ORIGINAL RESEARCH PAPER
Volume-9 | Issue-2 | February-2020 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

AMELIA LEFT LOWER LIMB - A RARE CASE REPORT WITH REVIEW OF LITERATURE

Orthopaedics

Umesh Yadav*	Assistant Professor, Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001 *Corresponding Author
Jay Dhariwal	Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001
Hitakshi Sharma	Consultant Radiologist , Synapsica, New Delhi
Ashish Devgan	Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001
Ajay Sheoran	Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001
Parth Singh	Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001
Kshitish Chandra Behera	Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001
Himanshu Bansal	Department of Orthopaedics, PGIMS, Rohtak, Haryana-124001

ABSTRACT

Congenital amputations in child are so-called true amputations arising from limb bud arrest. Manifestations vary from complete absence of a complete limb (Amelia) to partial absence of phalanges (Partial Aphalangia). It may present as an isolated defect or may be associated with other congenital abnormalities like cleft lip, cleft palate, diaphragmatic hernia.

Here we report a 3 month old child who presented to orthopedics outpatient department with complete absence of left lower limb. A lobule of fat was present over the lateral aspect of pelvis. No other congenital abnormality was noted in the child. Antenatal and intrapartum period was not significant.

With proper prosthesis and other orthopedic rehabilitation, baby may live normal life but deformities like this are a big challenge to orthopaedic surgeon and the prosthetist in that they may be entirely unsuitable for standard prostheses because of gross variations in limb contour, substandard muscle power, and under-lying skeletal deficiencies.

KEYWORDS

Congenital abnormalities, Amelia, Anomaly

INTRODUCTION

Amelia is characterized by complete absence of part of upper or lower extremity or all four limbs. It is derived from Greek word *"melos"* meaning limb. It refers to type of skeletal dysplasia and is generally thought to be a sporadic anomaly.[1] It may present as isolated defect or may be associated with other congenital malformations, particularly abdominal wall defects i.e. omphalocele and diaphragmatic hernias , cleft lip and/or palate, body wall defect, malformed head, defects of the neural tube, diaphragm and renal anomalies.[2,3,4] More than 50% of the cases are associated with major malformations in other organ systems.[2]It has an incidence rate of 0.04-0.15 per 10000 live births which makes it a very rare congenital anomaly.[5]

This severe limb deficiency can be caused by Teratogens such as alcohol, thalidomide (thalidomide embryopathy), vascular compromise by amniotic bands , maternal diabetes, Roberts syndrome or other cause which can lead to arrest in the formation of primordial limb bud(s) during the early phase (before the 8th week of gestation) of embryogenesis.[6,7]We report one such case of congenital anomaly presented at Department of Orthopaedics,PGIMS, Rohtak.

Case report

A 3 month old female child was brought to orthopaedic department for evaluation for absence of left lower limb since birth. The mother of the child was a 22 year-old healthy primigravida and father was a healthy 24 year-old man, labourer by occupation. There was no history of consanguinity or any other relevant family history. The mother was not diabetic and there were no known teratogenic exposure during her pregnancy. Intrauterine USG details were not available.

The child was delivered at 35 weeks of gestation normally by vaginal route at Health centre of village. Baby's weight at birth was 2200 gm. The height, weight, and head circumference were normal. Parents were informed about the absence of limb at time of birth and were advised to consult a tertiary centre regarding the condition of the child.

Parents brought the child to PGIMS at 3 months of age and were quite hesitant in telling the exact details and exposing the child. Child was examined clinically after adequate exposure and informed consent of the parents. On clinical assessment, there was complete absence of left thigh and leg with no rudimentary toes. A lobule of fat was present over the lateral aspect of pelvis. (Figure 1) Abnormal soft tissue swelling with normal skin wasfound just above the genital area. There were no obvious facial,chest or abdominal anomalies. The anus and spine were normal. Radiographs of pelvis with bilateral hip demonstrated absence of left lower limb along with absence of left hemipelvis. Overlying fat produced radiopaque shadows resulting in obscuration of further bony details. Echocardiography was found to be normal. Parents were advised MRI for further evaluation but parents refused.

