

Diagnosis of Cornelia de Lange Syndrome

Because the genetic causes of Cornelia de Lange syndrome have only recently begun to be understood, most diagnoses are made based on the [clinical features](#) of the syndrome. Clinical features include distinctive facial and other physical characteristics associated with the syndrome.

The signs of Cornelia de Lange syndrome can vary between individuals and are sometimes referred to as 'Classic', 'Mild' and 'Atypical' forms. There may be differences in the genetic mechanism underlying these different subtypes. However, this is not yet completely understood. To read more about the genetics of Cornelia de Lange syndrome, [click here](#).