

An unusual combination of primordial dwarfism with isolated cleft palate

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ABSTRACT

Primordial dwarfism is a rare anomaly with over 200 sub-types, which results in a relatively small body size in all stages of life, beginning at the intrauterine period. The life expectancy for these patients is around 30 years but very few survive their intra-uterine phase. Other features associated with primary dwarfism reported till date includes pigmentation of skin, cleft lip and palate, and semilobar holoprosencephaly. We report an unusual combination of primordial dwarfism associated with isolated cleft palate.

Key Words: Primordial dwarfism, cleft palate, dwarfism with cleft palate

Introduction

Primordial “dwarfism” is an autosomal recessive disorder and refers to patients who have severe intrauterine and postnatal growth retardation. Intrauterine growth retardation often leads to premature delivery, with mean delivery age of 35 weeks.^[1] Since, they have low birth weight

the diagnosis can only be made after birth by evaluating clinically their stunted growth rate, leaving primordial dwarfs perpetually years behind their peers in stature and in weight. Out of 200 types of primordial dwarfism, 5 sub types are more severe and it is estimated that there are only 100

individuals in the world afflicted with the disorder. The life expectancy for these patients is around 30 years but very few survive their intra-uterine phase.

Case Report

A 22 year old boy reported to our cleft centre with a chief complaint of hole in the palate. His parents revealed history of consanguineous marriage. He was born at term with uneventful gestation period and birth weight of 1.5Kg.

Clinically, the boy had a playful friendly nature that spoke few words and obeyed simple commands. His skin was dry, scaly and without hairs. His face was 'Bird like', triangular in shape with multiple scars and corneal haziness was noticed bilaterally (Fig. 1).

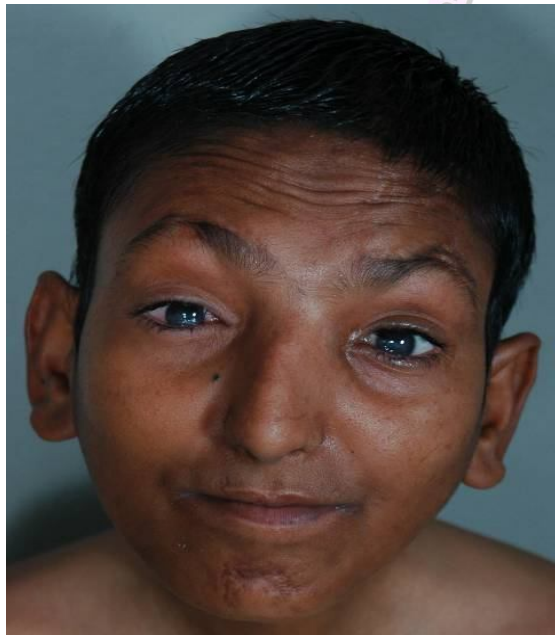


Fig.1 Bird like face with corneal haziness

with abnormal lengthening of both the arms (Fig. 2).



Fig.2 Physical appearance

He looked old with a 'Beak like' nose and receding chin. He had an unsteady and abnormal gait due to mild bowing of his legs

Intra oral examination revealed incomplete cleft palate with normal dentition (Fig. 3).



Fig.3 Incomplete Cleft Palate with normal dentition

Bilateral Hallux valgus was noticed with shortened great toe (Fig.4).



Fig.4 Bilateral Hallux Valgus

On general examination, he was short for his age. The height, weight, and occipito-frontal circumference were 130 cm (< 3rd centile), 24.7 Kg (< 3rd centile), and 47.5 cm (< 3rd centile) as compared with normal a range of 175 to 180 cm, 68 to 72 kg and 65 to 70 cm respectively for his age. He showed signs of global developmental delay with delayed bone age (>7< 8 years) and generalized decreased muscle tone. The external genitals were present but underdeveloped with absence of secondary sexual characters. CT scan of brain showed no intracranial abnormality and neurological examination was normal.

Blood investigations, thyroid function test, ultrasound of abdomen, electrocardiogram and echocardiogram were normal.

Discussion

Caroline Crachami was the first recognized individual in 1815 with primordial dwarfism.^[2] The five main sub-types of primordial dwarfism are Seckel Syndrome, Osteodysplastic Primordial Dwarfism Type I, Osteodysplastic Primordial Dwarfism Type II, Russel-Silver Syndrome and Meier-Gorlin Syndrome. Other osteodysplastic cases with short stature, microdontia, opalescent teeth, and rootless molars have also been described.^[3, 4]

Association of seckel syndrome with cleft lip and palate present together has been documented^[5] but as per our knowledge this is the first report of a combination of primordial dwarfism with isolated cleft palate. The features suggestive of primordial dwarfism in our case were primordial short stature, with height, weight, and occipito-frontal circumference less than 3rd centile and no mental/cranial nerve dysfunction.

In view of the large number of abnormalities reported, these patients need to be evaluated thoroughly to rule out systemic problems, particularly pituitary, thyroid, and adrenal dysfunction. Seckel and Russell-Silver syndromes are the most clearly defined syndromes within the category of primordial dwarfism while accurate diagnosis is more difficult with the osteodysplastic types of primordial dwarfism, even with the most experienced geneticists. The clinical features present in our case and as described in the literature, resembled osteodysplastic primordial dwarfism type I with associated unique features like skeletal changes in the legs, abnormal hair pattern and cleft palate.

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