Cornelia de Lange Syndrome (CdLS) Diagnostic Scoring System

Clinical diagnostic criteria for Cornelia de Lange Spectrum Disorder (CdLS) were compiled by the Scientific Advisory Counsel of the World CdLS Federation (Kline et al., Nat Rev Genet, 2018;19:649-66) and adapted by Antonie Kline, M.D., Medical Director of the CdLS Foundation, USA.

These are based on cardinal features, or the most typically seen in CdLS, that should carry the most weight, and suggestive features, or those less specific for CdLS and carrying less weight. If molecular testing has identified a pathogenic variant in one of the associated genes, the individual is likely to have CdLS, but these criteria can still be helpful.

Clinically, classic CdLS is present when three or more cardinal features are present with a total of 11 or more points. A diagnosis of non-classic CdLS can be made when there are 9 or 10 points of which two or more cardinal features are present. When there are 4-8 points, molecular testing is indicated to help make the diagnosis. And with less than 4 points, other diagnoses should be considered. If these criteria are met, the individual is diagnosed clinically with CdLS. The scoring system was developed to assist practitioners in diagnosing CdLS.

Please contact the Canadian CdLS Foundation for support.

Refer family to a genetic councillor and the Canadian CdLS Foundation.

CANADIAN CDLS FOUNDATION

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Adapted with permission from the US CdLS Foundation - 2023

Patient's name:

Features	Clinical Findings	Points Assigned	Points on Patient
Cardinal Signs	Synophrys and /or thick eye brows	2	
	Short nose and/or concave nasal ridge and/or upturned nasal tip	2	
	Long and/or smooth philtrum	2	
	Thin upper lip vermilion and/or downturned corners of mouth	2	
	Hand oligodactyly and/or adactyly	2	
	Congenital diaphragmatic hernia	2	
Suggestive Features	Global developmental delay and/or intellectual disability	1	
	Prenatal growth retardation (below 2 standard deviations)	1	
	Postnatal growth retardation (below 2 standard deviations)	1	
	Microcephaly (prenatal or postnatal)	1	
	Small hands and/or feet	1	
	Brachydyctyly 5th finger (s)	1	
	Hirsutism	1	

Legend:
Classic CdLS > 11 points with at least 3 cardinal features, Non-classic CdLS 9-10 points with at least 2 cardinal features.
For 4-8 points, with at least 1 cardinal feature, molecular testing indicated. For < 4 points, likely not CdLS.

from Management of Genetic Syndromes, 4th Ed Kline and Deardorff, Table 1