Acta Scientific PAEDIATRICS (ISSN: 2581-883X)

Volume 5 Issue 10 October 2022

Case Report

Phocomelia: A Case Report

Edwin Dias[1]*, Abu Farhaz Ali[2] and Kavya[3]

[1]Professor and HOD Department of Pediatrics, Senior Adjunct Professor, Srinivas University, Director of Research and Publication, Srinivas Institute of Medical Sciences and Research Centre, Mukka, Mangalore, India

[2]Junior Resident, Department of Pediatrics, Srinivas Institute of Medical Science and Research Center, India

[3]Intern, Srinivas Institute of Medical Science and Research Centre, India

***Corresponding Author:** Edwin Dias, Professor and HOD Department of Pediatrics, Senior Adjunct Professor, Srinivas University, Director of Research and Publication, Srinivas Institute of Medical Sciences and Research Centre, Mukka, Mangalore, India.

Received: August 08, 2022

Published: September 29, 2022

Abstract

To report a case of left upper limb phocomelia in a monochorionic, monoamniotic twin pregnancy. It is an extremely rare congenital disorder. On antenatal ultrasound examination it was not associated with any other major anomalies. The differential diagnosis of this condition includes thrombocytopenia-absent radius (TAR) syndrome and Robert syndrome. Twin A was affected, twin B was normal. On the basis of Normal karyotyping and with no other major structural anomalies the leading diagnostic case encompasses of either TAR syndrome or Robert syndrome.

Keywords: Upper Limb Phocomelia; Twin Pregnancy

Introduction

PHOCOMELIA is a type of congenital disorder which causes short limbs. It can vary in type and severity. It can affect all the four limbs either the upper or lower limbs. It usually affects the upper limbs. Phoco means "seal limb" or "flipper" and "Melos" means limbs. The particular pattern of inheritance has not been found so far. Phocomelic deformities was very rare initially till the widespread use of thalidomide drug. Phocomelia in comparison to other longitudinal deficiencies, is regarded as a part of transverse and intercalated segmental dysplasia. Many patients have severe, but not as such classifiable upper-extremity deformities [1]. Abnormalities in the limb is also present proximal and also distal to the segmental defect.

Anatomically, phocomelia is described into three (by Frantz and O' Rahilly) viz; 1. Complete phocomelia- presenting with absence of all bones of limb proximal to the hand, 2. Absence of humerus bone with bones of forearm and hand attached to the trunk, 3. Direct attachment of hand to the humerus.

Associated deformities can also be found including radial ray deficiencies (usually seen in phocomelia associated with thalidomide) and it also may be associated with cleft lip and cleft palate (Robert syndrome). Various degrees of humeral, forearm and hand deficiencies along with clavicle and scapula can also be present. Scapula may be deficient laterally; true elbow joint is absent. Usually, hand only have three or four digits with an absent thumb. As

the child grows there is considerable functional and psychologic impairment.

Case Report

A 22-year-old, gravida 2, para 1, living 1, antenatal USG scanning was done for the first time at 12 weeks of gestation. Findings were consistent with a monochorionic, monoamniotic twin pregnancy. It was a conception after a non consanguinous marriage, there was no positive family history from either side of both couples, and had 1 healthy son. In the first trimester of pregnancy there was no history of intake of any drugs by the mother or any history of fever in the mother. Genetic counseling was given about the increased risk of chromosomal abnormality and other adverse outcomes, follow-up was done with sonography and genetic amniocentesis at 17 weeks. On ultrasound scanning the femur lengths were appropriate for gestational age in both twins. The nuchal thickness measurements observed was also within normal limits for both the twins (2.5 mm for twin A and 2.8 mm for twin B). No other structural fetal anomalies were detected. Partial absence of humerus was noticed and complete absence of the radius and ulna of left side. The hands extended from a small soft tissue density at the level of the shoulder. Twin B had normal humerus, radius and ulna.

Discussion

Twin A was affected, while twin B was normal. No history of congenital anomalies in the family. The first child of the couple was also normal.

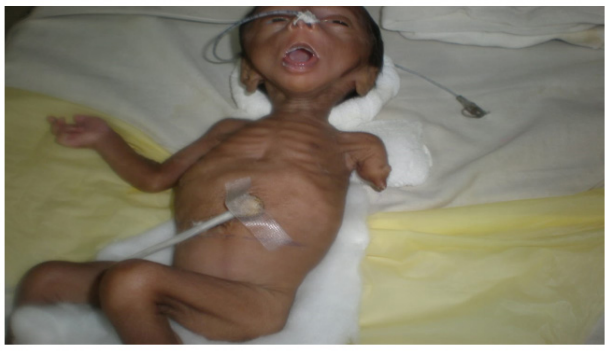

Figure 1: Absence of left distal humerus, radius and ulna with absent thumb and underdeveloped three fingers.

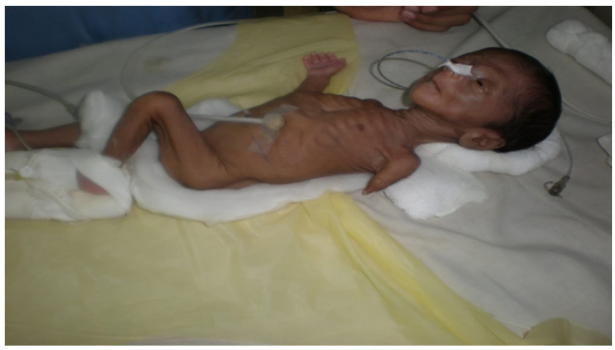

Figure 2: Phocomelia with absent distal humerus, radius and ulna.

