Gollop-Wolfgang Complex - A Rare Limb Deficiency Syndrome: Case Report And Review Of Literature

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Citation

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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 complex1 is a rarer malformation with highly variable manifestations. The first case with this pattern of malformations was reported by Sir Ambroise Pare way back in 1575.2 The full blown syndrome consists of aplasia of tibia and split hand/ split foot deformity.3

CASE REPORT

A 25 year old primigravida was referred at 36 weeks gestation for intrauterine growth restriction (IUGR). IUGR was confirmed by ultrasound and Doppler studies. She was induced with prostaglandin E2 gel. In view of nonreassuring fetal heart rate she was taken up for emergency cesarean delivery. A term small for gestational age (SGA), male baby, weighing 1990 grams was born with multiple congenital abnormalities – right hand ectrodactyly (Fig: 1a, 1b), right foot ectrodactyly with absent talus (Fig: 2), right tibial aplasia, right short femur with posterior subluxated knee and penoscrotal hypospadiasis (Fig: 3). The baby cried immediately, was active with APGAR score 8 and 10 at 1 and 5 minutes respectively. X-ray confirmed skeletal abnormalities (Fig: 4a, 4b). This was a sporadic case in absence of family history of malformations. There was no internal organ anomaly on ultrasonographic evaluation, echocardiogram was also normal, and karyotyping was 46, XY.

