

Cornelia de Lange Syndrome (CdLS)

Diagnostic Criteria Checklist



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.



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.

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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 5 th percentile for age
	Height/length below 5 th percentile for age
	Head circumference below 5 th percentile for age
Development (> one or more of the following)	
	Developmental delays 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 5 th finger
	Abnormal palmar crease
	Dislocated elbow/abnormal elbow extension
	Short 1 st knuckle/proximally placed thumb
	Bunion
	Partial webbing between 2 nd and 3 rd 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
	Deafness or hearing loss
	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