

## The Children's Hospital of Philadelphia has one of the world's best teams of specialists with expertise in Cornelia de Lange syndrome.



Andrew, CdLS Center Patient

### How many times will I visit?

If you live far away, likely one visit will be sufficient. After a complete evaluation we will provide a written plan to share with your regular pediatrician and specialists and encourage you to keep in touch by phone or e-mail and to visit again if necessary.

If you live nearby, we may see you more frequently. We will work with you and your doctors to make sure your care remains consistent and appropriate and is improving your child's life.

### What if we can't travel to Philadelphia?

We strongly encourage a visit to the center here in Philadelphia. If this is not possible, we offer a "virtual consult." We will request the same information as if you were coming in person and may interview you by phone. We will evaluate the information, offer our diagnostic impression, and answer any questions you may have about your child or the diagnosis in general.

### May I bring my adult child to the center?

Of course! We have evaluated patients in their 30s and 40s. We will put together a plan for care regardless of age.

### What are your research goals and will we be able to participate?

We have a large, and growing, database of genetic information from children and adults with CdLS and related diagnoses. This database allows us to gain a more in-depth understanding of the clinical issues associated with various diagnoses so as to better serve these populations medically. There are also many ongoing basic science studies being performed in laboratories to understand the molecular basis of these various diagnoses and how these changes cause the differences seen in the affected individuals.

The goal of this research is to better understand what role these genes play in development, with the hope of using this information to improve medical treatment and developmental outcomes for your family members.

Families are invited to voluntarily participate in the various research studies. This may require that you and your child contribute a blood sample. Participation in these research studies is voluntary and if you choose not to participate, the level of care we provide to your child will not be affected.

With the help of families, we will continue to make great strides in research.

### The Center for Cornelia de Lange Syndrome and Related Diagnoses: Our Mission

The Center for Cornelia de Lange Syndrome and Related Diagnoses at The Children's Hospital of Philadelphia exists to provide clinical care and to improve the lives of children and adults with Cornelia de Lange syndrome (CdLS) and related diagnoses, and their families. This permanent, self-sustaining center offers the opportunity to translate clinical and bench top research towards the development of improved management and therapeutics for affected individuals and to provide a setting in which they can receive coordinated care and comprehensive services.

### For Additional Information

We encourage you to contact the center directly if you have questions or would like more information. Please call Dinah Clark at 215-590-4248 or e-mail [cdlscenter@email.chop.edu](mailto:cdlscenter@email.chop.edu).

A source of information for families is [www.cdlsa.org](http://www.cdlsa.org). This is the Web site of the Cornelia de Lange Syndrome Foundation, an organization that is a great resource for family support and education.

**Thank you!**



The Children's Hospital  
of Philadelphia®

Founded in 1855, The Children's Hospital of Philadelphia is the birthplace of pediatric medicine in America. Throughout its history, a passionate spirit of innovation has driven this renowned institution to pursue scientific discovery, establish the highest standards of patient care and train future leaders in pediatrics. For a century and a half, Children's Hospital has served as a haven of hope for children and families worldwide.

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## The Center for Cornelia de Lange Syndrome and Related Diagnoses



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## The Best Team

The Center for Cornelia de Lange Syndrome and Related Diagnoses is composed of an internationally known team at The Children's Hospital of Philadelphia committed to children with conditions affecting multiple systems of the body, including structural birth defects, growth and cognitive delays. Our team has a high level of expertise specific to these rare conditions. To provide the best medical care, we work together, across many specialties and with primary care providers and families, so that every patient has a plan for care that optimizes cognitive development and quality of life.

The Center for Cornelia de Lange Syndrome and Related Diagnoses is also an international leader in research, committed to identifying the causes of these conditions and treatments to alleviate their effects. To learn, to play, to enjoy life — all children deserve to experience these simple joys, and this motivates our research.

## For the Children

While children with complex multisystem development diagnoses bring much joy and happiness into their families' lives — and our lives — they face many challenges. Our patients may look different. They often have a harder time communicating. They experience more medical difficulties, and more pain. However, with the support of a loving family and a knowledgeable healthcare team, they are capable of great progress.

