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Case Report

Dyssegemental Dyspalsia; Rolland-Desbuquois Type — A case report from Pakistan

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Abstract

Two different forms of dyssegmental dysplasia can be distinguished; the lethal Silverman-Handmaker type and less severe Rolland-Desbuquois type. Patients with Rolland-Desbuquois type often survive beyond neonatal period. The purpose of this paper is to report a rare case of Dyssegmental dysplasia, Rolland-Desbuquois type from Pakistan.

Introduction

Dyssegmental dysplasia (DD) is a rare autosomal recessive micromelic dwarfism with anisospondyly and pathogonomic features in spine.[1] Apparently, first case of DD was reported by Silverman in 1969 as a case of "unclassified dysostotic dwarfism."[2] Later in 1977 Handmaker et al coined the term "dyssegmental dysplasia."[3] This name was a better description of the condition highlighting the marked disorganization of vertebral bodies appearing as vertebrae of variable size and shape, which is the most important diagnostic radiological feature. Alerk et al described clinical, radiographic and morphological heterogeneity of DD in 1987.[4] In literature two distinct forms of DD have been demonstrated; the milder Rolland-Desbuquois type and the severe Silverman Handmaker type. After the review by Alerk et al, Gorlin RJ serendipitously discovered a case described by Dr. M. Simmonds dating from 1900-1901. Dr. M. Simmonds had actually described early use of x-ray for rare skeletal malformation. And this case was a clear example of Silverman-Handmaker type of DD.[5] This may be considered as the true first description of DD.

The purpose of this paper is to report a case of Dyssegmental Dysplasia Rolland-Desbuquois type from Pakistan. This is the first case report to the best of my knowledge describing DD Rolland-Desbuquois type from Pakistan.

Case Report

A male baby was born at 39 weeks of gestation to a gravida two, after a lower segment caesarean section due to breech presentation.

Parents were first cousins, mother was 22-year-old and father was 30-year-old. Mother's first pregnancy was terminated at 28 weeks of gestation, as ultrasound indicated the presence of short limb chondrodysplasia.

Mother received prenatal care throughout this pregnancy. An ultrasound study done at 24 weeks of gestation showed short limbs and a narrow thorax. The couple opted to continue the pregnancy this time and a male baby was born at 39 weeks of gestation with good apgars. At birth he was noted to have multiple birth anomalies. Birth weight was 3.2 Kg (50th percentile), length was 46 cms (2nd percentile) and head circumference was 36 cms (91st percentile)

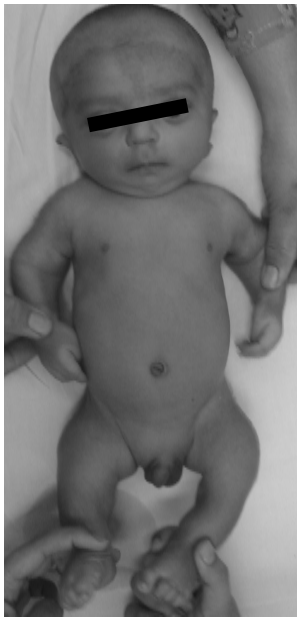

Figure-1: Clinical photograph showing prominent eyes, depressed nasal bridge, micrognathia, narrow chest, flexion contracture at both elbows.

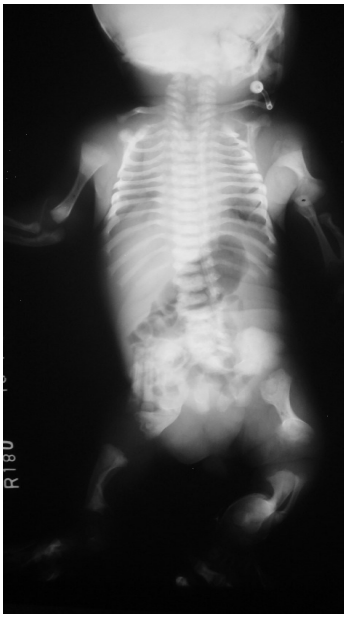

Figure-2: Babygram showing short dumbbell-shaped tubular bones and bowing of legs, short ribs and hypoplastic radii.

