CORNELIA DE LANGE SYNDROME : A CASE REPORT

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SUMMARY :

An eleven - month - old male patient with dismorphic face, thick and curly hair, abnormally low placed ears, cleft palate, short back, low hair line, as well as hypospadias, cryptoorchidism, bilateral inguinal hernia, and phocomelia of the upper extremites was considered to have Cornelia de Lange syndrome. He had the combination of patent ductus arteriosus (PDA), tricuspid valve insufficiency, and bicusbid aortic valve as the cardiac anomalies, which haven't been reported to our knowledge in the literature before.

Key Words: *Cornelia de Lange Syndrome, Children.*

INTRODUCTION

Cornelia de Lange syndrome (CDLS) consists of significant growth deficiency with intrauterine onset, failure to thrive, moderate to severe mental retardation, microbrachycephaly, characteristic face; confluent eyebrow (synorphrys), long eyelashes, small nose and anterverted nostrils, prominent philtrum, thin lips down turned angles of mouth, low set ears, micrognathia and micromelia, phocomelia and genital hypoplasia (1). Other common malformations are cardiovascular anomalies, ventricular septal defect (VSD), atrial septal defect (ASD) and patent ductus arteriosus (PDA) being the most common ones (2-4).

A case is presented with CDLS associated with tricuspid valve insufficiency, bicuspid aortic valve, and PDA, which haven't been reported before.

CASE REPORT

An 11-month-old male patient was admitted to our hospital with a history of dyspnea, caughing, and cyanosis for two days and convulsion. He was the first son of young healthy non consanguin parents and he was born with breech presentation. He had weighed 1500 g as a newborn and he also couldn't keep his head straight.

On physical examination, his fever was 38°C, his weight and height were 2450 gr, (below 3rd percentile) and 55 cm. (below 3rd percentile) respectively, and head circumference was 35 cm. His general condition was poor with a decreased attention and he had both peroral and peripheral cyanosis. He had cutis marmaratus, hemangioma at glabella, and hirsutism on the back. His face seemed dismorphic; he had thick and curly hair eyebrows meeting at the midline and he had long and curled eyelashes. His ears were abnormally low placed. He also had anteverted nostrils, a small nose, a prominent philtrum, thin lips with down turned angles of mouth, a cleft palate, bifid uvula, and micrognа-

tia. His back was short and hairline was low. He also had hypoplastic nipple, a systolic II° / VI° murmur at the pulmoner and aortic areas, disseminated crepitant ralls in both lungs, hypospadias, cryptorchidism, bilateral inguinal hernia, phocomelia of the upper extremites, and spastic lower extremities (Fig. 1-2).

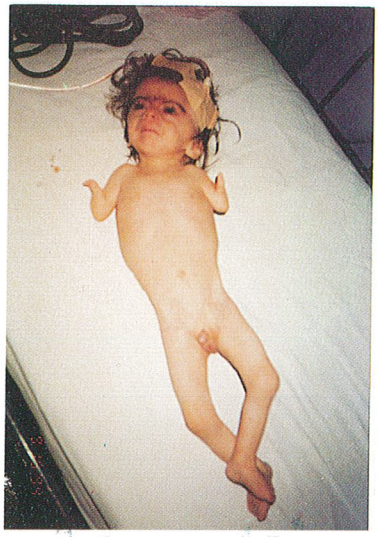

Fig - 1 : Clinical appearance of the patient.

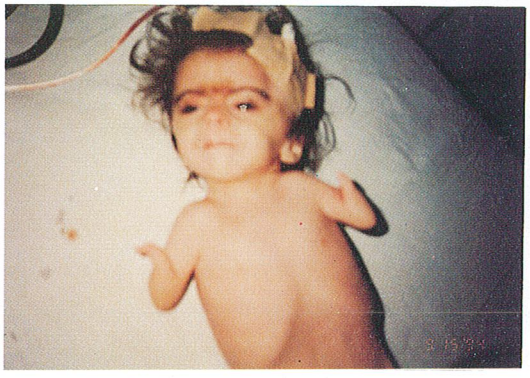

Fig - 2 : Clinical appearance of the patient.

Laboratory findings were as follows : Hb : 10.5 g/dl, WBC : 20400 / mm^3, platelets : 320000 / mm^3, CRP : 96 mg/dL. CSF examination revealed purulent menengititis. Chest X-ray : disseminated infiltration, Telecardiography : cardiomegaly. Coloured doppler echocardiography : PDA, tricuspid valve insufficiency (velocity of regurgitant jet : 2.4 m/s) and bicuspid aortic valve (velocity: 1.4 m/s). Caryotype analysis was found to be 46, XY.

With these clinical and laboratory findings, the diagnosis of CDLS, pneumonia, and purulant menengitis were made, and antibiotic and supportive treatment was begun. The patient died on the second day of his hospitalization.

DISCUSSION

Cornelia de Lange syndrome, first described in its full clinical presentation by Dr.Cornelia de Lange (1933), is a multisystem syndrome involving congenital malformations, growth retardation, and neurodevelopmental delay (1, 4-6). CDLS is seen in 1/10.000 newborn infants. The risk of recurrence is % 2-6. Also, familial recurrence, autosomal recessive and autosomal dominant inheritance have been reported. A new dominant mutation is suspected in sporadic cases. Its etiology is still unknown (4, 7-9). Its clinic findings include hirsutism due to hypertricosis, low posterior hair line, meeting of the eye brows at the midline and long eyelashes. The malformations of the face include broad and / or depressed nasal bridge, small and anteverted nostrils, prominent philtrum, thin lips down turned angles of mouth, cleft palate, bluish tinge around eyes, low set ears, micrognatia, short neck, and anomalies such as focomelia (especially upper extremites) and oligodactyly. The most important diagnostic criteria are the existence of face and extremity anomalies, but other findings that may be associated with CDLS are hypoplastic genitalia, cutis marmaratus, hemangioma, undescended testis, small umblicus and nipples (5-8).

In our patient, the growth retardation present at his birth continued after the postnatal period as a remarkable psychomotor retardation. The association of facial and extremity abnormalities led us to consider him to be type I classic CDLS.

The major cardiac abnormalities associated with CDLS are VSD, ASD, and PDA, but other complicated anomalies (single ventricle, pulmonary stenosis, TOF, aortic stenosis and bicuspid aortic valve) were also reported (1, 6, 9).

To our knowledge our patient is the first case in the literature with a complex cardiac abnormality defined as; PDA, bicuspid aortic valve and tricuspid valve insufficiency demonstrated by coloured doppler echocardiography. Patients below 2265 gr

birth weight, and with major extremity malformations, facial abnormalites and microcephaly have a poor prognosis (2, 8) and could die due to respiratory problems (3, 5).

Since cardiac abnormalites are major components of this syndrome, we wanted to point out that the cardiac evaluation of patients with CDLS must be done with special attention in order to reveal any associated undefined new abnormality, such as that reported in this paper.

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