Quest Journals
Journal of Medical and Dental Science Research
Volume 7~ Issue 9 (2020) pp: 22-24
ISSN(Online) : 2394-076X ISSN (Print):2394-0751
www.questjournals.org

Research Paper

Type-I Thanatophoric Dysplasia - A Lethal Skeletal Disorder

*Dr.A.Himabindu[1], Dr.B.K.Durgaprasad[2]
**Corresponding Author*
1.Professor & HOD of Anatomy, 2. Professor & HOD of Radiology,
GITAM Institute of Medical Sciences &Research, Rushikonda, Visakhapatnam.

ABSTRACT
Thanatophoric dysplasia is a rare lethal skeletal disorder, characterized by huge head, short limbs narrow thorax with bulged abdomen. It is of two types with typical radiological features of curved bones with telephone receiver pattern of femur and clove leaf skull. It is inhereited as autosomal dominant pattern due to mutation of fibroblastic growth factor receptor 3 gene located on short arm of chromosome 4. A typical case of Thanatophoric dysplasia was found in a destitute fetus with characteristic morphological features that were correlated radiologically. This skeletal disorder can be diagnosed by ultrasound in second trimester of pregnancy. As it has a genetic basis, genetic counseling of parents will help to reduce anxiety and plan further pregnancy due to its low chance of recurrence.

***KEY WORDS**: Skeletal, Autosomal dominant, curved bones, Ultrasound*

***R**eceived 14 December, 2020; **A**ccepted 28 December, 2020 © The author(s) 2020.*
P**ublished with open access at **www.questjournals.org

I. INTRODUCTION

Thanatophoric dysplasia(TD) is a congenital primary bone dysplasia inherited as autosomal dominant condition.It is characterised by severe shortening of the limbs,narrow thorax with distinctive facial features.Its incidence is 1/ 20,000 to 1/ 50,000.The term thanatophorus means death bringing and was first described by Maroteux for dwarf babies who died in first hour of life.(1,2)It is one of the rare, lethal osteochondrodysplasia.(3) Two types of TD have been described based on the bone deformity pattern.Type-1is characterized by marked underdeveloped skeleton and short curved long bones and type -2 is presented with straight bones with cloverleaf skull with premature closure of coronal and lambdoid sutures .(4) TD is due to mutation of fibroblastic growth factor receptor 3 gene located on short arm of chromosome 4.The present study describes about the rare lethal variety of bone dysplasia -Thanatophoric dysplasia.

II. CASE REPORT

Fetuses were collected from Victoria general hospital , Visakhapatnam as a part of departmental activity to develop the museum. An anomalous female fetus was collected that featured huge face , depressed nasal bridge, short neck, narrow thorax with protuberant abdomen. The limbs were markedly shortened and bowed.(fig.1) Fetogram revealed, markedly underdeveloped skeleton with flattened vertebral bodies(platy spondyly) .It had small scapulae and shortened ribs .The long bones were short and curved .Typical presentation of telephone receiver shaped bowing was seen in femora , a feature of type-1 Thanotophoric dysplasia .(fig.2)The domes of diaphragm were raised narrowing the thoracic cavity. The external features correlating with radiological features,that were more in favour of type-I TD.

