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Case Report

A rare case of limb deficiency syndrome: Gollop WolfGang syndrome

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ABSTRACT

Limb deficiency disorders are rare, etiologically heterogeneous skeletal dysplasias that occur as an isolated anomaly or as a part of a syndrome. The term limb deficiency incorporates both absence and size reduction of any of the 120 human limb bones, with around 205 identified abnormalities. Congenital absence of tibia is a rare and severe lower limb malformation with an incidence of approximately 1:1,000,000 live births. Absence of tibia with ectrodactyly (lobster claw deformity) or tibial hemimelia with split hand/foot malformation (TH-SHFM) or Gollop-Wolfgang complex is a rarer malformation with highly variable manifestations.

Introduction

Ectrodactyly is also known as split hand or foot malformation (SHFM), or Karsch Neugebauer syndrome or EEC (Ectrodactyly-ectodermal Dysplasia-clefting) syndrome [2]. First case of ectrodactyly was reported in 1936 [[1,2]] and first antenatal diagnosis of ectrodactyly was reported in 1980 [3]. Ectrodactyly develops due to failure of apical epidermal ridge to produce molecules that signals nearby cells to differentiate into digital rays. Pattern of inheritance for ectrodactyly may be autosomal dominant, autosomal recessive or X-linked [4]. Seven different genetic mutations are known to be associated with SHFM namely SHFM1 to 6 and SHFM/SHFLD [4]. Two forms of ectrodactyly occur, one with isolated involvement of limbs. It is known as the nonsyndromic form, which follows pattern of inheritance of a regular autosomal dominant gene with high penetrance. The second form is the syndromic form with associated anomalies such as tibial aplasia, mental retardation, ectodermal craniofacial findings and orofacial clefting and deafness [5].

Blauth and Borisch classified ectrodactyly into 6 groups based on the number of metatarsal bones: Type I have 5 normal metatarsals; Type II have 5 metatarsals, which were partially hypoplastic; Type III have 4 metatarsals; Type IV have 3 metatarsals; and Type V have 2 metatarsals. Type VI represents the monodactylous cleft foot. Two additional groups include: cleft feet with central polydactyly (polydactylous type) and monodactylous feet with lower-leg diastasis or tibial aplasia or both (the diastatic type) [6].

Case report

A case of a near term male baby (4-day-old) of a 22-year-old primigravida mother with limb deficiency. The second trimester scan was not done and in late third trimester scan

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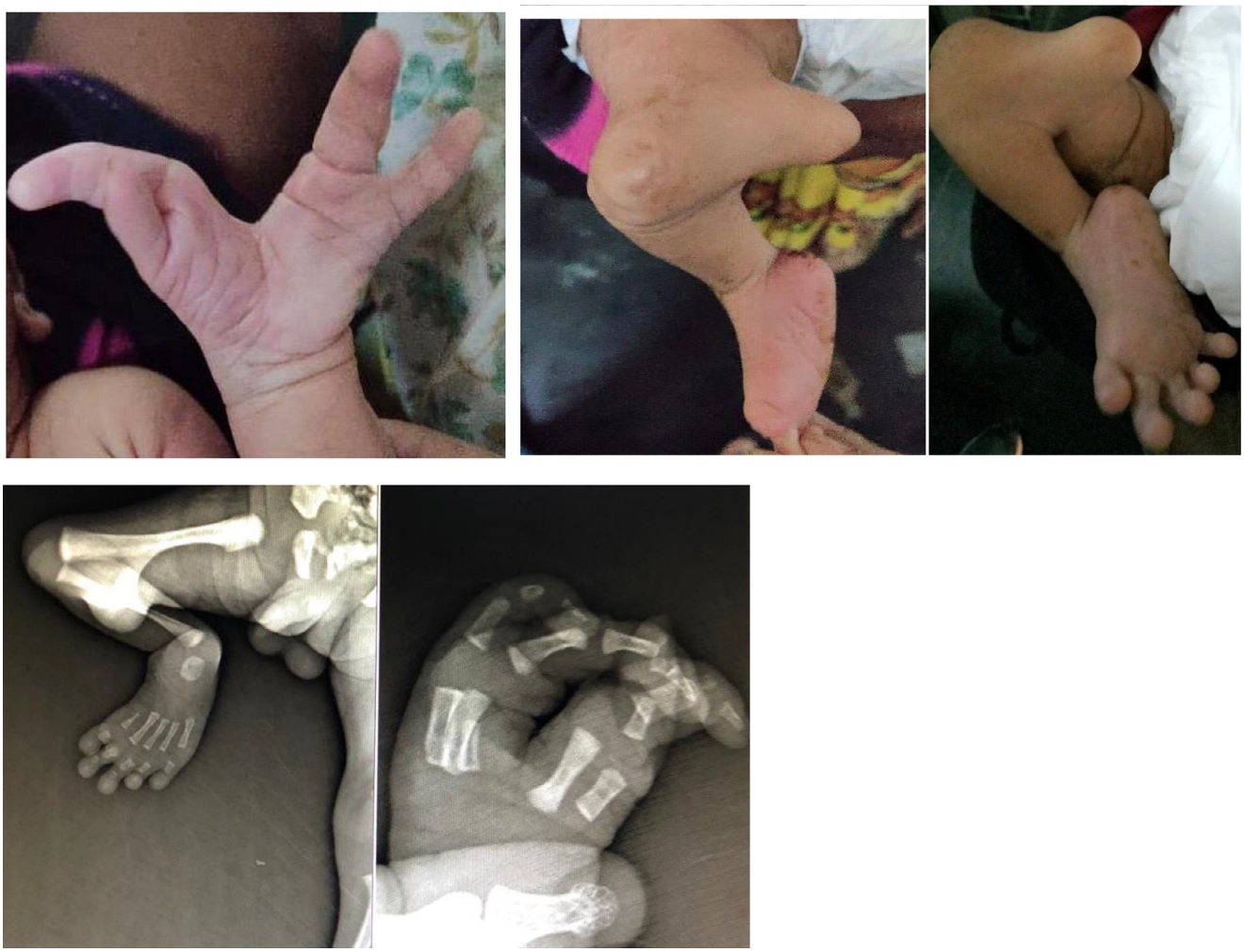

