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RESEARCH ARTICLE

A CASES OF LIMB BODY WALL SYNDROME (LBW): REPORT AND LITERATUREREVIEW

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Abstract

Limb body wall syndrome (LBW) is one of the rarest malformation complexes. It is also known by other names as « Abnormal stem of the body » « Congenital absence of the umbilical cord » and « cyllosome and Pleurosomus » [1]. This complex is a heterogeneous disease characterized by multiple severe congenital abnormalities of the fetus with exencephalon/thoraco-encephalo and/or abdominoschisis (anterior parietal malformations) and malformations of the limbs, with or without facial slits. The diagnosis remains difficult to establish in view of the clinical polymorphism the prenatal ultrasound can detect this abnormality during the first trimester. The exact etiology of this condition is still uncertain, the theory of amniotic bands of Tropin and the vascular theory (early vascular accident) of Van Allen have not succeeded in explaining all the abnormalities observed in the LBWC [2]. Karyotype study are normal and there has been no evidence of correlation with sex, age of parents or teratogenic agents. Prognosis is fatal death occurring in antenatal or early neonatal period. We are describing two cases of Limb body wall syndrome (LBW).

Introduction:-

Limb body wall syndrome (LBW) is one of the rarest deadly polymalformative syndrome. This complex is a heterogeneous disease characterized by multiple severe congenital abnormalities of the fetus.

The diagnosis remains difficult to establish in view of the clinical polymorphism and the prenatal ultrasound can detect this abnormality during the first trimester. The exact etiology of this condition is still uncertain.Karyotypes are normal and there has been no evidence of correlation with sex, age of parents or teratogenic agents.Prognosis is fatal death,We are describing two cases of Limb body wall syndrome (LBW).

Case report 1:-

S.A : A 19-year-old woman with 1 para 1 gravida, married to a 34-year-old man. The couple is in good health, related to the second cousin's relationship with a history of taking mushrooms during the first trimester and no other medical history was found.

It was sent to our department for suspicion of fetal malformations on a pregnancy estimated at 20 WA according to the date of the last menstruation on 22/06/2021.

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Ultrasound shows active monofetal pregnancy with irregular positive cardiac activity, cross-sectional presentation, placenta and background fluid were in normal amount the malformative evaluation found a huge occipital brain with problem of all intracranial content

Associated with the externalisation of the liver, stomach and small intestine by the defect of the abdominal wall Members without detectable abnormalities at the limits of the exam. The vaginal delivery took place without incident giving birth to an abnormal newborn , Apgar 02/10 with a birth weight of 1400 g.

The clinical examination revealed : Omphalocel caused by a hernia of the abdominal wall with which the intestines spread freely at the base of the umbilical cord.(Fugure 1). Abnormal insertion to the right of the umbilical cord.

Abnormal morpholigia of axial skelton with deformed rib cage and enlarged cervical tendril whose closure is incomplete probably suspecting of spina bifida complete (figure2) sex indeterminate , there is also a microcephaly associated.

Case report 2:-
DK : A 20-year-old woman with 1 para 1 gravida, married to a 30-year-old man. The couple is in good health, no medical history has been found , GAJ negative serology negative. It was sent to our department for suspicion of fetal malformations on a pregnancy estimated at 23 +6 WA according to the date of the last menstruation on 25/05/2021

Ultrasound reveals active monofetal pregnancy with positive cardiac activity ,Fetus in transverse presentation placenta background excess amniotic fluid.

Assessment of malformations : spinal column with major angulation in the dorsal regionUpper and lower celeosomy (externalisation of the heart and liver) Club foot varus

The vaginal delivery took place without any incident giving birth to an abnormal newborn male , Apgar 02/10 with a birth weight of 1600 g.

The clinical examination found :-
A huge defect of the thoracoabdominal wall with congenital protrusion of the abdominal and thoracic viscera and gastroschisis with a total absence of ribs, sternum and closure of the abdominal wall (figure 3/4)

An abnormally inserted umbilical cord on the abdominal wall defect .

