ejpmr, 2022,9(1), 620-623

SJIF Impact Factor 6.222

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Report

ISSN 2394-3211

EJPMR

CASE SERIES ON RARE AND LETHAL CONGENITAL ANOMALY: PRENATAL DIAGNOSIS OF LIMB BODY WALL COMPLEX

Vivek Kumar Garg[1], Manjula Sharma*[2], Monika Karpa[3] and Tanu Verma[4]

[1]Department of Radiodiagnosis, NSCB Zonal Hospital Mandi, Himachal Pradesh, India.
[2]Medical Officer, Civil Hospital, Sundernagar, Himachal Pradesh, India.
[3,4]Department of Gynaecology, NSCB Zonal Hospital Mandi, Himachal Pradesh, India.

***Corresponding Author: Manjula Sharma**

Medical Officer, Civil Hospital, Sundernagar, Himachal Pradesh, India.

Article Received on 21/10/2021 **Article Revised on 11/11/2021** **Article Accepted on 01/01/2022**

ABSTRACT

Limb body wall complex (LBWC) is one of the rare clinicopathological entity in which an abdominal wall defect with limb defect is major fetal anomaly. We report case series of two cases of sonographically diagnosed LBWC. The parents opted for termination of pregnancy. Physical examination of the two abortus revealed a large abdominal and thoracic wall defect with contents protruding from the large ventral defect, fetuses being adherent to placenta. Early sonographic diagnosis of this anomaly is critical so that parents can make informed decision about continuation or termination of pregnancy well with in time. Therefore, expertise in prenatal detection of this congenital anomaly is of utmost importance for better antenatal care.

KEYWORDS: Limb body wall complex, early sonography, congenital anomaly.

INTRODUCTION

Limb body wall complex(LBWC) also known as body stalk syndrome is a rare and lethal congenital anomaly with a prevalence between 1 in 14,000 to 1 in 42,000.[1,2] Criteria for diagnosis of LBWC, also known as Van allen et al,[3] criteria has three major criteria, out of which least at two to should be present.

1. Exencephaly/ encephalocele and facial cleft
2. Thoraco- and/or abdominoschisis
3. Limb defects

Two adhesion phenotypes have been identified, "placentocranial" and "placentoadhesion" type. LWBC is also known as "body stalk syndrome". This lethal malformation has no sex or familial predilection and is invariably fatal.[4] Due to extremely lethal nature of this malformation, early antenatal diagnosis is of utmost importance to enable the patient to decide on the options of continuation or termination of pregnancy. Prenatal sonographical screening is the imaging modality of choice used in the diagnosis of LBWC. We report a fetus at 20 weeks of gestation with features of LBWC diagnosed at Department of Radiodiagnosis at NSCB Zonal Hospital Mandi.

CASE REPORT

A 24 years old primigravida, presented for routine level II ultrasound examination in the Department of Radiodiagnosis. She had no known medical condition. No familial history of genet predilections. No history herbal or non-prescribed allopathic drugs intake was noted. No history of alcohol or tobacco intake during the duration of pregnancy was noted. Antenatal ultrasound showed normal fetal head displaced towards the lower uterine segment, in close approximation to the placenta. Fetal thorax appeared compressed and embedded in the placenta. Fetal abdomen and limbs could not be distinguished from placenta. A large amniotic cavity was noted with single largest pocket measuring > 10 cm (Fig.1, 2) Colour doppler showed single umbilical artery. No anomaly was detected in the eyes, palate, lips, face. Provisional diagnosis of LBWC was made. The patient was referred back to Department of Gynaecology and Obstetrics for management of LBWC. After counselling the patient about lethality of LBWC, final informed decision was made by the patient to terminated the pregnancy. Post abortion, delivered foetus showed, a large thoraco-abdominal defect with herniation of internal contents through the defect. The thoraco-abdominal portion was in very close approximation to the placenta. Further investigation showed right club foot (Fig 3,4,5&6).

