Antenatal Diagnosis of Fetal Micromelia with Thoracic Deformity by Ultrasonogram: A Case Report and Literature Review

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Abstract

Fetal micromelia or limb shortening is a relatively common entity among the skeletal dysplasia. It is related to heterogenous group of disorder, can be mild to severe in form and sometimes may be lethal in nature if associated with other congenital defects. To detect this complex problem, a robust systemic sonological assessment is needed in the late first and early second trimester of pregnancy. Here we reported a case of fetal micromelia with chest deformity and polyhydramnios detected by ultrasonogram at 27 weeks of gestation and also discuss about its association with others congenital defects, influencing factors and screening methods for better outcome.

Keywords: Congenital anomaly, Fetal limb abnormality, Micromelia, Ultrasonogram

1. Introduction

Congenital anomalies are a worldwide problem comprising a wide range of abnormalities causing perinatal and infant deaths as well as postnatal physical disabilities. It can be defined as structural, functional or biochemical abnormalities that occur during intrauterine life. The most common anomalies are heart defects, neural tube defects, Down syndrome and skeletal dysplasia. Although approximately 50% of all congenital anomalies cannot be linked to a specific cause, but mostly there are multifactorial causes related to some known genetic factor, environmental predisposition and some risk factors. Many studies observed that these are associated with maternal age, consanguineous marriage, maternal obesity, teratogenic drugs, nutritional deficiency such as folic acid and iodine, infections like TORCH and HIV [1]. The etiology of skeletal dysplasia is very complex, it involves single gene disorders, chromosomal abnormalities, intrauterine factors, vascular events, maternal diseases and maternal exposures to various stimuli, but in many cases, it remains unknown [2, 3]. Multidisciplinary team approach involving obstetrician, sonologist, clinical geneticist, neonatologist and orthopedic surgeons are necessary for prenatal diagnosis of such abnormalities as well as prediction of future prognosis and management planning. Ultrasonogram is a readily available non-invasive screening tool to detect these abnormalities however proper assessment depends upon various factors like skilled sonologist, quality of the ultrasound machine, gestational period of scanning and patient body habitus [4].

2. Case Report

A 20 years female primigravida of 27 weeks gestational age was referred to ultrasound department of Institute of Nuclear Medicine & Allied Sciences, Dhaka for sonological assessment of pregnancy profile. She was normotensive, mildly anaemic (haemoglobin level: 9.9 gm/dl) with no history of previous medical or surgical comorbidity. She did not go through regular antenatal check-up and had no previous ultrasonogram (USG) of her present pregnancy. In per-abdominal examination, uterine height corresponded to her gestational period with detectable fetal heart sound. USG showed a single live intrauterine fetus of 27 weeks duration (according to biparietal diameter and abdominal circumference). Features of micromelia were evident in the form of shortening and curving of all the long bones with approximately 12 weeks of discordance. Femoral length was about 16 mm which corresponds to 15 weeks of gestation (Fig. 1). Fetal chest was narrowed, tubular in shape with reduced thoracic circumference (Fig. 2) with hyper extended curved cervical spines (Fig. 3), relatively thicker scalp tissue (8mm) and evidence of polyhydramnios with amniotic fluid index of about 26cm. Cardiac assessment was within the normal limits with a regular heart rate. No other remarkable abnormality was noted. There was no history of consanguineous marriage, no skeletal abnormality was evident in mother and father and they were of average normal height. Patient was referred to obstetric department at Dhaka medical college hospital and counselled about the poor fetal prognosis. She underwent medical termination of pregnancy. The fetus was still born, post-delivery fetal examination revealed shortening of limbs, narrowed chest deformity and relatively protuberant abdomen (Figs. 4-5).

3. Results and Discussion

Approximately, the prevalence of limb abnormalities is six in 10,000 live births, the incidence is higher in the upper limbs compared with the lower limbs [5]. Micromelia is a form of skeletal dysplasia characterized by abnormally small and imperfectly developed extremities resulting from malformation and disruption of developmental process. Fetal micromelia can be classified under three major divisions: mild, mild with bowing and severe micromelia. They are often a part of underlying syndromes like ATD (Jeune syndrome), chondroectodermal dysplasia (EVC syndrome) and short-rib polydactyly syndromes (SRPS). Shortening of the limbs with long and small thorax, polydactyly, polyhydramnios and sometimes renal and liver diseases are the presenting features in these

