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CASE REPORT

Atypical Foetal Limb Dysplasia: An Anatomical Insight into the Uncommon Lower Limb Congenital Defects

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ABSTRACT

The global birth prevalence of congenital malformations is about 2-3%. Congenital heart defects are the most common followed by the limb anomalies. Congenital limb deficiencies have many causes and often occur as a component of various congenital syndromes. During routine examination of a foetus of 18 weeks gestation obtained for foetal repository in the department of anatomy, some atypical limb anomalies were noted on gross examination which were further evaluated by detailed dissection. This case was identified to be a case of unilateral fibular hemimelia with tibial bone spur and oligodactyly in the same limb. The knowledge of such atypical anomalies is of tremendous clinical relevance for radiologists, obstreticians, paediatricians, paediatric surgeons and orthopaedic surgeons who need to be aware about the presence of such defects at this gestational age for its timely detection, appropriate counselling of parents and timely intervention for surgical planning.

KEYWORDS

• Fibular hemimelia • Oligodactyly • Embryology • Congenital anomalies

INTRODUCTION

Congenital malformations are single or multiple defects of the morphogenesis of organs or body parts, identifiable at birth or during the intrauterine life. The global birth prevalence of congenital malformations is about 2-3%. Most of the congenital defects are due to primary intrauterine growth inhibition, or disruptions secondary to intrauterine destruction of normal embryonic tissues. The upper extremities are more commonly affected while the anomalies of lower limb are less common, non-fatal and are the leading causes of deformities and disabilities amongst the general population.[1]

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Congenital limb deficiencies have many causes and often occur as a component of various congenital syndromes. Teratogenic agents (eg, thalidomide, vitamin A) are known causes of hypoplastic/absent limbs. The limb abnormalities could be because of failure of formation(transverse or longitudinal), failure of differentiation, polydactyly, overgrowth, undergrowth, amniotic band syndrome and generalized skeletal syndromes.[2,3] One or more limbs may be affected, and the type of defect may be different in each limb.[4] The present report describes the various lower limb congenital malformations along with its possible embryonic basis in a foetus observed during foetal autopsy done at a central Government Medical College.

CASE REPORT

The observations were made in the Department of Anatomy, Lady Hardinge Medical College, New Delhi on a foetus of approximately 18 weeks gestational age. The foetus was obtained from the department of Gynaecology and Obstetrics of Smt. Sucheta Kriplani hospital, New Delhi for the foetal repository of Department of Anatomy. The autopsy was conducted on this foetus with gross atypical left lower limb malformation to study the various other congenital anomalies associated with it. The biparietal diameter, abdominal circumference, crown-rump length and foot length were also measured.

On examination, the Crown-rump length of the foetus was measured as 15.6 cm, foot length 3.4 cm, abdominal circumference as 12.9 cm and biparietal diameter as 4.8 cm. Some unusual features were noted grossly in the foetus in the unilateral lower limb. On gross examination, the left leg was grossly shortened and a bony spur arising from the tibia was seen 1cm below the knee joint. The detailed dissection of the leg revealed the absence of fibula depicting fibular hemimelia. On dissection of the left foot, the fourth and fifth digital rays were observed to be absent.

The first, second and third metatarsals were present in their normal anatomical positions. Grossly, the right leg and foot were normal and detailed dissection revealed that both tibia and fibula bones along with all five digital rays were present in normal anatomical positions. On foetal autopsy, the chest and abdomen were dissected and all chest viscera and abdominal organs were normal in development and no other congenital anomaly was observed in the foetus. This case presentation represents unilateral hemimelia with oligodactyly without any other syndromic features.

DISCUSSION

The congenital malformations observed in this case are uncommonly detected on gross examination and detailed dissection of the foetus. Similar findings have been reported earlier in lower limb but either on the right side or bilaterally, rarely it is seen on the left side.[5,6] Such cases may be associated with other congenital disorders like Adams-Oliver syndrome (aplasia cutis congenita with partial aplasia of the skull bones and terminal transverse limb malformations), Holt-Oram syndrome, VACTERL syndrome (vertebral anomalies, Anal atresia, cardiac malformations, tracheoesophageal fistula, renal anomalies and radial aplasia, and limb anomalies) etc.[7,8]

Also, fibular hemimelia may be associated with focal femoral dysplasia.[9] In some cases, fibular Aplasia, Tibial Campomelia(bending of tibial bone) and Oligo-Syndactyly are seen together in FATCO syndrome.[10] In the present case, no such associated anomalies or syndromic features were identified.

