What is Cornelia de Lange Syndrome?

Cornelia de Lange syndrome is a developmental disorder that can affect multiple body parts. Although the exact prevalence of Cornelia de Lange syndrome is unknown, it is estimated that the disorder affects 1 in every 10,000 to 30,000 newborn children. The features of Cornelia de Lange can be expansive and can manifest on a continuum from mild complications to profound complications.

Cornelia de Lange syndrome is commonly characterized by some combination of the following features:

- slow growth before and after birth
- intellectual disability that is usually severe to profound
- skeletal abnormalities involving the arms and hands, including the absence of arms, forearms
- arched eyebrows that often grow together
- long eyelashes
- low set ears
- small, widely spaced teeth
- a small upturned nose
- behavioral problems similar to those observed in children with autism
- excessive body hair (hypertrichosis)
- a small head (microcephaly)
- hearing loss
- short stature
- digestive system problems
- cleft palate
- seizures
- heart defects

Causes of Cornelia de Lange

A linkage has been established between Cornelia de Lange syndrome and specific human genes. Three specific gene mutations have been found to correlate to the presence of Cornelia de Lange syndrome. These mutations occur in the NIPBL, SMC1A, and SMC3 genes. More than 50% of all individuals with Cornelia de Lange syndrome have a mutated NIPBL gene. All of the identified genes produce proteins that contribute significantly to directing prenatal physiological development. Specifically, the proteins produced by these three genes organize and structure chromosomes, help repair damaged DNA, and contribute to the development of the limbs, face, and other body parts. It is therefore unsurprising that mutations in the aforementioned genes can manifest as a syndrome that has such detrimental effects on a wide variety body parts as those found in Cornelia de Lange syndrome.

Prognosis & Considerations

Most children with Cornelia de Lange Syndrome do survive to adulthood. However, most of these children, especially those with more severe complications, will require assistance and supervision throughout their lives.

As a child with Cornelia De Lange ages, the following areas related to developmental and educational goals should be considered:

- the child’s medical and health status
- the child’s stamina
- the child’s ability to manage sensory-motor demands
- the child’s levels of arousal
- communicative status of the child
- The child’s need for structure and organization
- relevance of goals and objectives to be pursued
- what activities are motivating and understandable to the child
- what adaptations and modifications can be made to make activities accessible to the child

References:


