

Events & Fundraising

Special events such as golf tournaments, walks/runs, bowl-a-thons, and bake sales are held throughout the country to raise funds in support of the CdLS Foundation. Foundation-sponsored events include Team CdLS and 5K Club, golf tournaments and Cornelia's Garden.

To learn more or to host an event in your area, contact events@CdLSusa.org.



Volunteers

A core component of the Foundation is its volunteers. Hundreds of parents, relatives and friends give their time and energy to help improve the lives of people with CdLS. Volunteers can sit on committees that plan and implement Foundation services and events, or participate in Awareness Coordinator and/or Regional Coordinator programs.



Research

The Foundation has a vision and researchers have the determination to understand CdLS and find treatments to help people with the syndrome overcome the many challenges. Through its CdLS Grants Program, the Foundation has funded research projects including studies on behavior, immunodeficiency, aging, and blood disorders.

Centers of Excellence

The Foundation has established relationships with researchers at prominent medical and scientific institutions. CdLS Centers of Excellence are Greater Baltimore Medical Center, the University of California at Irvine, the Children's Hospital of Philadelphia, Saint Louis University School of Medicine, and Stowers Institute for Medical Research in Missouri.

Public Outreach

Thousands of men, women and children remain undiagnosed and/or without proper support services. In order to raise awareness of CdLS among the public, the Foundation produces awareness materials such as bookmarks, fliers, brochures, and public service announcements. Materials are available upon request.

Additionally, National CdLS Awareness Day is observed on the second Saturday of May each year.

About the CdLS Foundation

The CdLS Foundation is a 501(c)(3) nonprofit organization founded in 1981. It's the only organization in the country devoted to the syndrome. The Foundation 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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Facts about CdLS and the CdLS Foundation

 **CdLS Foundation**
Cornelia de Lange Syndrome Foundation, Inc.
REACHING OUT. PROVIDING HELP. GIVING HOPE.

www.CdLSusa.org

What is Cornelia de Lange Syndrome?

Cornelia de Lange Syndrome (CdLS) is a congenital syndrome, meaning it is present from birth. It causes a range of physical, cognitive and developmental disabilities, and occurs in approximately 1 in 10,000 live births. The effects of CdLS range from mild to severe. It affects both genders equally and is seen in all races/ethnic backgrounds.

Signs & Symptoms of CdLS

As with other syndromes, individuals with CdLS strongly resemble one another. Common characteristics include low birth weight (often under 5 pounds), slow growth, small stature, and small head size. Typical facial features include eyebrows that meet in the middle, long eyelashes, a short upturned nose, and thin downturned lips. Other features may include excessive body hair and small hands and feet.

Common medical issues include:

- Gastroesophageal reflux disease
- Heart defects
- Seizures
- Feeding difficulties
- Vision problems
- Hearing loss

Missing arms, forearms or fingers are also present in some individuals. Behavioral, communications and cognitive challenges often exist and may vary in severity.

Not all individuals with CdLS will have all the signs and symptoms mentioned above.



How it Happens

Researchers have identified multiple genes that, when altered, cause CdLS. These genetic changes (called mutations) occur in individuals with CdLS that *usually* are not present in their parents.

How it's Diagnosed

The diagnosis of CdLS is primarily clinical based on signs and symptoms observed through an evaluation by a physician, including a physical examination, medical history, and laboratory tests.

Genetic testing for most genes is available through the University of Chicago (call 773.834.2627). The Children's Hospital of Philadelphia offers select genetic testing on a research basis (call 215.590.4248).

For a complete list of laboratories that offer CdLS testing, please visit www.cdlsusa.org/research/genetic-information.htm.

Family Services

Family Service Coordinators provide support weekdays from 8:30 a.m. to 5 p.m., Eastern Time.

Services include:

- Ask the Expert (medical/educational)
- Connect with a Family
- Regional Family Gatherings

In addition, the Foundation holds a biennial national family conference and at least eight family gatherings throughout the country.

Publications/materials available:

- *Facing the Challenges: A Guide to CdLS*
- New Family and Professional Packets
- Education Handbook
- Transition Guide
- Grandparent Booklet
- *Reaching Out* (quarterly newsletter)
- Management and Treatment Guidelines
- Diagnostic checklist
- Bereavement resources



Reaching Out, Providing Help, Giving Hope

Clinics

Greater Baltimore Medical Center offers a twice-yearly Multidisciplinary Clinic for Adolescents and Adults. For information, call the Foundation at 800.753.2357.

The Children's Hospital of Philadelphia operates the Center for Cornelia de Lange Syndrome and Related Diagnoses. The clinic meets monthly. For information, call the Center at 215.590.4248.

The CdLS Clinic hosted by Shriner's Hospital in Salt Lake City, Utah, held annually, is for children 18 years old and younger. For information, call the Foundation at 800.753.2357.

Professional Development

The Foundation sponsors a Scientific and Educational Symposium in conjunction with its biennial family conference. The multi-day meeting brings together doctors, scientists and interested professionals from around the world to share current research and stimulate collaboration. Continuing Medical Education (CME) credits and Continued Education Units (CEU) are offered.

The Foundation also offers online self-study modules for professionals. Some offer CME credits at no cost.