

## Cornelia de Lange Syndrome *At a Glance*

Cornelia de Lange Syndrome (CdLS) is a genetic condition that causes a range of physical, cognitive, and medical challenges. CdLS can be caused by one of a group of genes, some of which are more associated with more severe forms and some are more associated with milder forms. It affects both genders equally. CdLS is seen in all races and ethnic backgrounds. About 1 in every 10,000 babies is born with CdLS.

There is a classical form of CdLS and a milder form. Many of the descriptions of physical features and cognitive and behavioral problems refer to the more classical form and may be much milder in the mild form. Because of advances in genetic testing, a child may have been given the diagnosis of CdLS and *not have* as many of the physical features listed below. The cognitive challenges and behavioral issues may also not be as prominent.

Individuals with CdLS strongly resemble one another, mostly due to characteristic facial features. The resemblance will not be as strong in the more mildly affected individuals. Typical facial features include:

- Thin eyebrows that meet in the middle
- Long eyelashes
- A short upturned nose
- Thin downturned lips

Cornelia de Lange can be caused by one of a group of genes, some of which are more associated with more severe forms and some are more associated with milder forms.



*Meet Ben on Page 10.*

### ***Things to Think About:***

#### **1. Medical / Dietary Needs**

##### **What You Need to Know**

##### **Gastrointestinal problems**

- Gastroesophageal Reflux Disease (GERD) or reflux is an almost universal problem in individuals with CdLS and may be severe
  - Symptoms range from occasional vomiting, belching, heartburn to intermittent poor appetite
  - If undiagnosed, reflux can lead to problems with pneumonia, esophagitis (irritation of the throat), general irritability and failure to thrive.
- The first symptom may be irritability.
  - GERD symptoms, if recognized may be easily managed
- Pyloric stenosis (a narrowing of the valve between the stomach and the intestines) or intestinal malrotation (kinking of the intestines)
  - Not as common as GERD
  - Surgery is needed
  - Most often present in infancy

### Other medical issues

- Cardiac defects, found in about 25% of individuals with CdLS may require surgery and close follow-up
- Radio-ulnar synostosis (a fusion of the forearm bones together) may be present and may limit movement
- Standard treatment is appropriate for other medical issues (i.e. hearing loss, cardiac defects, seizures)
- Cognitive and communication challenges may make it harder to identify cause of problem.
- Puberty may occur slightly later than unaffected children
  - Average age is 13 years for females with CdLS
  - Average age is 14 years in males with CdLS

### *Physical characteristics and/or symptoms:*

*Not all people with Cornelia de Lange have all of these characteristics.*

### **Individuals with Classic CdLS have:**

- Distinctive facial features in more than 95%
  - Thin eyebrows that meet in the middle
  - Long eyelashes
  - A short upturned nose
  - Thin downturned lips
- Small stature  
Height and weight remain below 5<sup>th</sup>%
- Upper limb abnormalities occur in 25% of individuals and can be severe

- More severely affected individuals may be missing fingers and/or have shortened bones in their arms
- Surgery may be needed in order to make the arms and hands more functional
- Radio-ulnar synostosis (a fusing of the forearm bones) may be present
- In the absence of structural differences, the arms and hands are often smaller than usual for their age and even for their overall size
- Hair
  - The hair on the head may be thick and may extend down onto the face or in front of the ears
  - Body hair may be thicker and more extensive than usual
- Degree of intellectual disability ranges from mild to profound
  - IQ ranges from 30-86 (mean 53) in most individuals with CdLS
  - Expressive communication is often poor
- Many individuals demonstrate autistic and self destructive behaviors
  - Individuals may avoid or reject social interactions and physical contact
  - Behavior problems may be caused by frustration at inability to speak
  - Some temperature intolerance and decreased pain sensation

### Other features

- Cardiac (25%)
- Gastrointestinal dysfunction (85%)
- Seizures (25%)
- Hearing loss (sensorineural hearing loss in 80% with 40% being profoundly affected)
- Vision
  - Nearsightedness (myopia) (60%)
  - Upper eyelid drooping (ptosis) (50%)
  - Involuntary eye movement (nystagmus) (37%)
- Genitourinary
  - Absence of one or both testes (73%)
  - Underdeveloped genitalia (57%)
  - Abnormal flow of urine from bladder (12%)

### Mild CdLS

- Individuals with this type of CdLS have many of the characteristic facial features but with less cognitive and limb involvement.