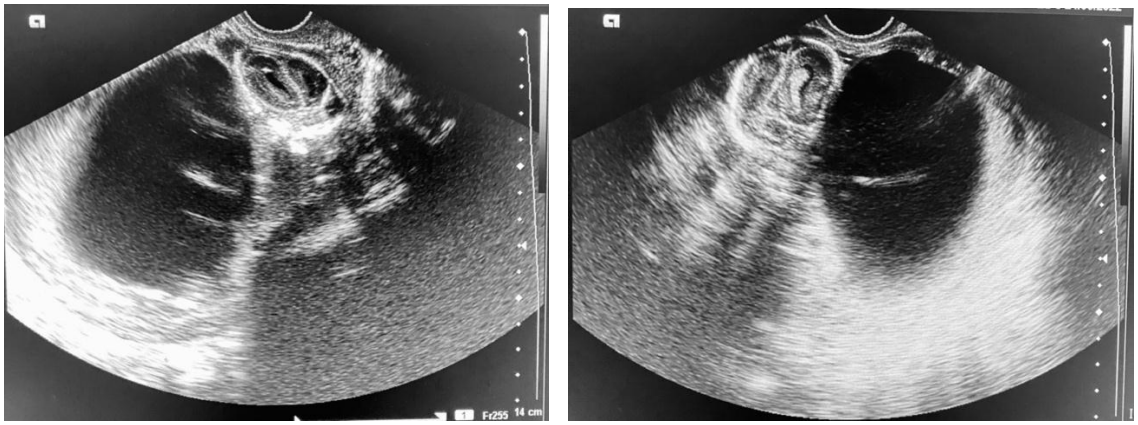

Figure 1,2: Antenatal ultrasound showing fetal head and compressed thorax in close approximation to the placenta with large amniotic sac with polyhydramnios.

