

## **Cornelia de Lange Syndrome (CdLS)**

### ***For Healthcare Providers***

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

#### Physical characteristics and/or symptoms

*Note: not all people with CdLS will have all of these features.*

**Cornelia de Lange Syndrome (CdLS) can be subdivided into Classic and Mild. The physical features as well as the underlying cognitive impairment is much milder in the mild form.**

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

- Microcephaly
- Distinctive facial features in more than 95%
  - Highly arched eyebrows with synophrys
  - Long eyelashes
  - Short upturned nose
  - Small widely-spaced teeth
  - Long philtrum
  - Thin upper lip with down-turned corners of the mouth
- Growth restriction (usually prenatal in onset and persists throughout life)  
Height and weight remain below 5<sup>th</sup>%
- Upper limb abnormalities occur in 25% of individuals and can be severe
- Hirsutism
- Degree of intellectual disability ranges from mild to profound (mean IQ= 53)
- Autism
- Self-destructive behaviors
- Cardiac defects in 25% (primarily septal defects)
- Gastrointestinal dysfunction
- Seizures in 25%
- Sensorineural hearing loss in 80%
- Myopia
- Cryptorchidism occurs in 73%
- Immunologic abnormalities occur in some leading to frequent infections

### **Mild CdLS:**

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

## **Recommended Routine Surveillance**

- Management of gastroesophageal reflux
- Routine eye and hearing evaluations
- Monitor for heart and kidney abnormalities
- Monitor for seizures

## **Emergency Protocols**

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:

[https://www.aap.org/en-us/Documents/Seizure\\_Action\\_Plan\\_for%20School.pdf](https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf)

## **Specialists Who May Be Involved**

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Screening for cardiac septal defects
- Developmental evaluation
  - Autistic and self-destructive tendencies
  - Speech therapy with emphasis on nonverbal methods of communication
  - Physical therapy
  - Occupational therapy
- Gastroenterologist
  - Gastroesophageal reflex
  - Constipation
- Geneticist / Genetic Counselor
  - Diagnosis
  - Coordination of care
  - Genetic risk for family

- Clinical trials
- Immunologist
  - Antibody deficiency
- Nephrologist
  - Renal anomalies
- Neurology
  - Monitor seizures
- Ophthalmologist
  - Myopia
  - Ptosis
  - Nystagmus
- Orthopedists
  - Upper limb reduction defects
- Otolaryngologist
  - Sensorineural hearing loss
- Urologist
  - Cryptorchidism

## Sample Forms

Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

*My patient \_\_\_\_\_ has been diagnosed with Cornelia de Lange syndrome. Cornelia de Lange is characterized by developmental delay, growth retardation, upper limb reduction defects. Medical complications with Cornelia de Lange syndrome include management of immune deficiencies, gastrointestinal reflux, hearing loss, and congenital heart disease. Because of these, \_\_\_\_\_ needs the following accommodations.*

## Seven Important Aspects of School Life

“[Cornelia de Lange Syndrome at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



## Resources

### **Gene Reviews**

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

### **Genetic Home Reference**

<https://ghr.nlm.nih.gov/condition/cornelia-de-lange-syndrome>