Journal of Clinical Imaging Science

Editor-in-Chief: Vikram S. Dogra, MD
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CASE REPORT

Fibrochondrogenesis, an Antenatal and Postnatal Correlation

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Received : 03-01-2012
Accepted : 28-01-2012
Published : 18-02-2012

ABSTRACT

Fibrochondrogenesis is a rare, neonatally lethal osteochondrodysplasia, with autosomal recessive inheritance. It differs from other lethal dwarfisms in that it leads to broad, long-bone metaphyses (dumb-bell shaped) and pear-shaped vertebral bodies. We report a case of fibrochondrogenesis with severe pear-shaped platyspondyly, suspected antenatally, and give a comprehensive pictorial review of the antenatal ultrasound and postnatal radiographic findings. Only few cases of fibrochondrogenesis are diagnosed before the termination of pregnancy.

Key words: Antenatal diagnosis, fibrochorogenesis, platyspondyly, ultrasonography

INTRODUCTION

Fibrochondrogenesis is a genetically inherited form of osteochondrodysplasia that occurs in neonate. This lethal variant results from the inheritance of an autosomal recessive gene and causes abnormal development of cartilage and fibrous connective tissues. Fibrochondrogenesis presents several features that distinguish it from other forms of lethal dwarfism. These features are broad, long-bone metaphyses (dumbbell shaped) and shortened ribs with pear-shaped vertebral bodies. We report a case of fibrochondrogenesis with severe pear-shaped platyspondyly (flattened spine), suspected on antenatal ultrasound examination. Only few cases of fibrochondrogenesis are diagnosed before the termination of pregnancy.[1] This case gives a comprehensive pictorial review of the antenatal ultrasound and postnatal radiographic findings, which are not available in current literature.

Access this article online	
Quick Response Code:	Website: www.clinicalimagingscience.org
	DOI: 10.4103/2156-7514.93037

CASE REPORT

A 20-year-old primipara, underwent a routine antenatal ultrasound checkup in the third trimester, at our Institute. Ultrasound images showed a molded fetal head, with bilateral temporal bulging and decreased mineralization of the fetal skull vault, with disparity in the fetal limb

This article may be cited as:
Kundaragi NG, Taori K, Jathar C, Disawal A. Fibrochondrogenesis, an Antenatal and Postnatal Correlation. J Clin Imaging Sci 2012;2:5.
Available FREE in open access from: http://www.clinicalimagingscience.org/text.asp?2012/2/1/5/93037

parameters, bronchopulmonary dysplasia (BPD), and gestational age. A rhizomelic type of dwarfism showing underdeveloped limbs was noted. Metaphyseal flaring with irregularity was observed in the long tubular bones of both the upper and lower extremities [Figure 1]. The metaphyseal ends of the small tubular bones of the hand and foot were normal. Bilateral club foot was noted [Figure 2]. The fetal spine demonstrated severe platyspondyly with unossified posterior vertebral elements, suggestive of defective mineralization [Figure 3]. Other additional features were dorsolumbar scoliosis, short and narrow fetal thorax, and mild protuberance of the abdomen. The 3D fetal face sonogram revealed a depressed nasal bridge and a long philtrum [Figure 4]. After considering all the findings, a provisional diagnosis of fibrochondrogenesis was made.

The patient delivered a live female child within a few days. The antenatal findings were confirmed postnatally. The postnatal infant examination [Figure 5] revealed a flat mid face, with a depressed nasal bridge, flat nose with anteverted nares, long philtrum, short and narrow thorax, scoliosis, deformed extremities, with enlarged joint regions, contractures and restriction of joint movements, and bilateral club foot.

An infantogram showed [Figure 6] shortening of all the long tubular bones of the extremities predominantly affecting the proximal segments (rhizomelic type of dwarfism). Metaphyseal flaring, giving rise to the dumb-bell appearance, with irregularity and peripheral spurs were noted. Ribs were short with wide cupped anterior and posterior ends, long and thin clavicles, and small scapulae. Metaphyses of small tubular bones of the hands and feet squared off without obvious flaring and appeared normal in length. The spine demonstrated scoliosis. A lateral radiograph of the spine revealed severe pear-shaped platyspondyly, with increased intervertebral disk spaces [Figure 7]. Short broad iliac bones with caudally bump-like configuration (bordered by spurs) and flattened acetabulae with medial spikes and narrow sacrosciatic notches were observed. The baby died on day three after birth, due to respiratory distress.