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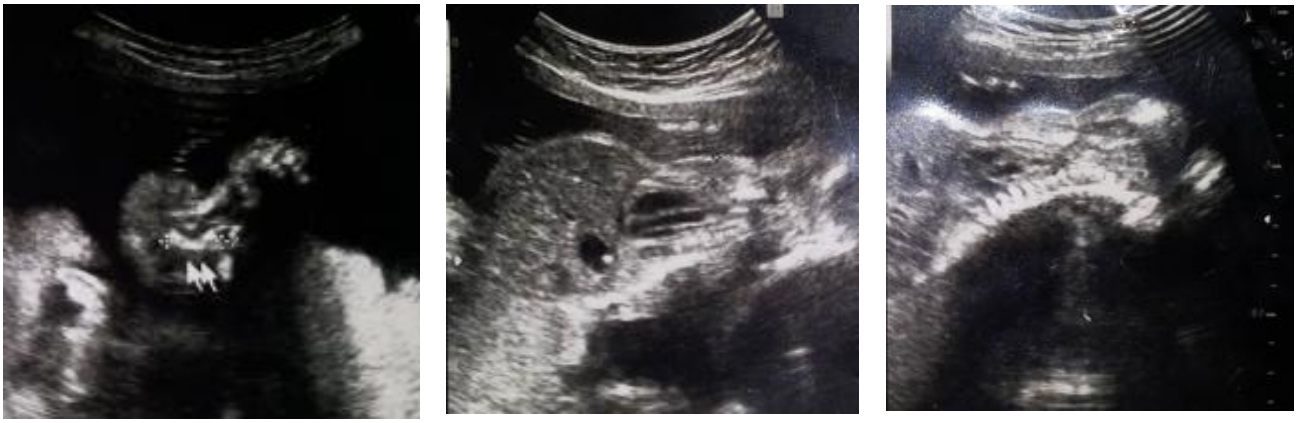
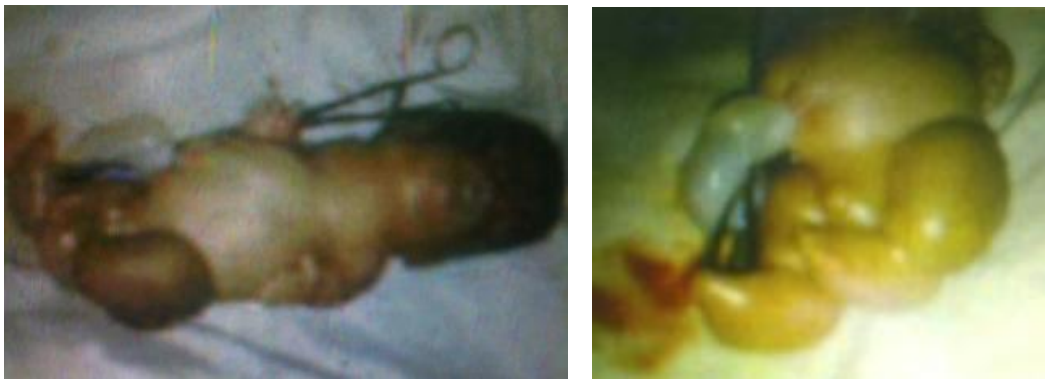


(1) (2) (3)

Fig. 1-3: Antenatal 2D ultrasound images of the fetus showing short and curved femur, (1) evidence of polyhydramnios, (2) tubular shape narrow chest deformity and (3) hyperextended cervical spines



(4) (5)

Fig. 4-5: Post-delivery photographs of still-born congenital anomalous fetus showing shortening of limbs, chest deformity and protuberant abdomen

syndromes detected on antenatal USG scan. The disease is often lethal, owing to its narrow thorax leading to respiratory failure. Chromosomal mutations and autosomal recessive pattern of inheritance are also evident, showing 25% recurrence in the subsequent pregnancies [6-7]. Screening test of maternal blood sample or free fetal DNA screening can help to predict the risk of recurrence in subsequent pregnancy. Diagnostic tests such as chorionic villus sampling and amniocentesis can be used to detect chromosomal abnormalities and infections in women with high risk. These tests are not practiced routinely in our country due to lack of skilled personnel and limited laboratory facility. Usually, assessment of musculoskeletal system is a part of routine fetal ultrasound examination. Genetic USG scan at 11-13 weeks+ is an important readily available cost effective and noninvasive tool for fetal anatomy assessment. Here Nuchal translucency (NT) assessment is more sensitive in early gestation and abnormally increased nuchal fold may associate with major skeletal abnormalities [8]. In second-trimester as fetal skeletal system is completely developed, anomaly scan provides standard morphologic evaluation by 2D conventional ultrasound images. More precise diagnosis can be made by using 3D static images with surface rendering mode or skeletal mode if facilities are available in USG machine. Though there is a high patient load in limited time schedule in the busy laboratories, prior to USG scan it is important to know about history of maternal diseases such as diabetes mellitus, hypercoagulability, autoimmune diseases like systemic lupus erythematosus, myotonic dystrophy, presence of high blood pressure, exposure to teratogens, medications, infections, alcohol, cigarette smoking, history of recurrent miscarriage, stillbirths, mental retardation, inherited conditions, consanguinity and lastly positive family history of congenital anomaly. By correlating with the clinical assessment note by the referring physician, a sonologist can play an important role in diagnosing a congenital defect as well can explain the pathophysiology. Systemic sonological approach may guide the clinicians for management planning thus unwanted recurrence can be easily prevented in future pregnancies.

4. Conclusion

Congenital anomalies are important causes of neonatal mortality and morbidity. In Bangladesh, there is limited laboratory facility for genetic or chromosomal screening of

birth defects and also limited access for amniocentesis in gestational period. In this point of view, sonological assessment of congenital anomalies is the most cost effective and noninvasive procedure. Expert sonological assessment of anomaly scan during routine antenatal checkup helps to create maternal awareness as well ensure early detection and prompt management for better pregnancy outcome.

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