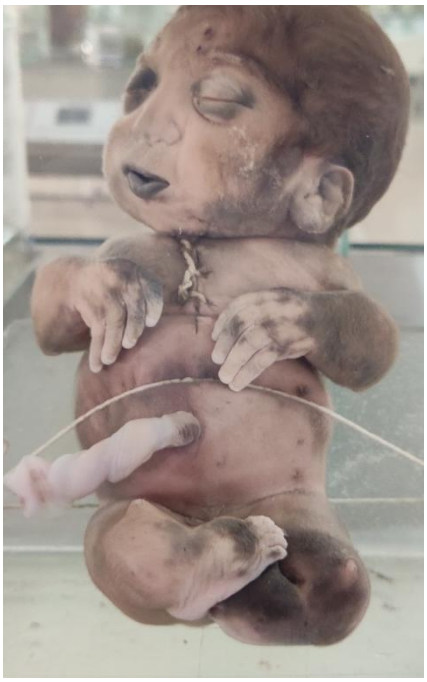

Fig.1 Fetus of Thanotophoric dysplasia –external features

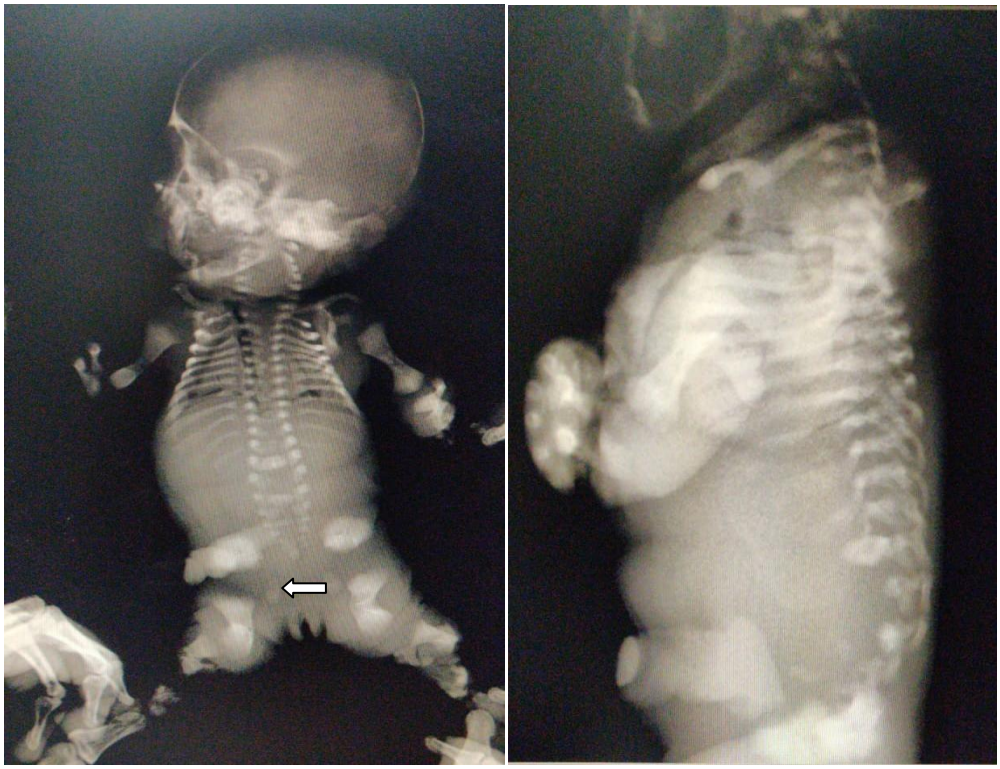

Fig.2 Radiological features of type-I TD-Telephone receiver like femur

III. DISCUSSION

Thanototrophic dysplasia is a lethal skeletal dysplasia, characterized by short and curved limbs, narrow thorax with protruded abdomen. It presents with large head, frontal bossing , prominent eyes and depressed nasal bridge. Based on the skeletal deformity , type-I and type-II TD were described ,TYPE –I is more common and death occurs within minutes after birth usually due to respiratory failure due to hypoplasia of lungs. Both sexes are equally affected with no ethnic or racial predisposition(4)

Generally in this condition, the ribs are short and horizontal .Scapulae are small. The extremities of long bones are short and curved. The femora show characteristic telephonic receiver configuration.(4)

Radiological features of bowing of extremities,large skull base ,reduced interpeduncular spaces of lower lumbar vertebrae, small and squared iliac wings and horizontal roof are also present in achondroplasia which is a close differential diagnosis. (5,6) The distinguishing features in TD are platyspondyly and absence of posterior scalloping of vertebral bodies.TD is associated with characteristic telephonic receiver appearance of femora.

