

LIMB BODY WALL COMPLEX IN TWO HETEROZYGOTIC TWINS: A CASE REPORT

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Introduction. Limb-Body Wall Complex (LBWC) is a congenital defect which includes at least two of the following characteristics: abdominal and/or thoracic body wall defects, exencephaly or encephalocele with or without craniofacial defects (56%) and spinal defects associated with marked vertebral or sacral defects (95%).

Case Report. We present a case report of an infant with LBWC, born by diamniotic twin pregnancy. A prenatal ultrasound reported an healthy fetus and a fetus with multiple malformations. At birth we found a big abdominal wall defect, absence of scrotal sac and testicles, asymmetric chest and no major deformities in craniofacial region. At 2 hours of life, we removed the amniotic sac, we put the stomach and the spleen in the abdominal cavity, which is virtual, and positioned a spring-loaded silo. At 15th day of life we had complete reduction of the intestinal loops and liver and we closed the wall defect with a prosthesis and a cryopreserved skin. The general conditions of the patient, very severe since birth, became progressively worse and he died in the 21st day of life.

Discussion. Once the diagnosis is done the physician should offer the parents a therapeutic abortion, and above all, in case they want to carry the pregnancy to term, we need to prepare them to the severity of the malformation and the high probability of death.

Keywords: Limb-body wall complex, heterozygotic twins, prenatal counseling.

INTRODUCTION

Limb-Body Wall Complex (LBWC) is a descriptive name for a congenital defect which usually includes at least two of the following characteristics: abdominal and/or thoracic body wall defects (in almost all patients), exencephaly or encephalocele with or without craniofacial defects (56%) and spinal defects such as scoliosis or kyphoscoliosis associated with marked vertebral or sacral defects (95%). (1) It is also associated with defects of the interior organs, such as diaphragmatic atresia (74%), renal agenesis, intestinal atresia, gallbladder atresia, heart defects, lung hypoplasia, hydrocephalus and limb defects and genitalia anomalies(2,3).

This severe and rare malformation complex has a prevalence ranging from 1/7.000 to 1/42.000(2,4,5).

The pathogenesis of this malformation is not clear and it has been a matter of debate in the literature (2,3,6,7,8).

We present a case report of an infant with Limb Body Wall Complex, born by diamniotic twin pregnancy.

CASE REPORT

Child born from diamniotic dichorionic twin pregnancy by caesarean section at 33 weeks. The first prenatal ultrasound reported an healthy fetus and a fetus with omphalocele. An ultrasound, performed at our hospital at 31 weeks +5, confirmed the presence of two

fetuses in cephalic presentation, posterior placenta grade II and the presence of multiple malformations in the fetus placed on top and left. In particular, it reported a large abdominal wall defect (37 mm), imperforate anus, neural tube defects and abnormalities of the urinary tract. It was done an initial diagnosis of OEIS complex (omphalocele, bladder exstrophy, imperforate anus and spinal defects). It was also noted probable cardiac malformation and anomalies of the lower limbs, clubfoot and arthrogyposis.

The birth weight was 1000 grams and the twin weighed 1820 grams. The APGAR at 1st and 10th minute was 4 and 5. Vital signs were stable (PA 44/22, SatO2 92% FC 139 bpm). The patient has been intubated and sedated. At the physical examination we found a big defect in the abdominal wall (4.5 cm), with evisceration of ileum, stomach, spleen, liver and part of the colon, the anus appeared to be well positioned and patent. The external genitalia were characterized by absence of scrotal sac and testicles, that have been found in the abdomen later on during the operation, a normal penis with normal urethral meatus. We placed a vesical catheter (3.5 Fr) without any problem. The chest was asymmetric, and no major deformities appear in craniofacial region. The lower right limb had pterygium of the knee and only 4 toes, clubfoot with extreme valgus is evident on the left (Fig. 1a-b).