Figure 1 showing clinical image of child with Amelia of left lower limb.

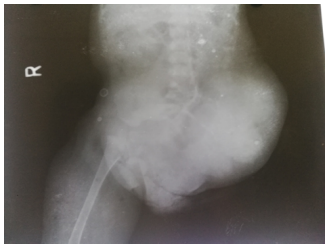

Figure 2- Radiograph of the child showing absent limb and hemipelvis and abnormal white shadow due to fat.

DISCUSSION

This report presents an infant with Amelia of left lower limb with no striking dysmporhic features noticed elsewhere. Traditionally Amelia was considered to be a sporadic anomaly which was associated with little risk of recurrence, or evidence of genetic origins. However, now there are indication for the genetic heterogeneity of this condition which can be due to different modes of inheritance that could be involved in its etiology which include autosomal recessive, X linked dominant and autosomal mode of inheritance.[8]

Likelihood of amelia in the pregnant mother may be indicated by certain signs like Abnormal vaginal bleeding, diabetes mellitus, and toxaemia even prior to clinical observation of absent limbs. Neural tube defects may be indicated by increased alfa fetoprotein in the maternal blood that can accompany limb defects.[9] Intrauterine ultrasound can be used to assess presence or absence of limbs, and abdominal wall and organs. Amelia cases maybe show breech and other non-cephalic presentations at birth, a single artery in the umbilical cord, low placental weight and extremely low birth weight, not accounted for by the lack of limbs.[10]

The prognosis depends upon the severity of other associated anomalies. More than 60% of newborns die early postnatally due to complicated Amelia. Prenatal diagnosis including detailed ultrasound and amniocentesis examination play a major role in counselling parents with fetal anomalies including amelia.

The case described in this report had only isolated lower limb amelia and no other malformations as described above which makes this case rare and academically important.It does not seem to be familial. With proper prosthesis and other orthopedic rehabilitation, baby may live normal life.

REFERENCES-

1. Mastroiacovo P, Källén B, Knudsen LB et-al. Absence of limbs and gross body wall defects: an epidemiological study of related rare malformation conditions. Teratology. 1992;46 (5): 455-64.
2. Lenz W. Genetics and limb deficiencies.Clin Orthop Relat Res 1980; 148: 9-17.
3. Froster-Iskenius UG, Baird PA. Amelia: incidence and associated defects in a large population. Teratology 1990; 41:23-31 10.
4. Mastroiacovo P, Källén B, Knudsen LB, Lancaster PA, Castilla EE, Mutchinick O, et al. Absence of limbs and gross body wall defects: an epidemiological study of related rare malformation conditions. Teratology 1992; 46: 455-464 10.
5. Froster-Iskenius UG, Baird PA. Amelia: incidence and associated defects in a large population. Teratology. 1990; 41: 23-31.
6. Smithells RW, Newman CG. Recognition of thalidomide defects. J Med Genet 1992; 29:716-723.
7. Bruyere HJ, Jr, Viseskul C, Opitz JM, Langer LO, Jr, Ishikawa S, Gilbert EF. A fetus with upper limb amelia, "caudal regression" and Dandy-Walker defect with an insulin-dependent diabetic mother. Eur J Pediatr 1980;134:139-143.
8. Al Riyami N, Ahmed A, Tanzeem S, Abdul-Latif M. Fetal amelia: A case report. Oman medical journal. 2012; 27: 54.
9. Eghbalian F, Sharif A, Monsef AR. Amelia: A case report and literature review. Iranian journal of pediatrics. 2015; 25(6).
10. Song SY, Chi JG. Tri-amelia and phocomelia with multiple malformations resembling Roberts syndrome in a fetus: is it a variant or a new syndrome? Clin Genet. 1996;50(6):502–4.