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A CASE REPORT OF LIMB-BODY WALL COMPLEX MALFORMATION.

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Abstract : During routine antenatal checkup, a 24 year old primi-gravida was observed to have a 20 weeks old fetus with abdominoschisis with liver and intestinal contents herniating through the midline defect, amelia (absence of right upper limb) absence of sternum, thoracic scoliosis and congenital talipes equinovarus of right foot were noted. There was no history of consanguinity or family history of any malformations. There was no history of intake of drugs other than iron and calcium supplements during the pregnancy. After medical termination of pregnancy, on gross examination of the expelled fetus the above mentioned conditions were confirmed and a diagnosis of Limb Body Wall Complex Malformation was given.

Keyword : Limb Body Wall Complex Malformation, Abdominoschisis and Amelia.

INTRODUCTION

Limb Body Wall Complex Malformation (LBWC) also known as the body-stalk syndrome is a rare sporadic fetal polymalformation of unknown origins. This entity is most commonly characterized by thoraco-abdominoschisis associated with variable spectrum of limb and visceral anomalies. Two phenotypes have been described the “placento-cranial” and “placento-abdominal” adhesion phenotypes. Estimated incidence is at around 0.32 in 100,000 live births.1 This condition has no sexual or familial predilection, carries an extremely poor prognosis and is invariably fatal.

CASE REPORT

During routine antenatal checkup, a 24 year old primi-gravida was observed to have a 20 weeks old fetus with congenital poly-malformative condition. There was no history of consanguinity or family history of any malformations. There was no history of intake of drugs other than iron and calcium supplements during the pregnancy. After confirmation of the diagnosis by ultrasound examination parental counseling followed by medical termination of pregnancy was done.

OBSERVATION

At dissection, the aborted fetus weighed 215 gms with a Crown-Rump Length (CRL) of 145mm. On examination right sided abdominal wall defect with protrusion of liver and intestines not encased by membrane, skeletal abnormalities of marked right kyphoscoliosis, amelia of right upper limb, absence of sternum and congenital talipes equinovarus (CTEV) of right foot was noted.(Fig.2)

Fig.2: 20 week old aborted fetus with LBWC

DISCUSSION

Limb Body Wall Complex Malformation belongs to the spectrum of fetal midline disruption syndrome along with Pentalogy of Cantrell. The exact cause of the Limb Body Wall Complex Malformation has not yet been determined. This complex of ventral wall anomalies has been classified under changing nomenclature that has included limb-body wall complex, simply body wall complex amniotic band disruption complex, and amnion rupture sequence. Traditionally diagnosis has been based on the Van Allen et al., criteria, i.e. Exencephaly or encephalocele with facial clefts, Thoraco or abdominoschisis and Limb defects (the presence of two out of three confirms the diagnosis) Several hypothesis including Tropin’s amniotic band theory, Allen’s vascular disruption theory and Hartwig’s theory of embryonic dysplasia were suggested.2 Non-development of the lateral wall mesoderm leading to midline defects like ectopia cordis, gastroschisis and bladder extrophy may also be a variant of this complex. Abnormal embryonic folding with malfunctional migration of body wall ectodermal placodes may also lead to such poly-malformations. Experimental studies conducted in animals revealed primary migration defect in the ectoderm of the embryonic disc during early gastrulation.3 Abnormalities commonly associated with this complex include cranio-facial abnormalities, scoliosis, ventral body wall defect (thoraco-abdominoschisis), limb deformations, short umbilical cord, placenta previa , neural tube closure defects and others. In our case report, abdominoschisis (Fig.1) with amelia was noted.

Fig.1 : Abdominoschisis

USG Scan Image - 20 week old fetus

LBWC is a lethal anomaly hence it has to be differentiated from treatable causes like omphalocele or gastroschisis using non-invasive monitoring of fetus. Autopsy is the gold standard for diagnosis. Karyotyping is usually normal.4,5 Diagnosis of LBWC using 3D ultrasound allows for identification of the complex more clearly and also for making the information more understandable to parents

CONCLUSION

LBWC is a lethal poly-malformative fetal syndrome which is to be considered during differential diagnosis of anterior abdominal wall defects in-utero. Risk of recurrence is very minimal hence proper education by the medical team regarding antenatal assessment of fetal well being will help families plan their reproductive future with better medical care.

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