|| Print ISSN: 2589-7837 || Online ISSN: 2581-3935 ||
International Journal of Medical Science and Diagnosis Research (IJMSDR)
Available Online at www.ijmsdr.com
NLM (National Library of Medicine ID: 101738824)
Original Research Article
Volume 4, Issue 9; September: 2020; Page No. 35-37

UNILATERAL LOWER LIMB PHOCOMELIA DURING THE COVID-19 PANDEMIC: ETHICAL DILEMMA FROM AN OBSTETRICIAN'S PERSPECTIVE

Avir Sarkar[1], Snigdha Kumari[2], Seema Sheokand[3]

[1]MBBS, Junior Resident, Department of Obstetrics and Gynaecology, Post Graduate Institute of Medical Education and Research, Chandigarh

[2]MS, DNB, Assistant Professor, Department of Obstetrics and Gynaecology, Post Graduate Institute of Medical Education and Research, Chandigarh

[3]MD, Senior Resident, Department of Obstetrics and Gynaecology, Post Graduate Institute of Medical Education and Research, Chandigarh

Conflicts of Interest: Nil

Corresponding Author: Snigdha Kumari

DOI: https://doi.org/10.32553/ijmsdr.v4i9.674

Abstract:

Isolated unilateral phocomelia is a rare congenital disorder. Historically, the most famous association with consumption of thalidomide in first trimester of pregnancy resulted in babies born with limbs resembling flippers of a seal. Genetic inheritance in the form of autosomal recessive manner is also postulated. It can be sporadic, familial or syndromic.

On account of the deadly COVID-19 pandemic, suspension of non-emergency services including OPDs in majority of the hospitals led to diversion of resources for combatting emergency medical care. This temporary pause and containment was detrimental to non-COVID patients. We present a rare case of unilateral phocomelia where lockdown due to COVID-19 pandemic has resulted in denial of medical termination of pregnancy (MTP).

Introduction:

Fetal limb abnormality is seen in every 6 of 10,000 live births (1). Congenital defects of the limbs can be caused by teratogenic drugs, genetic syndromes, familial diseases and sporadic events. Maternal risks factors of limb defects include advanced maternal age (> 35 years), overt diabetes in first trimester, fetal infections like toxoplasma, rubella, cytomegalovirus and herpes (TORCH) and familial history of other congenital disorders (2). Phocomelia is transmitted as an autosomal recessive disorder. It is hypothesised that mutation in the centromeric region of chromosome 8 hinders proper replication of genome and division of cells. As a result, they cease to develop or die preventing proper development of limbs, eyes and palate (3). True phocomelia refers to complete absence of mid-segment of limb whereby the extremities like fingers and toes are attached directly to the torso (4).

Case Report:

A 23-year-old primigravida presented to the antenatal clinic (ANC) at 28-weeks period of gestation (POG) with an anomaly scan revealing short length of right femur and absence of right tibia and fibula with foot appearing directly attached to the torso. No other malformation was noted. Her marriage was non-consanguineous with no history of limb deformities in both the families. Blood sugar was normal during pregnancy. There was no history of consumption of teratogenic drugs, radiation exposure and no viral prodrome with rash in the first trimester.

Fetal echocardiography was grossly normal. Patient was counselled regarding the future outcome of such a functional limb defect. Considering the advanced gestation, MTP could not be offered. She was advised serial antenatal monitoring. She lost to follow-up due to COVID-19 pandemic and revisited gynae casualty at 38 weeks POG in spontaneous labour. Ultrasound (USG) revealed right femur length of 6.04cm corresponding with 31weeks 3 days (Figure 1).

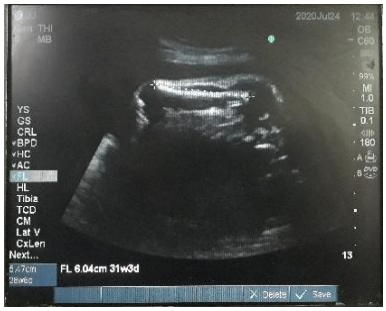

Figure 1: USG of right femur.

