Cornelia de Lange syndrome (CdLS) is a rare genetic disorder that is apparent at birth (congenital). Associated symptoms and findings typically include delays in physical development before and after birth (prenatal and postnatal growth retardation); characteristic abnormalities of the head and facial (craniofacial) area, resulting in a distinctive facial appearance; malformations of the hands and arms (upper limbs); and mild to severe intellectual disability. Many infants and children with the disorder have an unusually small, short head (microbrachycephaly); a prominent vertical groove between the upper lip and nose (philtrum); a depressed nasal bridge; upturned nostrils (anteverted nares); and a protruding upper jaw (maxillary prognathism) with small chin (micrognathia). Additional characteristic facial abnormalities may include thin, downturned lips; low-set ears; arched, well-defined eyebrows that grow together across the base of the nose (synophrys); an unusually low hairline on the forehead and the back of the neck; and curly, unusually long eyelashes. Affected individuals may also have distinctive malformations of the limbs, such as unusually small hands and feet, inward deviation (clinodactyly) of the fifth fingers, and webbing (syndactyly) of certain toes. Less commonly, there may be absence of the forearms, hands, and fingers. Infants with CdLS may also have feeding and breathing difficulties; an increased susceptibility to respiratory infections; a low-pitched "growling" cry and low voice; heart defects; delayed skeletal maturation; hearing loss; or other physical abnormalities. The range and severity of associated symptoms and findings may be extremely variable from person to person. CdLS is a very rare disorder that is apparent at birth (congenital). Males and females appear to be affected in equal numbers. It has been estimated that CdLS occurs in approximately one in every 10,000 live births in the United States. More than 400 cases have been reported in the medical literature, including affected individuals within several families (kindreds). Multiple affected siblings have been reported in some families. It is estimated that there is a 1-2 % rate of recurrence within affected families.