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Case Report

Cantu syndrome - A case report.

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Abstract

Cantu syndrome is a rare congenital disorder characterized by hypertrichosis, osteochondrodysplasia, cardiomegaly, macrocephaly, short body stature, prominent or enlarged forehead, wide set and bulging eyes, loose skin with wrinkled palms and soles of the feet, hyperextensible joints, wide ribs and small vertebrae. We describe a newborn with features of Cantu syndrome.

Key words: Cantu syndrome, Hypertrichosis, Macrocephaly, Hyperextensible joints, Coarse facies

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Introduction

Cantu syndrome is a rare congenital disorder characterized by hypertrichosis, osteochondrodysplasia, cardiomegaly, and dysmorphism (1,2). This syndrome is also known as Hypertrichotic osteochondrodysplasia. So far less than 20 cases have been described in literature. To the best of our knowledge this is the first report of Cantu syndrome from India.

Case Report

A preterm (36 weeks) male baby born by spontaneous vaginal delivery to a primi mother with normal Apgar score was admitted for preterm care. The parents were third degree consanguineous and the antenatal ultrasonography revealed polyhydramnios. On examination he had macrocephaly with prominent forehead, short body stature, coarse facial features, small nose, narrow shoulder

Table 1. *Comparison of clinical features between earlier cases and this baby*

Clinical features	Frequency among reported csases (n=18)	Clinical features in the present case
Coarse facial features	18	✓
Congenital generalized hypertrichosis	17/18	✓
Long philtrum	17/18	✓
Abundant eyebrows and/or curly eyelashes	15/18	✓
Epicanthal folds	14/18	
Prominent mouth	14/18	
Flat broad nasal bridge	13/18	
Cardiomegaly/cardiopathy	13/18	✓
Narrow thorax	12/18	✓
Anteverted nares	11/18	✓
Macrosomy at birth	11/18	
Narrow shoulders	10/18	✓
Mild mental retardation	9/18	
Macrocephaly	8/18	✓
Short and broad first toe	8/18	✓
Small nose	6/18	✓
Pericarditis with effusion	2/18	
Lymphedema	2/18	

