Thanatophoric Dysplasia Type I with Bilateral corneal opacities: A Case Report

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Abstract— *Thanatophoric dysplasia is the most common form of lethal skeletal dysplasia, diagnosed by its characteristic clinical and radiographic features. It has two types distinguished by radiological findings. Specific ocular features associated with Thanatophoric dysplasia have not known to be reported. A baby with clinical findings of a large head, short stature , low set ears, short and curved limbs, small thorax, bilateral corneal opacities and radiographs showing short and curved long bones, femurs like telephone- receiver look, H- shaped vertebrae, small iliac wings was reported. This rare case was thoroughly examined and investigated which came out to be a case of Thanatophoric Dysplasia Type I with corneal opacities. So when a case of Thanatophoric dysplasia Type 1 with bilateral corneal opacities was attended at pediatric department, a detailed report was prepared to publish such a rare case.*

Keywords: Skeletal Dysplasia, Corneal Opacities.

I. INTRODUCTION

Thanatophoricdysplasia(TD) is the most common lethal chondrodysplasia with a birth prevalence of 1 in 35000 births.[1] It is inherited in Autosomal Dominant manner and is due to heterozygous mutation of genes encoding FGFR 3 (Fibroblast growth factor receptor 3).[2]

Two types of TD have been described.[3,4] The radiologic features in Type 1TD are shortness and bowing of the long bones, femurs curved like telephone-receiver, hypoplasia in pelvic bones, flattening in acetabular skeleton and hypoplasia in vertebral corpus.[5]

In Type 2 TD, there is no shortening and bowing of long bones, femurs being straight and clover – leaf deformity of skull is significant.[6]

Cases of TD Type I have been reported in the past[3,4,5,6] but none have mentioned any specific ocular finding . Here present a case of TD type 1 which had all the characteristic features[6] but differed in having bilateral corneal opacities.

II. METHODOLOGY

A rare case of Thanatophoric dysplasia Type 1 who was having all characteristic featers for its type but different in having bilateral corneal opacities was admitted in NICU, Mahila Chikitsalaya, SMS Medical College, Jaipur (Rajasthan) India. So case study was done thoroughly and case report was prepared to publish this rare case.

III. CASE REPORT

A Preterm (30-32 wks) appropriate for gestational age (AGA) male neonate having birth weight 1.3 kg was born vaginally to a 25 year old primigravida mother. There was no consanguinity, no family history of previous such babies. Mother had short stature but father was normal in height. Pregnancy was uneventful. Mother had regular antenatal checkups. Antenatal ultrasound examination done which

was suggestive of all limb bone shortening suggestive of skeletal dysplasia. Parents knowing all the facts willingly continued the pregnancy.

At birth, baby had a poor cry, was given bag and mask ventilation for 30 seconds, Apgar being 6/10 at 5min. Baby had severe respiratory distress for which was admitted in NICU and started on Continuous Positive Airway Pressure (CPAP), Intravenous fluid (IVF), inotropes and antibiotics.

Postnatal examination showed a large head, low set ears, short and curved limbs, small thorax, micropenis, bilateral corneal opacities. Postnatal bone radiographs showed short and curved long bones, femurs like telephone- receiver look, H- shaped vertebrae, small iliac wings. (Figure 1& 2)

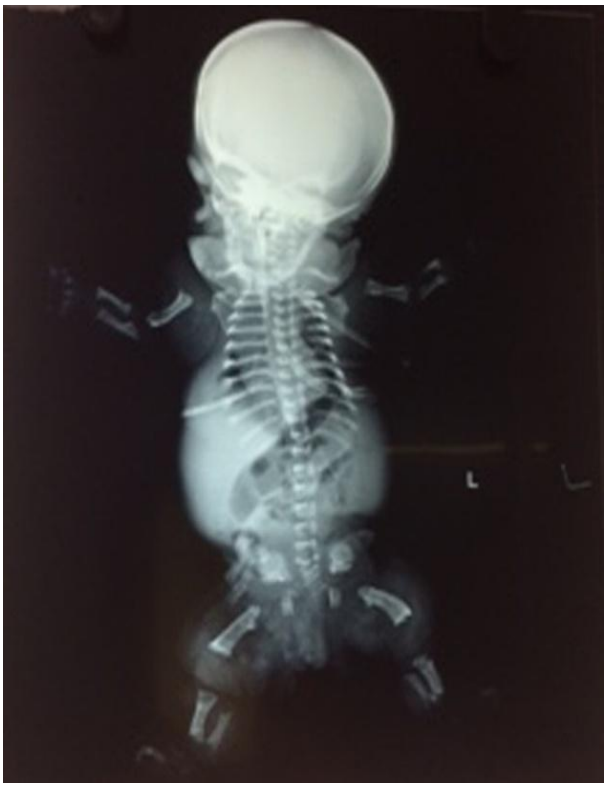
In other routine investigations done, CBS was suggestive of leucopenia and thrombocytopenia. RFTs, LFTs and USG abdomen were normal. USG brain was suggestive of tiny right choroid plexus cyst.

Based on clinical examination and radiographic features, a diagnosis of Thanatophoric Dysplasia Type I was made.

Baby deteriorated on day 4th of life (LD4), was taken on ventilator and other supportive measures but expired on day 5th of life (LD5) due to respiratory failure most probably due to pulmonary hypoplasia. Autopsy was not done in our case as parents did not give consent.