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Fig.1a-b. *physical examination at birth.*



with bidirectional shunt and mild circumferential pericardial dissection, in the absence of other major anomalies. At 2 hours of life the patient has been operated. We removed the amniotic sac above the eviscerated organs, we put the stomach and the spleen in the abdominal cavity, which is virtual, and positioned a spring-loaded silo to protect the ileal loops, the sigmoid colon and liver. He passed meconium in the 2nd day of life. The brain ultrasound showed intraventricular hemorrhage 3rd grade on the left and 2nd grade on the right, with hyperechoic areas near the ventriculi and at the level of the basal ganglia. The genetic tests were in the norm, the QF-PCR showed normal 13, 18 and 21 chromosomes, the karyotype was XY and the array-CGH was negative. We reduced daily the silo. At 15th day of life we had complete reduction of the intestinal loops and liver in the abdomen (Fig.3).

Fig.3. *15th day of life: complete reduction of the intestinal loops and liver in the abdomen.*



The patient was transferred to the Neonatal Intensive Care Unit, with wet gauzes wrap to protect the eviscerated organs. The x-ray confirms the severe asymmetry of the two costal arches, multiple costal-vertebral dysostosis with the pelvis misaligned from the sacrococcygeal spine and femur, tibia and fibula of the right leg hypoplasia (Fig.2)

Fig.2. *x-ray at birth: severe asymmetry of the two costal arches, multiple costal-vertebral dysostosis with the pelvis misaligned from the sacrococcygeal spine and femur, tibia and fibula of the right leg hypoplasia.*



Because of the high risk of sepsis we decided to close the abdominal wall defect with a prosthesis made of a pure collagen membrane with holes and placing on the top a cryopreserved skin. The general conditions of the patient were very severe since birth, they became progressively worse, and he developed mixed acidosis, thrombocytopenia, increased creatinemia, anuria, severe jaundice, generalized edema and pulmonary edema. The patient had a blood culture positive for candida parapsilosis as well. The baby died in the 21st day of life.

DISCUSSION

Congenital malformation of the ventral abdominal wall occur in many forms, ranging from exomphalos to more complex malformations, depending on the severity of the defect and associated malformations(8). The case we presented is part of a more severe malformation called Limb Body Wall Complex (LBWC) characterized by a defect of the abdominal wall associated with serious spinal, skeletal defects and external genitalia malformation.

The pathogenesis of LBWC has been a matter of debate in the literature and three mechanisms have been proposed:

- a) *early amnion rupture, often because of a trauma(6);*

The echocardiogram indicated patent Botallo's duct

b) *embryonic circulatory failure leading to vascular disruption*(2,3);

c) *embryonic dysgenesis due to malfunction of ectodermal placodes*(7).

Russo et al. and Cusi et al. afterwards propose two different phenotypic expression of LBWC depending on the placental adhesion(9,10,11):

1. *Fetus with craniofacial defects such as exencephaly or facial cleft. In these cases usually there is cranioplacental attachment characterized by an adhesion between the skull defect and the amnion that is intact and covers the cerebral tissue*(12).

2. *Fetus without craniofacial defects, but with thoracic and/or abdominal ventral body defects combined with limb defects. In these patients there is thoraco- and/or abdominoplacental attachment characterized by an amniotic sheet that connects the skin margin of the ventral body wall defect to the placental surface. This sheet forms a hollow tube containing the eviscerated organs and represents a persistent connection between the intra- and extra-embryonic celomi cavity. The umbilical cord is not well-formed*(12).

Our patient did not have craniofacial defects, and he had all the phenotypic characteristics of patients with abdominal placental attachment. He presented, as in most of these fetuses, abnormalities of the abdominal wall and spinal cord, in association with defects of the lower limbs, feet and fingers. During the caesarean section it was seen the strict connection between the placenta and the amniotic sac that covers the abdominal organs, so that the gynecologist was forced to take away the baby with the placenta with minimal disruption of the amniotic sac.

As shown in Literature this malformation is not associated with specific genetic abnormalities, as well as in our case the genetic tests are perfectly normal(2).

In Literature there are no specific references to cases of dizygotic twins with LBWC, but it seems to have a 12.5 times risk in monozygotic twins(2).

The diagnosis of this severe malformation could be done with prenatal ultrasound and it is possible since the second trimester of pregnancy(12,13). The prenatal ultrasound shows the parietal defect with eviscerated abdominal organs and spinal defects, although it is not easy to distinguish it from other malformation such as OEIS complex, which is characterized by omphalocele, bladder exstrophy, imperforate anus and spinal defects. In most cases the pregnancy is not completed, due to mother's decision or to fetal problems that lead abortion. In our case, even if the prenatal diagnosis was made early, the mother decided to bring their pregnancy at the end, because the other fetus was perfectly healthy. Caesarean section at 33 weeks was necessary to avoid further risks.

