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Thanatophoric Dysplasia : A Rare Form of Skeletal Dysplasia

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ABSTRACT

Thanatophoric Dysplasia is an inherited(AD) rare but severe form of skeletal dysplasia that may require Medical Termination of Pregnancy as the disease usually leads to perinatal death or in less severe forms lifelong disability. Two variants have been identified. Radiology in the form of pre-natal USG provides an excellent tool for suggesting the diagnosis even though confirmation of diagnosis requires genetic testing.

Keywords: Thanatophoric Dysplasia; Cloverleaf skull; telephone receiver femora, Autosomal Dominant, FGFR3

Introduction

Thanatophoric dysplasia (TD) is a short-limb skeletal dysplasia that is usually lethal in the perinatal period. TD is divided into subtypes:

- TD type I is characterized by micromelia with bowed femurs and, uncommonly, the presence of craniosynostosis of varying severity.
- TD type II is characterized by micromelia with straight femurs and uniform presence of moderate-to-severe craniosynostosis with cloverleaf skull deformity.

Other features common to type I and type II include: short ribs, narrow thorax, relative macrocephaly, distinctive facial features, brachydactyly, hypotonia, and redundant skin folds along the limbs. Most affected infants die of respiratory insufficiency shortly after birth. Rare long-term survivors have been reported. We report here two cases confirmed by genetic testing.

Case Report

Case 1

A 32 year old female presented to the USG clinic at 21weeks of gestation for routine Congenital Anomaly Scan . The only abnormality suspected clinically was polyhramnios ..Fig 4(fundal height greater than expected Gestational Age). USG revealed: Growth deficiency with limb length below fifth centile**(Fig 2),** polyhydramnios**(Figure 4),** bowed femurs**(Figure 2)**, platyspondyly, narrow rib cage, Macrocephaly with Cloverleaf skull**(Figure 1**)(Craniosynostosis involving coronal, lambdoid, and sagittal sutures) resulting in a trilobed skull shape. In consultation with Department of Obstetrics, MTP was advised,and the patient willingly accepted. Post MTP molecular genetic testing proved the presence of heterozygous pathologic variant of FGFR3 proving the diagnosis of Thanatophoric Dysplasia (Type II)

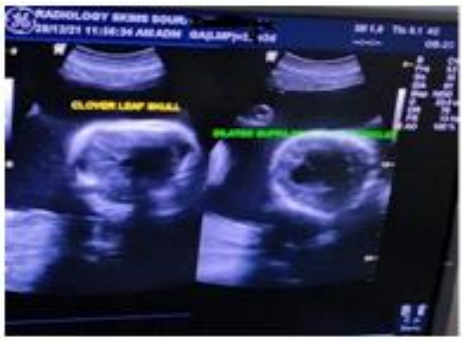

Figure1: Clover Leaf Skull with dilated ventricles

Figure 2: Telephone handle femur(Bowing)

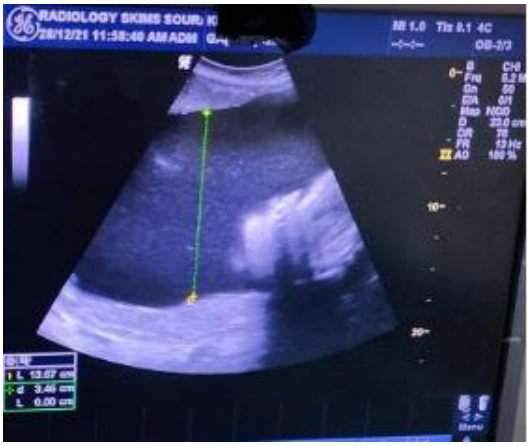

Figure 4: Increased Liqor with polyhydramnios

Case 2

A 26 year old Kashmiri female presented to USG Clinic for her first pregnancy Scan at 18 weeks of gestation, with no previous scan done. Clinically the patient was normal. USG revealed normal liquor levels,normal skull and normal brain. Both femurs were short with "telephone handle" deformity and bowing along with both humeri(**Figure 5).** One of the femurs was so severely bent that it appeared to be on the verge of fracture. Thora was normal, heart was normal, abdominal organs normal.. No other abnormality could be detected. A presumptive diagnosis of Rhizomelic shortening of Bones ? Skeletal Dysplasia was made. Patient was explained the outcome of Child and voluntarily asked for early MTP. Post abortion molecular testing revealed Thanatophoric Dysplasia(Type 1.)

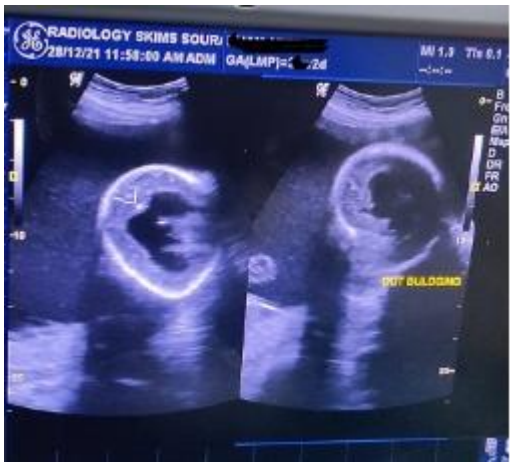

Figure 3. Dilated ventricles with out bulging

Figure 5: Short humerus

Discussion

Thanatophoric Dysplasia is a rare cause of skeletal dysplasia as compared to Achondroplasia and others and many times difficult to distinguish. Some of the key findings for a Radiologist to consider the diagnosis include "Clover Leaf Skull", telephone handle deformity of Femurs, and narrowing of thorax.

Potential pregnancy complications include prematurity, polyhydramnios, malpresentation, and cephalopelvic disproportion caused by macrocephaly from hydrocephalus or a flexed and rigid neck, This necessitates early detection of the anomalies by a radiologist , to ensure planned management in the form of Cesarean Section or MTP is instituted. In our cases both the women chose to Medically Terminate the pregnancies.

Diagnosis can only be confirmed by genetic testing, but role of USG cannot be overemphasized. Antenatal sonography in second trimester not only confirms the diagnosis but also differentiates it from the other non lethal dysplasias.

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