Cornelia de Lange Syndrome

(de Lange Syndrome, Brachmann-de Lange Syndrome) - Submitted by Paula Forney

What is Cornelia de Lange Syndrome (CdLS)?
Cornelia de Lange Syndrome is a congenital syndrome, meaning it is present from birth. Most of the signs and symptoms may be recognized at birth or shortly thereafter. A child need not demonstrate each and every sign or symptom for the diagnosis to be made. As with many other syndromes, individuals with CdLS resemble each other. Common characteristics include: low birthweight (usually, but not always, under five pounds), delayed growth and small stature and small head size (microcephaly). Typical facial features include arched, well-defined eyebrows which grow together above the nose (synophrys), an unusually low hairline, long eyelashes, a short upturned nose, and thin, downturned lips.

Other frequent findings include excessive body hair (hirsutism), small hands and feet, partial joining of the second and third toes, incurved fifth fingers, gastrointestinal reflux, seizures, heart defects, cleft palate, bowel abnormalities, feeding and breathing difficulties, autistic features, self-injurious behaviors, and developmental delay. Found in some individuals are limb anomalies , including missing limbs or portions of limbs, usually fingers, hands ,or forearms. Sensory problems are also possible and may include: small, low-set ears, hearing loss, misaligned eyes (strabismus), shaky
eyes (nystagmus), nearsightedness (myopia), dry eyes (blepharitis), and droopy eyelids (ptosis). The range and severity of associated symptoms and findings may be extremely variable from case to case.

Why is it called Cornelia de Lange Syndrome?
In 1933, Dr. Cornelia de Lange, a Dutch pediatrician, described two children with similar features, one 17 months and the other 6 months, who were admitted within weeks of each other to the same hospital with common medical problems. The syndrome is sometimes referred to as Brachmann-de-Lange Syndrome after Dr. W. Brachmann who also had described a similar patient in 1916.

How many people have CdLS?
The exact incidence is unclear, but it is thought to be between 1:10,000 and 1:30,000 live births. For example, this would suggest that a population the size of Canada might experience 18 births of children with CdLS per year, in Ohio one might expect 8, and in the United Kingdom, 38 births per year.

Is there always cognitive disability?
Usually a level of cognitive disability is present, ranging from mild to profound. The majority of individuals with CdLS fall in the moderate to severe range.

What causes CdLS?
At present, the cause is not clearly known, although it is suspected that a gene may be responsible. At present there are several research programs underway which are attempting to find answers to the cause of CdLS. In most individuals with the disorder, CdLS appears to occur randomly for unknown reasons. However, there have been some familial cases, suggesting autosomal dominant inheritance. According to investigators, the disorder may be caused by changes (mutation) of a gene or genes on the long arm (q) of chromosome 3 (3q26.3) or 17 (17q23).

How is diagnosis made?
A thorough medical evaluation including a history and physical exam, family history, lab tests, X-rays and chromosome analysis is usually conducted before a diagnosis is made. Since there is no specific test for CdLS, this is best accomplished through a referral to a genetics specialist or clinic. No definitive prenatal detection tests are currently available, but prenatal ultrasound can assist families who have already had a child with CdLS to evaluate subsequent pregnancies.

What are the expectations for a child with CdLS?
Each child will develop at his/her own rate, depending on the presenting problems and their severity. Generally a slower than average rate of development can be expected. Speech and communication are often significant areas of delay. Early intervention programs and other appropriate developmental and therapeutic interventions are recommended to optimize outcomes.

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