Left femur was 6.9cm in length corresponding to 36 weeks POG. She delivered a live baby boy of 2.5 kg with good APGAR scores. Gross examination revealed extremely

shortened right lower limb with foot attached to the same (Figure 2).

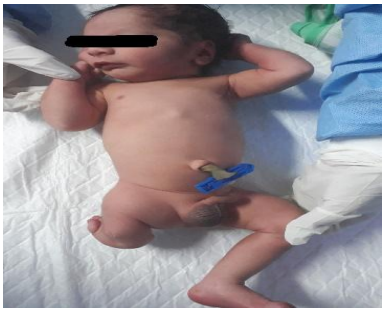

Figure 2: Picture showing right lower limb phocomelia.
It was oligodactyly with wide sandal gap (Figure 3).

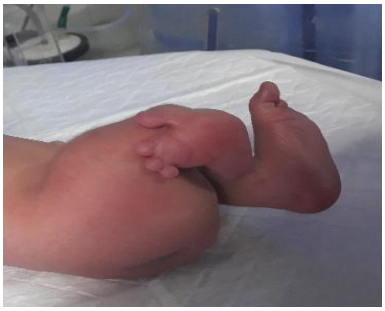

Figure 3: Picture showing oligodactyly.
Left lower and bilateral upper limbs were grossly normal. Infantogram in the post-natal period suggested short femur with absent tibia and fibula of right limb with no other long bone abnormalities (Figure 4).

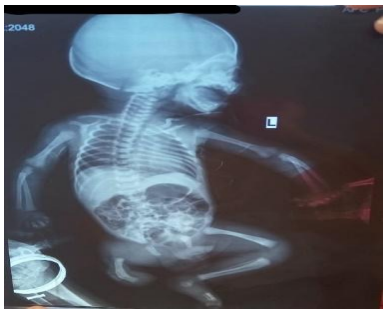

Figure 4: Infantogram showing right short femur with absent tibia and fibula.

Ultrasound of cranium and kidneys were also normal. Baby was started on exclusive breastfeed. Genetic and orthopaedic counselling was done regarding physiotherapy and follow-up.

Discussion:

Being a part of appendicular skeleton, limbs arise out of the lateral plate mesoderm differentiating on 26 -55th day of life (4). Any teratogenic insult in this window manifests as limb deformities. Extensively studied in this category is thalidomide. CPS49, an anti-angiogenic analogue of thalidomide is responsible for limb defects (5). Increased cell death can be due to direct activation of the caspase pathways or due to antiangiogenic effect of thalidomide (5).

Stumbling upon the diagnosis of foetal limb defects we should try to probe around the following possibilities of chromosomal abnormalities, mutant genes, teratogens, a combination of environmental and genetic factors and vascular defects (6).

A study conducted by Sanchez et al with 144 phocomelia cases showed that in 50% cases, phocomelia was an isolated defect (2). Another study conducted over 7 years including 1300 pregnancies with limb defects stated that there is 59% association between limb defects and aneuploidies. Trisomy 18 which is a lethal chromosomal abnormality frequently presents with limb defects such as club foot (7). 9.9% cases were syndromic with Roberts syndrome being of highest incidence followed by Holt-Oram and Al-Awadi/Raas-Rothschild (AARR) syndrome (4). A complete pathological study (including radiological examination) of the fetus is mandatory in order to precisely define not only phocomelia but all the defects present in the fetus which is essential for an accurate counselling of the parents regarding recurrence risk and possibility of other malformations (4).

Al-Awadi/Raas-Rothschild (AARR) syndrome is a rare phocomelia syndrome characterized by limb and pelvic bone hypoplasia, renal anomalies like horseshoe and polycystic kidney, abnormal facial features including cleft palate, hypertelorism and retrognatia (8). Our baby had none of these features on antenatal USG and post-natal evaluation. The absence of symmetrical limb defects with silvery blond hair and hemangiomas ruled out Robert's syndrome, yet another cause of syndromic phocomelia.

