

Case Report

Limb body wall complex or body stalk complex or cylosomas: a case report

S. Saritha*, Gouri, Sumangala

Department of Anatomy, Medical College (KIMS), Hyderabad, Andhra Pradesh, India

Received: 20 February 2013

Accepted: 22 March 2013

***Correspondence:**

Dr. S. Saritha,

E-mail: kmr.saritha@gmail.com

© 2013 Saritha S et al. This is an open-access article distributed under the terms of the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.

ABSTRACT

Limb body wall complex (LBWC) is also called Body stalk complex and Cylosomas. We present this rare congenital malformation complex highlighting the importance of early sonographic imaging findings in LBWC along with differentiation from other anterior abdominal wall defects. Limb body wall complex / Body stalk anomaly refers to a rare complicated polymalformative fetal malformation syndrome of uncertain etiology and results in head, heart, lung, diaphragm, kidney or gonadal abnormalities. LBWC was first described by Van Allen et al; in (1987). The two of the three following anomalies must be present to establish the diagnosis: 1. Exencephaly / Encephalocele with facial clefts, 2. Thoraco-Abdominoschisis / ventral body wall defects and 3. Limb defects. LBWC arises as a result of early amnion disruptions or error in embryonic development. If all components of the syndrome are present, the condition is lethal. LBWC is invariably fatal and incompatible with life. No case of postnatal survival is reported so far. Serum alpha-fetoprotein measurement and ultrasonography examination is the key to the prenatal diagnosis and followed by medical termination of pregnancy. It presents two distinct phenotypes described by Russo et al (1993) and later Cusi et al in (1996), according to the foetoplacental relationships: 1. Placento-cranial and 2. Placento-abdominal types. Among the 168 live births at S.V.S. Medical College & hospital Mahabubnagar (INDIA) during the period of 2010-2011 we came across an aborted female fetus. It was weighing 1800gms, 30 weeks of gestation diagnosed by antenatal ultrasonography as ventral body wall defect. It was associated with omphalocele, severe scoliosis and limb defects. Its confirmation of the diagnosis of Limb body wall complex with Placento-abdominal type was done by postmortem fetography.

Keywords: Limb body wall complex (LBWC), Ultrasonography, Abdominal defects, Fetal malformations

INTRODUCTION

The LBWC pathogenesis is probably heterogeneous. Mechanisms included germ disc disruption, vascular disruption and amniotic disruption. LBWC refers to a rare combination of disruptive and lethal abnormalities which starts early in the gestational process. It is characterized by the presence of an abdominal wall defect associated with variable spectrum of limb and visceral anomalies. Russo et al² have proposed two clearly

distinguishable phenotypes: the "Placento-cranial" and "Placento-abdominal" adhesion phenotype. The first phenotype shows craniofacial defects and amniotic bands while the second phenotype is without craniofacial defects and presents urogenital anomalies, anal atresia and abdominal placental attachment with a persistence of the extra-embryonic coelom. LBWC has no sex or familial predilection or known recurrence risk. Karyotype study has been reported to be normal in all LBWC. The present case was diagnosed by prenatal ultrasonography

and later followed medical termination of pregnancy. The aborted female fetus 30 weeks old, diagnosed as LBWC of placento-abdominal type with several unusual features. In our index case, apart from thoracoabdominosis and limb defects presents an interesting feature the two lobes of liver are separated by the stomach and duodenum.

CASE REPORT

A 22 years old primi with 32 weeks of gestation presented to S.V.S. Hospital Mahabubnagar (A.P. INDIA) came for routine antenatal checkup. Ultrasonography scanning was done (Figure 2). She was G1POA0, at the time of scanning. She gave a history of non- consanguineous marriage, did not take any drugs or chronic medication. Ultrasound scans revealed live female fetus with 30 weeks of gestation. The scan also revealed a large abdominal wall defect through which abdominal contents herniated into the extra embryonic coelom, with marked scoliosis and limb defects. The patient was informed of poor prognosis and after counseling she was selected for termination of the pregnancy. Subsequently she aborted female fetus completely along with placenta and membranes. The diagnosis of LBWC was confirmed after delivery and detailed autopsy was done in Anatomy dissection hall.



