Genetic Education Materials for School Success

Cornelia de Lange Syndrome

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Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

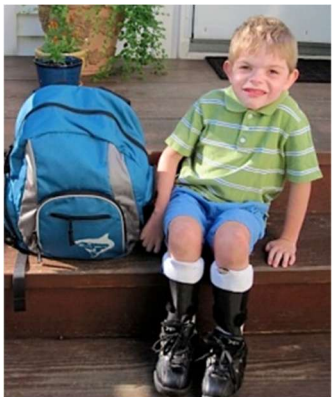
Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

Cornelia de Lange Syndrome

At a Glance

Cornelia de Lange Syndrome (CdLS) is a genetic condition associated with a wide range of physical, cognitive, and medical challenges. It affects both males and females. CdLS is caused by variants in one of several genes that control development before birth. There is a classical form of CdLS and a milder form, but even among children with the same gene variant, symptoms can range widely. It is likely that additional genes and environmental factors are also involved. This condition almost always occurs spontaneously, i.e., there is no family history.



Meet Ben on Page 11.

Common Features of Cornelia de Lange Syndrome:

- Short stature
- Abnormalities of the bones in the arms, hand, or fingers
- Gastrointestinal problems such as gastroesophageal reflux (GERD)
- Distinctive facial features that cause them to strongly resemble one another
    - Arched eyebrows that meet in the middle, long eyelashes
    - Low-set ears
    - Small, widely spaced teeth
    - Upturned nose
    - Thin downturned lips
- Excessive body hair
- Hearing loss
- Cleft palate
- Developmental delay

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In Depth

Medical and Dietary Considerations

What You Need to Know

An interprofessional team is necessary to manage a patient with CdLS effectively. It may include medical and surgical specialists, PT/OT, speech therapy, and special education. For all children with neurodevelopmental disorders, hearing and vision screening are highly recommended. The severity and presentation of symptoms will vary by child. Of note, there is a mild form of CdLS in which the child has many of the characteristic facial features but with less cognitive and limb involvement. As for all children, communicate with the parents to understand the unique needs of the child. Some common issues include the following:

Intellectual challenges

- Degree of intellectual disability ranges from mild to profound
    - IQ ranges from 30-86 (mean 53)
    - Expressive communication is often poor
- Many individuals demonstrate autistic and self-destructive behaviors
    - Variety of interventions may be used, might include medications
    - 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

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
        - The first symptom may be irritability.
    - The pain from GERD can interfere with appetite, social activities, and sleep.
    - If untreated, reflux can lead to pneumonia, esophagitis (irritation of the throat), general irritability and failure to thrive.
- 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

- Cognitive and communication challenges may make it harder to identify cause of medical problems.

- 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
- Seizures – may be on medication to manage
- Puberty may occur slightly later than unaffected children

What you can do

- Follow recommended diet and medications for GERD management
    - Elevating the torso after eating may help minimize GERD symptoms
    - Consultation with a nutritionist may be necessary to address feeding difficulties
- Provide prescribed medications for any cardiac or seizure disorders if indicated.
    - Know if there are specific signs to watch for and what actions to take if observed
- If radio-ulnar synostosis (fusion of the forearm bones together) is present, physical therapy may be prescribed
    - Exercise care during PT and other physical activity to avoid fractures
- Communicate any changes in activity, behavior, mood, or appetite to parents

Education Supports

It is important to have high expectations in educating children who have Cornelia de Lange syndrome, utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

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 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. 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

What you can do

General

- Parents and special education should work together to develop and revise plans as needed ensure relevant goals and objectives
    - Provide motivating and understandable activities

    - Recognize the 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
- Many children with CdLS thrive on routines because they know how to predict and anticipate favorite activities throughout the day.

Communication

- Consult an expert who knows how to teach the school team and family about non-verbal communication
    - Consider Alternative or Augmentative Communication (AAC) to ensure the child can communicate effectively in all environments
- Many children remain non-verbal into adulthood, but they will understand sign language, receptive language, and still have intact literacy skills

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
- Investigate 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.

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.

