De Lange Syndrome

Pages with reference to book, From 20 To 21

Sadia Akram,Shamim A. Qazi (Children Hospital, Pakistan Institute of Medical Sciences, Islamabad.)

Introduction

de Lange syndrome of unknown etiology was first described by Cornelia de Lange in 1933[1]. It is also known as Brachinann de Lange syndrome[2]. A survey in Denmark implied the minimum incidence to be one in 50000'. Till now more than 150 cases have been reported[1]. The presentation of this syndrome includes shortness of stature of a prenatal onset; mental retardation and sluggish physical activity; initial hypertomcity; low pitched, weak, growling cry in infancy; microbrachycephaly; bushy eye brows and synorpluys; long curly eyelashes; small nose; anteverted nostrils; characteristic lips and mouth; micrognathia; hirsutism; simian crease; proximal implantation of thumbs; flexion contracture of elbows; ni.icrornelia and syndactaly of second and third toes[1]. A literature search on medline, did not show any reported case of Cornelia de Lange syndrome from Pakistan.

Case Report

Noman two months old male baby, resident of Dera Ismail Khan was admitted on August 27, 1996, with complaints of regurgitation of feed through the nose and mouth. Cry was weak and he had cyanotic spells during feeding and crying. There had been watery discharge from eyes since birth. The mother complained that the child regurgitated milk from the nose and mouth nearly after every feed, usually a few minutes after the feed. The child fatigued easily during feeding and would become exhausted and cyanosed. The cry was weak and noiseless since birth, the facial features were atypical and the discharge from the eyes was not associated with pus, redness of eyes or stickiness of eye lashes. The infant had developed urinary retention at the age of five days which was relieved by catheterization locally. The mother had concieved the child despite reported regular use of injectable and oral contraceptives. The baby was born after seven months of gestation, cried half an hour after birth and was cyanosed, He was the twelfth issue of a consanguineous marriage. There had been three male still births and one male intrauterine death at seven months gestation. This infant is the only surviving male offspring. All seven alive female siblings are normal. The mother is presently 35 years old and was married at 16 years of age. On admission, the patient was weak, irritable with dysmorphic features. He was afebrile with normal heart rate and respiration, his length was 45.0 cm, weight was 2.0 kg and occipitofrontal circumference was 30.5 cm, all less than the third percentile according to the reference measures. His antenorfontanelle was patent and wide measuring 3x3 cm and posterior fontanelle measured 0.5x0.5 cm. Patient had a low pitched weak cry, microbrachycephaly, depressed nasal bridge, anteverted nostrils, swollen eyelids, thin lips, high arched palate, micrognathia, hypertrophied gums, hypoplastic nipples and malformed umbilicus, single palmar crease,
polydactaly in left hand, micromelia, short stubby feet and bilateral undescended testis.

Discussion

This child has most of the features of de Lange syndrome. These children show retardati9n of growth and severely limited intellectual performance[3]. The child may avoid or reject social interactions[2]. Most of them have been reported to have difficulties in distinguishing between a stranger and their mothers or mother's substitutes[2] . Their intellegent quotient varies from 4 to 85 with majority being below 35[1]..

Prognosis is not favourable. Susceptibility to infections due to aspiration in infancy is a major1hazard and early death may result due to repeated infections[1]. We could not find any published association of this syndrome witi? the use of contraceptives. Although the etiology is unknown[1], all male still births and intrauterine deaths in this family does indicate some genetic predisposition, possibly X-linked transmission for this family, Duplication of the Q25-29 band region Of chromosome some1imes yields a phenotype similar to de Lange syndrome[1]. Unfortunately specialized chromosomal studies could not be performed. This child needs to be evaluated in future for assessment of his growth and development, although the long term prognosis is not optimistic.

References

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