Figure 1: Limb body wall complex (LBWC).



Figure 2: Ultrasonography scan.

Ultrasonography (Figure 2)

- Live female fetus corresponding to 30 weeks gestation.
- BPD 74 mm- corresponding to 30 weeks.
- Femur length- 58 mm corresponding to 30 weeks- 3 days.
- Head circumference- 268 mm corresponding to 29weeks- 2 days.
- Abdominal circumference- 216 mm corresponding to 26 weeks.
- Marked scoliosis of the spine.
- Left upper limb was shorter than the right upper limb.
- Malformation of fetal abdominal viscera and abdominal organs with evidence of exomphalous.
- Placenta- posterior, grade II- upper uterine segment.
- Weight of the fetus- 1185 gms approximately.

External features

The present female fetus with LBWC showed placento-abdominal attachment. It revealed large ventral body wall defect through which abdominal contents herniated (Figure 1). The eviscerated organs formed a complex mass covered with membranes. The umbilical cord was short adherent to the eviscerated mass. On clear examination herniated mass included left lung, liver, stomach, spleen, small intestine and large intestine. The abdominal circumference was markedly diminished (Figure 9). No anomalies were seen at the cranial region. Facial profile, fetal eyes and ears, palate and lips, and neck region was normal (Figure 3). Right hand with syndactyly between 3rd and 4th fingers (Figure 4). Left shoulder joint hyperextended, forearm short and with only two digits (Figure 5). The left great toe was small with constriction and right foot showed syndactyly of 4th and 5th toes (Figure 6). Back of the trunk showed marked scoliosis towards the right lateral side and highly prominent nuchal region (Figure 7, 8). The external genitalia were ambiguous with small anal orifice (Figure 10).

Internal features

Brain was soft. No cardiac anomalies found (Figure 12). Right lung normal, within the thoracic cavity but left lung hypoplastic and present in the extra embryonic coelom (Figure 11). Diaphragm was absent.

The intra-abdominal anatomy was in apparent disarray. While the fetal stomach and urinary bladder were readily visualized, the exact location of remaining anatomy (i.e. kidney, liver, and bowel) could not be established.

Abdomen showed evidence of malrotation of stomach (Figure 13&14). The two lobes of liver separated by stomach and duodenum (Figure 13&14). Spleen was normal, small intestine and large intestine filled with

meconium present outside the abdominal cavity and covered by membranes (Figure 14).

Kidneys were large and malrotated, ureters inserted laterally into the kidney and urinary bladder was normal

(Figure 15). There was partial lack of pelvic organs and perineum, with presence of one ovary, one fallopian tube on the left side and uterus rotated to the left (Figure 16). Plain radiographs showed marked scoliosis and limb defects (Figure 8).



