MEDICAL SCIENCE

A case report of sporadic (tetra) phocomelia and unilateral isolated upper limb phocomelia: An antenatal ultrasonography of an extremely rare congenital anomaly

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ABSTRACT

Phocomelia is a severe and a rare form of limb defect in which the limbs are not formed completely. It affects the growth and development of musculoskeletal system leading to grossly underdeveloped or absent limb. It may be inherited as autosomal-dominant or recessive-disorder. We are presenting two cases of phocomelia, Case 1: A 28-year-old primigravida presented to our hospital with the history of irregular menstrual cycles for which she was advised ultrasound-pelvis which revealed a 21-week 6-days fetus. An antenatal-ultrasound was performed to rule of anatomical anomalies which showed symmetrical shortening of all four limbs with no other congenital anomalies. Because of her irregular cycles, the patient was unaware of her last-menstrual date and she reports spontaneous conception and also gives a history of first-degree consanguineous marriage. Due to the lack of significant history or any other associated symptoms, our case did not fit into any syndromic phocomelia and appears to be the result of sporadic phocomelia involving all four limbs or an isolated limb. Case 2: An ultrasonogram was done at 12 weeks 2 days gestational age in 24-year-old with obstetric history of Gravida-3 Para-1 Abortion-1 who presented to our hospital for regular first-trimester antenatal check-up. Ultrasound finding showed absent left radius and ulna with the deformed hand attached to the left humerus. Both patients had no family history of any congenital anomaly or infections during pregnancy, no history of any drug intake or exposure to fertilizers or pesticides. There is no relevant medical or surgical history.

Keywords: Phocomelia, tetra phocomelia, limb defect, congenital anomaly, flippers, ultrasonography.

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1. INTRODUCTION

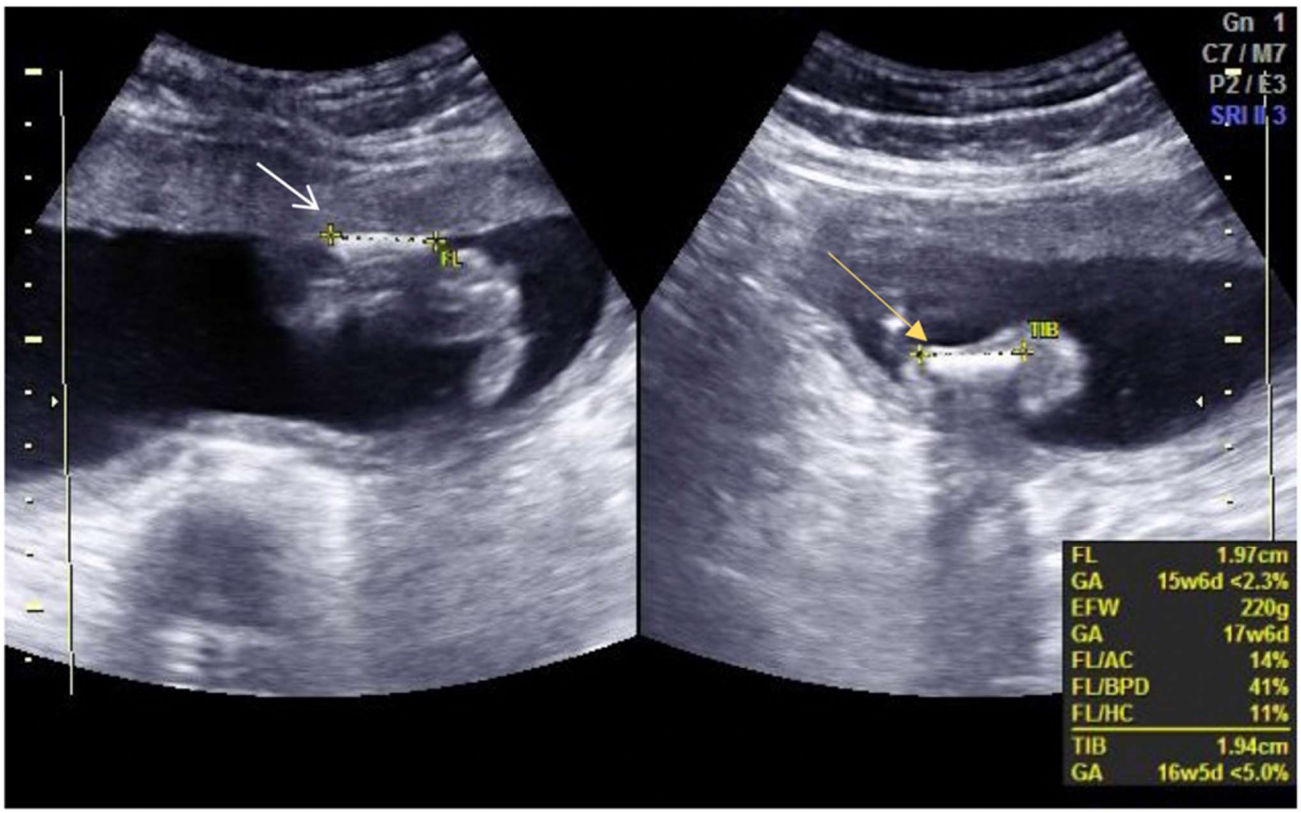
Phocomelia is a severe and a rare form of limb defect in which the limbs are not formed completely. It affects the growth and development of musculoskeletal system leading to grossly underdeveloped or absent limb. Limbs may be totally absent or severely hypoplastic with hand or foot attached to the truck of the foetus and number of toes and fingers might also vary. Greek terminology for phocomelia which refers to the appearance of patients' limb to flipper like appendages. Sometimes phocomelia may be associated with craniofacial malformation and other visceral system involvement (multisystem involvement). However, phocomelia is considered as a limb defect to be produced by the drug thalidomide. The exact cause of most of the phocomelia is not well understood yet. Incidence of phocomelia reported is 0.62 per 100000 live births (Lavanya et al., 2020; Samal et al., 2015). Sporadic phocomelia is either inherited as a result of spontaneous mutation or autosomal recessive trait (Tia and Al-Ghafri, 2018).

