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A case report of an unusual case of Tibial Hemimelia with syndromic association

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ABSTRACT:
Introduction: Tibial hemimelia is reported to occur in 1 in 1 million cases. It is usually associated with a congenital syndrome. Fibula may be normally formed or absent. It is a very rare entity and the treatment modalities are very aggressive including knee disarticulation and prosthetic fitting.
Case Presentation:A one month old male child presented with deformed head, ears, legs and feet. The x-ray of the patient showed proximal tibial hemimelia, tibial vara and normal fibula on both sides. This case is being presented because of its rarity & association with other anomalies. Because of these associated anomalies this case does not fit into many of the syndromes quoted in literature. The child is still under follow up. Here, we will discuss about a review of literature of Tibial Hemimelia, clinical presentation, radiological appearance, and management of our case
Keywords: Tibial Hemimelia, Syndromic congenital disorder

I. INTRODUCTION:

Tibial hemimelia is a very rare disorder occurring in 1 in 1 million cases. It is defined as complete absence of tibia or its hypoplasia. The fibula may be normally formed or may be deficient [1]. There is only one case report with similar clinical findings which has been reported in the English literature [2].This is the first case report of bilateral variant of Tibial hemimelia micromelia trigonal brachycephaly syndrome. Its association has been seen with many syndromes including Werner's syndrome, CHARGE Syndrome, Tibial Hemimelia Diplopodia Syndrome, Langer–Giedion syndrome, tricho–rhino–phalangeal syndrome (TRPS II), tibial hemimelia diplopodia syndrome, tibial hemimelia and split hand and foot syndrome etc.

II. CASE PRESENTATION:

A one month old male child (Fig.1) presented to us by his parents with bowing and shortening of the legs and deformity of the hands and feet. He is the third child of a 27 year old female and her 32 year old husband born through normal vaginal delivery at term. Birth weight was 2.7 kg and the height was 60 cm with complete immunization done till date. The two elder siblings had no history of such complaints. The child was exclusively breast fed till date. There is no history of any medication during pregnancy. On general physical examination, the child had trigonal brachycephaly (Fig.2a) and microcephaly (Fig.2b) along with low lying and malformed ears. There was grade I microtia and the ear lobule was small with auricular cleft and had 2 small prominent tubercles (Fig.2c). The hearing, however was normal since the child reacted in response to any sound and closed his eyes when loud noise stimulus was given. The anterior fontanelle was palpable and was not closed. In the limbs, there was posterolateral bowing of the legs with bilateral genu vara (Fig.3a). Relative shortening of the limbs was noticed. The child was extending and flexing the knee joint actively during crying which implies he had an active quadriceps mechanism. Equinovarus was noticed at the foot and ankle joint (Fig.3b). There was bilaterally symmetrical (mirror) preaxial polydactyly of hands (Fig.3c) and postaxial polydactyly of the feet with syndactyly (Fig.3d). Milestones were also checked at frequent intervals in which there was no delay which implies there was no neurological defect. The x-ray of lower limbs showed proximal tibial hemimelia. There was deficiency of proximal tibial diaphysis with thickened portion proximally and tibial vara. The proximal tibial physis is absent (Fig.4). The hip joint, femur and fibula were all normal. The skull and spine X-rays were normal (Fig.5,6). The child was haemodynamically stable and afebrile. Haematological investigations showed normal blood count, no raised infective marker and normal vitamin D3 levels. OtoAcoustic Emission (OAE) studies and Brainstem Evoked Response Audiometry (BERA) studios were conducted which came out to be normal. Treatment was aimed symptomatically. We decided to treat the equinovarus deformity with Serial corrective castes at weekly interval using Ponsetti method. The equinovarus deformity of the feet got corrected in three casts and after that, Dennis Browne splint was applied (Fig.7). The child is still under follow up and the further course of action including

corrective osteotomies and soft tissue releases will be done as and when required.

III. DISCUSSION:

The child is a rare case of ?Tibial hemimelia micromelia trigonal brachycephaly syndrome. Based on the classification systems, It belonged to Jones type III and Weber type IV. Due to rarity of combination of anatomical findings present in our case, these cases are very difficult to find in literature [3]. The chances of these syndromes occurring together are very rare about 1 in $\mathbf{10^{12}}$ and we couldn't find any case in the literature which could match the exact findings. Only 1 case could be found which was reported by Hans Rudolf Wiedemann and John M. Opitz in 1983 in which there was unilateral involvement of the lower limb [4] while in our case, bilateral involvement is present. Tibial hemimelia is usually congenital and occurs in about 1 in 1 million patients. It is usually unilateral and is bilateral in about 30% of the cases (mirror). It is usually associated with polydactyly and syndactyly. It can be autosomal dominant or recessive associated with changes on 18q chromosome or sporadic also. It is sporadic in our case since no elder sibling suffers from the same complaints. The tibia may be aplastic or completely absent. The fibula is always present and may be normally formed or dysplastic and, in some cases, even duplicated [5]. The foot also may be normally formed, deformed or duplicated. The movement at ankle joint may range from normal to fixed equinovarus. It is associated with several syndromes including Werner's syndrome, CHARGE Syndrome, Tibial Hemimelia Diplopodia Syndrome, Langer–Giedion syndrome, Tricho–Rhino–Phalangeal syndrome (TRPS II), Tibial Hemimelia Diplopodia Syndrome, Tibial hemimelia and split hand and foot syndrome etc [6]. Other known potential cause is mother assumption of thalidomide during pregnancy. The baby was breast fed in our case and the mother had no history of taking any medication in the past year. The boy had to be treated to provide early ambulatory status at the cheapest cost since the family belonged to lower class status. Due to the less severe nature of the disease with active quadriceps mechanism (the most important prognostic indicator), Our main goal was to screen the child for all congenital defects since syndromic association was found but the child did not have any organic defects.

