IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 16, Issue 9 Ver. I (Sep. 2017), PP 04-06
www.iosrjournals.org

Case Report of A Rare Congenital Anomaly – Thanatophoric Dysplasia

[1*]Dr.brinda, [2]Dr.saraswathi
Corresponding Author: [*]Dr.brinda

Abstract: *Congenital anomalies areimportant causes of infant and childhood deaths,chronic illness and disability. They are characterized by metabolic , structural ,functional and behavioural disorders present at the time of birth .The etiological factors include chromosomal abnormality and mutation of various genes ,which gives rise to structural and metabolic defects .Thanatophoric dysplasia is a lethal skeletal muscle abnormality incompatible with life due to severe pulmonary hypoplasia .*

Keywords: *congenital anomaly, skeletal muscle dysplasia, thanatophoric dysplasia, pulmonary hypoplasia*

I. Introduction

Thanatophoricdysplasia is a disorder involving transmembrane receptors , it manifests as a lethal osteochondro dysplasia as a result of autosomal dominant mutation ,occurring at fibroblast growth factor receptor 3 ,resulting in accumulation of cysteine residues at various transmembrane receptors. Depending on skeletal radiographs there are two types of thanatophoric dysplasia which includes typeI and typeII.This anomaly can be identified as early as 12 to 14 weeks of gestation by ultrasound examination.

II. Case Report

A 26 yr old primigravida was referred from outside for medical termination of pregnancy at 20 weeks of gestation, in view of anomalous fetus, anomaly being thanatophoric dysplasia. Ondetailed clinical history ,there was no significant past history or any history of genetic disorder in the family ,no history of anyteratogenic drug intake during 1st trimester ,nuchal translucency scan was not done at 12 weeeks of gestation. Inview of lethal skeletal tissue abnormality ,patient was informed and explained about the abnormality and was counselled for termination of pregnancy .MTP proceeded with medical methods. After expulsion of the female fetus ,weighing 250 gms,the fetus were examined .There was significant shortening and bowing of the long bones ,narrow thorax, clover leaf appearance of skull ,frontal bossing ,femur had a typical telephone handle appearance .The gross pathological features correleated with thanatophoricdysplasia.The fetus was sent for autopsy , the report of which was conclusive of thanatophoric dysplasia.

III. Discussion

Thanatophoric dysplasia involves defect of transmembrane receptor resulting from mutation of genes encoding Fibroblast growth factor receptor3 and parathyroid hormone receptor .It is divided into two types typeI and type II based on radiological findings. There's no sexual preponderance noted

Thanatophoric dysplasia can be detected as early as 12 to 14 weeks with predictive signs such

- femur length >5 mm below 2 standard deviations for gestational age ,
- femur:foot<1,
- femur length:abdominal circumference<0.16,
- lethality of skeletal dysplasia depends on decreased thoracic circumference and pulmonary hypoplasia ,calvarial demineralization(mild transuder pressure over the skull resulting in flattening of skull bone).

Ultrasongraphic findings includes :

- increased nuchal translucency ,
- reverse flow in ductus venousus due to narrow thorax compressing vascular flow,
- Shortening of limbs noted
- 2nd/3rd trimester ultrasound findings includes
- growth deficiency of limbs ,less than fifth percentile by 20 weeksof gestation ,
- well ossified spine and skull ,
- platyspondyly ,
- ventriculomegaly,
- narrow thorax with short limbs,
- bowed femur,

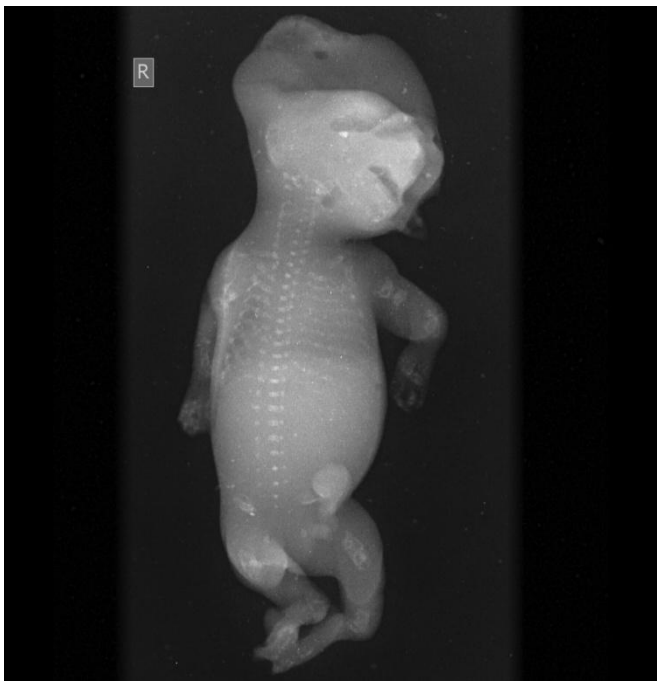
- encephalocele ,
- clover leaf skull
- ,relative macrocephaly .

Morphological features include

- macrocephaly,
- large anterior frontannel,
- frontal bossing ,
- flat facies with depressed nasal bridge ,
- ocular proptosis,
- micromelia (marked shortening of limbs)
- redundant skin folds ,
- trident hand with brachydactyly,
- narrow bell shaped thorax with shortening of ribs,
- protuberant abdomen ,
- generalized hypotonia,
- bowed femur ,
- clover leaf skull ,
- other findings include cardiac defect with renal abnormalities.

Management of this condition is ideally done by termination of pregnancy. Since survival is seen only till infancy because of severe pulmonary hypoplasia which gives rise to severe respiratory distress,as the thoracic cage is narrow. Investigation of this condition includes ultrasonographicex amination. Karyotyping is inconclusive because of decreased sensitivity and specificity. Mutation of FGFR3 genes gives rise to similar skeletal abnormalities with variable deformity such as a chond roplasia, hypochondroplasia, SADDAN (Severe achondroplasia with developmental delay) and osteogenic imperfecta .

Type 1 Thantophoric Dysplasia	Type Ii Thanatophoric Dysplsia
1 In20,000- 50,000	1 In 50,000
Head Circumference >95 Percentile,Frontal Bossing	Head Circumference >95 Percentile,Clover Skull Appearance
Normal Trunk Length,Decreasd Thoracic Circumference	Normal Trunk Length,Decreasd Thoracic Circumference
Short Ribs	Short Ribs
<<3 Rdpercentile,Telephone Receiver Shaped Femur	Straight With Flared Metaphysis
Polyhydramnios Present	Polyhydramnios Present



IV. Conclusion

With recent advances, congenital skeletal abnormalities such as thanatophoric dysplasiacan be screened and the appropriate decision can be taken in time . Hence routine antenatal ultrasound screening for structurally evident ,gross congenital abnormalities is an effective way of diagnosing an anomaly early , thereby avoiding lethal abnormalities being carried till term ,benefiting the mother too .Hence routine screening for anomalies in a women with no risk factors is validated as known .

References

[1]. Nelsons Book Of Paediatrics.
[2]. the prevalence of thanatophoricdysplsia and lethal osteogenesis imperfecta typeII in northern Ireland –a complete population study ,
[3]. Deride E Donnelly,VivienneMcConrll,PatrickJMorrison Thanatophoric dysplasia @gene reviews NCBI bookshelf

*Dr.brinda. "Case Report of A Rare Congenital Anomaly – Thanatophoric Dysplasia." IOSR Journal of Dental and Medical Sciences (IOSR-JDMS) 16.9 (2017): 04-06