2. CASE REPORT

Case 1

A 28-year-old primigravida presented to our hospital with the history of irregular menstrual cycles for which she was advised ultrasound-pelvis which revealed a 21-week 6-days fetus. An antenatal ultrasound was performed to rule of anatomical anomalies. She is teacher by occupation and husband is a 38-year-old bus driver. There's no family history of any congenital anomaly or infections during pregnancy, no history of any drug intake or exposure to fertilizers or pesticides. There is no relevant medical or surgical history. On routine antenatal ultrasound imaging we found there is symmetrical shortening of all four limbs-bilateral lower limb shortening (Figure 1) and upper limb shortening (Figure 2), otherwise foetal brain and abdomen did not show any abnormality with no other congenital abnormality (Figure 3). 3D volumetric acquisition imaging showing markedly shortened upper and lower extremities (Figure 4).

Imaging findings



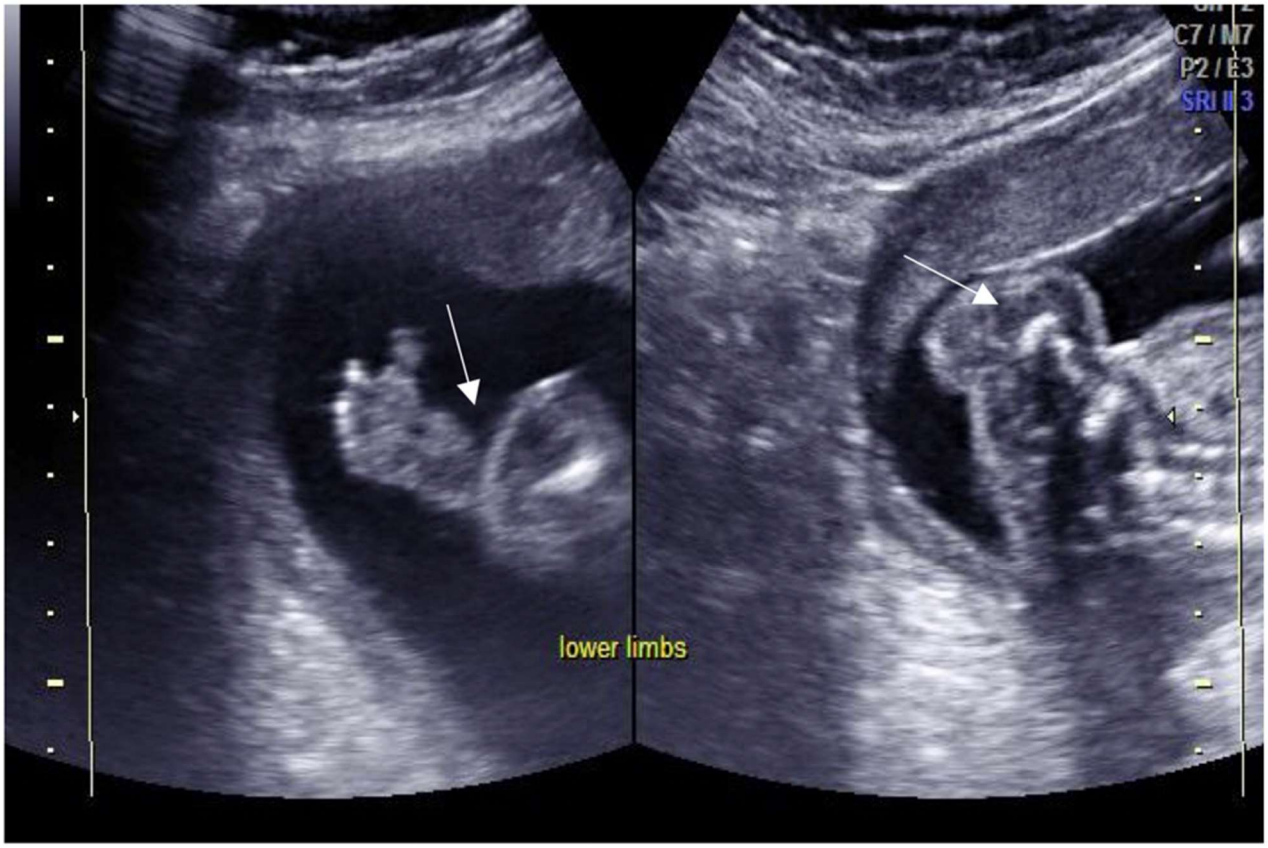
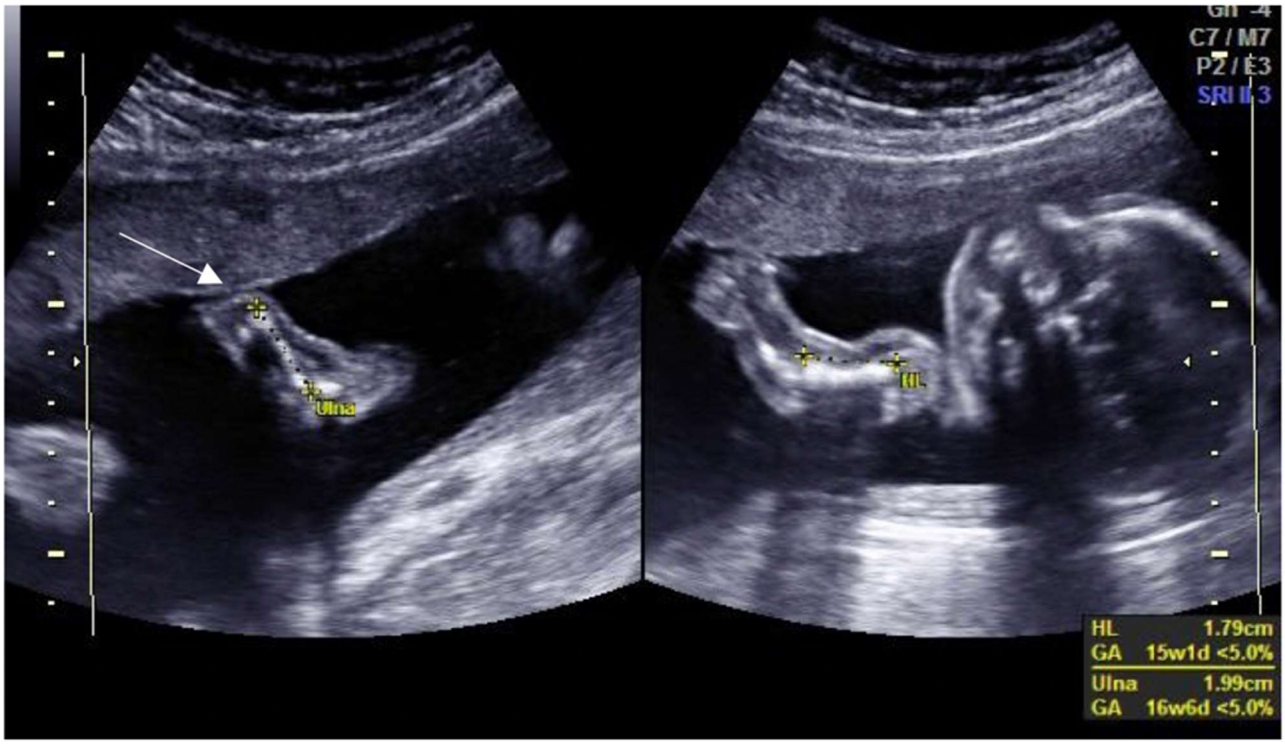


