Osteochondrodysplasia: A Heritable Disorder

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Case Report

ABSTRACT

We report a case of a 25 years old pregnant lady in her 2nd pregnancy reported for prenatal Ultrasonography, which revealed an estimated fetal weight of 428 grams alongwith shortening of the long bones, short neck, cloverleaf skull deformity and thoraco abdominal disproportion. The case was diagnosed as Osteochondrodysplasia.

KEY WORDS: achondrogenesis, clover leaf skull, osteogenesis imperfecta Type 2, ostochondrodysplasia, somatic mosaicism, thanatophoric dysplasia

INTRODUCTION:

The skeletal dysplasia also termed as osteochondrodysplasia is a heritable group of disorders which primarily affect bone and cartilage, but can also involve muscle, tendons and ligaments. These are inherited as either autosomal dominant, autosomal recessive or X linked disorders and some result due to imprinting errors, somatic mosaicism, and teratogen exposure during pregnancy. Recent advances in imaging modalities has improved our abilities to recognize osteochondrodysplasia in the prenatal period.[1,2]. Some dysplasias are lethal in perinatal period and detected on antenatal ultrasound scans, while the nonlethal dysplasia present early in infancy or childhood with disproportionate short stature, failure of linear growth or with other physical deformities. The three most common lethal skeletal dysplasias are thanatophoric dysplasia, osteogenesis imperfecta Type 2, and achondrogenesis. Many pregnant women are offered an array of noninvasive tests to determine if their fetuses are at risk for genetic disorders. [3, 4] Recent advances in imaging modalities has improved our abilities to recognize osteochondrodysplasia in the prenatal period.[1,2]

CASE REPORTS:

A 25 years old pregnant women in her 2nd pregnancy with one previous normal delivery was referred for prenatal USG scan in 26th week of pregnancy according to her last menstrual period. Her family history was normal. A prenatal USG biometry revealed an estimated fetal weight of 428 grams along with shortening of the long bones, short neck and cloverleaf skull deformity. A thoraco abdominal disproportion was also evident which suggested a high probability of severe pulmonary hypoplasia. Preterm normal vaginal delivery occurred at 32 weeks and infant died within 30 minutes after birth.

DISCUSSION:

Dysplasias are the conditions associated with bone and/ or cartilage growth or texture.[5] The appropriate diagnosis of skeletal dysplasia is dependent upon the integration of clinical and family history, physical examination, radiological examination and molecular and biochemical tests. Among these, a radiological evaluation is an integral part of the diagnostic workup of a dysplasia. For the early diagnosis of skeletal dysplasia a well organized scan of each and every fetus starting from head to toe should be done. In case of any anomaly, extensive scan along with karyotyping and if required consultation with a pediatric surgeon should be sought. Some dysplasias are lethal in perinatal period whereas others presents in infancy or childhood with disproportionate short stature, failure of linear growth or with other physical deformities. Ultrasound helps to discriminate between lethal and non lethal forms of skeletal dysplasias and if a lethal anomaly is detected termination of pregnancy is considered or can be closely monitored with parents fully counseled about the prognosis. Extensive examination of the parents and family members is needed to predict the risk in the

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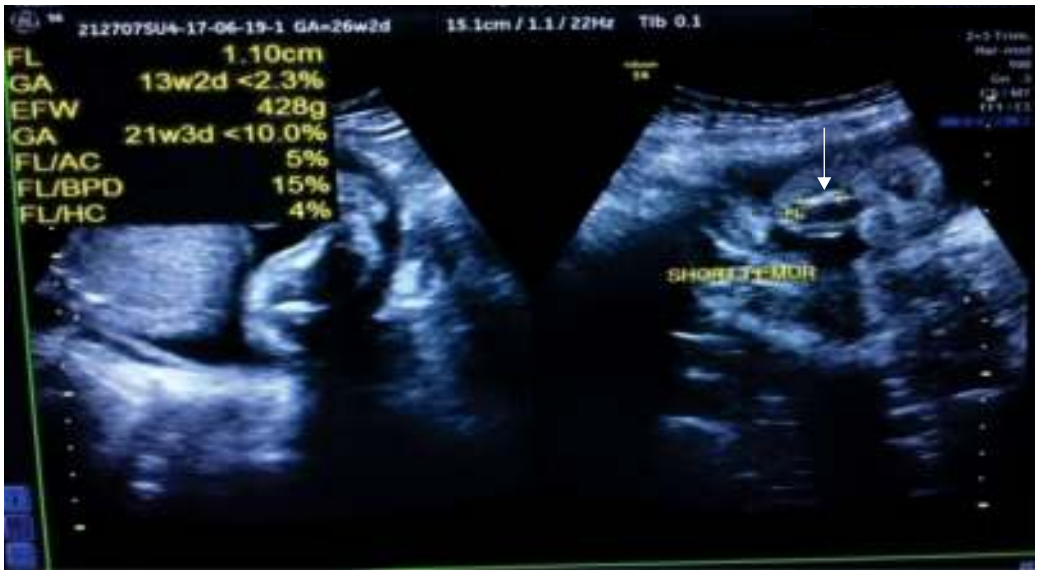

Figure 1: USG image showing short femur.

