Cornelia de Lange syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Cornelia de Lange syndrome. ORPHA:199

Cornelia de Lange syndrome (CdLS) is a multisystem disorder with variable expression marked by a characteristic facial dysmorphism, variable degrees of intellectual deficit, severe growth retardation beginning before birth (2nd trimester), abnormal hands and feet (oligodactyly, or sometimes an even more severe amputation, and constant brachymetacarpia of the first metacarpus), and various other malformations (heart, kidney etc.).