Limb body wall complex - report of an unusual case

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Abstract

Limb body wall complex is an extremely rare birth defect and a foetal polymalformative syndrome. A case of Gravida-2, Para-0, Abortion-1 with 20 weeks of pregnancy who attended the antenatal clinic in the month of May 2012, at Konaseema Institute of Medical Sciences and Research foundation (KIMS&RF), Amalapuram, Andhra Pradesh, underwent routine anomaly scan which showed a polymalformative fetus. The pregnancy was terminated. The physical examination findings of the aborted fetus were suggestive of limb body wall complex (LBWC).

Key words: Body wall defects, foetal polymalformative syndrome

Introduction

Limb body wall complex (LBWC) is very rare anomaly with a large abdominal wall defect and protrusion of abdominal viscera; this anomaly is due to failure of development of body stalk and is associated with other limb anomalies. Incidence of LBWC is 0.32/100000 births. Because of the associated malformations which are incompatible to life, it is important to diagnose prenatally by maternal serum AFP & USG followed by termination of the pregnancy.

Embryology

After fertilization, human zygote turns into Blastocyst. Second week after conception, blastocyst will separate into 3 layers- ectoderm, mesoderm, endoderm from which, all the organs will develop. When the portion of embryonic formation is disrupted, chances of multiple massive deformities are inevitable.

LBWC is due to the defect and interruption of development of very early developing embryo between 24th & 36th day after fertilization. Small intestines developing from midgut remain out side the anterior abdominal wall and fail to return in to the abdominal cavity due to failure of development of body stalk; embryonic malfunction of body wall ectoderm and mesoderm leads to defective closure of embryonic abdominal wall and abnormal development of umbilical cord. So there is a large abdominal wall defect and limb malformations. Though limb defects are present in vast majority of cases absence of a limb is seen in less than tenth.

Case report

A case of Gravida-2, Para-0, Abortion-1 with 20 weeks of pregnancy came for routine antenatal care in the month of May 2012 to our KIMS hospital. There was no H/o consanguinity; no H/o taking any drugs; no H/o exanthamatous fever during early pregnancy; no significant family history of congenital malformations. Antenatal period was uneventful. Routine USG was done which showed fetal anterior parietal wall defect along with herniation of liver and loops of bowels; there was also presence of sacral meningocoele measuring 3x3 cms. Family counseling was done regarding the anomalies which are incompatible with life and pregnancy was terminated by medical methods viz., misoprostol and misoprostol. A female fetus weighing approximately 500gms was expelled.

The following anomalies were noted in the aborted fetus:

1. Large abdominal wall defect with herniated abdominal viscera (liver, stomach, intestine loops, bladder) covered by a thin membrane (figure 1). There was no umbilicus but the short umbilical cord (figure 2) was attached to the thin membrane directly nearer to the edge of abdominal wall defect.

2. Other features were sacral meningocoele, low set ears, right sided complete absence of lower limb.
X-Ray findings (figure 3):
1. Severe scoliosis.
2. Central type of longitudinal defect of right lower limb (i.e. absent).
3. Anterior abdominal wall defect with internal organ malformations: gastrochisis on Right side with herniation of the liver and intestines.
4. Bladder and cloacal exostrophy seen.
5. No significant cranio facial anomalies.

![Image](image1)
**Fig. 1**: Showing the aborted fetus with abdominal wall defect, gastrochisis on right side with herniation of the liver and intestines, bladder and cloacal exostrophy and absence of right lower limb suggestive of LBWC defect without craniofacial defects.

![Image](image2)
**Fig. 2**: Showing short umbilical cord directly attached to the thin membrane nearer to the edge of abdominal wall defect.

![Image](image3)
**Fig. 3**: Skiagram of the aborted fetus showing Severe scoliosis, absent right lower limb, anterior abdominal wall defect with internal organ malformations: gastrochisis on right side with herniation of the liver and intestines, bladder and cloacal exostrophy.

**Discussion**

Limb body wall complex (LBWC) is an unusual developmental abnormality. It was first described by Van Allen et al in 1987. It has Uncertain aetiology, no genetic cause and low recurrence rate. But some authors have commented that there is connection between LBWC and teratogenic drugs like anticonvulsants & cocaine abuse. Theories of pathogenesis of limb body wall complex: 1. Germ disc defect with early embryonic maldevelopment. 2. Primary rupture of the amnion leading to the formation of amniotic bands (Torpin,1965). 3. Vascular disruption (van Allen, 1987) and 4. Disturbance of embryonic folding process (Hartwig et al.1989,1991).

Review of literature over 100 cases clearly indicates possible phenotypic variation of LBWC. These data add further evidence of support for existence of two different phenotypic variants of limb body wall complex. First variety of phenotype has features like "foetus with craniofacial defects"; presence of encephalocele or exencephaly with associated facial clefts and amniotic bands (broad amniotic adhesions between cranial defects and placenta) characterizes the first variety. The first phenotype is caused by early vascular disruption and
amnion rupture with formation of amniotic bands. Second variety of phenotype has features like "foetus without craniofacial defects". In this urogenital anomalies, anal atresia, lumbosacral meningocoele, short umbilical cord, persistence of extra embryonic coelom and intact amnion are the significant positive findings. The second phenotype is associated with body stalk anomaly and embryonal maldevelopment with malfunction of body wall ectoderm which leads to defective closure of embryonic abdominal wall\(^1\). LBWC is associated with multiple anomalies with essential features of encephalocle with facial clefts, thoraco abdominoschisis and limb defects. To diagnose LBWC, two of the three above features should be present\(^2\). Other anomalies associated with LBWC are cardiac, renal, bowel, urogenital anomalies, limb defects.

In our case there were abdominoschisis, complete absence of right lower limb (Amelia), sacral meningocoele, scoliosis of the dorsal lumbar spine, 11 pairs of ribs, no umbilicus, a short umbilical cord, placenta attached to the thin membrane covering the organs through the cord. The above features fit into second variety of phenotype.

**Conclusion**

LBWC is a rare and lethal anomalous disease with very poor prognosis which should be diagnosed prenatally with high Maternal alpha feta proteins & USG, followed by termination. It may require chromosomal analysis since it is associated with trisomies 13, 18, 21. It is mandatory to do TIFFA (targeted image for foetal anomalies) scan for all antenatal cases to diagnose anomalies and to initiate appropriate treatment to prevent pregnancy carrying till term.

**Consent**

Written informed consent was taken from the mother for publication as well as figures of this case report.

**References**


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