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.

**Cornelia de Lange Syndrome Foundation**
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