Phocomelia is seen with significant neurocognitive derangement if associated with syndromic entity. Trisomies are usually associated with microcephaly and poor survival rates. Robert's phocomelia is associated with learning disabilities (9). DK phocomelia syndrome presents with impairment in adaptive skills and poor school performance (10). Since our baby had no evident syndromic features, a close watch on his milestones could be the earliest clue to predict neurological prognosis.

As the emaciated healthcare system is attempting to break the tide of the novel coronavirus pandemic across the globe, the highest cost of this fight is being borne by the non-COVID-19 patients everywhere. For about 6 months, non-emergency services including OPDs have been suspended in majority of the hospitals in order to divert resources for combatting emergency medical care during this deadly pandemic. This temporary pause and containment proved detrimental to non-COVID-19 patients like the index case. Fate of thousands of patients who used

to travel to towns to receive medical care seems to hang by a fine thread now. Had she visited the healthcare setup before 20 weeks POG, she could have been counselled regarding the outcome of the baby thereby giving her an option of MTP. Limb defect like phocomelia cannot be cured and results in a permanent limp. Orthopaedic prosthesis is a viable option to reduce disability in this baby although utilisation of same may be difficult due to lack of soft tissues to provide support to the prosthesis. For her future pregnancies, we have asked her for early antenatal booking with strict follow up of aneuploidy screening and timely anomaly scan.

Conclusion:

After reviewing the literature for causes of phocomelia, our patient seems to be a sporadic case. The genetic analysis of the baby is still pending and delayed owing to the COVID pandemic. Although crippled health system during this COVID-19 pandemic was unable to cater to the agony of the couple in index pregnancy, they have been advised for pre-natal genetic counselling and assessment before their next conception to plan a better outcome.

References:

1. Santina Ermito, Angela Dinatale, et al. Prenatal diagnosis of limb abnormalities: role of foetal ultrasonography. *Journal of prenatal medicine.* 2009;3:18–22.
2. Shi Y, Zhang B, et al. Prenatal limb defects: Epidemiologic characteristics and an epidemiologic analysis of risk factors. *Medicine (Baltimore).* 2018;97: e11471.
3. Dar IH, Dar MI, et al. Phocomelia: case report of a rare congenital disorder. Egypt J Dermatol Venerol. 2020;35:82-84.
4. Bermejo-Sánchez E, Cuevas L, et al. Phocomelia: a worldwide descriptive epidemiologic study in a large series of cases from the International Clearinghouse for Birth Defects Surveillance and Research, and overview of the literature. *Am J Med Genet C Semin Med Genet*. 2011;157:305-320.
5. Sharma A, Chaudhary R, et al. Nonsyndromic Phocomelia: A Case Series. Transl Biomed. 2016;7:1-49
6. Pan P. Antenatal ultrasonography should be for all. Phocomelia: An extremely rare congenital anomaly-A case report. Glob J Medical Clin Case Rep. 2019;6:001-2.
7. Rice KJ, Ballas J, Lai E, Hartney C, Jones MC, Pretorius DH. Diagnosis of fetal limb abnormalities before 15 weeks: cause for concern. Journal of Ultrasound in Medicine. 2011;30:1009-19.
8. Alp E, Alp H, Atabek ME, Pirgon Ö. Al-Awadi/Raas-Rothschild Syndrome in a Newborn with Additional Anomalies. Journal of clinical research in pediatric endocrinology. 2010;2:49.
9. Goh ES, Li C, Horsburgh S, Kasai Y, Kolomietz E, Morel CF. The Roberts syndrome/SC phocomelia spectrum—a case report of an adult with review of the literature. American Journal of Medical Genetics Part A. 2010;152:472-8.
10. Antonini TN, Van Horn Kerne V, Axelrad ME, Karaviti LP, Schwartz DD. Neurocognitive profile of a young adolescent with DK phocomelia/von Voss phocomelia/von Voss Cherstvoy syndrome. American Journal of Medical Genetics Part A. 2015;167:1632-6.