Achondrogenesis is a type of skeletal dysplasia. Skeletal dysplasias are the heterogeneous class of bone growth disorders resulting in abnormal shape and size of the skeleton. Here, we present a rare case of achondrogenesis which was delivered by induced abortion at 6½ months of gestation. The physical, radiological, and ultrasonographic examinations done raised the possibility of this very rare anomaly. Achondrogenesis is characterized by extreme micromelia and marked discrepancy between the relatively large head and the decreased trunk length. This rare condition has got genetic mutations associated with it. Achondrogenesis resembles other chondrodystrophies, therefore, its diagnosis needs to be made promptly and accurately.

Keywords: Achondrogenesis, Investigations, Skeletal dysplasias, Type 1 and Type 2 achondrogenesis

INTRODUCTION

Achondrogenesis is a lethal chondrodystrophy characterized by extreme micromelia, short trunk, and a disproportionately large head. It has an autosomal recessive pattern of inheritance.1 It occurs in approximately 1 in 40,000 births.2 Achondrogenesis has been sub classified into Type 1A, 1B, and Type 2, based mainly on clinical and radiological criteria such as pattern of ossification, deficiency, and deformity of ribs.3 The prevalence of this disease is 0.23/10,000 live births, and lethality among perinatal deaths is 1:539.1

CASE REPORT

A male child was born to the mother at 6½ months of gestation. The maternal history revealed that there was no fetal movement on her clinical examination. Antenatal ultrasonography diagnosed fetal demise. Fetal limbs were severely shortened, severe micromelia. The diagnosis of a lethal musculoskeletal dysplasia was made most probably achondrogenesis. Abortion was, therefore, induced, and still birth fetus was delivered. The specimen was then sent to Department of Anatomy, Bundelkhand Medical College, Sagar. The child measured 24 cm in length. The head circumference was 29 cm. On examination, the baby had normal facies. He had spade-like hands. There was no syndactyly or polydactyly. All the four limbs were extremely shortened. Therefore, we further planned to carry X-ray of the child. The X-ray of the case is depicted in this report. Skull bones were well-formed, but there is slight micrognathia. There is shortening of upper and lower limb bones. No fractures of bones were seen (Figures 1-6).

DISCUSSION

There are two main types of achondrogenesis, out of which Type 1 is divided into A and B. Achondrogenesis Type 1 (Parenti – Fracaro type) is characterized by poor ossification of the skull, multiple rib fractures and very short, broad bones of the extremities with marked bowing. In short rib polydactyly syndrome and thanatophoric dysplasia, there is ossification of the vertebral column and pelvis.3 Histologically, achondrogenesis Type 1 is probably a primary disorder of connective tissue and shows severe retardation of chondrocytic proliferation in the physeal cartilage. On the other hand, achondrogenesis Type 2 is probably a disorder representing degeneration of proliferating epiphyseal cells with extremely deficient matrix.4 It also shows varied calcification of calvarium and spine. There are no rib fractures in this type.1 Type 2 has hypercellular cartilage with markedly deficient cartilaginous matrix, primitive mesenchymal chondrocytes with abundant clear cytoplasm.5 There is an increase in vascular channels with abundant perivascular fibrous tissue.4 Achondrogenesis should be differentiated from other forms of lethal neonatal dwarfism which comprise of thanatophoric dysplasia Type 1 and type 2 (the clover leaf skull), short rib polydactyly syndrome Type 1, Type 2, and Type 3, asphyxiating thoracic dystrophy, chondroectodermal dysplasia, severe hypophosphatasia, congenital osteopetrosis.

CONCLUSION

Whenever a skeletal dysplasia is suspected, a proper work-up plan should include a history that is both family and maternal, clinical examination, radiographic, genetic, and morphological examinations. Skeletal anomalies are rare
among birth defects and life threatening to the newborn. Therefore, the present report attempts to throw light on this rare anomaly, its gross and radiological appearances and its differential diagnosis.

REFERENCES

2. Spranger J, Beighton P, Giedion A, Hall J, Horton B, Kozlowski K,

How to cite this article: Yadav A, Yadav M. Achondrogenesis: A lethal chondrodystrophy. IJSS Case Reports & Reviews 2014;1(2):1-3.

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