BODY STALK ANOMALY: A CASE REPORT

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HOW TO CITE THIS ARTICLE:
Annapurna Bose, Mukul Yadav, Sudha Shrivastava, Abhijeet Yadav. "Body stalk anomaly: a case report". Journal of Evolution of Medical and Dental Sciences 2013; Vol2, Issue 38, September 23; Page: 7256-7262.

ABSTRACT: The present case is the report of singled out case of still born 34 weeks old, grossly abnormal, male fetus, with gross left sided anterior abdominal wall defect, which was donated and sent to the Department of Anatomy, Bundelkhand Medical College Sagar (MP), by the Department of Obstetrics for educational purpose. The still born's mother was 21 years old, primi gravida, with history of 34 weeks amenorrhoea. Detailed maternal history was taken to elicit any relevant information. After careful examination, observations were recorded. In the fetus the stump of umbilical cord was missing. Left half of anterior abdominal wall was totally absent with abdominal organs herniating out from left side. Detail examination of the viscera was done. After careful observation it was diagnosed that this is one of the types of anterior abdominal wall anomaly and this was concluded to be a rare case of body stalk anomaly. The findings of dissection and its embryological correlation are discussed in this report.

KEY WORDS: Short umbilical cord, anterior abdominal wall defect, body stalk anomaly, malrotation of gut, kyphoscoliosis, herniating organs.

INTRODUCTION: The present case with gross left sided anterior abdominal wall defect was donated by the parents to the Bundelkhand Medical College Sagar (MP), for educational purpose. The rationale of the study was to throw light upon type and genesis of the malformation and its possible prevention.

In the malformed fetus stump of umbilical cord was missing, left half of anterior abdominal wall was totally absent with abdominal organs herniating out from left side, the fetus was bent and twisted to the right side (kyphoscoliosis of the spine with the legs twisted posteriorly and to the right side.) The diaphragm was incompletely developed with heart and left lungs herniated in abdominal cavity, left thoracic cavity empty and malrotation of gut with some of the organs in rudimentary condition. As this was a type of anterior abdominal wall anomaly and our purpose was to classify it and find out embryological basis and fatality.

Body stalk anomaly is defined as a severe and rare (generally 1 per 14, 000 births) [1] abdominal wall defect caused by the failure to form a body stalk and is characterized by an enlarged abdominal wall defect, severe kyphoscoliosis and a rudimentary or the absent umbilical cord and was described for the first time by Van Allen et al. in 1987 [2, 3] as a set of disruptive abnormalities characterized by a severe body-wall defect (thorax, abdomen or both), evisceration of the abdominal organs into an amnio-peritoneal sac and a shortened or absent umbilical cord. It is also labeled by other authors as congenital absence or aplasia of the umbilical cord, limb-body-wall complex, cyllosomas, or tethered fetus syndrome. [4]

In body stalk anomaly, the lateral, cranial and caudal folds fail to form, leaving the abdominal content exposed often complicated by associated anomalies of head, face and extremities etc [1]. The etiology of body stalk anomaly is unknown, no chromosome defects identified with this anomaly. Three possible etiologic mechanisms have been proposed: 1) Mechanical obstruction secondary to

compression by amniotic bands, 2) abnormalities of the germ disk, 3) vascular disruption of the fetoplacental circulation. [5]

MATERIAL AND METHODS: The abortus was send by the Department of Obstetrics to the Department of Anatomy, as the parents had donated the fetus for research and study purpose to Bundelkhand Medical College Sagar (MP).The mother was 21 years old, primi gravida, with history of 34 weeks amenorrhoea with no history of any addiction or systemic disease as diabetes or hypertension, no history of any infective disease during first 3 months of pregnancy or taking any drugs or medicines during the period, no parental history of any congenital malformation in the family tree of either side. She works in farm/field. The mother was feeling less fetal movements for 1 week, after which she aborted a malformed fetus.
Observations were recorded after embalming and careful dissection.

OBSERVATIONS & RESULT

1. **OBSERVATIONS:** The case was a still born, grossly abnormal, male fetus.

