ISSN: 2576-2842

Case Report

Journal of Gynecology & Reproductive Medicine

Prospective Prenatal Diagnosis of Congenital Anterolateral Tibial Bowing with İpsilateral Hallux Duplication: Case Report

Nejla Gultekin

Department of Obstetrics and Gynaecology

*Corresponding author
Nejla Gultekin, Department of Obstetrics and Gynecology, Mersin City Hospital, Turkey, Tel: 5327995516, Email: nejlagultekin@yahoo.com

Submitted: 08 Nov 2018; **Accepted**: 30 Dec 2018; **Published**: 23 Jan 2019

Abstract

The precence of any skeletal deformity can be a patognomonic feature of a syndrome. Tibial bowing is defined as bowing of tibial diaphysis, some degree of angulation is physiologic in neonates, more severe forms are frequently associated with various syndromes . The congenital anterolateral tibial bowing and polydactyly is a rare condition. Posteromedial and anteromedial bowing may resolve in time spontaneusly . Anterolateral bowing should be followed up due to the possibility of development of pseudarthrosis of the tibia and risk of fracture. Congenital anterolateral bowing of the tibia in association with a bifid ipsilateral great toe is a much more rare entity.

Background

The precence of any skeletal deformity can be a patognomonic feature of a syndrome. However some skeletal deformities can be misleading and cause unnecessary investigations. Which in turn can cause parental anxiety. Tibial bowing is defined as bowing of tibial diaphysis . Although some degree of angulation is physiologic in neonates, more severe forms are frequently associated with various syndromes including skeletal dysplasia and neurofibromatosis [1,2]. Congenital bowing of the long bones can be feature of the campomelic dysplasia, achondroplasia metabolic bone disease as well as neurofibromatosis (NF) [2-4]. In NF, the café-au-lait macules should be observed only congenital bowing of long bones, should be diagnosed for neurofibramatosis type 1 (NF-1) and the symetrical skeletal involvement should observed in the metabolic bone disease [4,5].

Prenatal recognition of tibial bowing can enable timely investigation and counseling of parents. However as for most skeletal dysplasias, a straightforward diagnosis is usually not possible and meticulous prenatal investigation for additional sonographic clues in combination with genetic testing is necessary. The congenital anterolateral tibial bowing and polydactyly is a rare condition and can be readily distinguishable from the NF-1 by the abcense of neurocuteaneous signs, and from camptomelic dysplasia by involvement of unilateral extremities and hallux duplication [5].

Pathological congenital bowing of the tibia in the newborn is a rare condition. Posteromedial and anteromedial bowing may resolve in time spontaneusly whereas anterolateral bowing should be followed up due to the possibility of development of pseudarthrosis of the tibia and risk of fracture [6]. In contrast to other types of tibial bowing, anterolateral bowing in association with congenital tibial pseudarthrosis is rarely diagnosed at birth and usually develops during the first decade of life [7]. Congenital anterolateral bowing of the tibia in association with a bifid ipsilateral great toe is a much more rare entity which is not related with neurofibromatosis and/or pseudoarthrosis of the tibia and fibula is mainly not affected. Surgical intervention may be needed for hallux duplication and leg length discreapencies [8-10].

34 years old gravida1 parite 0 pregnant woman visited our obstetrics clinic for routine midtrimester anatomy scan at 20 weeks of pregnancy. At that time unilateral congenital bowing of tibia was detected (Figure 1) but we initially failed to detect ipsilateral hallux duplication. No additional abnormalities were evident on prenatal sonographic evaluation.

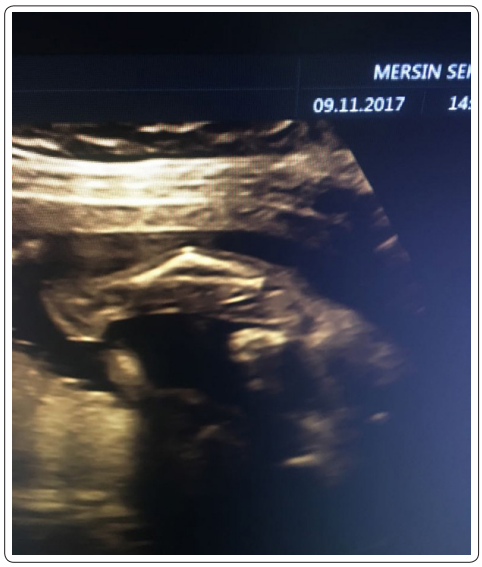

Figure 1: The ultrasound image of the tibial bowing at the 20 weeks of gestation

Amniocentesis and a whole exome sequencing was performed which returned negative results for NF-1 and campomelic dysplasia gene mutation. Despite negative results parents were counselled for a possible skeletal dysplasia. During antenatal follow up ipsilateral hallux duplication was detected. Based on this new finding the presumptive diagnosis was congenital anterolateral tibial bowing with ipsilateral hallux duplication. Antenatal follow-up was uneventful. At 38th gestational week labor ensued and the patient was delivered via cesarean due to positive contraction stres test (Figure 2). A 3600 gram male baby was delivered. Postnatal features was consistent with congenital anterolateral tibial bowing with ipsilateral hallux duplication (Figure 3).