Figure 3



Figure 4



Figure 5



Figure 6



Figure 7

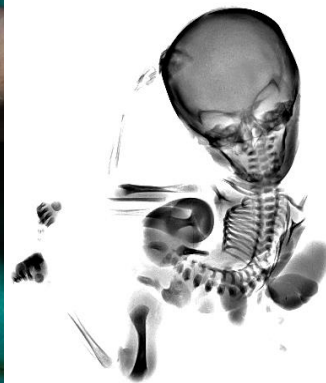


Figure 8



Figure 9



Figure 10

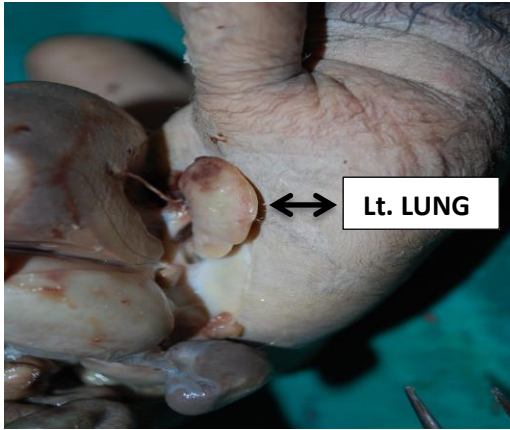


Figure 11

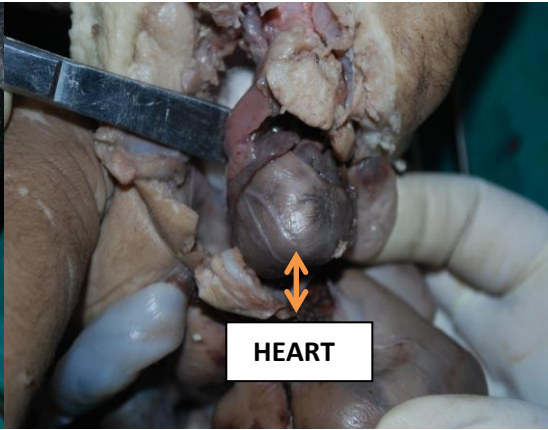


Figure 12

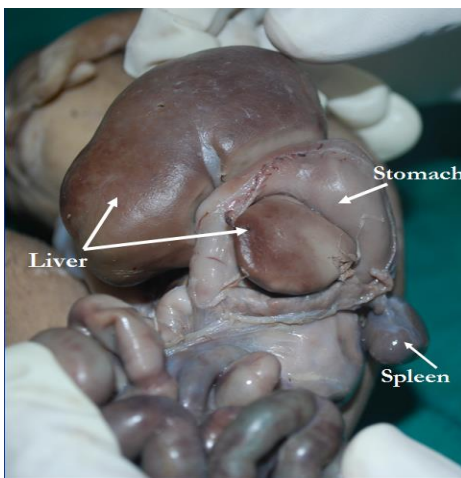


Figure 13

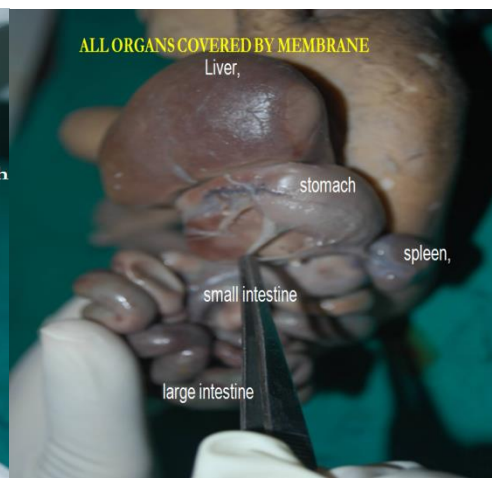


Figure 14

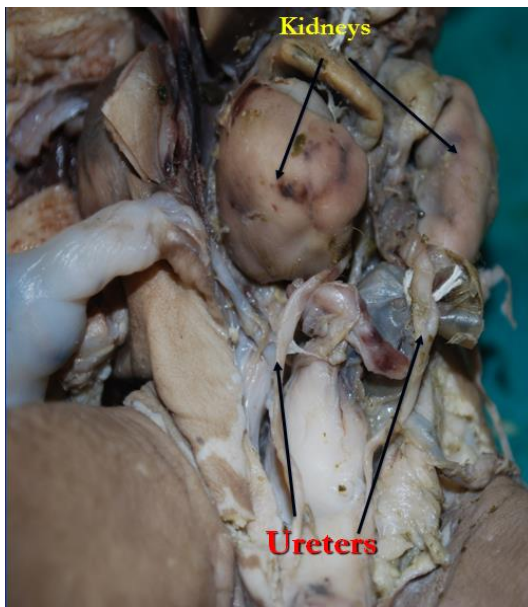


Figure 15: Kidneys and Uterus.

