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A rare case of isolated congenital bilateral femoral agenesis: Antenatal diagnosis and postnatal follow-up

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Abstract
Congenital absence of bilateral femur is a rare congenital anomaly and usually there is a strong association with other skeletal conditions and maternal diabetes. We present an unusual case of isolated congenital bilateral femoral agenesis in non-diabetic female. A 28 years old multigravida presented for level III obstetric scan. USG demonstrated single live fetus of EGA~ 32 weeks 2 days with oligohydramnios and non-accessibility of bilateral femur. Post delivery clinical examination showed shortening of bilateral lower limb and plain x-ray confirmed absence of bilateral femur. We report this case because it does not meet the existing classification criteria for congenital femur deficiency. Proper evaluation and management of this case can help the patient and the parents to accept the condition and be able to live a good social and economically productive life.

Keywords: bilateral lower limb, congenital, femoral agenesis, PFFD, USG

Introduction
Fetal growth and development of lower extremities begins with a genetic template followed by a cascade of various cell signaling factors which occur in the embryonic period and later continue to develop in fetal and childhood along with various environmental and biological factors. Embryogenesis of the extremities occurs between 4 and 8 weeks after fertilization. Most limb deficiencies occur in this period of time, especially during rapid proliferation and differentiation of cells and tissues, any insult during this phase can lead to abnormal lower limb development peak during the 5th and 6th weeks after fertilization [1, 2]. Congenital defects in femur can vary from simple hypoplasia to its complete absence. Bilateral femoral agenesis is a rare congenital disorder with the incidence 1 case per 200,000 population [3]. The clinical distinction between the various types of the femoral defect is important as a guide to the prognosis of limb development [4]. We report this case because it is rare and needs proper evaluation and management.

Case Report
A 28-year-old multigravida patient was referred for a level III scan at 35 weeks of gestation. There was no history of consanguinity in the parents. No congenital abnormality was detected in the previous pregnancy of the patient or in any of the family members before. Patient had history of hypothyroidism for which she was under medication consuming oral levothyroxine 25 mcg per day. The patient denied any exposure to radiation, intrauterine infections or any drug intake. She had taken iron, folic acid and calcium supplements as prescribed.
Ultrasonography was done which showed a single, live, intrauterine fetus corresponding to 32 weeks 2 days of gestational age with normal cardiac activity and oligohydramnios (AFI~ 5 cm). Biometric of the fetus showed biparietal diameter ~ 32 weeks 1 day, head circumference ~32 weeks 1day, abdominal circumference ~ 33 weeks 6 days and Humerus length was ~31 weeks and 2 days. On USG there was non-visualization of B/L femurs and tibia appeared to be articulated with pelvis / acetabulum. No other congenital malformation was seen in USG. Depending upon the biometry average gestational age was 32 weeks 2 days.
The baby boy was delivered by lower segment cesarean section due to oligohydramnios at 38 weeks of gestation. APGAR scores at 1 and 5 min were 7 and 9 respectively. Physical examination after delivery showed bilateral shortened lower limbs. He also had retrognathia, low set ears and overlapping toes. There was absence of both thigh and knee joint but both the feet were normal (Figure 1). The height of the baby was 36 cm (<1st percentile), weight was 2300 (<3rd percentile), OFC was 32 cm. We did not observe any other anomalies in other skeletal organs. The diagnosis was confirmed by a skeletal radiograph which showed absent bilateral femoral bones. (Figure 2). Microarray, karyotyping, whole- exome sequencing (WES) could not be performed due to unavailability.

