



# CdLS Foundation

Cornelia de Lange Syndrome Foundation, Inc.  
*Reaching Out, Providing Help, Giving Hope*

## Cornelia de Lange Syndrome Diagnostic Criteria

### **DIAGNOSIS:**

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

### **Facial Features:**

Synophrys (eyebrows that meet at the midline) and  $\geq 3$  of the following:

- Long eyelashes
- Short nose, anteverted nares
- Long, prominent philtrum (area between upper lip and nose)
- Broad or depressed nasal bridge
- Small or square chin
- Thin lips, down-turned corners
- High palate
- Widely spaced or absent teeth

### **Major Criteria**

**Growth:**  $\geq 2$  of the following:

- Weight below 5th centile for age
- Height/length below 5th centile for age
- OFC below 5th centile for age

**Development:**  $\geq 1$  of the following:

- Developmental delays or mental retardation
- Learning disabilities

**Behavior:**  $\geq 2$  of the following:

- Attention deficit disorder  $\pm$  hyperactivity
- Obsessive-compulsive characteristics
- Anxiety
- Constant roaming
- Aggression
- Self-injurious behavior
- Extreme shyness or withdrawal
- Autistic-like features

**Minor Criteria**

- Musculoskeletal:**  $\geq 1$  or more of the following
- Reduction defects with absent forearms
  - Small hand and feet and/or Oligodactyly (missing digits) with  $\geq 2$  of the following:
    - 5<sup>th</sup> finger clinodactyly (curved 5<sup>th</sup> finger)
    - Abnormal palmar crease
    - Radial head dislocation/abnormal elbow extension
    - Short 1<sup>st</sup> metacarpal/ proximally placed thumb
    - Bunion
    - Partial 2,3 syndactyly (webbing between 2<sup>nd</sup> and 3<sup>rd</sup> toes)
    - Scoliosis (curvature of the spine)
    - Pectus excavatum (chest or sternum deformity)
    - Hip dislocation or dysplasia

- Three or more of the following:
- 5<sup>th</sup> finger clinodactyly (curved fifth finger)
  - Abnormal palmar crease
  - Radial head dislocation/abnormal elbow extension
  - Short 1<sup>st</sup> metacarpal/ proximally placed thumb
  - Bunion
  - Partial 2,3 syndactyly (webbing between 2<sup>nd</sup> and 3<sup>rd</sup> toes)
  - Scoliosis (curvature of the spine)
  - Pectus excavatum (chest/sternum deformity)
  - Hip dislocation or dysplasia

- Neurosensory/ Skin:**  $\geq 3$  of the following:
- Ptosis (droopy eyelid)
  - Tear duct malformation or blepharitis (inflammation of eyelid)
  - Myopia  $\geq -6.00$  D
  - Major eye malformation or peripapillary
  - Deafness or hearing loss
  - Seizures
  - Cutis marmorata (mottled appearance to skin)
  - Hirsutism, generalized (excessive body hair)
  - small nipples and/or umbilicus

- Other major systems:**  $\geq 3$  of the following:
- Gastrointestinal malformation/malrotation
  - Diaphragmatic hernia
  - Gastroesophageal reflux
  - Cleft palate or submucous cleft palate
  - Micropenis
  - Hypospadias (abnormally placed opening of urethra on penis)
  - Cryptorchidism (undescended testes)
  - Renal or urinary tract malformation