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## Achondrogenesis: A Case Report.

R Sidhu Ganesh\*, and K Kanakaraj.

Department of Radiodiagnosis, Sree Balaji Medical College & Hospital, Chennai, Tamil Nadu, India

### ABSTRACT

Achondrogenesis is a group of severe disorders that affect cartilage and bone development. Achondrogenesis may be differentiated from other skeletal dysplasias by having the most severe degree of limb shortening. Characterized by extreme micromelia – (Short trunk, large cranium)

**Keywords:** Skeletal dysplasias, Micromelia, Ultrasound, Types of Achondrogenesis.

*\*Corresponding author*

## INTRODUCTION

Lethal osteochondrodysplasia characterized by hypoplasia of bone, resulting in marked limb shortening and associated with severe pulmonary hypoplasia. There may be other anomalies, which vary in severity [1].

The prenatal diagnosis of achondrogenesis is possible with ultrasound beginning at 13 weeks gestational age. The major ultrasound findings associated with this lethal osteochondrodysplasia include extreme micromelia, short trunk and neck, poor vertebral ossification, normal to poorly ossified cranium (which appears enlarged relative to the limbs and trunk), and a protuberant abdomen.

### Case report:

21 years old patient with 20 weeks amenorrhea came to radiology department for regular antenatal checkup. Last menstrual period (LMP): 19/12/2014. Previous ultrasound done outside showed single live intrauterine gestation corresponding to 8 weeks 4 days.

### Previous Obstetric History:

First pregnancy – Spontaneous conception followed by spontaneous abortion in one and half month.

Second pregnancy – Spontaneous conception followed by spontaneous abortion in one and half month (Medical method of MTP done on both pregnancies)

### Past History:

Not a known case of diabetes mellitus, hypertension, tuberculosis, bronchial asthma. No previous history of surgery. No history of drug intake (except folic acid). Non-consanguineous marriage.

### Family History:

Family history of congenital anomaly noted and patient's mother had poorly formed ears.

### On Examination:

Patient had polydactyly of left hand (Figure 1) and divergent squint. All blood parameters were under normal limits (HIV, HBsAg, RFT, LFT)



Figure 1: Polydactyly of patient's left hand

**Ultrasound:**

II Trimester ultrasound was performed at 21 weeks of gestation, which showed multiple limb deformities. Right upper limb showed absence of both forearm bones (radius and ulna) (Figure 2). Both lower limbs showed absence of femur with absence of tibia and fibula on right side. Both foot were present with the right feet directly attached to the right hip and the left feet attached to the left ankle (tibia and fibula). Both foot showed clubfoot deformity (Figure 3).

Thoracic circumference was smaller than the abdominal circumference (Figure 4) with associated crowding of ribs. Bi-parietal diameter and head circumference corresponded to the gestation age.

All above features were suggestive of skeletal dysplasia - Achondrogenesis.

Following the diagnosis based on ultrasound, patient was advised termination of pregnancy. Mifepristone and Misoprostol were administered. Dead born female fetus weighing ~ 300gms was expelled (Figure 6). Amniotic fluid was clear and placenta appeared healthy.



**Figure 2: Right upper limb with absence of both forearm bones (radius and ulna).**



**Figure 3: Both femur were absent with presence of left tibia and fibula. Bilateral clubfoot with right directly attached to the hip.**



Figure 4: Thoracic circumference was smaller than the abdominal circumference



Figure 5: Expelled fetal specimen.



Figure 6: Radiographs of dead fetus.



## DISCUSSION

Micromelia – Shortening involving entire limb.

Rizomelia – Shortening involving proximal segment.

Mesomelia – Shortening involving intermediate segment (eg. Radius & Ulna).

Acromelia – Shortening involving distal segments (eg. Hand) [2]

Achondrogenesis may be differentiated from other skeletal dysplasias by having the most severe degree of limb shortening and is characterized by extreme micromelia – (Short trunk, large cranium). Defective production of cartilage matrix by chondrocytes, resulting in poor ossification is the pathogenesis [3].

### Types of Achondrogenesis:

Type IA (Houston-Harris) (Autosomal recessive). Achondrogenesis type IA is characterized by minimal ossification of the skeleton. The skull is poorly ossified. There is no ossification of the vertebral bodies, and the iliac bones appear crenate. The ribs have cupped metaphyses with multiple fractures. The epiphyseal cartilage contains many vascular channels, which are dilated. The growth plate demonstrates disorganized endochondral ossification with lack of columnization in the proliferative and hypertrophic zones[5].

Type IB (Parenti-Fraccaro disease) (Autosomal recessive). Type IB is differentiated from sub-type IA by cranial vault ossification and the absence of rib fractures. Generally, the long bones are shorter in type IB than those seen in type IA.

Type II – Langer saldino disease (autosomal dominant mutations of the type II collagen gene). It is identified by defective enchondral ossification, good ossification of skull vault, non-ossification of lower lumbar vertebra and sacrum, short and stubby horizontal ribs without fracture, short trunk with narrow chest and protruding abdomen. Polyhydramnios is common[5].

Differential diagnosis include: Achondroplasia, Asphyxiating thoracic dystrophy (Jeune Syndrome), Hypophosphatasia, Osteogenesis Imperfecta and Thanatophoric dysplasia[4, 6].

## REFERENCES

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