CdLS Foundation – Genetic Information:

<http://www.cdlsusa.org/research/genetic-information.htm>

### What you can do

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[www.negenetics.org](http://www.negenetics.org)

- The pain from GERD can interfere with appetite, social activities, and sleep.
  - A person with CdLS may show changes in behavior that reflect chronic pain, such as irritability or self-injurious behaviors, but may not be able to verbally report symptoms.
- People with CdLS, who present with chronic pain that is thought to be related to the GI tract, should undergo a standard acid-reflux evaluation.
  - The treatment for reflux usually consists of special diets, medications, and elevating the torso after eating.
- If radio-ulnar synostosis (fusion of the forearm bones together) is present, care may be needed with physical therapy and physical activity in order to avoid fractures.
- It is important to monitor growth, hearing, vision, ear, and kidney issues.

### Management Guidelines

<http://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.31757/pdf>

## 2. Education Supports

**It is important to have HIGH EXPECTATIONS in educating children who have Cornelia de Lange syndrome.** Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

### What you need to know

Children with CdLS may have global delays in their development. The range of intellectual disability in the classical form of CdLS may be quite significant to moderate. The range in the mild form is from normal to moderate. All individuals with CdLS may progress at their own rate, but they will continue to learn throughout their lives. The parent/caregiver and the school team needs to work together to be sure everyone knows the child's unique gifts and talents. Parents can be involved with assessment, individualized education plan (IEP) development, educational placement, curriculum adaptation and necessary therapies for their child.

Factors to consider in educational planning include:

- The child's medical and health status
  - Stamina
  - Ability to manage sensory-motor demands
  - Social engagement and interest
- Ensuring relevant goals and objectives
  - Motivating and understandable activities
  - Need for structure and organization
- Appropriate adaptations/therapies.
  - Physical, speech and occupational therapies are important to optimize

- psychomotor development and communications skills
- Alternative communication methods may be useful to facilitate communication if verbal skills are inadequate

## What you can do

Individuals with CdLS have a wide range of intellectual abilities. Many of the individuals with the classical form have difficulty with verbal communication. It is important to evaluate each child to determine his or her academic strengths and weaknesses. Remember to have high expectations and determine what supports they need to benefit from the general curriculum.

### Communication

- Consult an expert who knows how to teach the school team and family about non-verbal communication;
  - Experts can usually be found at the state Deafblind Project
  - Consider Alternative or Augmentative Communication (AAC) to ensure the child can communicate effectively in all environments.
- Many children remain non-verbal to adulthood, but they will understand sign language, receptive language, and still have intact literacy skills.
- Many children with CdLS thrive on routines because they know how to predict and anticipate favorite activities throughout the day.

### Functional Skills

- Teach functional, daily living skills that make the child as independent as possible.
- In high school facilitate community experiences to learn about everything they will need to know when they are no longer in school.
- Try out many opportunities for volunteer work around school and the community;
- Determine what likes and dislikes they have in the world of work.

### Social

- Foster friendships with neighbors, schoolmates and relatives so they will have friends and companions for their entire life.

### ***For additional information on Transition Strategies from the CdLS Foundation***

[Winter 2013: Making Sense of Special Needs Planning & a Closer Look at Transition: Learn. Work. Live](http://www.cdlsusa.org/docs/ro/ro_winter13_web.pdf)

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## 3. Behavioral and Sensory Support

## What you need to know

Many people with CdLS have difficulties with vision and hearing. These systems should be checked annually as they affect the child's success in school. When a child cannot see or hear well, they depend on their other senses—touch, taste and smell to explore their world. Home and school modifications and assistive technology become necessary for the child to learn.