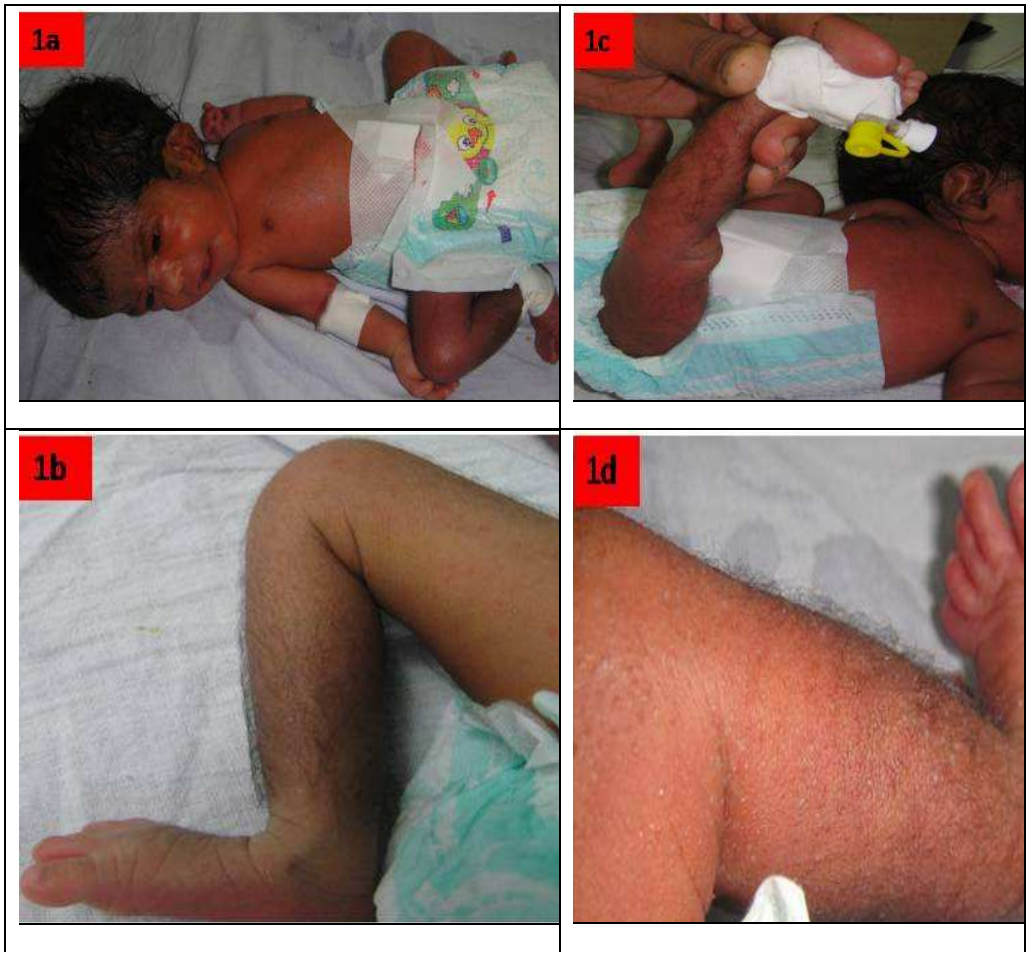

Figure 1. *a.Macrocephaly,coarse facies,abundant eyebrows,small nose,narrow shoulder. b. Hypertrichosis. c. Hyperextensibility of joints. d. Short and broad big toe with hypertrichosis*

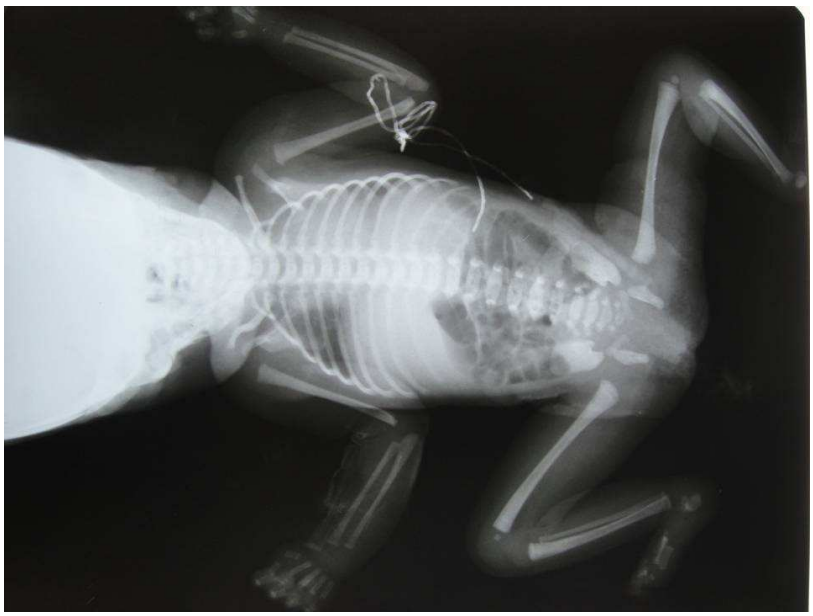

Figure 2. *Macrocephaly with thick calvarium, wide ribs, irregularities of vertebral body surfaces, hypoplastic ischiopubic branches, broad first metatarsal and cardiomegaly.*

and thorax with short neck (Fig. 1a). Generalized hypertrichosis especially in the legs (Fig. 1b), hyperextensibility of joints (Fig. 1c), and small hands and feet with short and broad big toes (Fig. 1d) were also noted. Anthropometry revealed a weight of 1.6 kg (<3rd centile), head circumference of 34 cm (90th centile), chest circumference of 23 cms and length of 40 cms (<3rd centile) with upper segment lower segment ratio of 1.5:1. Ophthalmological evaluation was normal. Systemic examination was normal except a short systolic murmur in the left parasternal area.