The diagnosis is by antenatal ultrasound examination and confirmed by molecular analysis on aminiocytes (7) Prenatal diagnosis of TD has been well established with ultrasound examination during second trimester. (8)The 3D anatomy scan and molecular confirmation may be helpful in early diagnosis and genetic counseling of this lethal condition.(9)Recently cell free fetal DNA analysis in maternal plasma is a diagnostic tool of TD. (10)

The genetic basis is due to autosomal dominat mutation affecting FGFR3 gene .The different ends of same gene involvement is observed in hypochondroplasia, achondroplasia and thanatotrophic dysplasia.The mutation of gene in TD is severe form. (11)Genetic counseling is important to the parents of affected individuals to prevent the anxiety about the recurrence .The occurrence of this condition is sporadic and recurrence risk is only 2%.

IV. CONCLUSION

Thanotrotrophic dysplasia is a lethal form of skeletal defect featured with shortened limbs, huge head, narrowed chest , hypoplastic lungs with abdominal protuberance. Death of the baby occurs within minutes of birth due to respiratory failure. Both sexes are equally affected with low chances of recurrence. It can be detected by antenatal ultrasound examination in second trimester. The chances of recurrence is less and genetic counseling helps in preventing this autosomal dominant condition.

REFERENCES

[1]. Ldr Sq, Sahu S, Cdr Wg, Kaur P. Thanatophoric Dysplasia: Antenatal Diagnosis. Med J Armed Forces India 2009;65:87-8.
[2]. Martínez-Frías ML, Ramos-Arroyo MA, Salvador J. Thanatophoric dysplasia: An autosomal dominant condition? Am J Med Genet 1988;31:815-20.
[3]. Orioli I, Castilla EE, Barbosa-Neto JG. The birth prevelance rates for the skeletal dysplasias. J Med Genet 1986;23:328-32.
[4]. NeelimaTirumalasetti Case report of Thanatophoric dysplasia: A lethal skeletal dysplasia : 2013 Volume : 2(4) Page : 275-277
[5]. Fink AM, Hingston T, Sampson A, Ng J, Palma-Dias R. Malformation of the fetal brain in thanatophoric dysplasia: US and MRI findings. Pediatr Radiol 2010;40 Suppl 1:S134-7.
[6]. Edeiken J, Rhodes PJ. Thanatophoric dysplasia. In: Roentgen Diagnosis of Diseases of Bone. 2nd ed., Vol. 1. Baltimore: The Williams and Wilkins Company; 1973. p. 75-6.
[7]. Lam AC, Lam YY, Tong TM, et al. Thanatophoric dysplasia type 1 (TD1) without "telephone receivers". HK J Paediatr 2006;11:320-323
[8]. Machado LE, Bonilla-Musoles F, Osborne NG. Thanatophoric dysplasia. Ultrasound Obstet Gynecol 2001. Jul;18(1):85-86
[9]. Wong HS, Kidd A, Zuccollo J, Tuohy J, Strand L, Tait J, et al. A case of thanatophoric dysplasia: the early prenatal 2D and 3D sonographic findings and molecular confirmation of diagnosis. Fetal Diagn Ther 2008;24(1):71-73.
[10]. Chanabasappa V. Chavadi, Nina Mahale1 , Goutham Jasthi, Sonali Prabhu, Swetha Kory2Thanatophoric dysplasia: Antenatal to postmortem Journal of the Scientific Society, Vol 42 / Issue 3 / September-December 2015 pg:210-212
[11]. Cohen MM Jr. Achondroplasia, hypochondroplasia and thanatophoric dysplasia: Clinically related skeletal dysplasias that are also related at the molecular level. Int J Oral Maxillofac Surg 1998;27:451-5.