At birth, after a meticulous examination, we proceeded to stabilize the patient and to perform the surgical correction of the abdominal defect. The defect of the abdominal wall was very large, about 4.5 cm and the abdominal cavity was completely inadequate to contain the eviscerated organs. It was necessary to use a

spring-loaded silo and finally close the abdominal wall with a prosthesis. The medical and surgical procedures, directed toward the stabilization of the patient, did not however permit the survival of the newborn. The general conditions of these patients are often very severe and even the literature shows how poor the prognosis is(12,14,15). Most of these patients die during the antenatal period and the babies born alive usually die few days after birth.

CONCLUSION

LBWC is a very serious malformation, which can be diagnosed prenatally. Once the diagnosis is done the physician should offer the parents a therapeutic abortion, and above all, in case they want to carry the pregnancy to term, we need to prepare them to the severity of the malformation and the high probability of death. Therefore, in these particular cases it is very important the prenatal counseling done in cooperation by the gynecologist and pediatric surgeon, to make parents aware of the low probability of survival these fetuses have. In our case, the parents, aware of the seriousness of the malformation, decided to bring their pregnancy to term and this has allowed the other baby to be born healthy and without any problems.

REFERENCES

1. Colpaert C, Bogers J, Hertveldt K, Loquet P, Dumon J, Willems P "Limb-body wall complex: 4 new cases illustrating the importance of examining placenta and umbilical cord" *Pathol Res Pract* 2000; 196: 783-790
2. Van Allen MI, Curry C, Walden CE, Gallagher L, Patten RM. "Limb body wall complex: I. Pathogenesis" *Am J Med Genet* 1987; 28:529-48
3. Van Allen MI, Curry C, Walden CE, Gallagher L, Patten RM. "Limb body wall complex: II. Limb and Spine defects" *Am J Med Genet* 1987; 28:549-65
4. Kurosawa K, Imaizumi K, Masuno M, Kuroki Y "Epidemiology of limb-body wall complex I japan" *Am J Med Genet* 1994; 51: 143-146
5. Martinez-Frias ML "Clinical and epidemiological characteristics of infants with body wall complex with and without limb deficiency" *Am J Med Genet* 1997 Dec 12; 73 (2): 170-5
6. Torpin R "Amniochorionic mesoblast fibrous string and amniotic band" *Am J Obstet Gynecol* 1965; 91: 65-75
7. Hartwig NG, Vermeji-Keers CHR, De Vries HE, Kagie M "Limb body wall malformation complex: an embryologic etiology?" *Hum Pathol* 1989; 20: 1071-7
8. Mann S, Blinman TA, Douglas Wilson R "Prenatal and postnatal management of omphalocele" *Prenat Diagn* 2008; 28(7): 626-32
9. Russo R, Vecchione R "Limb body wall complex: craniofacial defects as a distinctive factor" *Birth Defects* 1996; 30: 157-64
10. Russo R, D'Armiento M, Angisani P, Vecchione R "Limb body wall complex: a critical review and a nosological proposal" *Am J Med Genet* 1993; 47: 893-900
11. Cusi V, Torrens M, Villa J, Antich J, Carrera JM "Limb body wall complex: analysis of eight fetuses" *Birth Defects* 1996; 30: 165-70
12. Deruelle Ph, Hay R, Subtil D, Chauvet MP, Duroy A, Decocq J, Puech F "Diagnostic antenatal du limb body wall complex" *J Gynecol Obstet Biol Reprod* 2000; 29: 385-391
13. 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" *Genetic Counseling* 2007; 18(1): 106-112
14. Shah HR, Patwa PC, Tanik AV, Nayak C, Garasia R "A case report of foetus having combined features of LBWC + OEIS Complex" *Indian J Radiol Imaging* 2005; 15(1): 85-88
15. Prasun P, Behera BK, Pradhan M "Limb Body Wall Complex" *Indian J Pathol and Microbiology* 2008; 51(2): 255-256