IV. CONCLUSION:

A one month old child with Tibial Hemimelia Micromelia Trigonal Brachycephaly syndrome was diagnosed on the basis of clinical and radiological findings and careful review of the literature and treated conservatively which gave satisfactory results.

Clinical Message:

Diagnosing the cause of tibial hemimelia can be challenging. Certain congenital syndromes may present in an unusual fashion. Tibial heimelia is rare and a detailed evaluation is essential.

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Figures:

Figure 1: Gross appearance of child

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Figure 2a: Trigonal brachycephaly

Figure 2b: Microcephaly

Figure 2c: Low lying and malformed ears

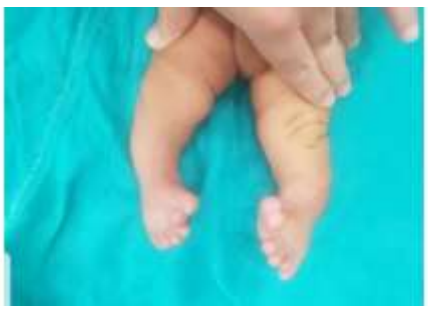

Figure 3a: Posterolateral bowing of the legs

Figure 3b: Equinovarus

Figure 3c: Preaxial polydactyly (mirror) of hands

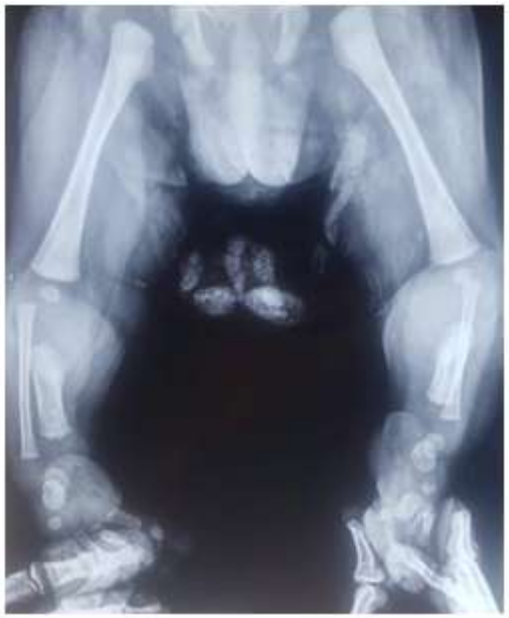

Figure 4: Proximal tibial hemimelia and normal fibula

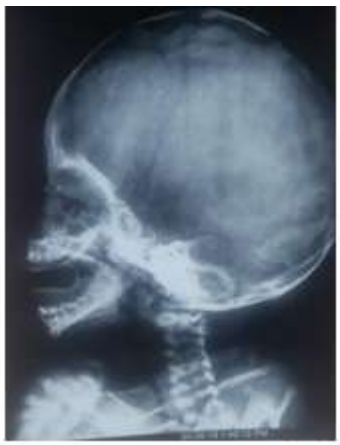

Figure 5: Normal skull Xray

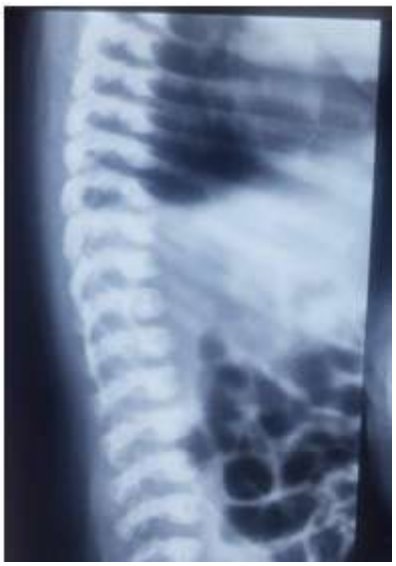

Figure 6: Normal spine Xray

Figure 7: Treatment

Competing Interests:
The authors declare that they have no competing interests.

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