MEASUREMENTS:

CR length 16cm, CH 25cm,	Head circumference: 26.5cm
Chest circumference 14.7cm	Abdominal circumference 17cm
Weight 300gm fetus	

GROSS: The fetus was bent and twisted to the right side (kyphoscoliosis of the spine with the legs twisted posteriorly and to the right side.) (Figure: 1 & 2) The umbilical cord was totally missing; left half of anterior abdominal wall totally absent with abdominal organs herniating out of the defect on left side, defect dimensions on left side being 8cmX 4cm, the width of intact right abdominal wall was 3cm. No anomaly found in head, neck, face, upper and lower limbs or external genitalia.

ON DISSECTION: Diaphragm was incomplete: partially present on right side, totally absent on left side, causing the heart and lung to get herniated. Thoracic wall was intact but upon dissection, left thoracic cavity was found empty. Heart and left lung were in abdominal cavity. Left lung (length -2.5cm, breadth -2.2cm), Right lung (length -3.9cm, breadth -2.2 cm) was in right thoracic cavity. There was no intra cardiac defect, thymus was small. Incomplete rotation of gut with appendix (length -1.2cm) found lying just below stomach (Figure: 3). Liver, gall bladder found displaced from its position, with no anomalies (Figure: 1).
Both kidneys were in normal position.

1. Left kidney: length-3.5cm, breadth-2.0 cm (smaller)
2. Right kidney nearly round/globular in shape: length -3.2cm, breadth -3.4 cm (Figure: 4)
   No anomalies found in Stomach, intestines (except mal rotation), pancreas, spleen, ureters, urinary bladder, trigone or opening of ureters, no exostrophy of the bladder or imperforate anus. (Figure: 5)

2. RESULT: The case under discussion comes under body stalk anomaly presenting as set of disruptive malformations with the failure of closure of the ventral wall, characterized by a severe

body-wall defect, evisceration of the thoracic organs into abdomen, abdominal organs into an amnio-peritoneal sac, malrotation of gut, an absent umbilical cord and kyphoscoliosis, but with no other associated anomalies. After careful observation we concluded to be a rare case of body stalk anomaly which is one of the types of anterior abdominal wall defect.

DISCUSSION: As the case was a type of anterior abdominal wall anomaly, its other types were referred for differential diagnosis.

Gastroschisis: Generally a full thickness right para-umbilical defect in the anterior abdominal wall (2-5 cm) through which the intestinal contents freely protrude. No covering membrane, cord inserts on abdominal wall, only loops of bowel herniated. [6]

Pentalogy of Cantrell: This is a cluster of defects first described by Cantrell in 1958 in five neonates. It is characterized by an upper midline omphalocele, anterior diaphragmatic hernia, ectopia cordis, sternal cleft and intracardiac defects. [6].

Omphalocele: midline defect of the abdominal wall, generally defect size is 2-15 cm, herniated content is covered with membrane, umbilical cord inserts on the top of membrane.

EMBRYOLOGICAL CORRELATION: Body stalk anomaly is a severe anterior abdominal wall defect consisting of a sac of amnion-mesoderm that contains the displaced abdominal organs resulting from a failure of cephalic, caudal, and lateral body folds to fuse and form an umbilical cord. Fusion of these infoldings (occurring at 22 to 28 days after conception) creates a cylindrical embryo from a flat oval germ disk. A predominantly cephalic defect results in a constellation of anomalies known as pentalogy of Cantrell. [6, 7, 8]. These foldings separate the intra-embryonic coelom from the extra-embryonic coelom. The amniotic cavity which was dorsal to the germ disk enlarges to encircle the embryo and obliterate the chorionic cavity. Body of the embryo closes and a body stalk (destined to become the umbilical cord) is formed with fusion of amnion and chorion at the umbilicus. [9].

A Faulty invagination process prevents obliteration of chorionic cavity and formation of umbilical cord. A wide-based cylinder is formed, bounded on one side by fetal retroperitoneal space, and chorionic plate of placenta at the other end. Its sac-like walls, consisting of amnion and foetal mesoderm, contain the abdominal organs. [10].

Defective formation / fusion of the pleuroperitoneal membrane(s) separating pleural and peritoneal cavities cause diaphragmatic hernia. Defect is in posterolateral part of diaphragm, usually unilateral with a large opening (foramen of Bochdalek), is more often on left due to an earlier closure of the right pleuroperitoneal opening [11]

Other associated anomalies which may be found**:** ectopia cordis, sternal cleft, intracardiac defects, exostrophy of the bladder, imperforate anus, intestinal and colonic agenesis, neural tube defects, skeletal defects, hypoplastic kidney, or absent external genitalia [1].