Figure 1 B- mode grey scale ultrasounds showing lower limb phocomelia



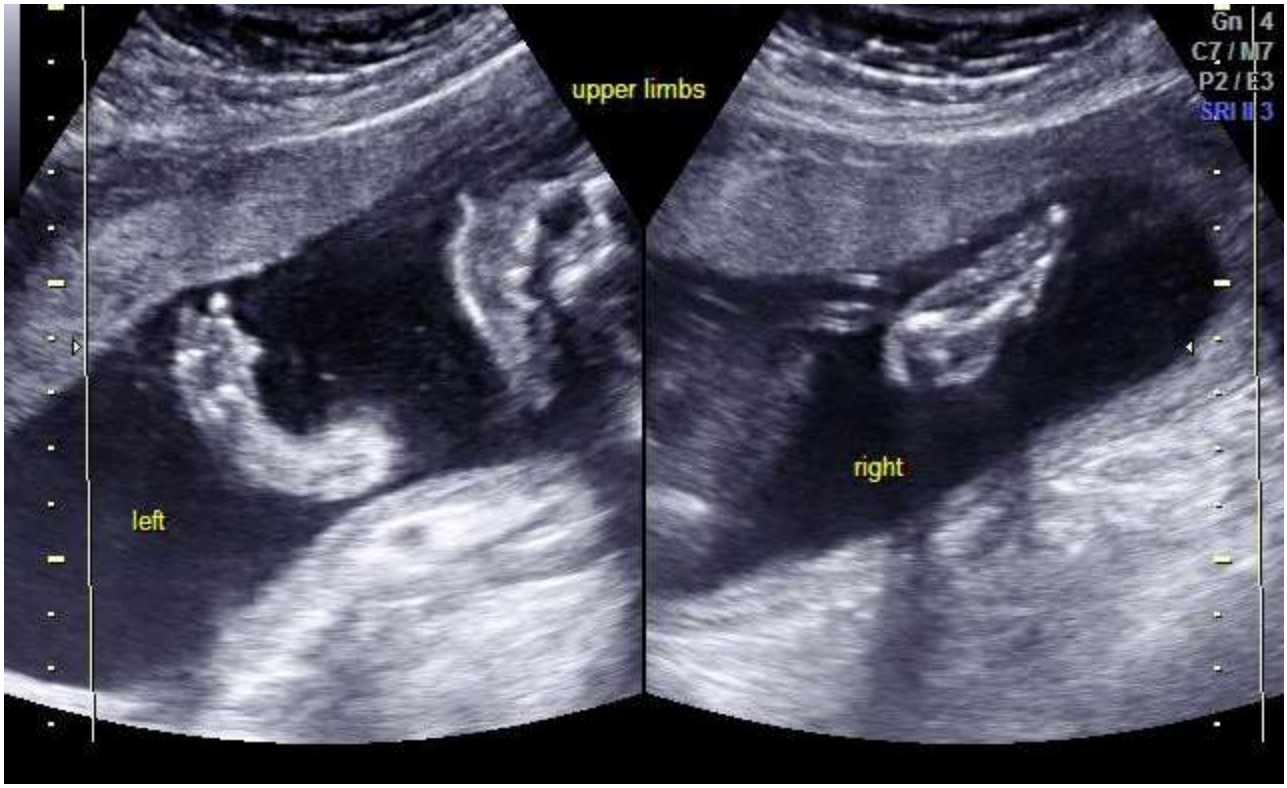
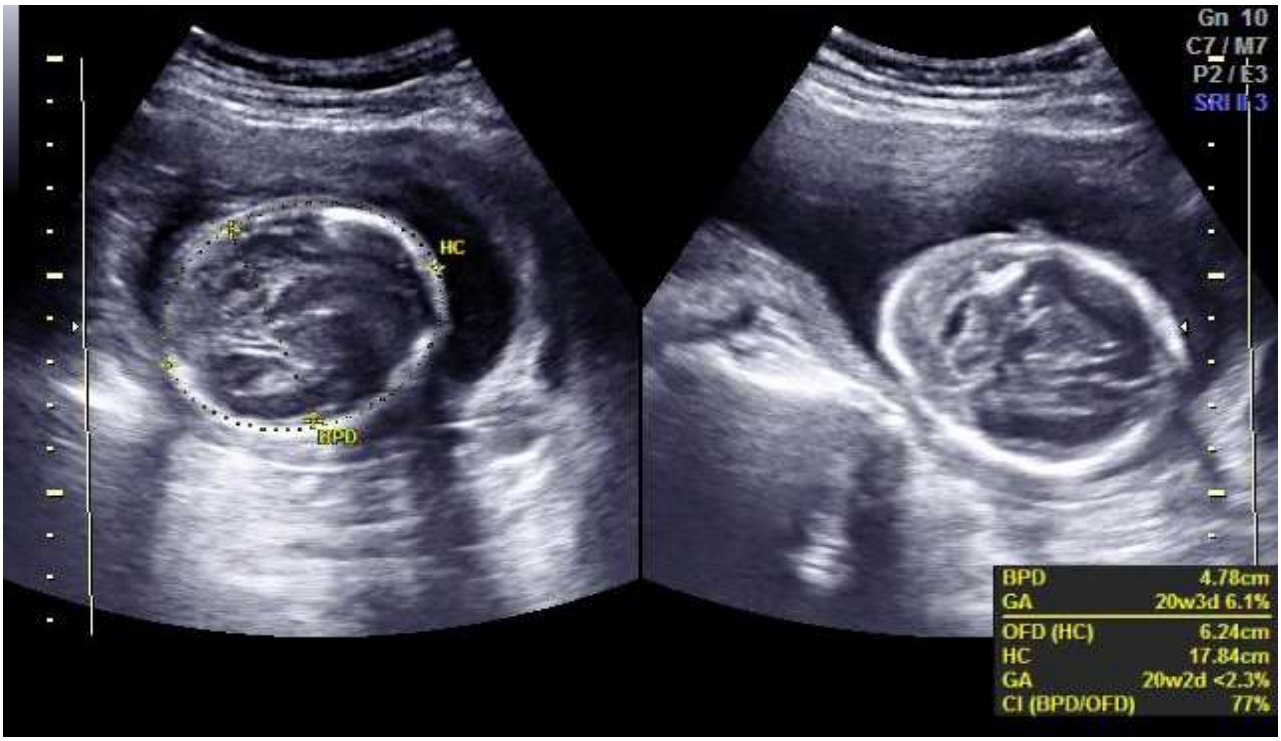


