Limb Body Wall Complex - case presentation and literature review

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Abstract: Limb Body Wall Complex (LBWC) is a combination of development abnormalities involving the neural tube, body wall and the limbs. There are few cases in literature, and our case is only the 2nd presented from Romania. The patient was a 31 year-old women G1P0A0 with a 33 week pregnancy which had no prenatal care. The ultrasound scan described several abnormalities, including: large abdominal wall defect, with difficult to identify pelvic organs and ambiguous genitalia; enlarged stomach with suspicion of intestinal atresia; scoliosis and spida bifida occulta with bilateral ventriculomegalia; one inferior limb absent; short umbilical cord with single artery. After therapeutic termination of pregnancy, the abnormalities were confirmed and polycystic liver and kidneys were also mentioned. Also bilateral cardiac ventriculomegalia and left superior pulmonary lobe hemorrhage were identified, and imperforated anus and pancreas agenesia. No abnormalities were found at karyotype examination (46, XY). The case presented is a placento-caudal phenotype of a LBWC syndrome, which had as a special element the polycystic kidney and hepatic disease.

Key words: Limb Body Wall Complex, neural tube, polycystic liver polycystic kidneys, karyotype

Limb Body Wall Complex (LBWC) is a fetal malformative syndrome which consists of neural tube defects, body wall disruption and limb abnormalities. The diagnosis is made by at least two of the above features [1, 2, 3]. The authors report a case of LBWC diagnosed in the late antenatal period. It is the 2nd case reported in Romania [4].

The particularity of this case consists in the association of the LBWC malformations with the polycystic disease of the liver and kidneys, not reported before, to our knowledge.

Case presentation
A 31 year old woman was referred for a 3rd level antenatal ultrasound because of a plurimalformative syndrome. She was G1P0A0, and she had 34 weeks gestational age at the time of the first scan. The ultrasound report showed a fetus of an estimate gestational age based on biometry of 36 weeks and 5 days (using BPD, HC, FL). The scan also revealed a large, ill defined abdominal wall defect through which the abdominal contents herniated into the extra embryonic celom (Fig. 1).
The protruded organs formed a complex with bizarre appearing and entangled with membranes. The diaphragm was intact. There was evidence of scoliosis and spina bifida occulta. The superior limbs were normal; the inferior left one presented club foot, while the right inferior limb was completely absent. The color flow Doppler showed evidence of a single umbilical artery with a short umbilical cord, and it also clearly designated the abdominal feto-placental attachment.

A provisional diagnosis of LBWC was made based on the ultrasound description. Several other abnormalities were described, which were later confirmed at the pathological exam results section.

The patient was informed of the poor prognosis and after counseling she accepted the termination of the pregnancy. Following the induction of labour, she delivered a stillborn fetus of 3000g.

The necropsy report confirmed the diagnosis of LBWC (Fig. 2) showing: herniation of the abdominal content through a large defect, exteriorisation of the liver and of the small and large bowel content, kidneys, stomach, cloaca, entangled with membranes, bilateral cardiac ventriculo-megalia with arterial duct persistence, polycystic liver, a type II polycystic kidney (the adult form) (Fig. 3, Fig. 4). One kidney was enlarged (4/6 cm), with a large pielon situated at its inferior pole; the other one was small (0.4 cm), localised on the surface of the big kidney.

The large bowel ended in a cul-de-sac manner, into a 3/4 cm pool with thick wall: the cloaca.

Other abnormalities were: pancreas agenesia, uncertain external genital organs, imperforated anus, lumbar neural tube defect-spina bifida occulta.
The microscopical examination described:
- multiple intrahepatic cysts (0.5-0.8 cm) with serosangvinolent content (Fig. 5);
- cysts of 0.8/1/1.5cm, disposed in the renal cortex and medulla, separated by a fibrous tissue;
- short umbilical cord (17 cm), with two vessels (one artery and one vein separated on a segment of 2 cm), which confirmed also the ultrasound description (Fig. 6).

The karyogram, prepared from the cord blood immediately after delivery was normal (karyotype 46XY).
**Literature review and case discussion**

Limb body wall complex (LBWC) is a rare, polimalformative fetal syndrome, appearing in 0.21-0.31/10000 deliveries, with only about 245 cases described in the literature until now [5, 6].


In general, the diagnosis is based on 2 of the above features.

There is no correlation with the fetal gender, parents’ age, or karyotype anomalies, as in our study too. The disease is invariably fatal [1, 7]

Serum alpha-fetoprotein measurement and ultrasonographic examination are the key of prenatal diagnosis. [8]

The ultrasound milestone of LBWC consists of thoraco-abdominal defect, spinal and cord abnormalities, positional limb deformities and abnormalities of umbilical cord and membranes. [7]

In this case report we highlight the ultrasound and colour Doppler flow findings in LBWC, demonstrated on routine antenatal scanning, along with the photographs of the still-born fetus and images of microscopic aspects.

The pathogenesis of LBWC is unclear. Four pathogenic mechanisms are proposed to be involved:

2. The early amnion rupture theory, discussed by Torpin in 1965 [9, 10, 11, 12]
3. The vascular disruption theory elaborated by von Allen [1, 13, 14].
4. The embryonic dysgenesis theory, proposed by Hartwig [3, 15, 16, 17]

Lately, different authors [18, 19] described the two different phenotypes, according to the placental attachment: placento-cranial and placento-caudal type.

1. the placento-cranial attachment, showing craniofacial defects and amniotic bands and/or adhesion; it seems to be related to early vascular disruption.
2. the placento-abdominal adhesion phenotypes presents urogenital anomalies, anal atresia, short umbilical cord, abdominal placental attachment and persistence of an extraembrionic celom. It seems to be due to an intrinsic embryonic maldevelopment.

Our case seems to be explained by the second mechanism.

**Differential diagnosis**

LBWC must be differentiated from abdominal wall defects classified according to their localisation: gastrochisis, localized in the paraumbilical area, omphalocele, localized in the umbilical area, ectopia cordis localized at the anterior face of the thorax, cloacal dystrophy -localized at the lower abdominal wall, urachal cyst, localized also at the lower anterior abdominal wall [6, 20, 21].

There are also some specific signs for LBWC: the presence of membranes covering the contents of the herniated sac, any associated bowel abnormalities, the presence or absence of urinary bladder, scoliosis and limb defects.
Conclusion

LBWC is a rare polimalformative syndrome possible to detect at an antenatal ultrasound. It can be divided in two phenotypes: placento-cranial, due to amniotic bands syndrome and placento-caudal, due to body stalk syndrome (short umbilical cord).

The syndrome is invariably fatal, but the couple must be reassured that there is no risk for recurrence. Therefore, the diagnostic during autopsy should be based upon prenatal assessments, accompanied by the pathological details revealed during the legal medicine examination.

Competing interests

The authors declare that they have no competing interests.

Authors contribution

All authors had equivalent contribution to this article.

List of abbreviations

LBWC- Limb Body Wall Complex
BPD- biparietal diameter
HC- head circumference
FL- femoral length

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