DISCUSSION

Fibrochondrogenesis is congenital disorder that was first described by Lazzaroni Fossati et al., in an infant who manifested many of the characteristics of thanatophoric dysplasia. However, marked methaphyseal flaring of long bones, clefting of the vertebral bodies, and a distinctive morphological lesion of the growth plate, distinguished fibrochondrogenesis from thanatophoric dysplasia.

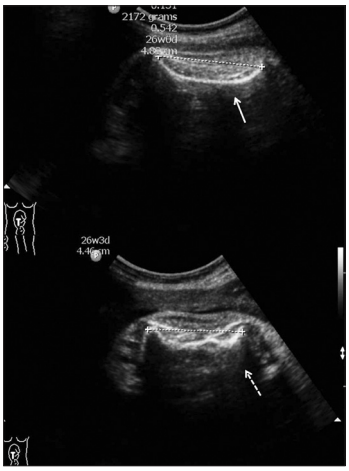

Figure 1: Antenatal ultrasound image showing short femur (white arrow) and humerus (dashed arrow) with mataphyseal flaring.

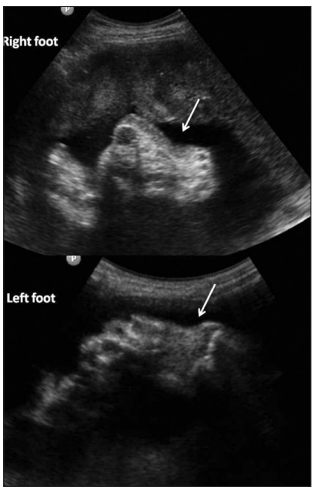

Figure 2: Antenatal ultrasound image showing bilateral club foot (white arrows).

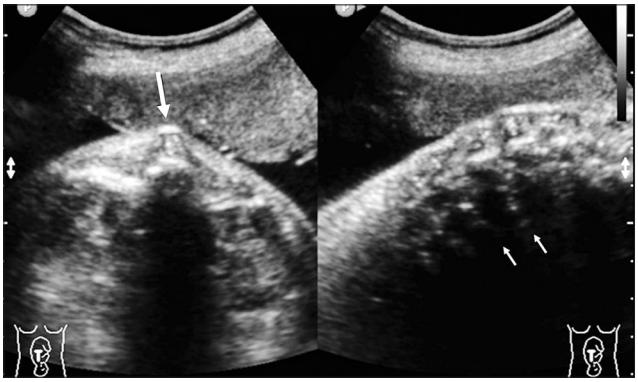

Figure 3: Antenatal ultrasound of the fetal spine showing severe platyspondyly (white arrow) with reduced mineralization of most of the posterior vertebral elements).

One of the studies showed prevalence of fibrochondrogenesis in the United Arab Emirates (UAE) was 1.05 : 10,000 births. This ratio appear to be far higher than the rates described in other world populations.[2] High prevalence of this disorder is noted in consanguinous marriages, it affects both sexes, and a concordance of affected male twins has been reported.

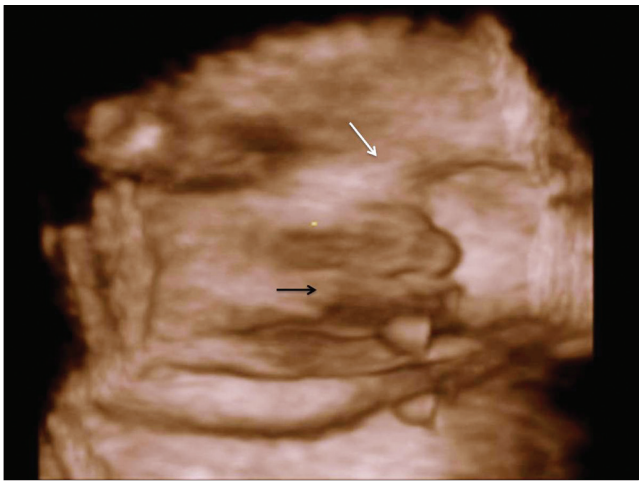

Figure 4: 3D ultrasound image of the fetal face showing depressed nasal bridge (white arrow) and long philtrum (black arrow).