Figure 2 B-mode grey scale ultrasound showing upper limb phocomelia



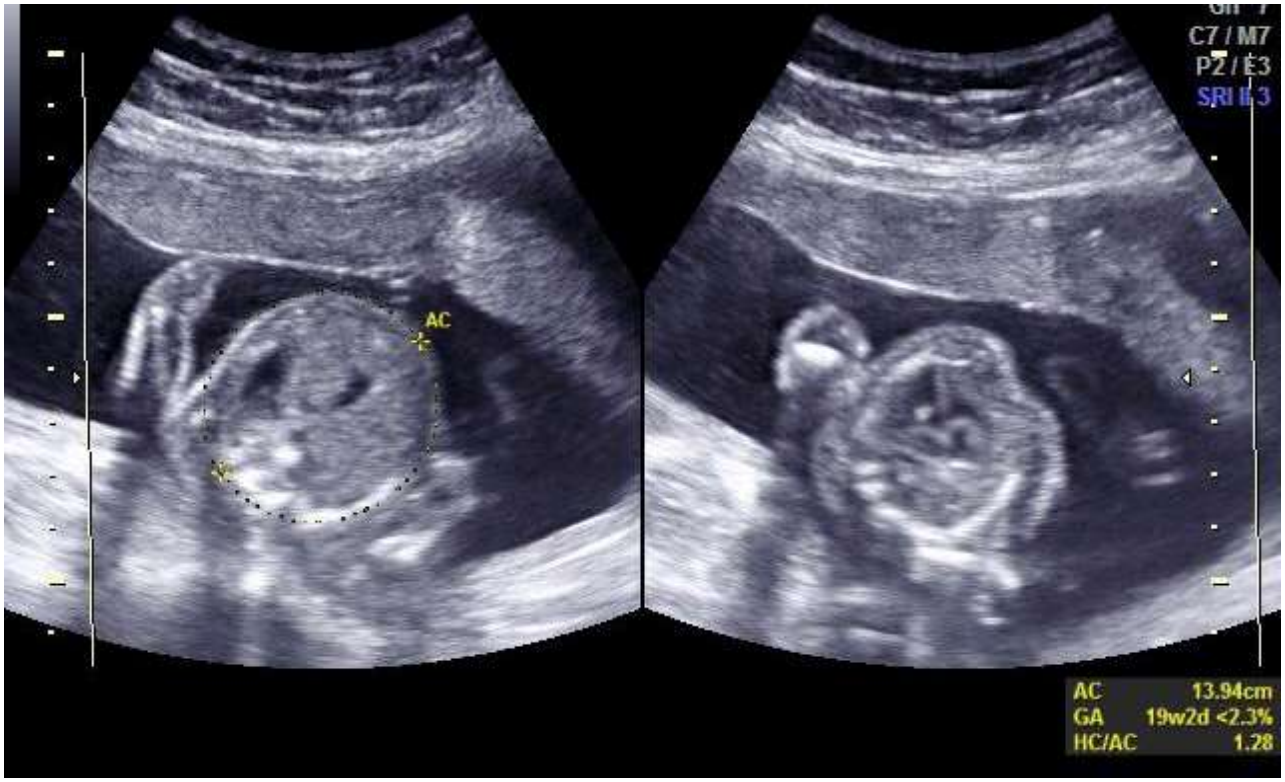


Figure 3 B- mode grey scale ultrasound foetal brain and abdomen did not show any abnormality