- Autistic-like behaviors may be seen:
  - Repetitive and ritualistic behaviors
  - Compulsive behaviors
  
- Other Behaviors that may be seen:
  - Self-injurious behavior
  - Pica
  - Moodiness
  - Obsessive compulsive disorder
  - Behaviors issues may appear and/or worsen during the onset of puberty.

## What you can do

- A special teacher of the visually impaired or deaf or hard-of-hearing or deafblind specialist should be brought in to work with the school team and family on how the child will be able to learn.
  - Materials and programs may need to be adapted.
  - Techniques may need to be taught so that the child understands what is happening around him.
  - Incidental learning may not happen without modification of materials.
- Make sure that the teaching strategies being used are appropriate for children who are already socially engaged.
- Be proactive with behavioral supports. Discuss involvement of behavioral or mental health professionals, or medications with the parents as needed.
  - Firm directions, rules, and clear expectations are helpful.
- Many children have difficulty regulating emotions and behavior. This is especially true when handling unplanned changes.
  - Talk through expected changes.
  - They usually thrive with consistency and routine. They can be easily upset with disruption.
  - Prepare for any change in schedule.
  - Provide a safe area to share emotions.
  - Teach and model use of words and/or pictures in sharing emotions.
  - Teach, emphasize, and reinforce behaviors you want to see.
  - Make sure they have an effective communication system.

- Quiet spaces when needed
- Small group instruction
- Calming activities
- Clear concrete plans and visual cues (i.e. visual sign for quiet)
- Proactive behavioral plans that include goals, rewards, and consequences for appropriate behavior.
- Structure and predictability
- Reduced level of environmental noise/sound, natural lightening, and avoidance of crowded areas.
- Predictable transitions and signal with visual cues
- Non verbal cues and feed back
- Alternative to stressful events
- Breaks and downtime if needed
- Rule out any medical problem that could be related to behavior
- May prefer routine, order, and sameness, which can be a coping mechanism for dealing with complexities of everyday life.
- Provide social cues and coaching.
  - Provide information to and discuss differences with the child's peers.
  - Help develop confidence and focus on strengths.
  - Work on conversational skills and friendships
  - Provide positive reinforcement.
  - Teach appropriate social behaviors/skills (e.g., how to ask a friend to play).
  - Teach how to recognize facial expressions, body language, and moods in others.
  - Teach how to regulate own body – sensory strategies may be helpful.
- Self Injurious behaviors and pica
  - Monitor
  - Distract individuals

## 4. Physical Activity, Trips and Events

### What you need to know

If a child has severe GERD (Gastroesophageal Reflux Disease), they may need certain considerations on trips and/or events.

- The child may feel better with constant movement. They may need to keep moving, perhaps even walking with little rest, all day and/or night.
- Frequent snacks/meals may be necessary to decrease the pain from the increased acid.

If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming ([www.nepassage.org](http://www.nepassage.org)).

### Field Trips

- Any change in routine may produce anxiety, fears, and/or worry.
- Anxiety may result in behavioral challenges.
- Crowds and loud noise may be overwhelming to some people.

### What you can do

- Caregivers need to anticipate these needs and have medication, food and drink available when they are travelling or at special events.
- Preventing discomfort is easier than trying to cure the flare-up when it occurs.
- Be proactive and discuss any change in schedule or setting with the child ahead of time.
- Use social stories and pictures to help them understand the change.
- Role play different social settings and appropriate behavior.

## 5. School Absences and Fatigue

### What you need to know

- Children with CdLS often miss school. The syndrome can impact every system in the body - from their eyes, heart, stomach, etc.
- When children are young, they may require various surgeries and a hospital stay.
- As they get older, children use their hands and mouths to explore their environment and they are very susceptible to picking up bacteria and viruses. This can result in colds, bronchitis, pneumonia, etc.
- Sleep disturbances are common in children with CdLS.
  - Sleep problems may increase in severity as an individual with CdLS ages and/or at the onset of puberty
  - Since the children with CdLS begin their school years at age three, they may fatigue as any 3-year-old child would and require a short nap at school.