Figure 1

Figure 1a

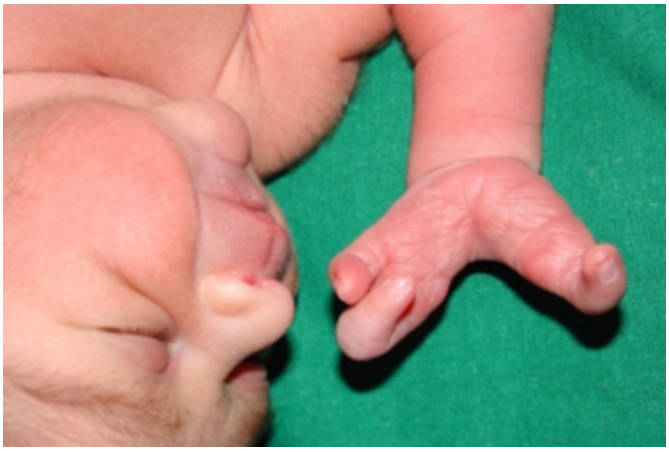


Figure 2

Figure 1b

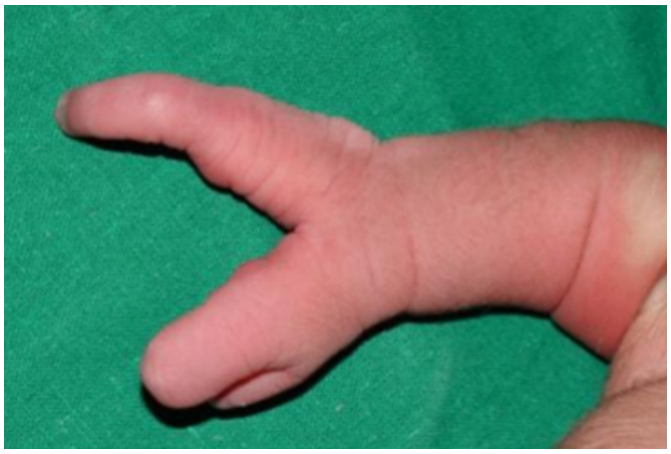


Figure 3

Figure 2

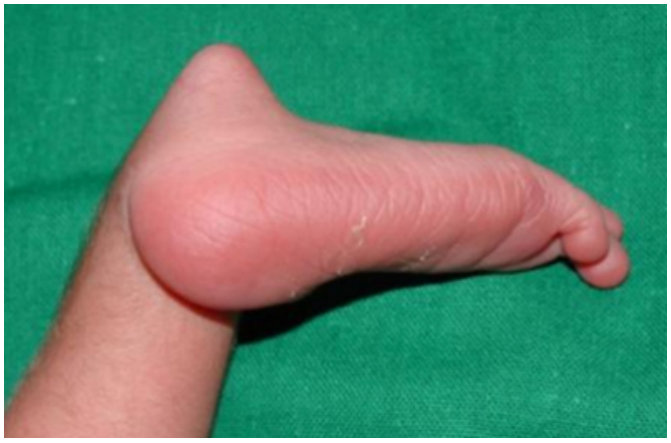


Figure 4

Figure 3

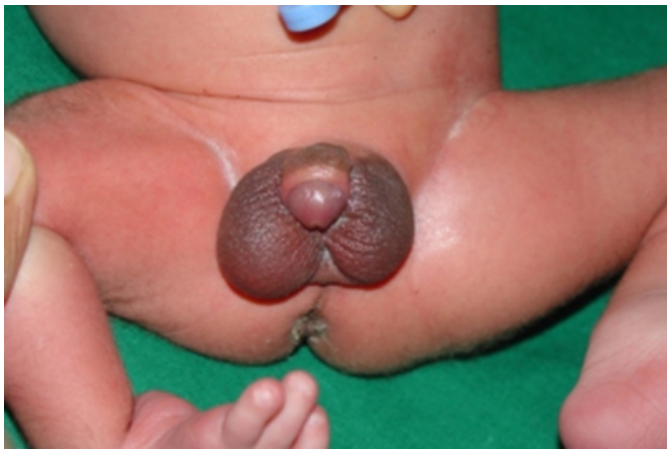


Figure 5

Figure 4a

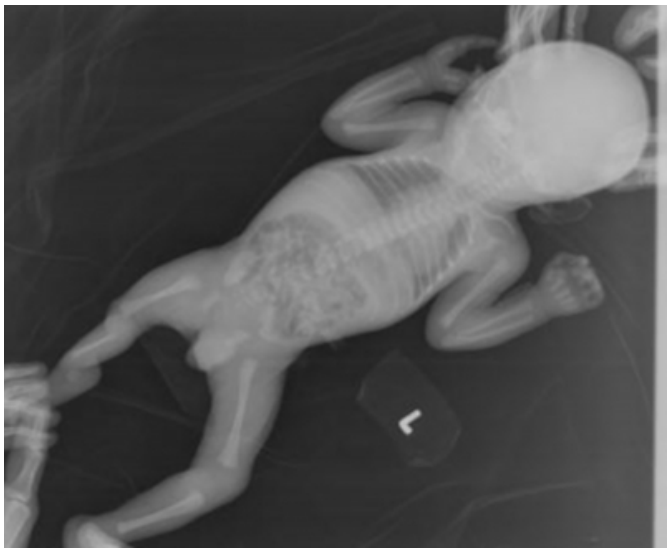
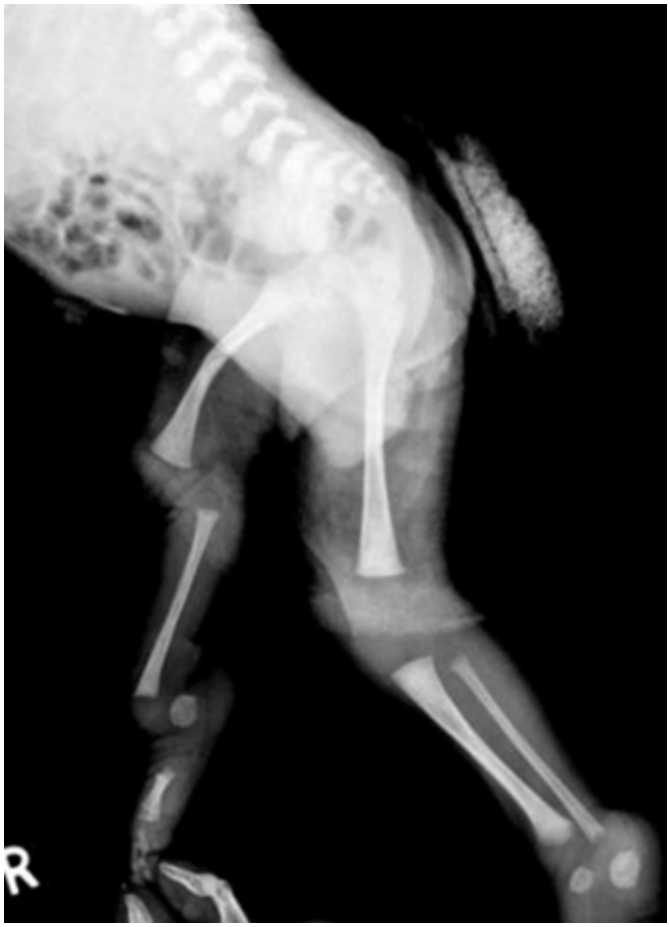