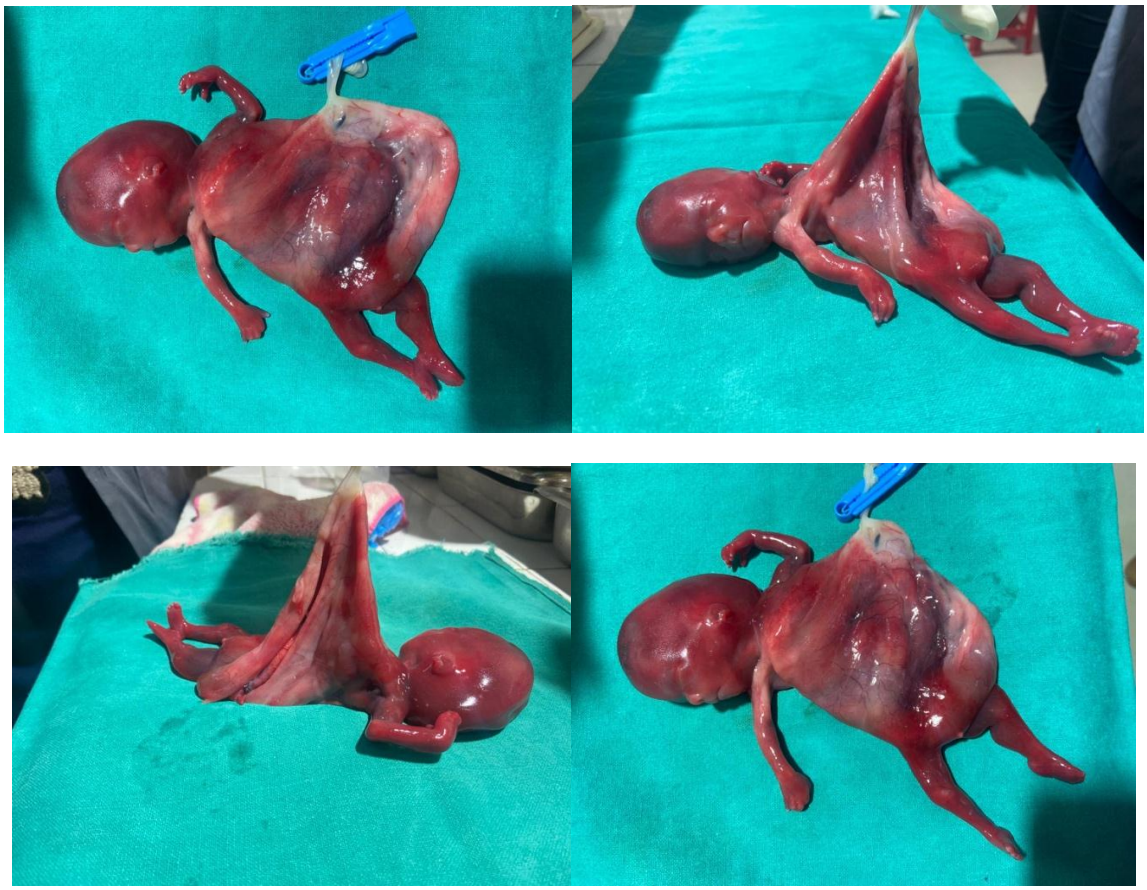

Figure 3,4,5&6: Abortus showing a large ventral thoraco-abdominal defect with herniation of bowel loops with close approximation of abdomen to the placenta and right club foot.

Case 2: A 22 years old multigravida with 6 months of amenorrhoea was referred from department of gynecology and obstetrics to the Radiology for routine antenatal ultrasound scan. She had no known medical condition. No familial history of genet predilections. No history herbal or non-prescribed allopathic drugs intake was noted. No history of alcohol or tobacco intake during the duration of pregnancy was noted. Furthermore, antenatal ultrasound revealed a large abdominal wall defect with herniation of bowel loops and liver through the defect. Spinal dysraphism was also noted. Colour doppler revealed single umbilical artery. No anomaly was seen in head and thoracic region. The female was counselled about the poor prognosis and fatal outcome of the foetus and pregnancy was terminated by the obstetrician. The findings were confirmed after delivery by the obstetrician

DISCUSSION

LWBC also known as " body stalk" anomaly is a sporadic and lethal anomaly with a prevalence between 1 in 14,000 to 1 in 42,000.[1,2] The incidence at birth is, however, lesser and it is about 0.2–0.3/100,000 live births because the majority of the affected fetuses

undergo intrauterine deaths.[4] Criteria for diagnosis of LBWC, also known as Van allen et al,[3] criteria has three major criteria, out of which least at two to should be present.

1. Exencephaly/ encephalocele and facial cleft
2. Thoraco- and/or abdominoschisis
3. Limb defects

The exact etiology of this condition is still unclear, Tropin's amniotic band theory, and Van Allen's vascular theory failed to explain all the anomalies in LBWC.