The team at the Center for Cornelia de Lange Syndrome and Related Diagnoses feels privileged to serve these beautiful children, and adults, and their families. We are committed to their well-being, now and for the future.

**With the support of a loving family and a knowledgeable healthcare team, our patients are capable of great progress.**



*(Above) Tyler, 1, with his mom, Heidi, and Center staff Ian Krantz, M.D., and Dinah Clark.*

## What is Cornelia de Lange syndrome?

Cornelia de Lange syndrome (CdLS) is a genetic condition present at birth and is characterized by numerous physical, intellectual and behavioral differences. Children with CdLS usually have low birth weight, are smaller in size and height and have a smaller head circumference (microcephaly). Most experience developmental delays that range from mild learning disabilities to profound mental retardation. Common physical characteristics of CdLS include: similiar facial features (which may include an upturned nose, eye-brows that meet in the middle, long eyelashes and low-set ears); gastroesophageal reflux disease (GERD), which can make eating painful and contribute to slow growth and other intestinal differences; and upper limb differences ranging from small hands to missing fingers or forearms. Cleft palate, diaphragmatic hernias, vision and hearing problems, excessive body hair (hirsutism), heart defects, seizures and dental issues are also common. Behavioral issues, such as self-injury or attention deficit hyperactivity disorder, might also be present.

The syndrome takes its name from the Dutch pediatrician who was one of the first to formally describe it, in 1933. It occurs in an estimated 1 in 10,000 live births.

## What causes CdLS?

To date there are several different genes that have been identified that result in CdLS when a change (mutation) occurs in one. There are still a small percentage of people with CdLS in whom a change in one of these genes cannot be identified; it is likely that additional causative genes have yet to be identified. In the majority of cases, the mutation is not inherited, but occurs spontaneously as a new (*de novo*) change in an egg or sperm or very early in fetal development.

A team at Children's Hospital, led by Ian Krantz, M.D., identified the first gene to cause CdLS when mutated. The gene is called *NIPBL*. Since that discovery in 2004, two additional CdLS genes have been identified.

## What is the life expectancy of a person with CdLS?

A person with CdLS can live a normal lifespan. However, if medical problems such as recurrent pneumonia, intestinal issues or congenital heart defects are not identified and properly treated, they may result in a shortened lifespan.

## What is the Center for Cornelia de Lange Syndrome and Related Diagnoses?

From a medical perspective, syndromes involving multiple systems of the body can be difficult to manage. A child with CdLS might see eight different specialists who might not communicate. As an additional barrier, many doctors aren't familiar with the syndrome.

The Children's Hospital of Philadelphia has one of the world's best teams of specialists with expertise in this, and related, syndromes. We have evaluated more than 1,000 children with CdLS, making this the largest program of its kind. Communication with one another, and with families, is a tenet of our care. Patients have traveled to see us from all over the United States and from countries including Italy, Bermuda, Iran, Egypt, Mexico, Hungary and the Dominican Republic.

*(Right) Cassidy, 5, with her mom, Pamela*

## What other syndromes does the center treat?

Our team also evaluates children and adults with other rare genetic diagnoses that involve cognitive impairment and problems with multiple systems of the body.

## What can I expect when I make an appointment at the center?

Before your first visit, you will be asked to send medical records, photos and your child's insurance information.

At your initial visit you will see a geneticist, gastroenterologist, developmental pediatrician and physical therapist. Based on your child's needs, we will schedule appointments with specialists from other areas of medicine, which may include neurology, cardiology, ophthalmology, dermatology, orthopedics, psychiatry and others. In all, you should expect to spend two to three days here.

After your visit, the team will discuss your child's needs and develop a comprehensive care plan that will be provided to you and your child's care givers.

You will be given a "Take Good Care of Me!" booklet with a summary of your child's clinical issues and medical needs to share with doctors and other health professionals. This booklet will educate them of your child's unique needs, in order to foster safer and more effective treatment.

In addition, we will give you a "care binder" to store and organize test results, doctors' reports, insurance paperwork and other important documents.