Figure 1
Coarse facies, mid facial hypoplasia, low set ears, short limbs, narrow thorex

Figure 2
Normal skull, flat vertebrae, telephone receiver Femur



IV. DISCUSSION

Thanato phoric dysplasia (TD) is a congenital skeletal dysplasia characterized by marked under development of the skeletal system and short limbeddwarfism.[1] It is the most common form of lethal skeletal dysplasia syndromes.[1,2]

Two types of TD have been described.[3,4] TD Type I is characterized by curved femora(telephone-receiver appearance), very flat vertebral bodies and usually normal skull.[5] While Type II TD shows relatively straight femora and tall vertebral bodies and is associated with a clover – leaf skull.[6]

This case justifies for TD Type I as it had all the characteristic features[3,4,5] like short and curved limbs, short neck, short narrow thorax, mid facial hypoplasia. Skeletal radiographs also added to the picture with very short and bowed tubular bones with flared metaphyses, femurs curved and shaped like a telephone- receiver, vertebrae were thin and flat like H shaped, very short ribs & hypoplasia of pelvic bones. TD type II was ruled out as there were no longer and straighter femurs and no clover- leaf skull deformity.[5,6]

The differential diagnosis includes the other causes of perinatal lethal skeletal dysplasia like Achondrogenesis type 1B, Atelo steogenesis type II, Campomelic dysplasia, Osteogenesis imperfecta type 2, Asphyxiating thoracic dystrophy (Jeune syndrome), homozygous Achondroplasia and Hypophosphatasia but each of these have their characteristic clinical and radiological features.[7,8,9]

In Achondrogenesis type 1 B, radiological features of extreme degree of deficient ossification in vertebral bodies is highly suggestive.

Atelosteogenesis types II have very short limbs, clubfoot, dislocation of elbows and knees. Femurs are hypoplastic with a club- shaped appearance.

Campomelic dysplasias have bowing of long bones, hypoplasia of scapulae and pelvic bone with other systemic anomalies.

Osteogenesis imperfecta type 2 has multiple intrauterine fractures of long bones which have a crumpled appearance on radiographs. Multiple rib fractures create a beaded appearance and sclerae are blue -gray.

Asphyxiating thoracic dystrophy (Jeune syndrome) has postaxial polydactyly, skeletal radiographs show tubular limb bones which are short with bulbous ends & cone shaped epiphyses occur in hand bones.

In Homozygous Achondroplasia, a family history of dwarfism makes differentiation possible. And Hypophosphatasia (congenital) is characterized by moth eaten appearance at the ends of long bones, severe deficiency of ossification throughout the skeleton, and marked shortening of long bones.

The reason for presenting this case is for the associated ocular finding of bilateral corneal opacities.

Case of TD Type I reported in past have described corneal haziness as an ocular finding[4] but there is no mention of corneal opacities. Reason might be that the condition itself is very lethal and such babies hardly survive so such minor features and variations might go unnoticed.

Ophthalmological reference was done in our patient and according to them the reason for corneal pacities could be due to congenital glaucoma which is one of most common causes of congenital corneal haziness.[10]

V. CONCLUSION

A baby having large head, short stature, low set ears, short and curved limbs, short neck, small thorax and bilateral corneal opacities and postnatal radiograph showing short and curved long bones, femurs curved like a telephone- receiver hook, H shaped vertebrae, small iliac wings should be worked up for Thanatophoric dysplasia (TD). As in this case which was investigated radiologicaly and found to be a case of Thanatophoric dysplasia (TD) type I?

CONFLICT OF INTEREST

None declared till now.

REFERENCES

[1] Herman TE . Siegel MJ. Thanatophoric dysplasia, type 1. J. Perinatol. 2012 Jun ; 32(6): 476-8

[2] Yang Y, Liu YN, LiDZ. Rapid detection of common mutations of the FGFR3 gene causing thanatophoric dysplasia type 1 : two case reports. Fetal Pediatr Pathol .2012 Jun ; 31(3) :128-33

[3] M.L. Kulkarni, C. Suresh Kumar, V. Venkataramana, Samuel Koshy, M. Bhagyavathi, G. Shekhar Reddy. Thanatophoric Dysplasia. Indian Pediatrics Vol 31 Nov 1994

[4] Vinayake Hegade P et al. Thanatophoric Dysplasia. Int J Contemp Pediatr.2015 Aug;2(3): 238-42

[5] Chang TK, Wang Y, Liu AM, Tung JC. Thanatophoric dysplasia type 1. Acta Paediatr Taiwan .2001 Jan-Feb;42(1):39-41

[6] Indu PS, Poothiode U, Augustine J, Pillai SB. Thanatophoric dysplasia : a case report and review of literature. Indian J Pathol Microbiol 2007 Jul;50(3):589-92

[7] Lemyre E, Azouz EM, Teebi AS, Glanc P, Chen MF. Bone dysplasia series, Achondroplasia, Hypochondroplasia and Thanatophoric dysplasia : review and update. Can Assoc Radiol J.1999 Jun;50(3): 185-97

[8] Sauer I, Klein B, Leeners B, Cotaralo C, Heyl W Funk A. Lethal osteochondrodysplasias : prenatal and postnatal differential diagnosis. Ultraschall Med. 2000 Jun;21(3):112-21

[9] Kolble N, Sobetzko D, Ersch J, Stallmach T, Eich G, Huch R, Huch A, Superti-Furga A, Wisser J. Diagnosis of skeletal dysplasia by multidisciplinary assessment : a case report of two cases of thanatophoric dysplasia. Ultrasound Obstet Gynecol.2002 Jan;19(1):92-8

[10] W Franks and D Taylor. Congenital Glaucoma-a preventable cause of blindness. Arch of Dis in Childhood,1989,64,649-50.