The evolution of the 2 cases was marked by a cardio-respiratory arrest after few min of life with in common.

Discussion:-

LBWC was first described by Van Allen et al. In 1987[2]. It is also known by other names as « Abnormal stem of the body » « Congenital absence of the umbilical cord » and « cyllosome and Pleurosomus » [1].

The complex (limb body wall) (LBWC) is characterized by multiple severe congenital abnormalities of the fetus with exencephalon/thoraco-encephalo and/or abdominoschisis (anterior parietal malformations) and malformations of the limbs, with or without facial slits.

The diagnostic criteria for LBWC are always questionable.

Traditionally, the diagnosis was based on the criteria of Van Allen et al. [2], namely the presence of two out of three of the following abnormalities :

1. Exencephaly or encephalocele with facial slits.
2. Thoraco and or abdominoschisis.
3. Anomalies of members.

Classification:-
Russo et al. In 1993[3] identified two distinct phenotypes :

1. Type I (cranio-facial defects, facial slits, amniotic adhesions and amniotic band syndrome)

2. Type II (absence of cranio-facial defects, urogenital abnormalities, anus imperforation, lumbosacral meningomyelocele, severe kyphoscolosis and placental abnormalities)

In 2007, Sahinoglu et al. [4] proposed a new classification that was established :

1. Type I (cranio-facial defect + intact thoraco-abdominal wall, placenta and umbilical cord often normal but rarely attached to malformed cranial structures)
2. Type II (The large abnormality of the chest-abdominal wall usually located laterally and abdominal organs wrapped in the amniotic leaf that connect to the cutaneous edge of the wall, without well formed umbilical cord and without normal cloacal structures).
3. Type III (Fetus has an abnormality of the infra-digital abdominal wall with intact chest. The placenta is widely attached to the skin at the site of the lesion The abdominal organs are dissected in the extra-embryonic coelomic cavity Cloacal structures are almost always malformed or absent.

Pathophysiology :-

The exact etiology of this condition is still uncertain, the theory of amniotic bands of Tropin and the vascular theory (early vascular accident) of Van Allen have not succeeded in explaining all the abnormalities observed in the LBWC [2].

The most accepted theory is early embryonic dysplasia presented by Hartwig et al. In 1989.

This leads to defective closure of the embryonic abdominal wall, umbilical abnormality and the persistence of an extra-embryonic coelom communicating with the abdominal cavity.

Karyotypes are normal and there has been no evidence of correlation with sex, age of parents or teratogenic agents.

Diagnosis :

LBWC is a heterogeneous disease and is associated with various internal abnormalities which are cited :

1. The central nervous system abnormalities observed are anencephaly, encephalocele and holoprosephalyalobaire.
2. Cardiovascular abnormalities include the primary ventricle, the common atrium, malformations of intraauricular communication, arterial trunk, membrane ventricular septal communications, hypoplastic right ventricle and ectopic heart disease.
3. Renal abnormalities observed are unilateral or bilateral aplasia/hypoplasia of the kidney, hydronephrosis, renal dysplasia, polycystic kidney and calcification of the kidney.
4. Genital abnormalities observed are abnormal external genitalia, absence of gonads and bladder extrophy.
5. Skeletal abnormalities are most common and include clubfoot, oligodactyly, arthrogryposis, absent limb, forearm bone, leg bone, pesudosyndactyly, radial/cubital hypoplasia, rotational malformations and polydactylia.

Other abnormalities include trilobulated liver, polysplenia, absent gallbladder, amniotic bands and single umbilical artery. Complex cranial and abdominal abnormalities have been observed that are not observed in the literature [6].

The diagnosis of this condition can be established by measuring the serum level of the mother's alpha feto protein.

Prenatal ultrasound (USG) can detect this abnormality in the first trimester.

Early antenatal ultrasound is possible and can be followed by early termination of pregnancy.

Ultrasound may show a large defect of the abdominal wall with the fetus adhering to the placenta, the insertion site of the umbilical cord is difficult to find or is absent and there is also a direct affixing of the membrane sac to the amniochorionic membrane.

