

RESEARCH

Research into the origin and treatment of CdLS is part of the Foundation's mission to serve individuals with CdLS and their families. Since its inception, the Foundation has supported research to better understand issues experienced by individuals with CdLS. It also supports research to better understand underlying causes of CdLS.

PUBLIC OUTREACH

Raising awareness and advocacy about CdLS to families, the medical community and general public is central to the Foundation's mission. Greater awareness can spur earlier diagnosis and more effective treatment. Advocacy helps increase awareness while raising funds for the work the Foundation does.

CdLS PUBLICATIONS

The Foundation also distributes a wide range of helpful and thoughtful publications. The Foundation's signature publication, *Reaching Out*, is filled with useful articles from CdLS experts and family members who share their triumphs and challenges. Communications in Spanish are also available.

QUICK FACTS ABOUT CdLS



CdLS is rare—it occurs in 1 in 10,000 live births



CdLS spectrum disorder is genetic & present from birth



CdLS affects both genders and all races equally

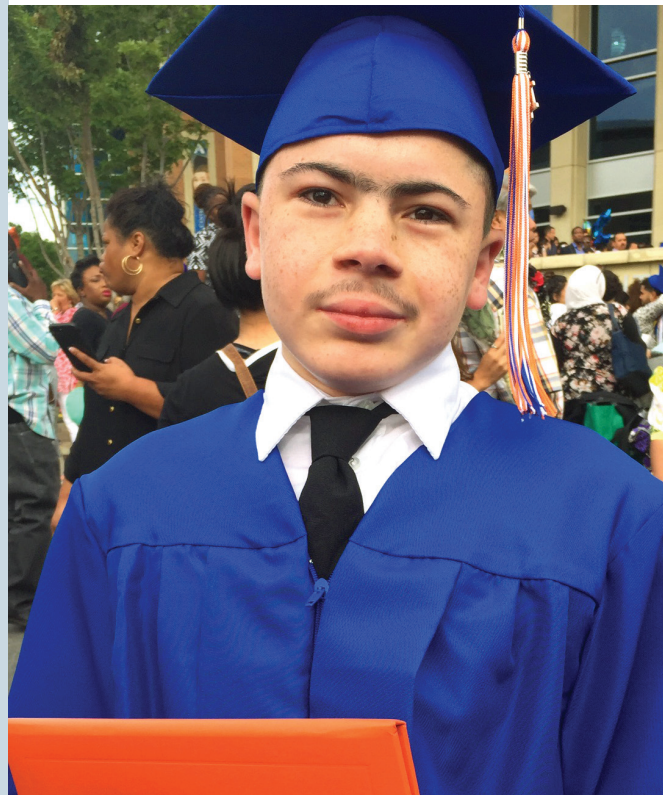


Common medical issues associated with CdLS:

- Gastrointestinal Issues (GI)
- Self-Injurious Behavior (SIB)
- Slow Weight Gain
- Seizures



Those living with CdLS are resilient #cdlsstrong



ABOUT THE CdLS FOUNDATION

The CdLS Foundation is a national 501(c)(3) non-profit organization that has served individuals with CdLS and their families since 1981.

The Foundation's mission is reflected in its slogan: *Reaching Out, Providing Help, and Giving Hope*. It is the only family support organization dedicated to CdLS in the nation. One of the goals is to establish community and enable advocacy for those experiencing CdLS and other isolating conditions.

FACTS ABOUT CORNELIA de LANGE SYNDROME & THE CdLS FOUNDATION



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 **CdLS Foundation**
Cornelia de Lange Syndrome Foundation, Inc.

WHAT IS CORNELIA de LANGE SYNDROME (CdLS)?

CdLS is a rare genetic disorder present from birth. It causes such a broad range of potential physical, cognitive and medical challenges that it is now known as the CdLS spectrum disorder. CdLS does not discriminate— it affects both genders equally and it's seen in all races and ethnic backgrounds. The occurrence of CdLS is estimated to be 1 in 10,000 live births, but because it is so variable, could remain undiagnosed.

CHARACTERISTICS OF CdLS

CdLS is not a “one size fits all” condition. Individuals with CdLS often strongly resemble one another. Typical facial features include thin eyebrows that meet in the middle, long eyelashes, a short-upturned nose and thin downturned lips.

Other characteristics include low birth weight (often under five pounds), slow growth, small stature, and small head size. Other features may include excessive body hair and small hands and feet.

Limb differences, including missing arms, forearms or fingers, are seen in about 25% of individuals with CdLS. Not all individuals with CdLS will have all the characteristics listed above.



HOW IT'S DIAGNOSED

The diagnosis of CdLS is primarily a clinical one based on signs and symptoms observed through an evaluation by a physician, including a medical history, physical examination and laboratory tests. However, genetic testing can be helpful in confirming the clinical diagnosis and assessing which gene is involved. Eighty percent of individuals can be diagnosed through genetic testing. If you suspect your child has CdLS, a genetic specialist can help.

Genetic testing is available at various locations throughout the United States. For a complete list of laboratories that offer testing for CdLS, please visit our website at www.cdlsusa.org/genetic-information.

FAMILY SERVICE

Our caring and knowledgeable Family Service Coordinators provide individuals with CdLS and their families a wealth of services, support and information. Calls placed to our support team are answered by caring professionals who listen, answer questions based on years of experience and offer individualized support over the phone and email.

Services Include:

- Ask the Experts: This service enables users to email their questions for response from members of our Clinical Advisory Board and Professional Development Committee
- Family Gatherings: These events are wonderful opportunity to meet with other families and share stories, resources and information.
- CdLS Clinics: Provides families with priceless head-to-toe evaluations by a variety of experts—all in one day. There are five CdLS Clinics in the United States:

- The Multi-disciplinary Clinic for Adolescents and Adults at Greater Baltimore Medical Center
- The Center for CdLS and Related Diagnoses at Children's Hospital of Philadelphia (CHOP)

- Shriners Hospital for Children-Salt Lake City, UT
- St. Louis Children's Hospital in St. Louis, MO
- Boston Children's Hospital, Cornelia de Lange Syndrome (CdLS) and Related Disorders Clinic

- National Family Conference: This life-enhancing, 4-day event bonds and strengthens attendees. Held every other year, Conference brings together the broad spectrum of the diverse CdLS community.
- Grief Support: Foundation staff is here to provide guidance on the complicated journey after losing a loved one with CdLS. Staff is here to listen, honor and remember your loved one.