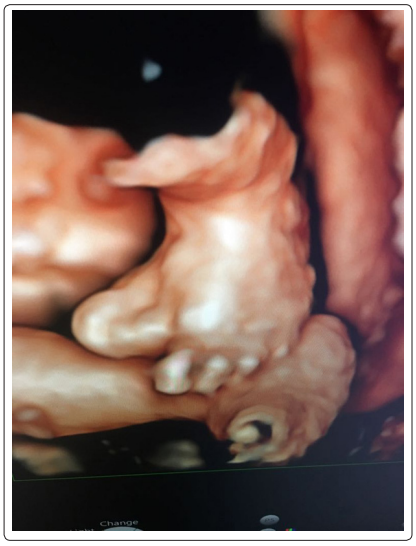

Figure 2: Hallux duplication detected at 28 weeks of gestation

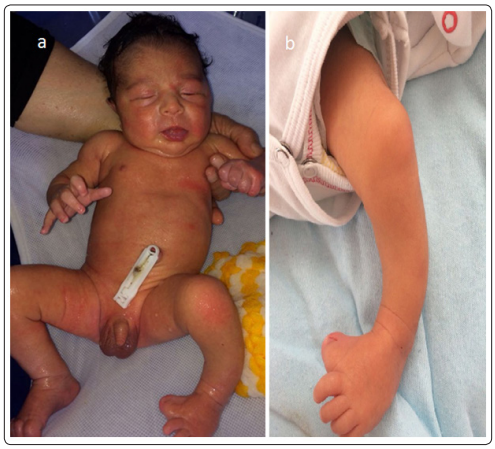

Figure 3: Image of otherwise normal neonate (a) with tibial bowing and ipsilateral hallux duplication (b)

Discussion

From a perinatal perspective congenital anterolateral tibia bowing with ipsilateral polidactyly can be considered as a benign disease with minimal implications on the offspring. Prenatal recognition of this condition is important to avoid unnecessary interventions as well as parental anxiety. A handful of cases have been reported so far and in all cases there were tibial bowing associated with duplication of hallux and normal fibula. In the present case initially neurofibramatosis (NF) and osteogenesis imperfecta (OI) was considered since polydactyly was missed in initial scans.

The neurofibramatosis type 1 (NF-1) is a relatively common genetic disease in general population [1]. Although the anterolateral tibial bowing is not an unusual feature to consider the neurofibramatosis type 1, it is not patognomonic for this condition [1,2]. Diagnostic criteria for NF-1 were established by a panel of experts at a National Institues of Health Consensus Development Conferance in 1987 [11]. The usual clinical presentation in infancy or early childhood is anterolateral bowing of the tibia [11]. Congenital anterolateral tibia bowing with ipsilateral polidactyly should be emphasized in the differential diagnosis of NF1. Other differential diagnosis to be considered are in case of tibial bowing are osteogenesis imperfecta which presents with associated fractures and hypomineralization of bones, skeletal dysplasias such as campomelic dysplasia which has associated features such as micrognathia, ambigious genitalia, achondroplasia and thanatophoric dysplasia. Besides congenital anterolateral bowing of the tibia in association with a bifid ipsilateral great toe is a much more rare benign condition, the surgical correction is needed for hallux duplication and leg length discreapencies [8-10].

Conclusion

In conclusion, congenital anterolateral tibia bowing with ipsilateral polidactyly is a beging disease without any systemic involvement and the prognosis is good with minimal morbidity which should be considered when tibial bowing is observed prenatally. In differential diagnosis, the consideration of neurofibromatosis and osteogenesis imperfecta and skeletal dysplasia disorders should be considered. The clues should be searched such as hypominerilization and fractures for osteogenesis imperfecta and micrognathia , amigious genitalia, achondroplasia and thanatophoric skeletal dysplasia for campomelic dysplasia and polydactily , ipsilateral hallux duplication for congenital anterolateral tibial bowing during ultarsound examination in perinatally. Because of stressfull condition for parents and incomplete phenotypic evalution, the congenital anterolateral bowing with ipsilateral polidactily must be in differential diagnosis in skeletal dysplasia cause of benign disease the spontaneusly resolution of posteromedial and anteromedial bowing will lead to unnecessary diagnostic tests.

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