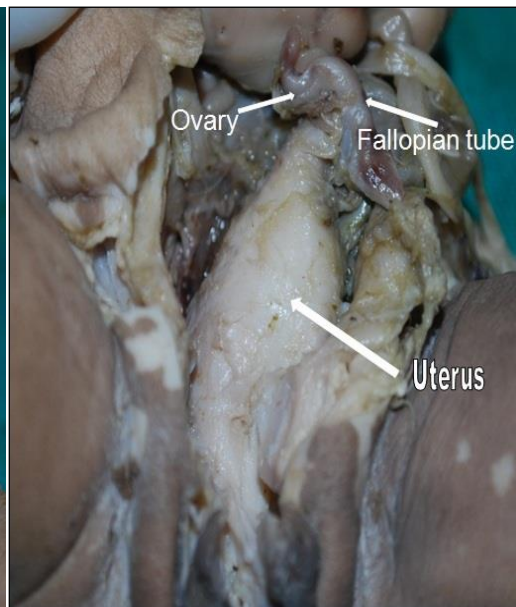


Figure 16: Uterus, Ovary and Fallopian tube.

DISCUSSION

Congenital malformations of the ventral abdominal wall occur in many forms, ranging from exomphalous to gastroschisis to more complexes, such as Pentalogy of Cantrell and LBWC. Limb body wall complex is an entity characterized by the presence of an abdominal wall defect associated with variable spectrum of visceral and limb anomalies. The diagnostic criteria for LBWC are still being discussed, but the most commonly quoted are those originally set forth by Van Allen et al in 1987.¹ The presence of two of the following three malformations: 1. Exencephaly/Encephalocele with facial clefts; (2) Thoraco and/or Abdominoschisis; (3) Limb defects.

Coelosomia is found in all cases, its variable coexists with encephalic, visceral or limb anomalies. LBWC results from defect in early embryonic folding process. The chance of recurrence in the next pregnancy is very low. This congenital anomaly has no sex or family predilection. LBWC in association with other malformations makes the prognosis poor.

Two main phenotypes have been described in the literature (Russo et al 1993)²:

1. *Fetuses with craniofacial defects (placento-cranial)*- whose characteristics are: (a) Encephalocele or exencephaly always associated with facial clefts and (b) Amniotic bands b/w the cranial defects and placenta.
2. *Fetuses without craniofacial defects (placento-abdominal)*- they often presents with urogenital anomalies, anal atresia, lumbosacral meningocele and placental anomalies like presence of short cord, persistence of extra embryonic coelom and intact amnion.

Pathogenesis of LBWC is unclear and uncertain. Three pathogenic theories have been proposed:

1. Early amnion rupture theory (Exogenic theory) leading to the formation of amniotic bands that interrupt embryogenesis and the fetus lies outside the amniotic cavity. It disturbs the normal development (Torpin et al, 1965).⁴
2. Vascular disruption theory (Endogenous theory) is described as events that negatively influence normal embryonic blood supply, thereby interrupting normal morphogenesis (Van Allan, 1987).¹
3. Embryonic dysgenesis or Germ disc defect with early embryonic maldevelopment. The fetal disruptions resulted from imperfect histogenesis, showing disturbances of the embryonic folding process (Streeter theory 1930).⁵

The placento-cranial phenotype is related to early vascular disruption, while placento-abdominal phenotype is attributable to embryonic maldevelopment.

The present case has the characteristics of the placento-abdominal phenotype, seems to favor with embryonic maldevelopment i.e. disturbances of the embryonic folding process, presenting with ventral body wall defect and limb anomalies (Hartwig et al 1989).⁵

The predominant feature of sonographic findings is evisceration of the abdominal contents. The eviscerated organs formed a complex mass covered with membranes floating in the persistent extra embryonic coelom. The umbilical cord was short adherent to the eviscerated mass. The herniated organs included are left lung, liver, stomach, spleen, small intestine and large intestine. The urinary anomalies are malrotated kidneys and genital abnormalities are one ovary, one fallopian tube on the left side and small rotated uterus was also present. The abdominal circumference was markedly diminished.

The internal malformations of the urinary and genital systems are usually associated with LBWC. They suggest abnormal mesodermal development. The placodes related to abnormal embryonic folding add cells to the intermediate mesoderm that forms the urogenital system in the trilaminar disc (Hartwig et al 1989).⁶

H. Saadi, K. Sfakianoudis, D. Thomas (2007)⁷ in their study reported anterior wall defect with herniation of the abdominal contents, skeletal defects in the upper limb and kyphoscoliosis.

