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CORNELIA DE LANGE SYNDROME: A CASE REPORT

Dermatology

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ABSTRACT

Cornelia de Lange syndrome is a multisystem developmental disorder with features of delayed developmental milestones and psychomotor retardation. This syndrome also comprises of facial dysmorphism, hypertrichosis, upper limb deformities, cardiac and gastrointestinal abnormalities. We report a case of hypertrichosis with features of facial dysmorphism and upper limb deformities suggesting a diagnosis of Cornelia de Lange syndrome[1].

KEYWORDS

Cornelia de Lange syndrome, Facial dysmorphism, Hypertrichosis

INTRODUCTION:

Cornelia de Lange is a congenital condition featuring distinct facial morphology, pre and post natal growth retardation, delayed motor and sensory milestones with behavioral abnormalities with upper limb malformation. Dr. Cornelia de Lange, a Dutch pediatrician was the first to report the case in 1933. Brachmann also reported a similar case in 1916 with upper limb deformity in addition to the other features. Hence, both the physicians' names are attached to the name of the syndrome2. This syndrome is inherited in an autosomal dominant pattern but the majority of the cases that are reported are sporadic in nature.

CASE REPORT:

An 18 year old female came to the Dermatology OPD with hyperpigmention over the angle of mouth with history of prior treatment for Herpes Labialis. She also complained of hypertrichosis over the face, arms and back. No complaints of hearing loss. No dental or gingival abnormalities. No visual impairments. No history suggestive of cardiovascular involvement. No neurological deficits. No gastrointestinal related complaints. No similar complaints in the family.

On examination, the patient is of short stature, with low set anterior and posterior hairlines, bushy eyebrows, long curled eyelashes, small upper lip in comparison to the lower lip, low set ears, depressed nasal bridge, micrognathia and clinodactyly of both little fingers.

On compiling the above features, a diagnosis of Cornelia de Lange syndrome was made.

DISCUSSION:

Cornelia de Lange syndrome is also called Brachmann Cornelia de Lange or Amsterdam dwarf or Typus degenerativus amstelodamensis. Cornelia de Lange syndrome is of an unknown etiology, but few cases have shown an association with the mutation of NPBL gene and duplication or partial trisomy of chromosome 3q. There are no differences in age or racial or gender distribution[3].

The syndrome affects a wide variety of systems in our body. Facial dysmorphism is the hallmark of this syndrome, which includes features of broad forehead, webbed neck, bushy eyebrows, long curled eyelashes, ptosis, small nose, prominent nares, thin upper lip, fish mouth, micrognathia, strabismus, long philtrum, cleft palate, prominent mandibular symphysis and hirsutism. Oral features include macroglossia, microdontia, partial anodontia and delayed tooth eruption4, 5. Skeletal abnormalities include short stature, clinodactyly of fingers and toes and proximally placed thumbs. Gastrointestinal symptoms include GERD, vomiting, heartburn, poor appetite and belching. Auditory symptoms include hearing loss of varying degree. Congenital heart disease is also a manifestation of the syndrome, of which VSD is the most common. Neurological deficits include mental retardation, seizures, hyperactivity, irritability, sleep disturbances and self mutilating behavior6, [7].

Van Allen et al classified this syndrome into three types. Type 1 or classic type; includes all facial dysmorphic features and skeletal abnormalities. Type 2 or mild type; includes all the facial features and minor skeletal abnormalities. Type 3 or phenocopies which includes patients who have the phenotypic features caused by chromosomal aneuploidies or teratogenic exposures8.

Management of these cases includes monitoring of systemic involvement other than the cutaneous manifestations. These will include imaging studies and a multi disciplinary approach involving an orthopaedician, neurologist, ophthalmologist, cardiologist and ENT specialist. Hair removal whether it may be temporary or permanent, can be advised for cosmetic purposes. Family support is of utmost importance especially when the child is diagnosed. Educating the family about the condition improves treatment compliance9.

CONCLUSION:

Cornelia de Lange poses a serious limitation to the life of the individual. The prognosis depends on the extent of involvement of various systems. This patient can be classified as Type 2 Cornelia de Lange syndrome. She and her parents were counseled about the various aspects that can be encountered in this disorder.

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CONFLICT OF INTEREST: The authors declare that they have no conflict of interest.

LEGENDS TO FIGURES:

Figure 1: Clinical photograph showing thin upper lip, micrognathia, low anterior hairline.

Figure 2: Clinical photograph showing bushy eyebrows and long curled eyelashes.

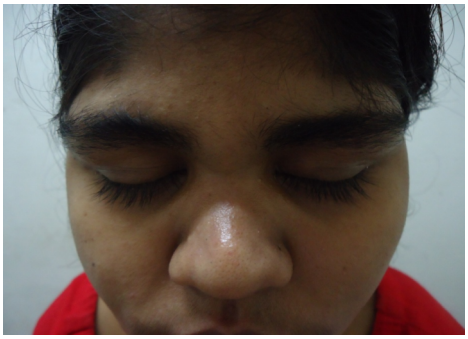


Figure 3: Clinical photograph showing depressed nasal bridge, low set ears and low hairline.

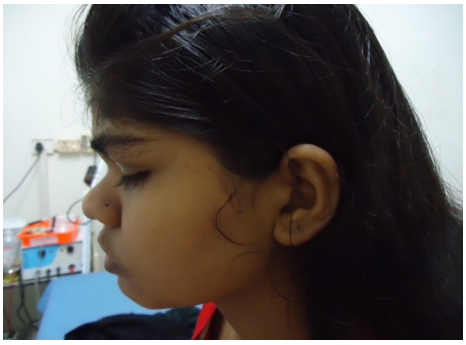
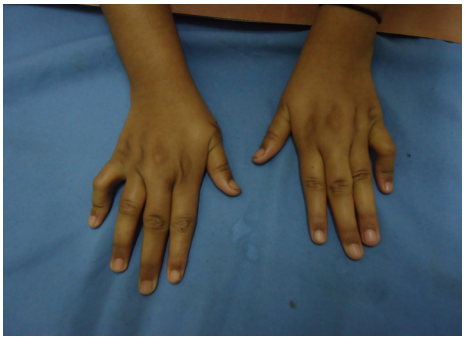


Figure 4: Clinical photograph showing clinodactyly.



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