Tropin's amniotic band theory suggested that once the amniotic sac is ruptured and fetus lies outside amniotic cavity, bands extending from chorionic side of placenta entraps various fetal body parts and disrupt normal fetal development. Period of gestation at which amniotic bands develop determines the severity of this condition. Early rupture of the amniotic sac would lead to severe craniofacial, abdominothoracic and limb deformities, whereas late rupture would lead to milder defects. Few other authors.[5] three pathogenetic mechanisms which are; abnormal folding of the trilaminal embryo during the first 4 weeks of development, early amnion rupture with amniotic band syndrome, and early generalized compromise of embryonic blood flow. The anomaly can be classified into two based on the malformation phenotypes. Type I is based on craniofacial defect whereas Type II is recognized based on the ventral wall defects.[5] Van Allen proposed vascular disruption theory (VDT) and suggests that multiple congenital malformations may generally result from vascular disruption. Vascular disruption is described as events that negatively influence normal embryonic blood supply during embryogenesis, thereby interrupting normal morphogenesis, or destroying previously existing structures. VDT is supported by experimental animal studies. Puncturing the amniotic cavity or ligating umbilical blood vessels have produced LBWC. In amnion puncture experiments, up to one-third of the total amniotic fluid was withdrawn, thereby greatly influencing fetoplacental circulation. Anomalies not explainable with VDT are anencephaly, absent orbit (one sided), oblique facial cleft, deformed nose (single nostril), rocker bottom feet, pulmonary hypoplasia, unlobulated liver, abnormal number of umbilical vessels, cardiac defect (Tetralogy of Fallot), skin tag, and facial dysmorphism. Usually, foetuses with LBWC have normal karyotype with a very low chance of recurrence in subsequent pregnancies.[6] Some risk factors have, however, been reported for the development of LBWC and these include tobacco, cocaine, nicotine or marijuana use, or abuse.[7] The condition is said to affect both sexes but is more common in males.[8] The diagnosis is usually made prenatally by ultrasound scan in the first half of pregnancy. As clearly indicated that LBWC is incompatible with life, it is important to diagnose the condition early in pregnancy to help the parents make an informed decision. Prenatal ultrasound scan in the second trimester does not only confirm the diagnosis but also differentiates it from the other nonlethal dysplasias such as gastroschisis, omphalocele, cloacal dystrophy, and urachal cyst as well as from other lethal poly-malformation complexes. Such complexes include pentalogy of Cantrell, omphalocele‐exstrophy‐imperforate anus‐spinal defects, amniotic band sequence, and omphalocele with cloacal–bladder exstrophy complex. Most of the pregnancies with LBWC end in miscarriages, termination of pregnancy or still birth. Leading cause of death is generally pulmonary dysplasia which leads to severe pulmonary hypertension and respiratory failure. Survival of foetus is almost rare, however literature reports a case where a foetus with LBWC survived for 84 days.[9]

CONCLUSION

LWBC is a rare and lethal congenital anomaly with most accepted theory of early embryonal dysplasia. An early antenatal diagnosis is extremely important in all pregnant patients, so as to counsel them for termination of pregnancy. It is important to differentiate this anomaly from more common gastroschisis and omphalocele as these conditions have favourable outcome in comparison to LWBC. Sonographic hallmarks of LWBC are large thoraco-abdominal defects with visceral herniation, neural tube defects, scoliosis and limb defects.

REFERENCE

1. Mann L, Ferguson-Smith MA, Desai M, Gibson AA, Raine PA. Prenatal assessment of anterior abdominal wall defects and their prognosis. Prenat Diagn, 1984 Dec; 4(6): 427–35.
2. Forrester MB, Merz RD. Epidemiology of abdominal wall defects, Hawaii, 1986-1997. Teratology, 1999 Sep; 60(3): 117–23.
3. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. Am J Med Genet, 1987 Nov; 28(3): 529–48.
4. Russo R, D'Armiento M, Angrisani P, Vecchione R. Limb body wall complex: a critical review and a nosological proposal. Am J Med Genet, 1993 Nov 1; 47(6): 893–900.
5. Daskalakis G, Sebire NJ, Jurkovic D, Snijders RJ, Nicolaides KH. Body stalk anomaly at 10-14 weeks of gestation. Ultrasound Obstet Gynecol Off J Int Soc Ultrasound Obstet Gynecol, 1997 Dec; 10(6): 416–8.
6. Sahinoglu Z, Uludogan M, Arik H, Aydin A, Kucukbas M, Bilgic R, et al. Prenatal ultrasonographical features of limb body wall complex: a review of etiopathogenesis and a new classification. Fetal Pediatr Pathol,2007 Jun; 26(3): 135–51.
7. Martinez JM, Fortuny A, Comas C, Puerto B, Borrell A, Palacio M, et al. Body stalk anomaly associated with maternal cocaine abuse. Prenat Diagn, 1994; 14(8): 669–72.
8. Chen CP, Lin CJ, Chang TY, Hsu CY, Tzen CY, Wang W. Second-trimester diagnosis of limb-body

wall complex with literature review of pathogenesis. Genet Couns Geneva Switz, 2007; 18(1): 105–12.

9. Lazaroni TLDN, Cruzeiro PCF, Piçarro C, Victoria ÁM, Botelho Filho FM, Tatsuo ES, et al. Body stalk anomaly: Three months of survival. Case report and literature review. J Pediatr Surg Case Rep, 2016 Nov 1; 14: 22–5.