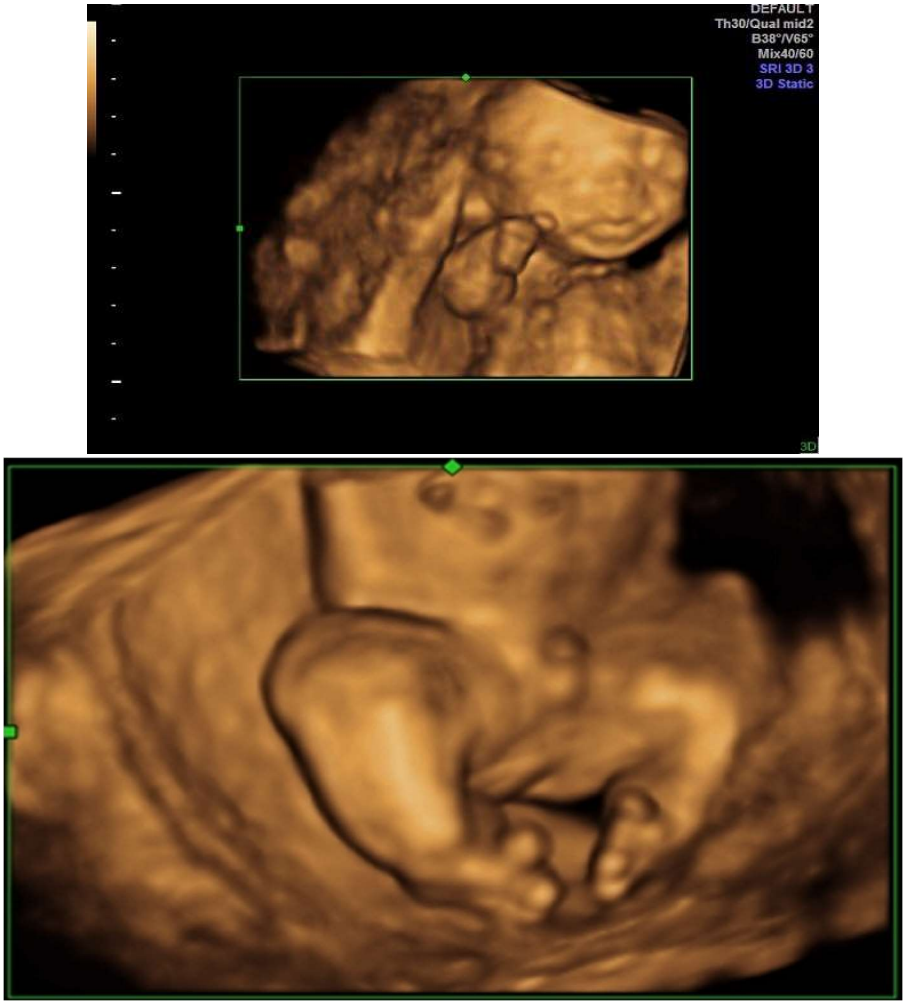


Figure 4 3D volumetric acquisition images shows markedly shortened upper and lower extremities

Case 2

An ultra-sonogram was done at 12 weeks 2 days gestational age in 24-year-old with obstetric history of Gravida 3 Para 1 Abortion 1 who presented to our hospital for regular first trimester antenatal check-up. Patient gives history of spontaneous conception, on regular perinatal vitamins with no other drug intake or substance abuse, no exposure to pesticides or fertilizer, no radiation exposure or infections during pregnancy. There's no family history of congenital anomaly, no history of consanguinity, diabetes or any other chronic medical illness. No exposure to pesticides or fertilizer, no radiation exposure or infections during pregnancy. She had previous history of spontaneous abortion for unknown reason at 10 weeks.

On routine antenatal, ultrasound images showed absent left radius and ulna with the deformed hand attached to the left humerus. Left fetal arm is seen with ossified distal part of humerus; radius and ulna (forearm) appear absent and the deformed hand is attached to the left arm (Figure 5). Otherwise, nuchal translucency: 1.3 mm (within normal limits) Nasal bone seen. Ductus venosus flow appears normal. Uterine artery PI is within normal limits.