Figure 6

Figure 4b



DISCUSSION

The human skeleton is a complex organ system comprising of 206 bones of different shapes and sizes. Its name is

derived from the Greek ‘skeletos' meaning dried up.

Skeletal dysplasia, affecting around 4 million people worldwide is a heterogeneous group of more than 200 disorders, characterized by abnormalities of cartilage and bone growth - resulting in abnormal shape and size of the skeleton and disproportion of the bones. A cumulative international incidence of at least 1:5000 newborns has been estimated.[4,5,6,7]

Over the past 50 years the classification of skeletal dysplasia has evolved from purely clinical-pathological descriptions to a complex nosology that reflects many of their underlying molecular etiology. For simplification, it can be broadly classified into two main groups: osteochondrodysplasias and dysostoses. In osteochondrodysplasias, there is generalized abnormality in bone or cartilage. Dysostoses refers to malformations or absence of individual bones singly or in combination. These are mostly static and the malformations occur during blastogenesis (1st 8 weeks of embryonic life). This is in contrast to osteochondrodysplasias, which often present after this stage, has a more generalized skeletal involvement and continues to evolve as a result of active gene involvement throughout life.[8]

Making a diagnosis of skeletal dysplasia can be extremely easy on clinical grounds but it may be very difficult and easy to miss on routine anomaly scans done in the early second trimester. Despite the advances in the prenatal ultrasonography, diagnosis of specific skeletal dysplasia remains difficult, with the largest study reporting an accurate prenatal diagnosis by the referring physicians in less than one third of cases.[9,10] Technical difficulties in performing the ultrasound owing to an inconvenient fetal position, decreased amount of liquor, maternal obesity and late gestational age at examination may interfere with an accurate diagnosis.

Although there are reports of prenatal diagnosis of skeletal dysplasia as early as the end of first trimester[11], diagnosis is usually made during the second and third trimester.[12,13] Now more cases are likely to be detected prenatally because the new American Institute of Ultrasound in Medicine (AIUM) performance guidelines for second trimester obstetric sonography include views of all extremities.[14] According to the guidelines, at the 18–23-week scan, the three segments of each extremity should be visualized, though it is necessary to measure only the length of one femur. The first clue to most skeletal dysplasias is the identification of a short femur.[15]

With the use of high resolution ultrasound, definitely there has been an increased awareness of exceptional, but detectable skeletal malformation syndromes. However an accurate diagnosis is essential for genetic counseling, for pediatric orthopedic counseling and management of the pregnancy.[16,17] So in high risk cases or if the abnormality is suspected in routine scan it is imperative to proceed with 3D ultrasound or even fetal MRI, as some of the skeletal dysplasias are lethal, so that the decision to terminate pregnancy can be taken at the earliest.

Tibial aplasia-ectrodactyly syndrome is one of the most severe defects involving the extremities. The mode of inheritance appears to be autosomal dominant with reduced penetrance.[18] As it is a non lethal condition, compatible with near normal life and a fair prognosis with orthopedic corrective measures, even after prenatal diagnosis, standard obstetrical management is not altered.

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