- Autistic-like behaviors may be seen:
    - Repetitive and ritualistic behaviors
    - Compulsive behaviors
- Other Behaviors that may be seen:
    - Self-injurious behavior
    - Pica – an eating disorder in which a person eats things not usually considered food
    - Moodiness

  - Obsessive-compulsive disorder
- Behavior issues may appear and/or worsen during the onset of puberty
- 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
- A person with CdLS may show changes in behavior, such as irritability or self-injurious behaviors, that reflect chronic pain if they are not able to verbally report symptoms

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/her
  - 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.*
- Address structure and predictability
  - Consistency and routine should be maintained
  - Firm directions, rules, and clear expectations are helpful
  - They can be easily upset with disruption. Talk through expected changes.
- Be proactive with behavioral supports, involving behavioral or mental health professionals, as needed.
  - Develop proactive behavioral plans that include goals, rewards, and consequences for appropriate behavior
  - Teach and model use of words and/or pictures in sharing emotions
  - Teach, emphasize, and reinforce behaviors you want to see
  - Non-verbal cues and feedback (e.g., visual sign for “quiet”)
  - Make sure they have an effective communication system
  - Provide a safe area to share emotions
  - Provide quiet spaces when needed
  - Promote calming activities
  - Reduced level of environmental noise/sound, natural lighting, and avoidance of crowded areas
  - Utilize predictable transitions and signal with visual cues
  - Provide small group instruction
  - Provide alternatives to stressful events
  - Allow breaks and downtime as needed
  - Rule out any medical problem that could be related to behavior
- Provide social cues and coaching.
  - Enlighten the child’s classmates about CdLS
  - Help develop confidence and focus on strengths

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  - 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 self-regulate – sensory strategies may be helpful
- Address self-Injurious behaviors and pica
  - Monitor
  - Provide distractions

Physical Activity, Trips and Events

What you need to know

Children with severe GERD (Gastroesophageal Reflux Disease) 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
- Frequent snacks/meals may be necessary to decrease the pain from the increased acid

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

- Have medication, food and drink available during trips 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

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 hospital stays.
- 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 become fatigued 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

Emergency Planning

What you need to know

Some people with CdLS do not understand danger. Adults are needed to keep them safe in dangerous situations, 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.

Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical.
https://medlineplus.gov/genetics/condition/cornelia-de-lange-syndrome/

Cornelia de Lange Syndrome Foundation

The CdLS Foundation enlists help from various experts in the medical, educational and professional fields to provide families with the most up-to-date information and advice. Here is their homepage. https://www.cdlsusa.org/

Please also checkout their **education resources available on this page
https://www.cdlsusa.org/resources/#Topic=Education

World Federation of CdLS Support Groups

If you are interested in what is happening internationally, check out the homepage of CdLS World. https://www.cdlsworld.org/

StatPearls

StatPearls on CdLS from the National Library of Medicine is a comprehensive description of CdLS and management guide designed for clinicians.
https://www.ncbi.nlm.nih.gov/books/NBK554584/

Info for School Nurse and Primary Care Staff

Cornelia de Lange Syndrome is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for Cornelia de Lange Syndrome provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. https://www.ncbi.nlm.nih.gov/books/NBK1104/

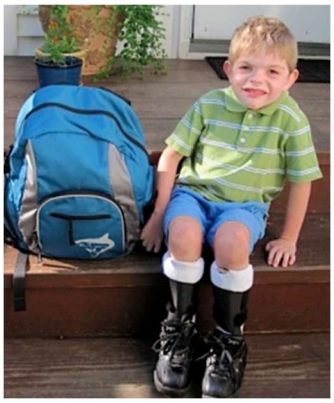
HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

"My patient_____ has been diagnosed with Cornelia de Lange Syndrome. This is a rare genetic condition that is evidenced by characteristic facies, developmental delay, and chronic gastrointestinal issues. Individuals may also have cardiac abnormalities, urogenital issues, bony abnormalities, or a seizure disorder. These children may have frequent absences from school. This condition necessitates an Individualized Education Program (IEP) or 504 plan.

Meet a Child with CdLS: *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. 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!"

Thank you so much to Ben and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.