Investigations revealed no evidence of sepsis and blood sugar and electrolytes were normal. Radiography revealed macrocephaly with thick calvarium, wide ribs, irregularities of vertebral body surfaces, hypoplastic ischiopubic bones, broad first metatarsal and cardiomegaly (Fig. 2). Ultrasonography of the cranium was normal while echocardiography revealed an Ostium secundum atrial septal defect of 8 mm with dilation of right atrium and ventricles and a small Patent ductus arteriosus. Ultrasonography of the abdomen and karyotyping were normal. At 3 weeks of age, he was on exclusive breast feeds, had hypertrichosis and same clinical feature as at discharge. Anthropometry revealed weight of 1.8 kg, length of 41 cms, head circumference of 34.8 cms .

Discussion

Cantu syndrome is a rare disorder characterized by congenital hypertrichosis, osteochondrodysplasia, cardiomegaly, and dysmorphism. This syndrome was first reported by Cantu et al in 1982. They described four patients with two siblings (brother and sister) and two unrelated cases, a male and a female with features of this syndrome (1,2). Since then occasional cases have been reported presenting with hypertrichosis and skeletal findings similar to those described by Cantu et al allowing further delineation of this new genetic syndrome. Initially autosomal recessive mode of inheritance was proposed for this syndrome, but Robertson et al by segregation analysis suggested an autosomal dominant inheritance or a microdeletion syndrome (3). This was later confirmed by the finding of Lazalde et al who recently described three affected members of a family (father, daughter and son) in which male-to-male transmission was evident suggestive of autosomal dominant inheritance (4) . All cases had apparently normal chromosomes and to date microarray analysis on several patients has not shown any sub-microscopic deletion or duplication in any region of the genome.

Cantu syndrome is characterized by generalized congenital hypertrichosis, coarse facial features, long philtrum, abundant eyebrows mild mental retardation, macrocephaly, wide posterior fossa in the skull, verticalized base of the cranium, narrow thorax, global cardiomegaly due to cardiomyopathy (sometimes complicated with pericarditis and effusion), hyperextensibility of joints (1,2,5-7). Cantu syndrome can sometimes become a cause or precursor of other diseases such as alopecia follicular hyperkeratosis and short stature. Due to this fact the syndrome is sometimes misdiagnosed. The other condition which is associated with hypertrichosis in newborn period is Congenital Hypertrichosis Lanuginosa (CHL) in which, at birth the entire body except the hands and feet are covered with excess fine blond hairs (lanugo hairs) (8). Though a subtype of CHL such as Brachmann-de Lange syndrome has distinctive faces, secondary to a low hairline, penciled but not bushy eyebrows, long eyelashes and thin lips and constant abnormalities like mental and growth retardation, Cantu syndrome can be differentiated from this due to characteristic clinical and radiological features.

The comparison in the clinical features along with their frequencies between 18 cases of Cantu syndrome described in literature and our case is shown in Table. It is evident that this baby had most of the common features of Cantu syndrome. It is to be noted that all features need not be present at birth and some can develop later.

The common radiological findings in Cantu syndrome described in literature include enlarged posterior fossa and vertical base of cranium, thick calvarium, wide ribs, irregularities of vertebral bodies surfaces, hypoplastic ischiopubic branches, Erlenmeyer flask'-shaped long bones ,short distal phalanx of first rays, broad first metatarsal and generalized osteopenia (9).

Mild intellectual deficiency has been described in several patients on long term follow up. There are no specific management strategies and most cases are managed symptomatically. Since recent evidence suggests an autosomal dominant mode of inheritance, genetic counseling can be given to parents of a child with Cantu syndrome.

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