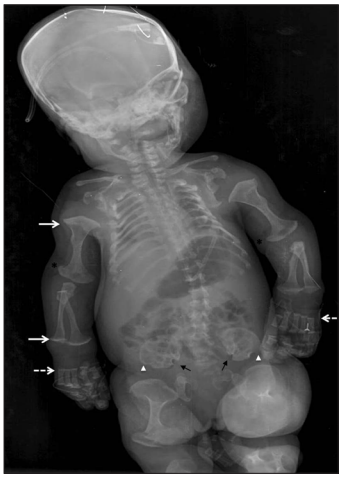

Figure 6: Postnatal infantogram AP view. Short, dumbbell shaped, long, tubular bones with metaphyseal irregularities (white arrows) peripheral spurs (asterisks), rhizomelic dwarfism, ribs are short and thin, and show anterior and posterior cupping, long and thin clavicles, and small scapulae. Metaphyses of small tubular bones of the hands and feet, squared off without obvious flaring (black arrow), with normal length. Pelvis showing short broad iliac bones noted with caudally bump-like configuration (bordered by spurs) (arrowhead), and flattened acetabulae with medial spikes and narrow sacrosciatic notches (dashed arrow).

Fibrochondrogenesis is characterized by limb and vertebral deformities including shortened dumbbell shaped metaphyses and pear-shaped vertebral bodies. The short limbs noted in fibrochondrogenesis are in a proximal or rhizomelic pattern. This is in contrast to dyssegmental dysplasia, which is characterized by micromelia or disproportionate shortening of the entire extremity.

A prenatal diagnosis of fibrochondrogenesis can be made if a fetus has short limbs and deficient ossification of the vertebral bodies [Table 1]. The major postnatal features described radiologically[3] are:

1. Defective ossification of the posterior parts of vertebral bodies
2. Short ribs with splayed ends

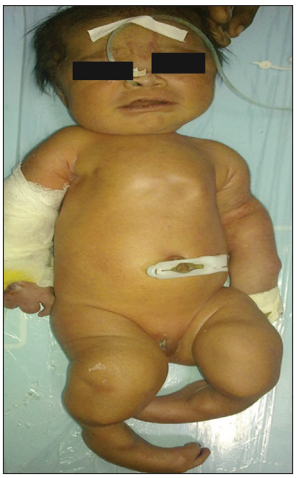

Figure 5: Clinical photograph of the infant showing flat mid face with depressed nasal bridge, flat nose with anteverted nares, long philtrum, short and narrow thorax, scoliosis, deformed extremities with enlarged joint regions, with contractures and restriction of joint movements, and bilateral club foot.

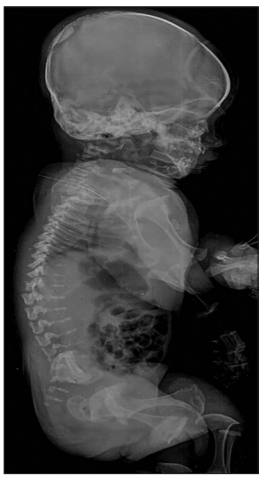

Figure 7: Lateral radiograph of the spine. Severe platyspondyly with increased intervertebral disk spaces noted involving the entire vertebral column. Bodies of the vertebrae appeared small and pear-shaped on lateral view, with more height anteriorly and tapering posteriorly.

3. Small ilia with spurs extending caudally from the acetabular roof
4. Short tubular bones with bulbous ends

The vertebral bodies are flat, with increased intervertebral spaces, and coronal clefts separate the diamond-shaped anterior portions of the vertebral bodies from the small dorsal ossification centers. Ossification of vertebral bodies can be reduced to thin, wafer-like structures.[4]

The face is round and flat with prominent eyes and anteverted nares. Small palpebral fissures with anti-mongoloid obliquity, low-set abnormally formed ears, small mouth, cleft palate, and hypertelorism are seen.

