CORNELIA DE LANGE SYNDROME: A CASE REPORT

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INTRODUTION

Cornelia de Lange syndrome is a developmental disorder that affects many parts of the body. The first ever documented case was in 1916 by W. Brachmann followed up by Cornelia de Lange, a Dutch pediatrician, in 1933 after whom the disorder has been named.[1,2]

Key Words: Cornelia de Lange, synophrys, hirsutism, SMC1A and SMC3 gene mutations, NIPBL gene

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Case Report

This 3 month-old Sudanese boy presented with his family, much concerned about his failure to thrive and dysmorphic features. Low pitched weak cry. On examination he was smaller for his age. Head circumference <3rd percentile. His face showed bushy eyebrows with central fusion, small nose anteverted nostrils, thin down ward upper lip and high arched palate. (Fig. 1) He also has Simian crease and proximally implanted thumbs. (Fig. 2) Cardiovascular system is normal. There is no disturbance of muscle tone or abnormalities of the feet or genitalia. Parents deny the presence of family history of similar malformations.

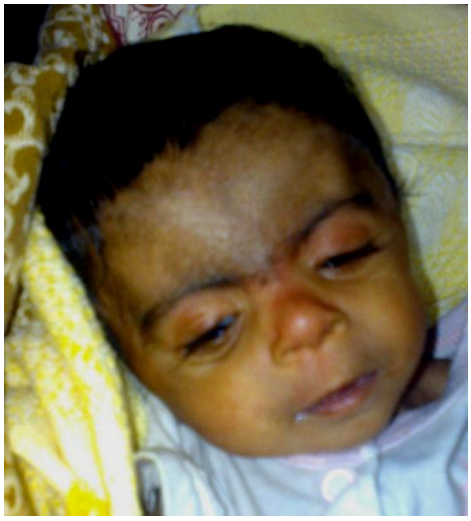

Fig. 1: Proximally placed thumb

Fig. 2: Synophrys, thin down turning upper lip

DISCUSSION

The diagnosis of CdLS is primarily a clinical one based on signs and symptoms and laboratory tests.[3] Distinctive facial features help to identify the syndrome. The features of this disorder vary widely among affected individuals and range from relatively mild to severe. The syndrome is characterized by slow growth before and after birth, intellectual disability that is usually severe to profound, skeletal abnormalities involving the arms and hands, and distinctive facial features.[4] The facial differences include arched eyebrows that often grow together in the middle (synophrys); long eyelashes; low-set ears; small, widely spaced teeth; and a small, upturned nose. Many affected individuals also have behavioral problems similar to autism. Additional signs and symptoms of Cornelia de Lange syndrome can include excessive body hair (hirsutism), an unusually microcephaly, hearing loss, short stature, and problems with the digestive tract and cleft palate. Seizures, heart defects, eye problems, and skeletal abnormalities also have been reported.

Although the exact incidence is unknown, Cornelia de Lange syndrome likely affects 1 in 10,000 to 30,000 newborns.

In about 35 percent of cases, the cause of Cornelia de Lange syndrome is unknown. Almost all cases result from new gene mutations and occur in people with no history of the condition in their family.

Mutations in the NIPBL at chromosome 5 have been identified in more than half of all people with this condition and are considered to have an autosomal dominant pattern of inheritance.[5] SMC1A mutation on X chromosome in (5%) and SMC3 on chromosome in (1%) also reported.[6] These mutations have been proposed to cause Cornelia de Lange syndrome by disrupting gene regulation during critical stages of early development. Studies suggest that SMC1A and SMC3 gene mutations tend to cause somewhat milder signs and symptoms than those seen with mutations in the NIPBL gene.[7]

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