Abstract:
Limb body wall complex is a rare anomaly with wide and varying spectrum of manifestations. We report a stillborn with thoracoabdominal schisis with limb deformities, renal abnormalities, ectopia cordis and meningomyelocele. We briefly discuss the differential diagnosis and review the literature.

Keyword: limb body wall complex, thoracoabdominoschisis, body stalk anomaly, cyllosomas

Limb body wall complex also known as body stalk anomaly, cyllosoma, short umbilical cord syndrome, aplasia of the cord, are rare compound anomalies characterized by severe limb defects and varying body wall defects mainly chest and abdomen. It needs to be differentiated from other treatable anomalies like gastrochisis. Using standard Mesh terms "Limb body wall complex OR cyllosomus OR body stalk anomaly OR short umbilical cord syndrome" and manual screening in PUBMED database retrieved only 133 case reports worldwide. Only 16 cases, including a case series are reported from India. Paucity in Indian literature and the need to distinguish it from treatable anomalies in antenatal scan made us to report this case.

Case report:
A 29 year old female with history of non-consanguinous marriage, G3P2L2, came for a second trimester antenatal check up. Her previous pregnancies were uncomplicated and now having two sons, 7 years and 5 years old. She has no medical illness and has stopped taking folic acid tablets early in the first trimester because of vomiting. On Antenatal scan, the fetus found to have multiple congenital anomalies with hydrocephalus, thoraco lumbar spinal rachischisis, hydroureteronephrosis (RT>LT), urinary ascitis and floating bowel loops. The Pregnancy was medically terminated and on mini autopsy, the following findings were noted (FIG 1).
A 24 weeks still born male, with 1) A large ventral abdominal wall defect exposing the abdominal contents, with loops of brownish black bowel lying on the right side of the abdominal wall. Both kidneys with ureter were enlarged (RT>Lt), urinary bladder not seen.Penis was present, but scrotum and testes were not developed.Also Pelvic organs like urinary bladder, rectum were absent and Anal orifice was not present.

2) Thoracic defects (Thoracoschisis) – Lower Sternal cleft with an anterior diaphragmatic defect was present .Cardiac herniation through the defect to epigastric region (Ectopia Cordis) was noted.(FIG 2)Cardiac fossa was filled with serous fluid. 3) spinal defects –dorsal curving of the spine with large meningomyelocele at thoracolumbosacral region and ventriculomegaly was present.(FIG 1.black arrow heads) 4) Limb defects- both lower limbs were malformed, externally rotated and extended at hip with clubfoot and radial polydactyly was present on Right hand.A diagnosis of Limbbodywallcomplex (LBWC) was made and mother advised to take folic acid tablets and to attend genetic counseling before planning next pregnancy.

Discussion:
Limb–body wall complex(LBWC) is a rare lethal anomaly with varying incidence reported from 0.21 to 0.31 cases per 10,000 births[ 1]. Majority of the cases are spontaneously aborted and the remainder are stillborn.

The diagnosis was based on two out of three (van Allen)criteria [2]: 1) exencephaly/encephalocele with facial clefts; 2)thoraco and/or abdominoschisis (a anterolateral body wall defect with evisceration of thoracic or abdominal organs into a persistent extra embryonic coelom) and 3) Limb defects.

In our case, there was no craniofacial anomaly and the other two criteria were present. Two distinct types were described by Russo et al [3]. Type I lumbosacral meningomyelocele, severe kyphoscoliosis and placental anomalies).our case comes under type 2 with additional limb defects. 95% cases of LBWC reported to have internal visceral anomalies .GI T anomalies(100%), diaphragm defect(74%), renal (65%),urogenital(56%) , cardiac (43%) anomalies and amniotic bands and single umbilical artery has been reported [2].our case had hydronephrosis probably due to lower urinary tract obstruction and also had non rotation of bowel loops, but had no amniotic bands or attached extraembryonic coelom.The proposed etiologic theories are
1) vascular disruption of developing embryo leading to facial clefts, neural tube–like defects, limb reduction defects, and visceral anomalies. Adhesion of the amnion to the necrotic areas due to vascular defect could lead to secondary amniotic adhesive bands.

2) Early amnion rupture leading to adhesive bands

3) Failure of the ventral body wall to fuse

4) Recently in 2011, Hunter et al. [4] concluded it as a defect in ectodermal placode at the embryonic disc stage. LBWC with neural tube defects in our case favors a vascular or ectodermal defect. In literature, karyotyping of LBWC cases reported to be normal and there were no reports of familial transmission. Lack of folic acid intake by the mother in our case needs to be investigated in further studies.

The Differential diagnosis includes

1) Gastroschisis – is characterised by an abdominal wall defect, located laterally to a normally closed umbilical ring, with herniation of organs that are uncovered by membranes.

2) Omphalocele – is characterized by the herniation of membrane-covered internal organs into the open base of the umbilical cord.

3) Pentology of Cantrell - a constellation of lower sternal defect, anterior diaphragmatic defect, ectopia cordis, absent parietal pericardium and intracardiac anomalies.

4) Amniotic band sequence; ABS Streeter anomaly – severe limb deformities including transverse limb defects, with or without encephalocele, facial clefts and abdominal defects[5].

5) Thoracoabdominal syndrome; (THAS) is another newer syndrome with Pentology of Cantrell like defect with varying abdominal defects affecting male fetuses, with X-linked dominant inheritance.[6]

The presence of extra intestinal abnormalities, limb defects and spinal cord malformations differentiate our case from the first three conditions and the absence of amniotic bands, amputations and previously normal male siblings in our case negate the remaining two.

Antenatal diagnosis can be made by measuring maternal serum alpha-fetoprotein level and ultrasonogram, as in our case. LBWC is a lethal anomaly and it has to be differentiated from treatable causes like omphalocele or gastroschisis. Early diagnosis although possible was not done in our case, since the mother has not taken regular antenatal checkup and Antenatal scan was not done in the first trimester.

**Carry home message**

Limb body wall complex is a lethal anomaly needs to be differentiated from treatable conditions like gastrochisis and omphalocele in antenatal scan.

Non familial inheritance should reassure parents and X linked inheritance should suggest an alternate diagnosis like THAS.

Lack of folic acid intake in our case suggests future studies to investigate its role in Limb body wall complex.

**References:**


