

Tibial Longitudinal Deficiency: Case Report and Literature Review

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Abstract: *Tibial longitudinal deficiency (TLD) is a rare congenital anomaly with an estimated incidence of 1 per 1,000,000 live births. This congenital defect can affect unilateral limb, which is more common, or it can present as bilateral defect of the tibiae. Inter-individual variability in the expression of the phenotype and inheritance with reduced penetrance has been reported. It may occur purely as an isolated anomaly of the tibia or it might associate with various skeletal and extra-skeletal malformations. Many authors have hypothesised the aetiology of TLD. There are four types of classification systems of TLD. Treatment of TLD is based on the functional impairment and type of anomaly present.*

Keywords: *Tibial deficiency, congenital anomaly, tibialhemimelia, tibial aplasia*

1. Introduction

Congenital longitudinal deficiencies of the upper and lower extremities have been reported by many authors in the literature. Tibial longitudinal deficiency (TLD) is considered a rare congenital defect [1]. The incidence of TLD is estimated to be approximately 1 per 1,000,000 live births [1]. Tibial Longitudinal Deficiency (TLD) (also known as long bone deficiency, tibialhemimelia, aplasia, hypoplasia, or dysplasia) is a rare condition and severe lower limb malformation. TLD is considered a rare congenital disease with an estimated incidence of 1 per 1,000,000 live births. This congenital defect can be unilateral or bilateral defect of tibiae. Inter-individual variability in the expression of the phenotypes and is inherited with reduced penetrance have been reported. It may occur purely as an isolated anomaly or be associated with various skeletal and extra-skeletal malformations. Many have hypothesised the aetiology of TLD. There are four types of classification systems of TLD. Treatment of TLD is based on the functional impairment and type of anomaly present.

2. Case Report

A 7-year-old male child, from Burkina Faso, was born at 39 weeks and 4 days to a 32 years old, Para 4+1. The pregnancy was not complicated by any medical illness, neither gestational diabetes mellitus nor hypertensive disorders. The mother is on no prescribed medications, over the counter medications nor a herbal medicines. She denies tobacco, alcohol and illicit drugs intake. The parents are of Africans descent with an evidence of consanguinity. The only event that happens during this pregnancy, i.e. at 20 weeks of gestation, was a 10 days history of fever and productive cough with yellow sputum. She underwent a chest X-rays and a clinical diagnosis of right lower lung lobular pneumonia was given. She

improved with a single course of amoxicillin. There is one abortion which was not investigated. The other three siblings were one healthy girl and two healthy boys.

Upon examination, a male patient looking well, there are no signs of pallor, cyanosis, jaundice or dysmorphic features. There are no signs of respiratory distress. Vital signs were as follow: temperature 36.9 celsius, heart rate 150/min, respiratory rate 40/min, blood pressure 70/50 mmHg. Chest examination revealed clear vesicular bilateral air entry with no added sounds and audible normal heart sounds with no added sounds or murmur. Abdomen is soft lax with no palpable masses. Normal pattern of male genital and descended testes. The musculoskeletal exam reveals the following: bilateral foot acheriria and left hand acheiria.

The plain radiograph reveals that there is a complete absence of bilateral tibiae and presence of both fibulae. The upper leg revealed absence of bilateral femoral bone.

3. Discussion

The original classic classification describing limb deficiency, classic terms include the following: amelia entails absence of a limb. Meromelia entails partial absence of a limb. Hemimelia entails absence of half a limb. Phocomelia entails flipper-like appendage attached to the trunk. Acheiria entails missing hand or foot. Adactyly entails absent metacarpal or metatarsal. Aphalangia entails absent finger or toe.

International Society for Prosthetics and Orthotics (ISPO) Classification System divides all deformities into transverse or longitudinal. Transverse deficiency has no distal remaining portions, whereas the longitudinal deficiency has distal portions. Transverse level is named after the segment beyond which there is no skeletal

portion. Longitudinal deficiencies name the bones that are affected. Any bone not named is present and of normal form [60].

Tibial Longitudinal Deficiency (TLD) (also known as long bone deficiency, tibialhemimelia, aplasia, hypoplasia, or dysplasia) is a rare condition and severe lower limb malformation. This condition is also known as split-hand/foot malformation with long bone deficiency (SHFLD) if it is associated with tibial anomalies. The incidence is estimated to be approximately 1 in 1,000,000 live births [3]. This congenital defect can be unilateral (common) or bilateral. It is a preaxial longitudinal deficiency with variable degrees of absence of the tibia with highly variable expressivity. Therefore, inter-individual variability in the expression of the phenotypes (i.e. clinical manifestations either skeletal or extra-skeletal anomalies) has been reported in the world literatures [29, 28, 30, 28]. It may occur as an isolated anomaly or be associated with various skeletal and extra-skeletal malformations [1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 13, 14, 15, 16, 17, 18, 19, 20, 21, 22]. Several reports indicate that it may also constitute part of a more complicated malformation complex or syndrome.

There are many aetiological hypotheses that have been postulated. These hypotheses are hereditary and familial (genetic) theory, [1, 2] sporadic theory, [27, 28] morphogenic theory (hypothesis), teratogenous theory (hypothesis), intoxication theory (hypothesis), and finally idiopathic [1]. The majority of cases are sporadic. There is an apparent autosomal dominance with reduced penetrance approximately 66% as well as autosomal recessive [2]. There might also be pseudo-dominance mode of inheritance [27].

There are four types of classification systems of TLD that are available in the literature. That is, Jones and Loyd-Roberts (1978) classification is the oldest and widely used [27], Henkel (1978) classification [2], and Kalamachi and Dawe (1985) classification [2], as well as the newest Werber (2008) classification system [2].

Treatment of TLD is based on the functional impairment and type of anomaly present. It traditionally consists of amputating the affected limb (e.g. Syme or Chopart amputations or knee disarticulation) to facilitate use of artificial limbs with or without reconstruction of the ankle and correction of leg-length inequality [30,31]. Historically, either knee disarticulation or (modified) Brown's procedure (fibular centralization) are performed if adequate quadriceps function [31]. In some cases, tibiofibular synostosis may be performed later. Other proposed surgical options is fibular transfer [32]. It is advocated by many authors to treat these patients on an individual basis [33].

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