Fig 1: clinical appearance

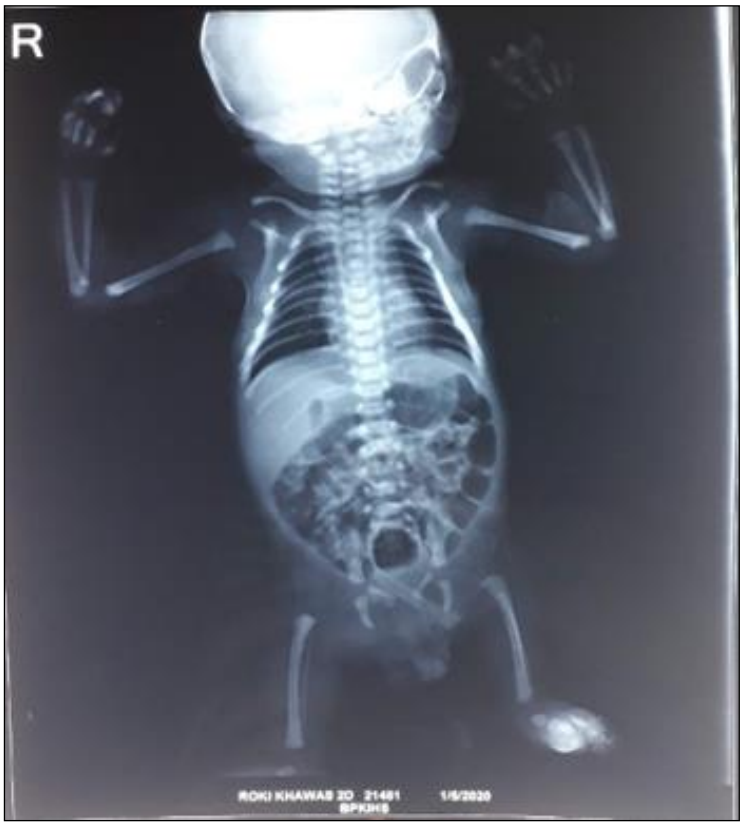

Fig 2: plain x-ray

Discussion

Bilateral femoral agenesis is rare and extreme form of congenital femur deficiency spectrum currently known as proximal femoral focal deficiency (PFFD). This disorder is more common in females. PFFD is a part of developmental field defect which may overlap with the clinical manifestation of femur-fibula-ulna syndrome, fibula aplasia-hypoplasia and femoral hypoplasia/unusual facies syndrome (5). The prenatal diagnosis of this condition can be done with transvaginal ultrasound earliest of which was reported at 14 weeks [6].

Although most of cases are sporadic, few are familial in which autosomal dominant mode of inheritance has been described. The exact etiology of congenital bilateral femoral agenesis is unknown; however, poor diabetic control, viral infections, drugs (thalidomide, alcohol, androgens, warfarin) radiation exposure, focal ischemia and trauma during 4th to 8th week of gestation may be possible etiological factors [7, 8, 9, 10]. However, none of the above mentioned factors were identified in our case.

In our case, there was absence of both the femur and was not associated with other limb anomalies. Generally, this condition is associated with other anomalies such as fibular hemimelia (most common), foot array abnormalities, club foot, acetabular dysplasia [11].

Few cases has reported to be associated with spinal dysraphism (spina bifida or caudal regression syndrome), absent cruciate ligament and microcephaly [12, 13, 14].

Only in absence of any other bony structural abnormality diagnosis of congenital femoral deficiency or PFFD should be made. Differential diagnosis should be made with femoral-facial syndrome which includes femoral hypoplasia, abnormal genitalia and kidneys, hypoplastic humeri with limitation in the range of motion of upper extremities. Early detection by prenatal ultrasound is important in these conditions.

Several classifications have been proposed for PFFD but the most widely used is the one proposed by Aitken and modified by Amstutz done on the basis of plain radiographs, which shows the anatomical relationship between the acetabulum and the proximal end of femur. There are five types in this classification; Type I – Femoral head is present and attached to shaft by femoral neck with coxa vara deformity; Type II – Presence of subtrochanteric pseudoarthrosis; Type III – Moderately dysplastic acetabulum with femoral head and no connection is seen between femoral head and shaft; Type IV – Severely dysplastic acetabulum with absence femoral head and no attachment with the shaft; Type V – Complete absence of acetabulum and proximal femur. Type V is the most severe form [15, 16].

In patient with type I or II, surgical procedures can be done correcting varus deformity, leg-length disproportion, pseudoarthrosis by which stable joint can be achieved [17, 18].

In type III, metaphysealepiphyseal synostosis can be done. In types IV and V, surgical procedures are done combined with above- or below-the-knee amputation along with correction of leg-length discrepancy either by surgery or by prosthesis.

The case presented here does not fit in any of the types mentioned above. There is complete absence of both the femur with no sign of ossification and tibia-complex is directly connected with the acetabulum. The proximal part of tibia appears like femur in shape. Since, this disease does not impair intelligence and is not found to be associated with chromosomal abnormalities or obstetric complication; parents decided to accept his condition. To prevent this condition, to the places where abortion is legal, early diagnosis before viability, option of termination of pregnancy can be offered. Microarray or karyotyping, whole-exome sequencing (WES) should be performed in the child and the parents to find the underlying cause for it.

Conclusion

Bilateral femoral agenesis a very rare congenital condition. To the best of our knowledge, it is the first reported case in eastern part of Nepal. This case is interesting as it does not fit in any category of PFFD classification. More cases like these might need a newer classification which covers this anomaly and to guide for new and better management for these patients so they can live good social and economically productive life.

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