### What you can do

- Washing hands and surfaces they are in contact with often can cut down on them contracting illness at home and school.
- If there is a large outbreak of flu or other illness, parents may choose to keep their child at home until the threat passes.



- If the child is showing fatigue at school, a shortened day may be considered until their stamina is increased.
  - Provide a rest or quiet time for student
- Evaluate for Medical Causes of sleep problems
  - GERD
  - Obstructive Sleep Apnea

## 6. Emergency Planning

### What you need to know

Some people with CdLS do not understand danger. Adults are needed to keep them safe in dangerous situation, especially if they have vision and hearing loss.

### What you can do

Create an emergency plan each year so that responsible adults are assigned to the child who has CdLS. They can assist them in staying safe and away from dangerous situations.

## 7. Resources

### The national headquarters of the Cornelia de Lange Syndrome Foundation

302 West Main St. #100, Avon, CT 06001

Phone: 800-223-8355 Fax: 860-676-8337

Web: [www.CdLSusa.org](http://www.CdLSusa.org) Email: [info@CdLSusa.org](mailto:info@CdLSusa.org)

### GeneReviews

<http://www.ncbi.nlm.nih.gov/books/NBK1104/>

### Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

### Medline Plus

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

<https://medlineplus.gov/genetics/condition/cornelia-de-lange-syndrome/>

## 8. Meet a Child with CdL



### Ben, Political Activist!

If you are ever in mid-coast Maine and see the Lobster Festival Parade marching by, you will probably see Ben with his grandparents strolling with the Democratic party! Ben, age 7, loves people and enjoys crowds. He goes to elections and house gatherings for the candidates and is known by so many people in this seacoast region in Maine. He loves the Art Walks, the Coastal Children's Museum and he loves school!

As a second grader, he understands far more than he seems to express. His mother hopes that he will be more challenged with his academic development. He is included in the typical classroom for French, art, PE, and field trips and learns other subjects in a Life Skills class where he enjoys working in small groups and receiving one-on-one attention. He is easily distracted, and this has been a great setting for him. He says a few words but mainly uses his Dynavox Maestro (augmentative communication device) to communicate. He is quite proficient at talking about himself says his mother, Dena! He loves to talk about places he has been, family members (especially cousin Isabel) and his friends and teachers at school.

"He is very social - a social butterfly, and is easy-going, smiley," Dena says.

Diagnosed just before his first birthday with Cornelia de Lange syndrome, Ben had an atrial-septal defect that closed but still has an enlarged aorta that is monitored yearly. He has mild astigmatism, will begin wearing glasses this summer, and has tubes in his ears. He has GERD but that is well controlled with Prevacid and diet, although sometimes certain foods can 'set him off', resulting in reflux and keeping him up at night. Ben is a light sleeper and sometimes wakes up with the birds at 4 am. He uses a walker to get around or walks independently on his knees. He has OT, Speech and PT at school and privately.

Dena has met other families who have a child with CdLS and remarks that the variability is so surprising. Their family is hosting a gathering for all New England families on August 24th. Although their state funding for family support/respite has evaporated, they are lucky to have

nearby family members who offer a great deal of support to the family, watching Ben and his little brother, Noah.

Dena describes her relationship with the school team as good. They chat in person about once a week and it is a very nurturing school of about 130 children in K-4. She remarks that both she and her husband are teachers, although she is now running two businesses from her home so she can be home with the children. She feels she has a good understanding of what happens in schools and works to be patient with changes.

Dena has a goal for herself to start speaking up more. Her advice to teachers is “Don’t assume that if a child can’t speak that they don’t understand! He really understands so much and is smart. Although he has developmental delays, he can still understand and learn. All kids are different. You can do research, but don’t assume you know all about the syndrome. Be sure to have conversations with families and be open to having your assumptions changed!”