
Pediatric Dietetics  
Pediatric Gastroenterology  
Pediatric Gastroenterology  
Pediatric Gastroenterology  
Pediatric Neurology  
Pediatric Neurology  
Pediatric Neurology  
Pediatric Ophthalmology  
Pediatric Ophthalmology  
Pediatric Ophthalmology  
Pediatric Otolaryngology  
Physical Therapy  
Professor of Pediatrics, Genetics  
& Genomic Medicine  
Radiology  
Research  
Speech-Language-Feeding  
Speech-Language-Feeding  
Speech-Language Pathology  
Urology



**CdLS Foundation**

Cornelia de Lange Syndrome Foundation, Inc.

30 Tower Lane, Suite 400

Avon, CT 06001-3681

800.753.2357 • [www.CdLSusa.org](http://www.CdLSusa.org)