Accompanying cyphoscoliosis/scoliosis may also be detected.

Early scintigraphy in the first trimester may also reveal an increase in nuchal clarity.

LBWC is a lethal anomaly, therefore it must be differentiated from treatable causes.

Prognosis :

is fatal death occurring in antenatal or early neonatal period.

Case report 1:-

Figure 1:-

Figure 2:-

Case report 2:-

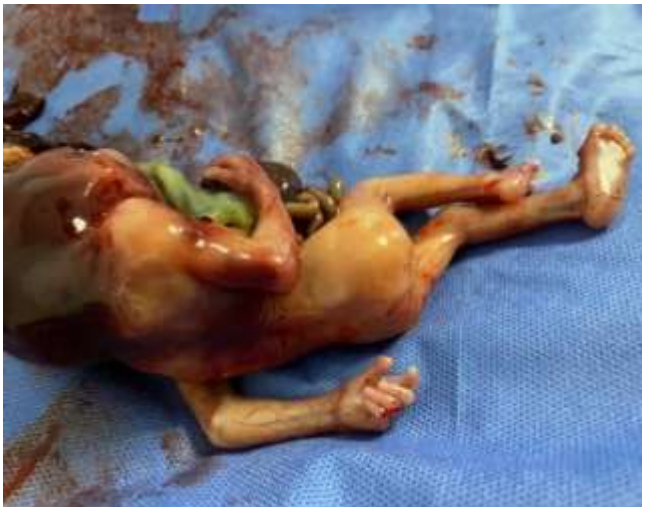

Figure 3:-

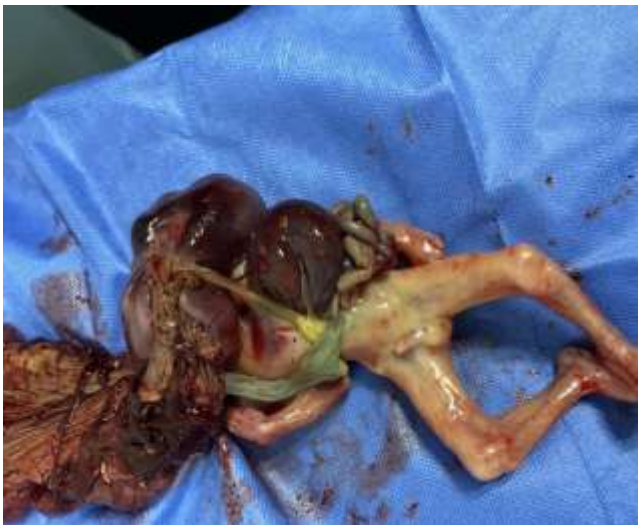

Figure 4:-

Conclusion:-

LBWC is a deadly polymalformative syndrome, which presents a rare combination of cranial and abdominal abnormalities

Antenatal ultrasound scans performed during pregnancy allow to detect deficiencies, and detect abnormalities.

References:-

1. Borade A, Prabhu AS, Prabhu GS, Prabhu SR. Limb Body Wall Complex (LBWC) [Last cited on 2009 Jul PediatricOncall. 2009 6 Art # 37. Available from: http://www.pediatriconcall.com/fordoctor/casereports/limb.asp .
2. Van Allen MI, Curry C, Gallagher L. Limb body wall complex: I. Pathogenesis. Am J Med Genet. 1987;28:529–48. [PubMed]
3. 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. [PubMed]
4. 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 PediatrPathol. 2007;26:135–51. [PubMed]
5. Hartwig NG, Vermeij-Keers C, De Vries HE, Kagie M, Kragt H. Limb body wall malformation complex: An embryologic etiology? Hum Pathol. 1989;20:1071–7. [PubMed]
6. Socolov D, Terinte C, Gorduza V, Socolov R, Puiu JM. Limb body wall complex-case presentation and literature review. Rom J Leg Med. 2009;17:133–8.