Ultrasonography can detect the typical features of body stalk anomaly by the end of first trimester. As body stalk anomaly cases are invariably fatal, so an early detection and termination of pregnancy can save parents from mental agony and prevent surgical intervention at later stage. [12]

CONCLUSION: Invariably fatal and severe body stalk anomaly results from a failure of cephalic, caudal, and lateral body folds to fuse and form an umbilical cord**.** As no direct etiologic factor is known, only prevention lies in its early detection and termination**.**

ACKNOWLEDGEMENT: To the parents and the department of obstetrics & gynaecology who donated the stillborn baby for research.

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PHOTOGRAPHS:

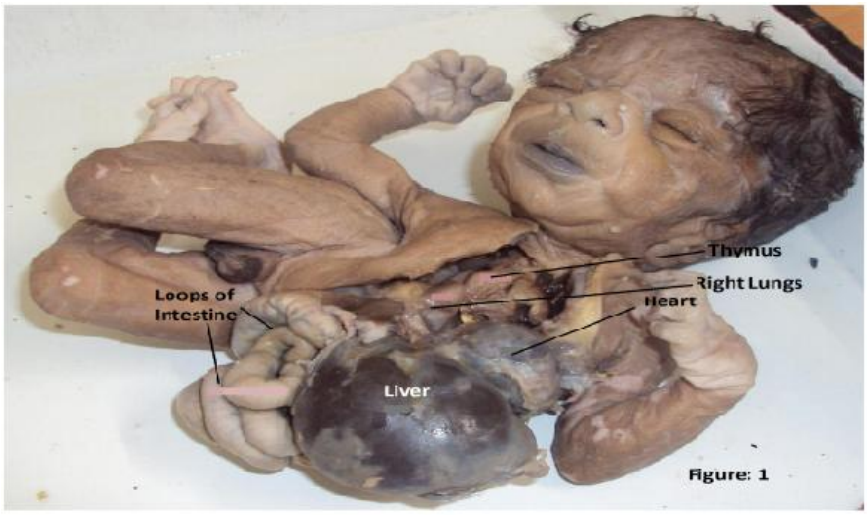

Figure 1: Liver was of normal size and displaced from its position many of the organs were found in rudimentary condition, figure also shows the abnormal positioning of the body & legs.

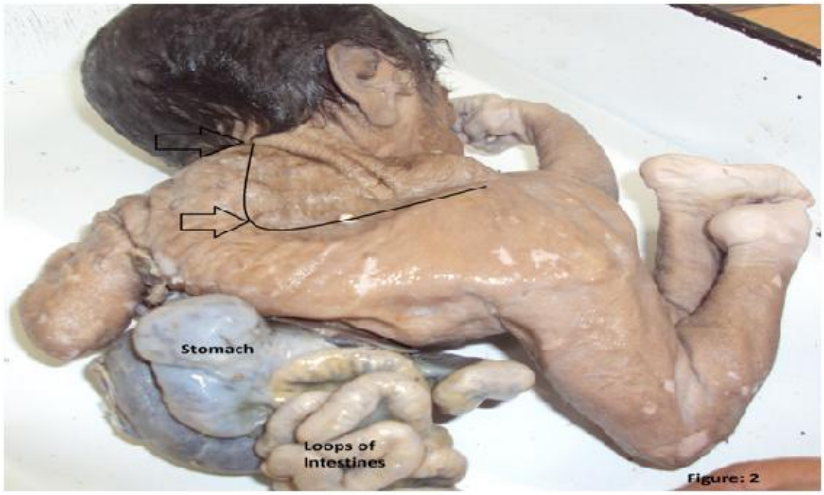

Figure: 2. The fetus was bent and twisted to the right side marked by bold line and arrow (kyphoscoliosis of the spine) also showing the abnormal position of the legs.

