Diagnostic criteria for Cornelia de Lange Syndrome (CdLS) were created by the CdLS Foundation's Medical Director Antonie Kline, M.D., in collaboration with members of the Clinical Advisory Board of the CdLS Foundation and the Scientific Advisory Committee of the World CdLS Federation.

If molecular testing has identified a mutation in one of the associated genes, the individual has CdLS. Otherwise, clinical findings should meet facial criteria, as well as criteria for two to three of six other system categories. At least one of the involved systems should be in the areas of growth, development or behavior. If these criteria are met, then the individual is diagnosed clinically with CdLS.

The checklist was developed to assist practitioners in diagnosing CdLS.

## Coming to a Diagnosis:

- Positive mutation on CdLS gene testing; OR
- Facial findings and meet criteria from two major categories (growth, development or behavior); OR
- Facial findings and meet criteria for at least one major categories and two additional categories (major or minor)

### Facial Features

- Eyebrows that meet at the midline and > three or more of the following:
  - Long eyelashes
  - Short nose, anteverted nostrils
  - Long, prominent area between upper lip and nose
  - Broad or depressed nasal bridge
  - Small or square chin
  - Thin lips, downturned corners
  - High palate
  - Widely spaced or absent teeth

### Major Criteria

- **Growth** (> two or more of the following)
  - Weight below 5th percentile for age
  - Height/length below 5th percentile for age
  - Head circumference below 5th percentile for age

- **Development** (> one or more of the following)
  - Developmental delay or intellectual disability, with speech more affected than motor skills
  - Learning disabilities

- **Behavior** (> two or more of the following)
  - Attention deficit disorder plus hyperactivity
  - Obsessive-compulsive characteristics
  - Anxiety
  - Constant roaming
  - Aggression
  - Self-injurious behavior
  - Extreme shyness or withdrawal
  - Autistic-like features

### Minor Criteria

- **Musculoskeletal** (> one or more of the following)
  - Absent arms or forearms
  - Three or more of the following or small hands and feet and/or missing digits with two or more of the following:
    - Curved 5th finger
    - Abnormal palmar crease
    - Dislocated elbow/abnormal elbow extension
    - Short 1st knuckle/proximally placed thumb
    - Bunion
    - Partial webbing between 2nd and 3rd toes
    - Scoliosis
    - Chest or sternum deformity
    - Hip dislocation or dysplasia

- **Neurosensory/Skin** (three or more of the following)
  - Droopy eyelid(s)
  - Tear duct malformation or inflammation of eyelid
  - Nearsightedness
  - Major eye malformation or peripapillary
  - Seizures
  - Mottled appearance to skin
  - Excessive body hair
  - Small nipples and/or belly button

- **Other major systems** (three or more of the following)
  - Gastrointestinal malformation/malrotation
  - Diaphragmatic hernia
  - Gastroesophageal reflux
  - Cleft palate or submucous cleft palate
  - Congenital heart disease
  - Micropenis
  - Abnormally placed opening of urethra on penis
  - Undescended testes
  - Renal or urinary tract malformation

---

**If you believe an individual you are caring for has CdLS, call the CdLS Foundation at 800-753-2357 or refer the family to a genetic counselor.**

---

**Cornelia de Lange Syndrome Foundation**

302 West Main Street, #100  Avon, CT 06001

800.753.2357 • fax 860.676.8337

www.CdLSusa.org

Supported by the Centers for Disease Control (under cooperative agreement Number U50/DP001863) and the American Legion Child Welfare Foundation Inc.