Martinez-Frias⁸ determined the presence of body wall defects with evisceration of thoracic and /or abdominal organs and other congenital abnormalities with or without limb deficiencies is considered to be the LBWC. The present study is in agreement with above authors because it also includes anterior body wall defects with evisceration of thoracic and abdominal contents, limb defects and severe scoliosis with prominent nuchal region.

Epidemiology of the LBWC syndrome is not always recognized. Its incidence is difficult to estimate. Kurosawa et al⁹ estimated the incidence in 0.21 to 0.31 cases per 10,000 births. He found no connection to fetal sex, parental age or any associated genetic anomalies. Luer et al¹⁰ reported higher incidence 3.3:10,000 births and presented series of cases with teratogenic causes, means environmental factors of natural or chemical origin may have come into contact with during pregnancy (tobacco, alcohol and certain drugs). There have been research studies done which suggest a connection with anti-convulsant drugs with the LBWC. Further evidence of cocaine teratogenicity is also associated with the LBWC (Viscarello et al 1992).¹¹ In this present case, it was found that karyotype was normal, 46xx. As reported by Van Allen et al¹ and Negishi et al¹², the etiology was unknown. No teratogen has been implicated. No genetic abnormality and familial recurrence has been reported. The pathogenesis of malformations related to LBWC of the present case was unknown.

CONCLUSION

LBWC is a syndrome with multiple malformations and with very poor prognosis. Pregnancy should be terminated on establishing correct diagnosis which requires careful ultrasonographic examination of the fetus whenever ventral body wall defect with other malformations is suspected. Exomphalous requires chromosomal analysis (seen in trisomies 13, 18 & 21) and isolated gastroschisis is the most benign of all. Thus it is important to distinguish between these entities, as it will influence the choice of the treatment. It is my hope that this case will stimulate lively discussion and give all of us an opportunity to learn something new about these rare disorders.

REFERENCES

1. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. *Am J Med Genet* 1987;28:529-48.
2. 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;47:893-900.
3. Cusí V, Torrents M, Vila J, Antich J, Carrera JM. Limb body wall complex: analysis of eight fetuses. *Birth Defects Orig Artic Ser* 1996;30:165-70.
4. Torpin R. Amniochorionic mesoblastic fibrous strings and amniotic bands. *Am J Obstet Gynecol* 1965;91:65-75.
5. Streeter GL. Focal deficiency in fetal tissues and their relation to intrauterine amputation. *Contrib Embryol* 1930;22:41.
6. Hartwig NG, Vermeij-Keers CHR, De Vries HE, Kagie M, Kragt H. Limb body wall malformation complex: an embryonic etiology? *Hum Pathol* 1989;20:1071-7.
7. Saadi H, Sfakianoudis K, Thomas D. Limb body wall complex associated with placenta previa accreta. Available at <http://www.sonoworld.com/fetus/page.aspx?id=2420>. Accessed 25 January 2013.
8. Martinez-Frias ML. Clinical and epidemiological characteristics of infants with body wall Complex with and without limb deficiency. *Am J Med Genet* 1997;73:170-5.
9. Kurosawa K, Imaizumi K, Masuno M, Kuroki Y. Epidemiology of limb body wall complex in Japan. *Am J Med Genet* 1994;51:143-6.
10. Luer B, Lipsett J, Quinlvan JA. Limb-body wall complex: A case series. *J Mater Fetal Neonatal Med* 2002;12:132.
11. Viscarello RR, Ferguson DD, Nores J, Hobbins JC. Limb-body wall complex associated with cocaine abuse: further evidence of cocaine's teratogenicity. *Obstet Gynecol* 1992;80(3 Pt 2):523-6.
12. Negishi H, Yaegashi M, Kato EH, Yamada H, Okuyama K, Fujimoto S. Prenatal diagnosis of limb-body wall complex. *J Reprod Med* 1998;43:659-64.

DOI: 10.5455/2320-6012.ijrms20130519

Cite this article as: Saritha S, Gouri, Sumangala. Limb body wall complex or body stalk complex or cyllomas: a case report. *Int J Res Med Sci* 2013;1:132-7.