Physical examination revealed prominent eyes due to hypoplastic supra-orbital ridges, a flat nasal bridge, microganathia, short neck, narrow chest and short curved limbs especially the femurs. In addition, flexion contractures were noted in both elbow joints and right hip joint. The baby also had significant hirsutism though there was no cleft palate (Figure 1 and 2). Cardiovascular, ophthalmological examination and renal ultrasounds were normal.

Radiographs showed short and thick tubular bones with bulbous ends. Right femur and left tibia showed significant bowing, right tibia was slightly bowed. Iliac bone appeared short and rounded. The trunk was short with short ribs having flaring of anterior ends. Cervical vertebral bodies appeared poorly ossified (Figure-3). The most striking

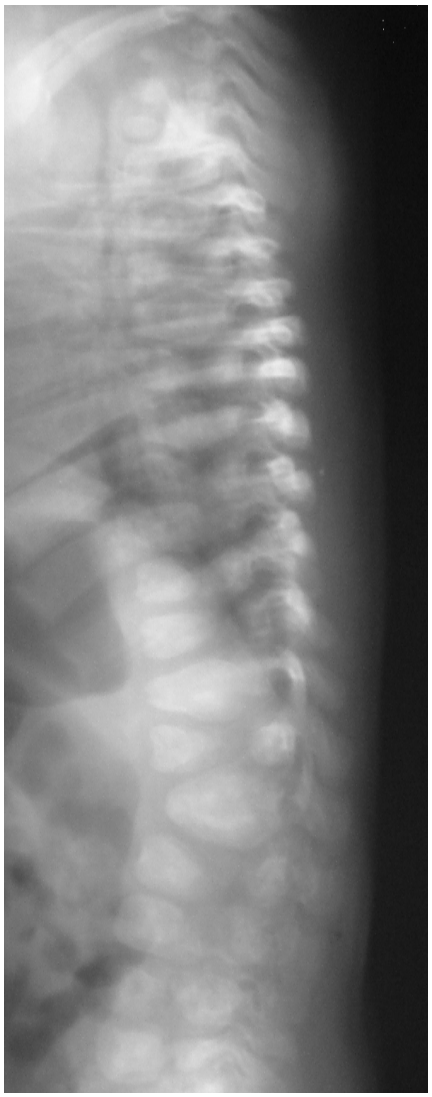

Figure-3: X-ray spines lateral view showing marked variation in size and shape of vertebral bodies with smooth margins.

radiological feature was seen on the lateral view of the spines, which showed vertebral bodies of variable size and shape having smooth margins.

Discussion

DDis a rare autosomal recessive, lethal anisospondylic camptomicromelic dwarfism.[5] Two types of DD are recognized; Silverman-Handmaker type, which is a lethal and severe form as most patients are either stillborn or die within first few days of life. Second type is Rolland-Desbuquois type which is the milder form; affected newborn may survive for as long as three years. In most cases death occurs secondary to pulmonary complications.[4,6]

Silverman-Handmaker type is characterized by markedly short stature, short bowed limbs, limited joint mobility, facial dysmorphism, cleft palate and equinovarus deformity of the feet. Radiographic examination of the spines show a peculiar type of malsegmentation characterized by delayed ossification of the vertebral bodies. Each vertebral body may contain two or more ossification centers of different sizes and shape. This type of DD is caused by mutation in the gene encoding perlecan.[7]

In Rolland-Desbuquois type both clinical and radiological features are less severe than the Silverman-Handmaker type. Rolland-Desbuquois type may show phenotypic resemblance to Kniest Dysplasia.[8] However, radiologically the two conditions are easily differentiated; malsegmentations seen in Rolland-Desbuquois type is not present in Kinest dysplasia whereas platyspondyly and kyphoscoliosis is a prominent feature of Kinest dysplasia and is absent in Rolland- Desbuquois type of DD.

At the time of writing this case report, the baby is surviving at 5 months of age, his clinical and radiological features are consistent with the diagnosis of Dyssegmental dysplasia, Rolland-Desbuquois type. In this case, short-limb dysplasia was made on antenatal ultrasound. Prenatal diagnosis by ultrasonic examination in early pregnancy has been reported in DD.[9] Though both types of DD are autosomal recessive but they must be distinguished so that correct prognosis regarding survival can be offered.

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