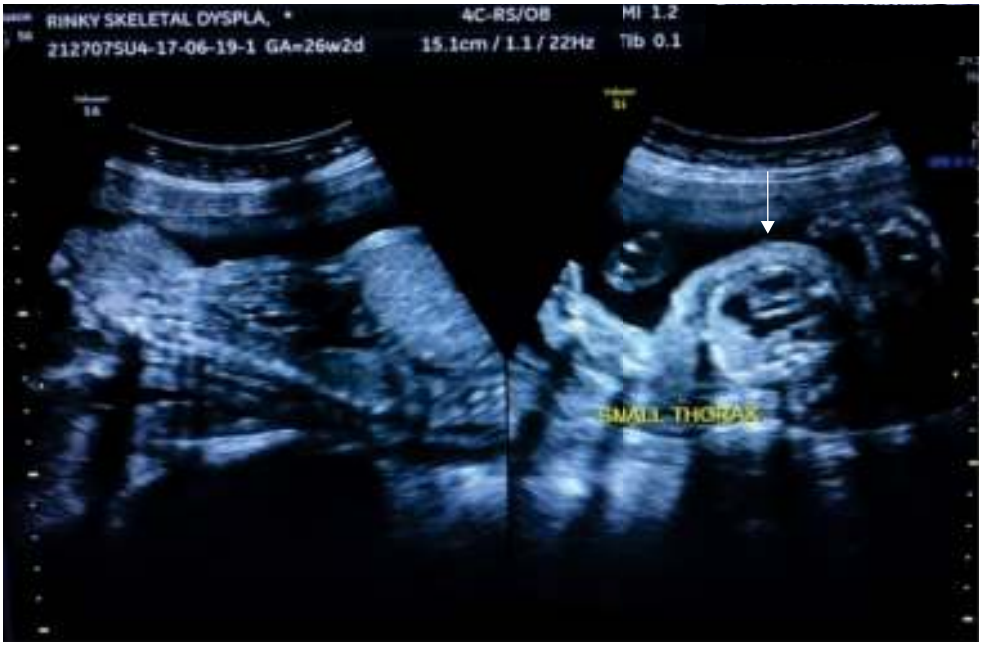

Figure 2: USG image showing small thorax.

next pregnancy. Majority of fetal skeletal malformations can be relatively easily visualised during ultrasound scans and hence ultrasound is the most sensitive way of prenatal diagnosis of these anomalies. However a specific diagnosis depends on the molecular genetics and post mortem examination. The advances in molecular genetics has allowed for gene identification in more than two thirds of the skeletal dysplasias. Chromosomal abnormalities (numerical & structural) are found in most of the patients, of which numerical abnormality (monosomy) is more common as compared to structural abnormalities.

CONCLUSION:

The complete group of osteochondrodysplasia, although individually rare, but it is an important group of disorders which present with significant morbidities due to destruction of bone and cartilage caused by defects in linear growth, bone

Figure 3: USG image showing clover leaf skull.

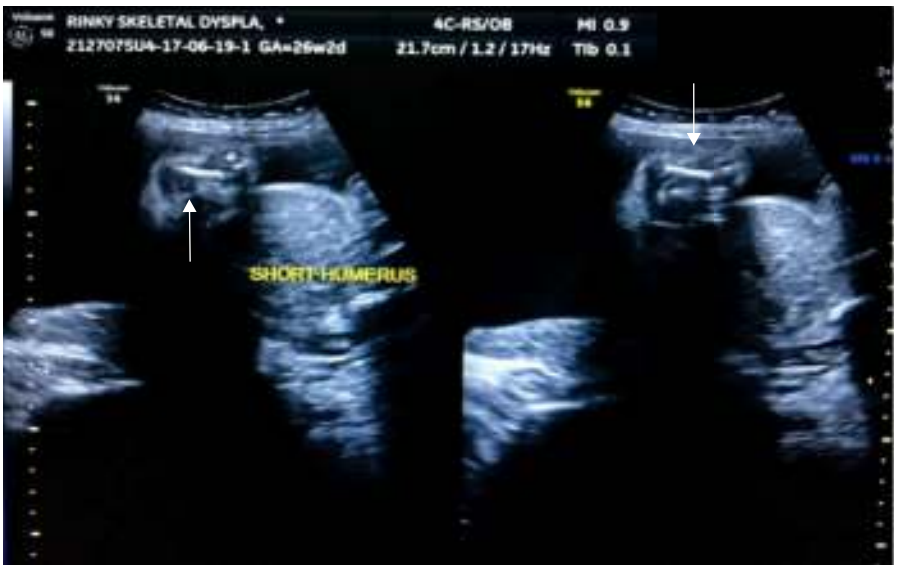

Figure 4: USG image showing short humerus.

modeling and regeneration. Regardless of the specific diagnosis, skeletal dysplasias in general share clinical and radiological findings helping us to group those in several ways. Hence, ultrasound of a fetus suspected to have a skeletal dysplasia is indicated and used to counsel the patient and plan further management.

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