The first case of phocomelia was described in Germany in 1956. Since then most of cases was studied and then related with intake of thalidomide which was readily available over counter for nausea and vomiting.

Thalidomide came to market of West Germany in 1957. Initially, the usage was as a sedative or hypnotic, it was also used to cure anxiety, insomnia and gastritis [7]. It was later used against morning sickness in pregnancy. Thalidomide was available over the counter in Germany around 1960, and was purchased without a medical prescription. Shortly after the drug becoming widely available, in Germany, around 5,000 to 7,000 infants were found to be born with phocomelic deformities [8]. Only 40% of these infants survived. Research proved that phocomelia which was nonexistent through the 40s and 50s, by the widespread use of the drug thalidomide in Germany in the 60s, cases of severe phocomelia was increased; and was attributed to the use of thalidomide [8]. Statistics suggested that '50% of the mothers with phocomelic children had taken thalidomide during the first trimester of pregnancy' [8,10]. Throughout Europe, Australia, and the United States, 10,000 cases of phocomelia in infants was reported; out of which 50% of the 10,000 survived [8]. Thalidomide was then recognized to be the cause of death or severe disabilities among babies. The fetus exposed to thalidomide antenatally experienced limb deficiencies as that the long limbs were either not developed or was seen as stumps. Deformities of eyes, hearts, alimentary, and urinary tracts, along with blindness and deafness were also observed [8,10].

Fetal karyotyping was necessary in this case as to rule out other conditions. The differential diagnosis of this includes various syndromes, of which Brachmann De Lange syndrome has also been reported with increased nuchal translucency [2-4] and the absence of the distal upper extremities. However, facial features in consistent with this syndrome were not seen in the fetus in our case. Holt-Oram syndrome is also ruled out as there is no cardiac defect, the severity of the upper limb abnormalities, and no triphalangeal thumb or extra carpal bones. Similarly, Fanconi anemia is also ruled out as it affects the thumb and radius where as in this case more severe limb abnormalities seen. On the ground of the severity of the upper limb phocomelia, karyotyping which has revealed to be normal, and there is no other structural defects, the leading diagnostic possibilities in this case includes TAR syndrome and Roberts syndrome.

In a well-documented studies, 90% of cases had tetra phocomelia, and 10% of cases showed phocomelia affecting only the upper limbs. Robert syndrome is usually associated with nuchal abnormalities, described as either increased nuchal translucency or cystic hygroma. So far, Robert syndrome has not been identified with a single gene defect, but 80% of cases with features of Roberts syndrome a cytogenetic phenotype ("centromere puffing") has been identified postnatally. Centromere puffing was seen in amniocytes and chorionic villus samples. Roberts syndrome phenotype has shown to have thrombocytopenia occasionally, which suggest an overlap between Roberts syndrome and TAR syndrome may exist.

Treatment is mainly conservative. Various devices have been designed and developed helping the child in achieving self-independence assisting in self-hygiene, feeding and dressing all of which plays a major role. Surgery though has only minor role and can be offered for shoulder instability, limb shortening or inadequate thumb opposition. Surgical specific techniques have not been well described but certain techniques improving thumb opposition like rotational osteotomy of one of the digits with web space deepening has been tried.

Conclusion

On the basis of upper limb severity and normal karyotyping and without any other structural abnormalities. The leading diagnosis of this case encompasses of either TAR syndrome or Roberts syndrome.

Bibliography

1. Charles A Goldfarb., *et al*. "Upper-extremity phocomelia re-examined: a longitudinal dysplasia". *JBJS* 87.12 (2005): 2639-2648.

2. Hyett J., *et al*. "Using fetal nuchal translucency to screen for major congenital cardiac defects at 10-14 weeks' gestation: population based cohort study". *BMJ* 318 (1999): 81-85.

3. Souka AP., *et al*. "Outcome of pregnancy in chromosomally normal fetuses with increased nuchal translucency in the first trimester". *Ultrasound in Obstetrics and Gynecology* 18 (2001): 9-17.

4. Snijjers RJ., *et al*. "UK multicentre project on assessment of risk for trisomy 21 by maternal age and fetal nuchal translucency thickness at 10-14 weeks' gestation". *Lancet* 352 (1998): 343-346.

5. Fisk NM., *et al*. "Transvaginal ultrasound recognition of nuchal oedema in the first-trimester diagnosis of achondrogenesis". *Journal of Clinical Ultrasound* 19 (1991): 586-590.

6. Soothill PW., *et al*. "Achondrogenesis type 2 diagnosed by transvaginal ultrasound at 12 weeks' gestation". *Prenatal Diagnosis* 13 (1993): 523-528.

7. Fukada Y., *et al*. "The prognosis of fetuses with transientnuchal translucency in the first and early second trimester". *Acta Obstetricia et Gynecologica Scandinavica* 76 (1997): 913-916.

8. Miller Marylin T. "Thalidomide embryopathy: A model for the study of congenital incomitant horizontal strabismus". *Transaction of the American Ophthalmological Society* 81 (1991): 623-674.

9. Pharmaceutical teratogens. Teratology society—birth defects research. 08 December (2007).

10. Ito T., *et al*. "Identification of a primary target of thalidomide teratogenicity". *Science* 327.5971 (2010): 1345-1350.

11. Cuthbert Alan. "Thalidomide". The oxford companion to the body. Oxford University Press, 2001, 2003. Answers.com 06 Dec (2007).