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ANTENATAL ULTRASONOGRAPHIC DETECTION OF LETHAL AND NON-LETHAL SKELETAL DYSPLASIAS ON ROUTINE ANOMALY SCAN

Radiology

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ABSTRACT

Skeletal dysplasias are a heterogeneous group of disorders characterized by abnormal fetal skeletal development .Early antenatal detection is essential for prognostication and counselling. This time-bound multicenter observational study screened 35 pregnant women undergoing routine anomaly scans between 16 and 24 weeks of gestation from December 2023 to March 2025. Fifteen fetuses demonstrated ultrasonographic features suggestive of skeletal dysplasia. Detailed assessment of fetal biometry, long bone morphology, thoracic dimensions, skeletal mineralization, and associated anomalies was performed to further provisionally classify dysplasias as lethal or non-lethal. Of the affected fetuses, 5 (33.3%) were classified as lethal and 10 (66.7%) as non-lethal. Routine second-trimester anomaly scans enable reliable antenatal detection and provide provisional but not definite differentiation between lethal and non-lethal skeletal dysplasias, aiding appropriate counselling and management.

KEYWORDS

Skeletal dysplasia, antenatal ultrasound, anomaly scan, lethal dysplasia

INTRODUCTION

Skeletal dysplasias, also known as osteochondrodysplasias, represent a heterogeneous group of inherited and sporadic disorders characterized by abnormalities in the development and structural integrity of the fetal skeleton.[1] These conditions commonly involve both the axial and appendicular skeleton.[2]

With advancements in ultrasonography, fetal skeletal structures can be reliably visualized by 14 weeks of gestation, making routine second-trimester anomaly scans an essential tool for early detection.[3] Skeletal dysplasias are broadly categorized into lethal and non-lethal forms.

Lethal skeletal dysplasias are frequently associated with severe thoracic hypoplasia, resulting in pulmonary insufficiency and respiratory failure in the neonatal period. Disorders such as achondrogenesis, osteogenesis imperfecta type II, and thanatophoric dysplasia are often incompatible with postnatal survival.[4] In contrast, non-lethal dysplasias, including achondroplasia and diastrophic dysplasia, permit survival into adulthood, although varying degrees of physical disability may occur.

Ultrasonographic evaluation of fetal biometry, particularly long bone measurements during the second trimester, remains central to prenatal assessment. However, definitive subtype diagnosis may require molecular testing due to overlapping imaging features.[5,6] Nevertheless, systematic ultrasonography plays a pivotal role in early detection, prognostication, and appropriate pregnancy management.

AIMS AND OBJECTIVES

Aim: To diagnose skeletal dysplasias during routine second-trimester anomaly scans.

Objectives

- To identify ultrasonographic features suggestive of skeletal dysplasia
- To provisionally classify skeletal dysplasias as lethal or non-lethal based on biometric and morphological criteria

MATERIALS AND METHODS

Source of Data:

- This was a multicenter, time-bound observational study.
- The primary study centre was the Department of Radiology, MGM Medical College, Aurangabad, Maharashtra, with additional collaborating centres following a standardized scanning protocol for second trimester ultrasound using
- Fetal biometry (BPD, AC, femur length, humerus length), long bone morphology, thoracic parameters, skeletal mineralization, digits, fetal profile, and associated anomalies were systematically evaluated.
- The study was conducted from December 2023 to March 2025.
- A total of 35 patients were included in this study.

Inclusion Criteria

- Pregnant women undergoing routine anomaly scans
- Gestational age between 16 and 24 weeks

Exclusion Criteria:

- Scans performed before 16 weeks or after 24 weeks
- Poor visualization due to technical limitations

RESULTS

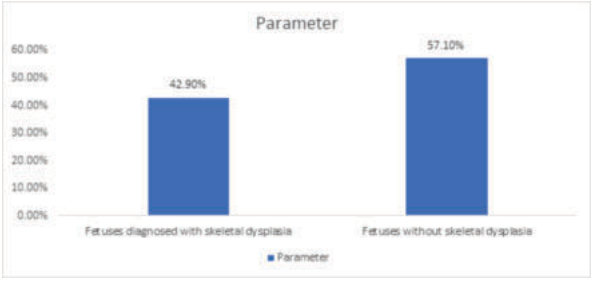


Figure 1-Out of the 35 pregnancies screened, skeletal dysplasia was identified in 15 fetuses (42.9%), while 20 fetuses (57.1%) showed no ultrasonographic evidence of skeletal abnormality.

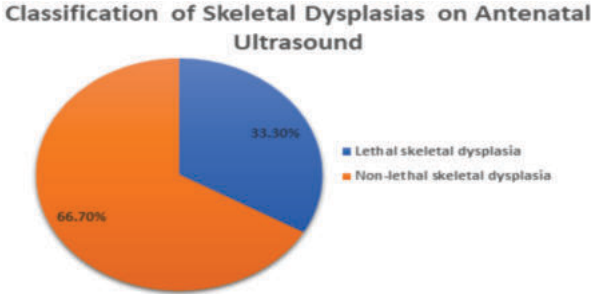


Figure 2: Classification of Skeletal Dysplasias on Antenatal

Ultrasound- Of the 15 fetuses diagnosed with skeletal dysplasia, 5 (33.3%) were categorized as lethal and 10 (66.7%) as non-lethal based on ultrasonographic criteria.