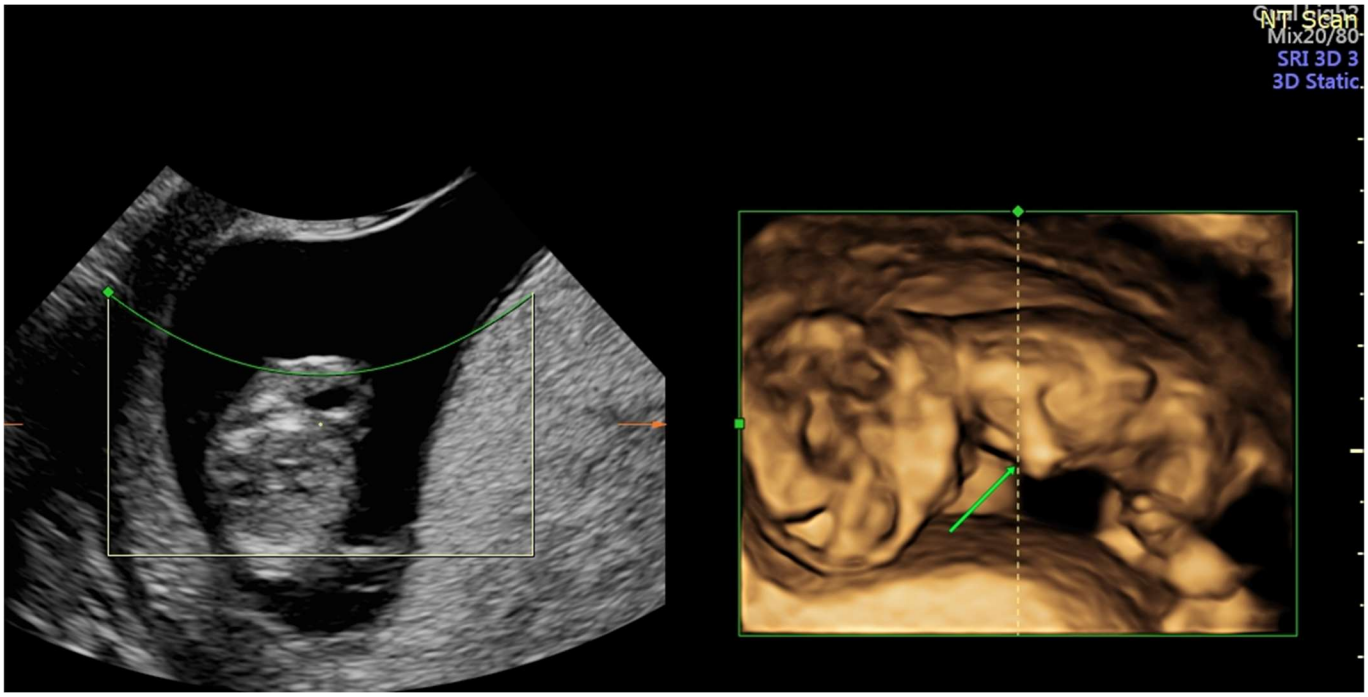


Figure 5 Shows isolated phocomelia of left upper limb. Left fetal arm is seen with ossified distal part of humerus; radius and ulna appear absent and the deformed hand is attached to the left arm.

3. DISCUSSION

Phocomelia is the rare congenital malformation and most severe form of limb defect affecting the normal growth and development of musculoskeletal system, the limbs are severely underdeveloped or absent in which long bones are shorter than normal size with upper portion of the limb being severely affected. Incidence of true phocomelia reported is 0.62 per 100,000 births. In Greek word phocomelia refers the patients' limbs similar to that of flippers on a seal. Various factors attributing the cause include genetic/consanguinity, any substance abuse or intake of teratogenic drugs (thalidomide), diabetes, radiation etc. in pregnancy (Lavanya et al., 2020; Samal et al., 2015; Shukla et al., 2015).

Fetal limb development begins between 24 and 36 days after conception. For upper limb involvement, the sensitive period for phocomelia was between days 24 and 33 following fertilisation and for lower limb involvement, between days 28 and 33 (Bermejo-Sánchez et al., 2011).

If the development process is disrupted at this early age, severe phocomelia may result. In 60% of cases, only one limb is affected and Amelia abnormalities make up about 25% of limb malformations. Mechanical disruption or vascular abnormalities during limb development are considered to be the cause of deformities (Shukla et al., 2015).

Phocomelia was classified into 3 types by Frantz and O'Rahilly in 1961.

1. Complete phocomelia (Type I): Hands or digits attached directly to trunk of patient.
2. Proximal phocomelia (Type II): Forearm bones between hand and trunk.

3. Distal phocomelia (Type III): Hands directly attached to humerus (Tytherleigh-Strong and Hooper, 2003).