Embryological basis: Exposure to teratogens (e.g., thalidomide, vitamin A excess) during 5th-7th week can interfere with limb bud development.[8] The fibula and lateral toes develop from the posterior (caudal) part of the limb bud which is regulated by the Zone of Polarizing Activity (ZPA), which secretes Sonic Hedgehog (SHH). Reduced or absent SHH signaling can lead to Fibular aplasia or hypoplasia, oligodactyly, especially affecting the 4th and 5th toes.[11,12] Also, the fibula and lateral digits arise from mesenchymal condensations in the lateral plate mesoderm. The disruption in mesenchymal proliferation or migration can result in failure to form the fibular anlage and absence or underdevelopment of lateral digital rays.[13]

CONCLUSION

The knowledge of such defects is also of importance to the radiologists who can detect them prenatally. It also has tremendous clinical relevance for radiologists, obstreticians, paediatricians, paediatric surgeons and

orthopaedic surgeons who need to be aware about the presence of such defects at this gestational age for its timely detection, appropriate counselling of parents and timely intervention for surgical planning.

Figure 1: Showing measurement of Crown-Rump length as 15.6cm

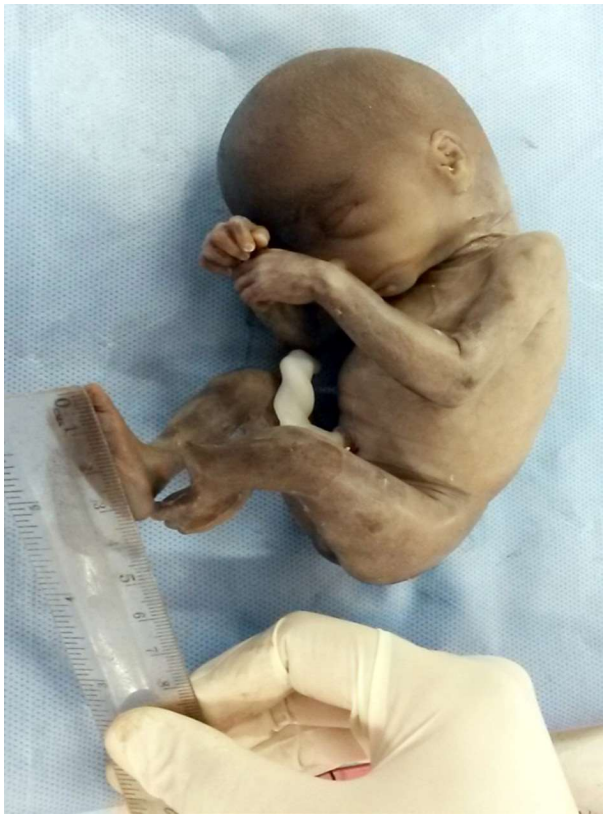

Figure 2: Showing measurement of foot length as 3.4cm

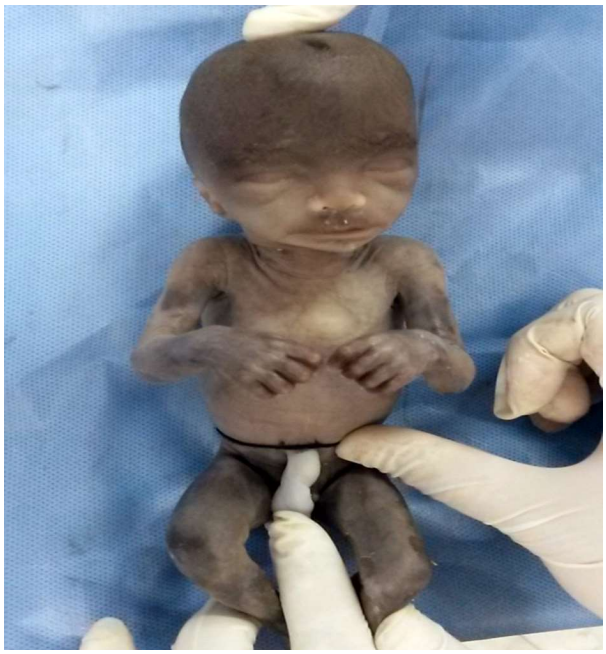

Figure 3: Showing measurement of abdominal circumference as 12.9cm

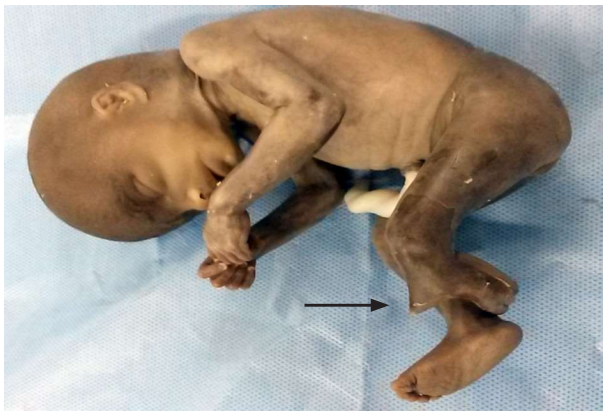

Figure 4: Showing bony spur arising from the left knee

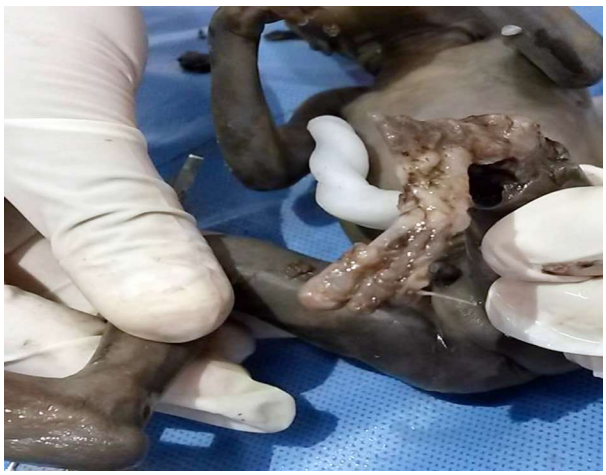

Figure 5: Showing dissected left leg and foot with bony spur arising from left tibia near the knee joint with absence of Fibula and absent 4th and 5th digital rays

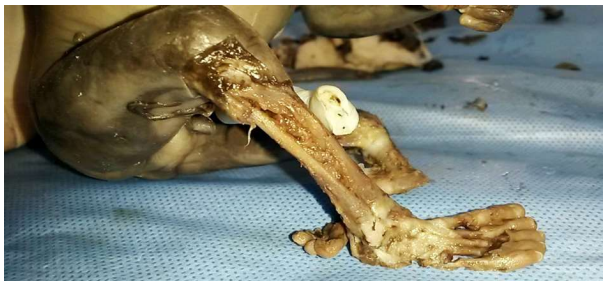

Figure 6: Showing dissected right leg and foot with normal anatomical structures

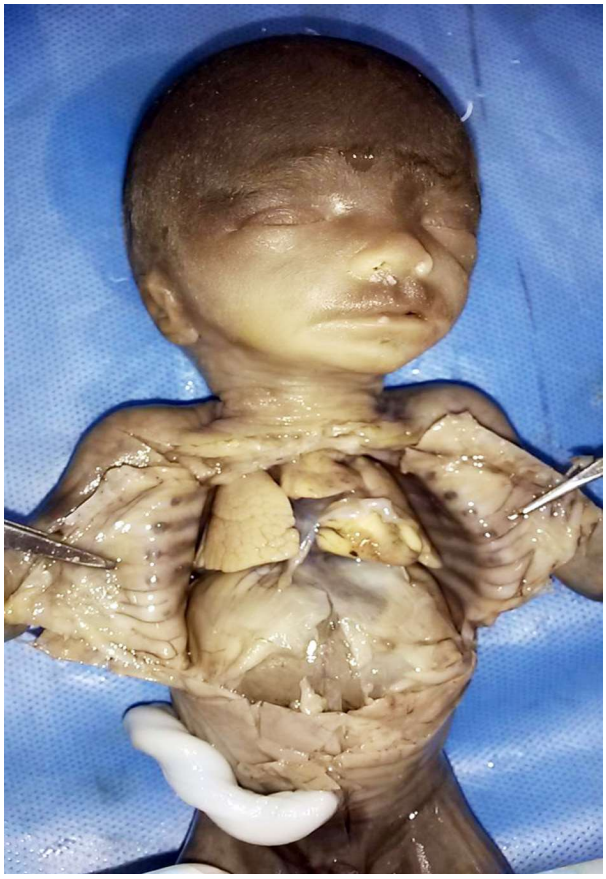

Figure 7: Showing normal viscera in the foetus

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