THANATOPHORIC DWARFISM

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Introduction

Thanatophoric dwarfism is lethal skeleton dysphasia; incidence is 6.9:1, 00,000 births. Can be diagnosed by ultrasound in early pregnancy.[1] Thanatophoric dwarfism is characterized by severe micromelic limb shortening, bowing of limbs, narrow thorax, severe platyspondyly,[2,6] polyhydramnios(71% of cases). Clover leaf skull-14% of cases & a small pelvis are also associated condition.[2]

Case Report

A female patient kalsoom w/o zafar presented with gestational amenorrhoea of 25 wks. She was married for 5 years age. G 5 P 1 A 3. Having three 1st trimester abortions, only one female alive issue with premature delivery in that pregnancy. She referred to radiology department for ultrasound examination for 1st time. A gray scale ultrasound finding reveals polyhydramnios, hydrocephalus, fetal skull edema, redundant soft tissue, hand and feet are attached with body wall through small bony structure, proper limb are not identified. Fetus has narrow thorax with bulging abdomen. No evidence of spina-bifida the heart was four chambered. No abdominal visceromegaly seen. On the basis of above mentioned findings possibility of thanatophoric dwarfism was suggested.

Discussion

In the past thanatophoric dwarfism has been mistaken clinically for achondroplasia, particularly heterozygous type in which both parents are of normal stature. Musculoskeletal anomalies are not common in prenatal life. They can be either sporadic or part of chromosomal syndrome causing prenatal mortality and morbidity. Among the images method ultrasonography is the most popular and cost effective for the evaluation in early stage of gestation. Now by sonography it's possible to determine which part of limb is severely shortened.[3] If polyhydramnios is present with dwarfism the bone dysplasia is mostly lethal, the polyhydramnios is thought to be due to esophageal compression by the small chest & abdomen balloons out in bell shape fashion below the rib cage.[4] Basic pathogenic mechanism proposed for thanatophoric dysplasia is persistence of abnormal fetal mesenchymal tissue leading to disorganized endochondral bone formation which transformed to abnormal bone and cartilage.[5]

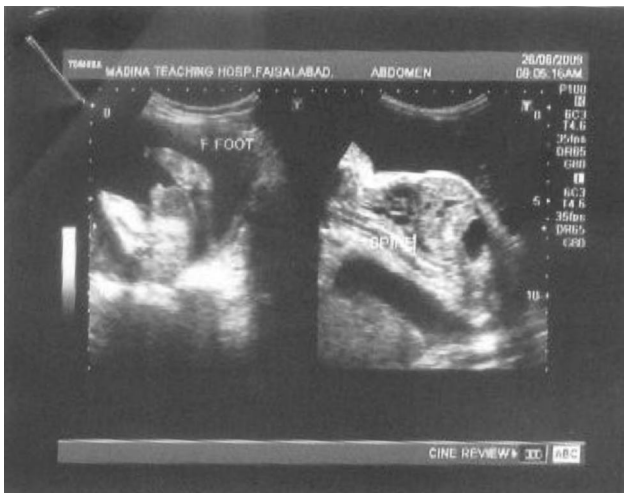

Figure 1: Fetal feet attached with the pelvic wall, narrow thorax with bulging abdomen

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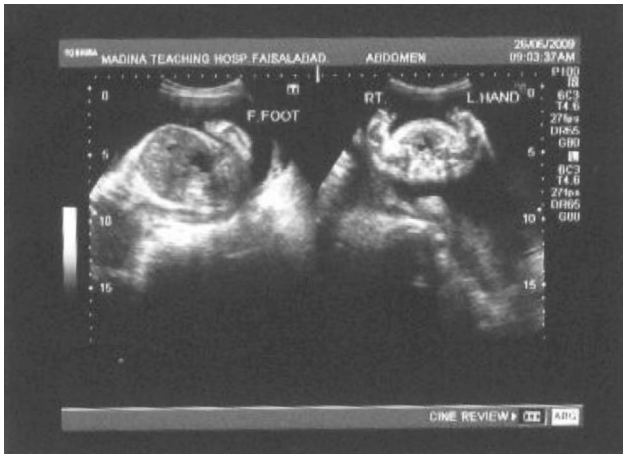

Figure 2: Fetal right and left hand attached with rudimentary bone with chest wall.

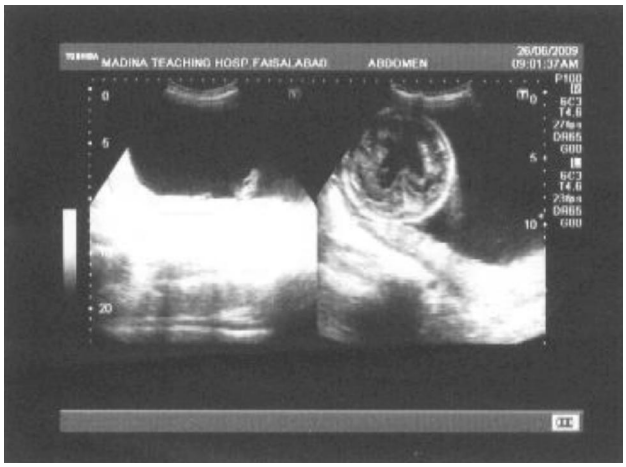

Figure 3: Polyhydromnios and hydrocephalus

References

1. Wolf Gang dahnert, Thanotophoric dwarfism in utero on ultrasound, Primer Radiology review manual 2nd edition; 1993.102.

2. Carol M Rumack, Stephanie R Wilson Charbonneau Fetal musculoskeletal system in diagnostic ultrasound volume 11 Mosby year back, 1991: 849-69.

3. Romero R.A Thanssiadis AP, Jeanty P. Fetal Anomalies RCNA 1989; **28(1):** 75-99.

4. Rogers C Sanders Prenatal Ultrasonic detection of anomalies with a lethal or disastrous outcome RCNA January 1990; **28(1):**163-75

5. MC Alister WH.Herman TE. Osteochondrodysplasia, dysostosis, chromosomal aberrations, Mucopolysaccharidosis In resnik Diagnosis of bone and joint disorders 3rd ed Philadelphia: WB Saunders 1995: 4136-244.

6. Fink IJ, filly RA, Callen PW et al. Sonographic diagnosis of Thanatophoric Dwarfism in utero J. ultrasound Med 1982; **1:**337.