It may be in the form of complete absence of proximal and distal bones of limb or may be incomplete when either proximal or distal bones are missing (Shukla et al., 2015). Based on other associated clinical abnormalities phocomelia was further sub classified into Al- Awadi/Raas-Rothschild syndrome (AARR syndrome), Zimmer and Schinzel phocomelia and Robert's/SC phocomelia (Samal et al., 2015). Differential diagnosis includes Robert's syndrome, Holt-Oram syndrome, Thrombocytopenia-absent radius syndrome, thalidomide-induced phocomelia and sporadic phocomelia (Osadsky, 2015).

Sporadic phocomelia is a rare genetic condition which is either due to spontaneous mutation or autosomal recessive trait. There is 25% of chance for the child to be affected if parents are found to be carriers. History of consanguinity has an important role in making diagnosis of sporadic phocomelia (Tia and Al-Ghafri, 2018).

4. CONCLUSION

Fetal ultrasound is into practice all over the world for last four to five decades. Antenatal ultrasound remains the hallmark for detecting all fetal anomalies including limb, thus by increasing knowledge and differentiation of limb defects few of the major limb defects can be visualised or suspected at time of evaluation of nuchal translucency and majority of it during anomaly scan and when detected before the period of viability, termination of pregnancy can be advice.

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Authors' contribution

Conceptualization, supervision, methodology, resources, data collection, writing and formal analysis: Baskar A and Murugan G.
Writing, investigation, resources, analysis, draft preparation, review and editing: Asokan Aradhana Shanmughan.
Writing, investigation, analysis, review and editing: Abhi Amit Shah.
All authors have read and agreed to submit the manuscript.

Informed consent

Written informed consent was obtained from the patients.

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Conflict of interest

The authors declare that there is no conflict of interests.

Data and materials availability

All data sets collected during this study are available upon reasonable request from the corresponding author.

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