Table 1. Ultrasonographic Features Observed In Lethal Skeletal Dysplasias (n = 5)

Ultrasonographic Feature	Number (n)	Percentage (%)
Thorax to abdomen circumference ratio < 0.7	5	100
Narrow thoracic cavity with protruding abdomen	5	100
Short tubular bones with bowing	5	100
Proximally bent femora (Thanatophoric pattern)	4	80.0
Cloverleaf skull	3	60.0
Reduced humerus length	5	100

Table 1 summarizes the ultrasonographic features observed in fetuses with lethal skeletal dysplasia

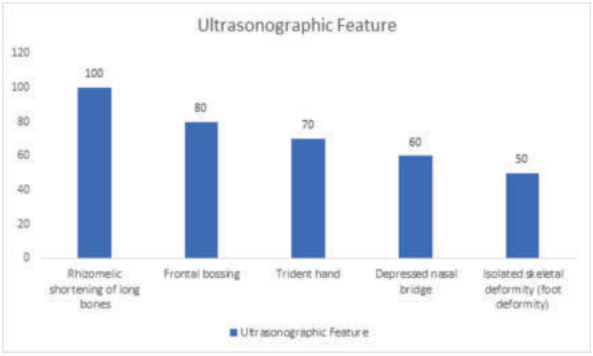


Figure 3: Ultrasonographic Features Observed In Non-Lethal Skeletal Dysplasias

Table 2 Ultrasonographic Parameters Used For Provisional Prediction Of Lethality In Skeletal Dysplasias (n = 15)

Prognostic Parameter	Cut-off	Number (n)	Percentage (%)
Chest circumference / Abdominal circumference ratio	< 0.7	5	33.3
Femur length / Abdominal circumference ratio	< 0.16	5	33.3
Heart to chest circumference	> 50%	5	33.3
Narrow thorax suggestive of pulmonary hypoplasia	Present	5	33.3

Table 2 depicts the ultrasonographic parameters used for provisional prediction of lethality in skeletal dysplasias.

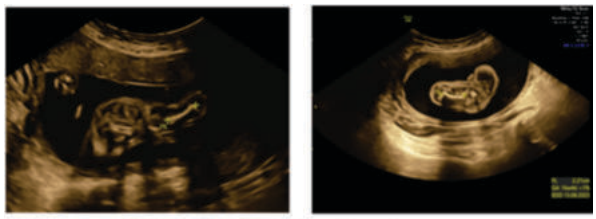
Lethal Skeletal Dysplasias Can Present With A Variety Of USG Findings:

1) Lethality is predicted by a reduced thorax to abdominal circumferene ratio (<0.7)
2) Protruding abdomen with narrow thoracic cavity
3) Short tubular bones with bowing of bones-classical bent femora in thanatophoric dysplasia
4) Cloverleaf skull

Reduced ratio between thorax and abdomen

Non Lethal Skeletal Dysplasias Can Present With Features Like

1) Rhizomelic shortening of long bones
2) Frontal bossing,
3) Trident hand
4) Depressed nasal bridge
5) Isolated skeletal deformities like foot deformity.



Rhizomelic shortening of long bones

Depressed nasal bridge Foot deformity Trident hand

Frontal bossing-increased AP diameter of head

DISCUSSION

Skeletal dysplasias constitutes a group of disorders characterized by abnormal development of the fetal skeleton. Many skeletal dysplasias can now be diagnosed prenatally, especially in the second trimester, due to advancements in ultrasonographic techniques[2] In the current study, 15 out of 35 pregnancies screened during routine anomaly scans had a prenatal diagnosis of skeletal dysplasia (Table 1). Two-thirds of the identified cases were provisionally classified as non-lethal, while one-third were categorized as lethal skeletal dysplasias (Figure 2).[1,2] Antenatal differentiation between lethal and non-lethal dysplasias is essential for further parental counselling and appropriate pregnancy management.

Characteristic ultrasonographic findings of lethal skeletal dysplasias have been outlined to help in provisionally diagnosing them on ultrasound. The primary cause of death in skeletal dysplasias is pulmonary insufficiency and thoracic hypoplasia[7,8]. The most prevalent lethal skeletal dysplasia is thanatophoric dysplasia.[9]

Non lethal skeletal dysplasias show a distinct imaging pattern (Figure 3). The most common non lethal skeletal dysplasia is achondroplasia.[2,7].

The precise type of skeletal dysplasia cannot be definitively diagnosed by ultrasonography alone. For definitive diagnosis and counselling, molecular or genetic testing is necessary since specificity is limited by overlapping characteristics and varied phenotypic expression.[9]

CONCLUSION

Antenatal ultrasonography , especially second trimester routine anomaly scan plays a key role in early detection of skeletal dysplasias and in provisional differentiation between lethal and non-lethal forms. This study demonstrates that a structured ultrasound approach-incorporating fetal biometry, long bone assessment, thoracic measurements, and evaluation of associated anomalies-enables effective detection and further counselling and management. Parameters such as the thorax-to-abdomen circumference ratio, femur

length-to-abdominal circumference ratio, and heart-to-chest circumference ratio are particularly useful in predicting lethality.

However, despite its value in screening and prognostic assessment, ultrasonography has limitations in establishing a definitive diagnosis. Therefore, confirmation often requires genetic testing, invasive diagnostic procedures, or postnatal evaluation. Early prenatal detection and provisional classification remain essential for appropriate counselling, referral, and management planning.

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