done elsewhere,the absence of limb was missed. X-rays of both upper and lower extremities with spine AP and lateral views taken.Right lower leg showed tibial hemimelia and bifid femur and left hand showed cleft hand(Lobster hand). Both lower leg showed club feet. Spine was unremarkable. Ultrasonography of abdomen and echocardiography were normal. There was no history of similar malformation in other family members.

Discussion

Skeletal dysplasia, affecting around 2 million people worldwide is a heterogeneous group of disorders, characterized by abnormalities of cartilage and bone growth. Two main groups: osteochondrodysplasias and dysostoses . In osteochondrodysplasias generalized abnormality in bone or cartilage. Dysostoses refers to malformations or absence of individual bones single or in combination.

Despite advances there is prenatal diagnosis of specific skeletal dysplasia remains difficult, with reporting an accurate prenatal diagnosis is less than 0.3% [7,8]. Technical difficulties in ultrasound owing to an inconvenient fetal position, poor liquor, maternal obesity and late gestational age at examination.

Review of literature

Ectrodactyly occurs as an isolated malformation or may coexist with other congenital anomalies. It has been shown to be associated with cleft lip or cleft palate, ectodermal dysplasia or developmental delay. In addition, it also occurs as a part of various syndromes such as EEC syndrome (ectrodactyly-ectodermal dysplasia-cleft lip/palate syndrome), EEC syndrome without cleft lip/palate, LADD syndrome (lacrimo-auriculo-dento-digital syndrome), ADULT syndrome (acrodermato-ungual-lacrimal-tooth syndrome), CHARGE syndrome (coloboma of the eye, heart defects, atresia of the nasal choanae, retardation of growth and/or development, genital and/or urinary abnormalities, and ear abnormalities and deafness) or VACTERL syndrome (vertebral defects, anal atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb abnormalities)[9].

A case of a 26-year-old healthy pregnant woman was reported where antenatal ultrasonography at 22 weeks period

of gestation detected V-shaped cleft in both feet of the fetus. Her first child also had median cleft in both feet [10].

A similar co-existence of SHFM in siblings has been reported by Ashi M et al [11]. Lévy J et al, reported a case of triplet pregnancy in a 31-year-old healthy primigravida where one fetus was detected to have ectrodactyly and radial agenesis on ultrasonography at 10 weeks' gestation. The abnormal fetus was selected for fetal reduction [12].

A retrospective review of sonographic studies was done at 2 fetal care centers in United States of America and China from 2002 to 2012. Ten pregnant women were identified with ultrasonography showing abnormalities in the fetus consistent with SHFM. The gestational age of the fetuses ranged from 15 to 29 weeks. Seven pregnancies were electively terminated due to the malformations, whereas 3 women had delivery at term [13].

Conclusion

Our case shows failure of antenatal detection of limb deformity making an important note to look for its presence, especially in mother with known deformity or with similar family history because if diagnosed early, medical termination of pregnancy can help in preventing the birth of babies afflicted with such debilitating malformation. When this anomaly is detected, prenatal counseling should be offered and genetic screening should be performed for all couples for present and possible future pregnancies. The disease-causing mutation must be identified in an affected relative. Pre-implantation genetic diagnosis should be offered so that only embryos that do not carry the disease-causing mutation are chosen for implantation.

Patient consent

Patient consent has been obtained

Declarations of competing interest

None.

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