A narrow chest, moderately severe micromelia, and markedly enlarged joints are usually noted in fibrochondrogenesis. The head and neck are proportionate. The skull is relatively

Table 1: Antenatal and postnatal radiologic and clinical feature associated with Fibrochondrogenesis

Antenatal
- Short limbs - Deficient ossification of the vertebral bodies[4] - Protuberant eyes - Flat midface - Flat small nose with anteverted nares - Small mouth with long upper lip - Cleft palate - Micrognathia - Bifid tongue
Postnatal
Clinical - Round and flat face with prominent eyes and anteverted nares - Small palpebral fissures with antimongoloid obliquity - Low-set abnormally formed ears - Small mouth - Micrognathia - Cleft palate - Hypertelorism - Narrow chest - Moderately severe micromelia - Markedly enlarged joints - Hand and foot contractures
Major radiological features described[3]
- Defective ossification of the posterior parts of the vertebral bodies - Short ribs with splayed ends - Small ilia with spurs extending caudally from the acetabular roof - Short tubular bones with bulbous ends - Other radiological features described - Large anterior fontanelle - Hypoplastic facial bones - Long and thin clavicles - Small scapulae - Short and wide ribs, cupped at both ends - Flat vertebral bodies with increased intervertebral spaces and coronal clefts - Short and broad (dumb-bell shaped) long bones with irregular metaphyses and peripheral spurs - Sometimes fibula is disproportionately short - Bones of hands and feet are usually normal, except for mild brachydactyly - Small, narrow, sacrosciatic notches and medial acetabular spike in iliac bones - Short and relatively broad ischium and pubis, have also been observed

large with large anterior fontanelle and hypoplastic facial bones. The clavicles are long and thin and the scapulae are small. Ribs are short and wide, cupped both anteriorly and posteriorly. The long bones are short and broad (dumb-bell shaped) with irregular metaphyses and peripheral spurs. The fibula is sometimes disproportionately short. Bones of the hands and feet are usually normal, except for mild brachydactyly. The vertebral bodies are flat, with increased intervertebral spaces, and the coronal clefts separate the diamond-shaped anterior portions of the vertebral bodies from the small dorsal ossification centers, and project a diagnostic pear-shaped silhouette on lateral view. Ossification of the vertebral bodies can be reduced to thin, wafer-like structures. The ilia are small with narrow sacrosciatic notches and medial acetabular spike. The ischia and pubic bones are short and relatively broad and the fibulae are short.[3-5] Hand and foot contractures have also been observed.

Schneckenbecken dysplasia is radiologically similar and may be etiopathogenetically related. The medial protrusion of ilia is more pronounced (snail-like appearance) and tubular shortening is less severe, with less bulbous ends, in Schneckenbecken dysplasia. Skeletal abnormalities in lethal metatrophic dysplasia are similar, but more severe than in fibrochondrogenesis, however, the metacarpals and phalanges are short and dumbbell shaped.[4] The differential diagnosis should include conditions associated with significant metaphyseal flaring, such as, metatropic dysplasia, Kniest dysplasia, and spondyloepiphyseal dysplasia congenita. Thanatophoric dysplasia should also be considered. Although few of them share radiological findings similar to those of fibrochondrogenesis, none of them show defective ossification of the posterior parts of the vertebral bodies or short ribs with splayed ends.

In conclusion, the antenatal and postnatal findings of defective ossification of the posterior parts of vertebral bodies with rhizomelic shortening and flaring of long tubular bone metaphyses, with peripheral spurs, and both anterior and posterior rib cupping, made us reach the diagnosis of fibrochondrogenesis. This rare case of fibrochondrogenesis gives a comprehensive pictorial review of the antenatal ultrasound and postnatal radiographic findings, which have not been shown in the current literature.

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Source of Support: Nil, **Conflict of Interest:** None declared.