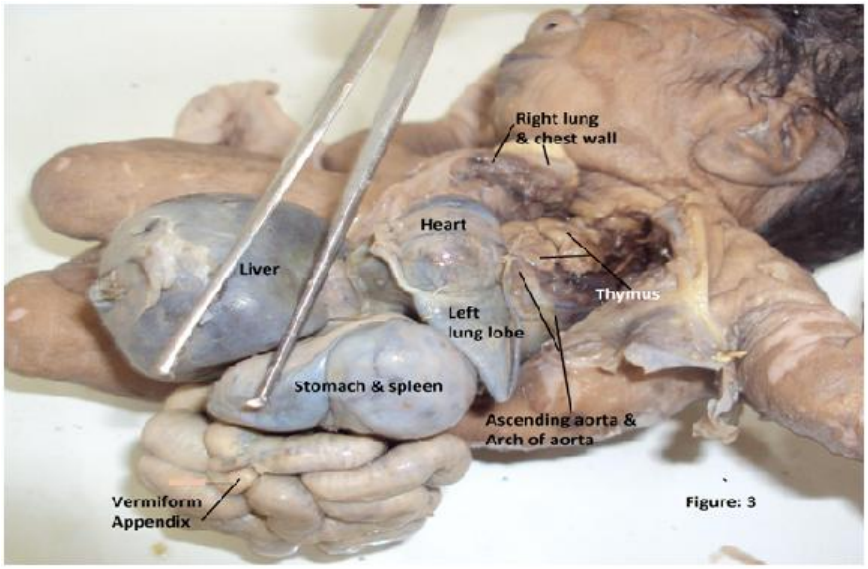

Figure: 3. Mal rotation of gut with appendix found lying just below stomach.

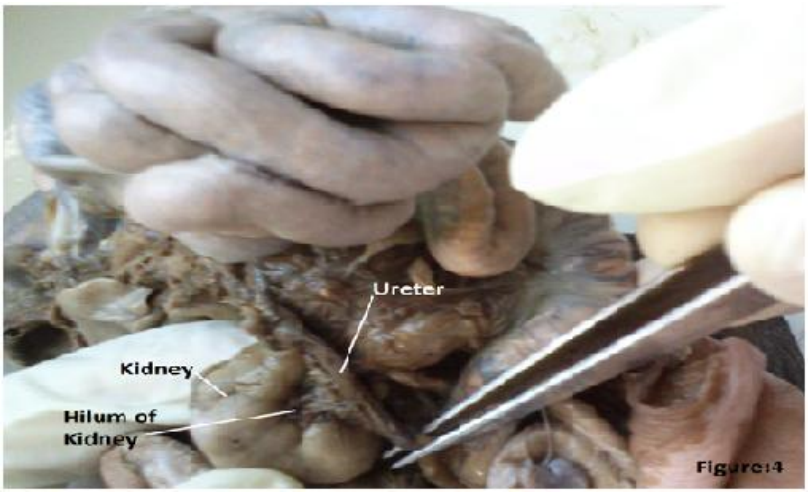

Figure 4: Showing the Left kidney, its hilum & ureter.

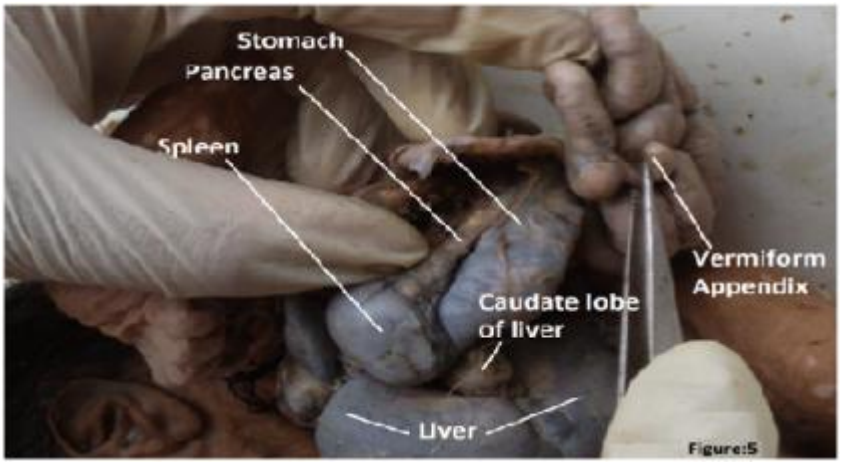

Figure 5: Abdominal organs of the foetus showing pancreas, lobes of liver & appendix.

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Date of Submission: 05/09/2013.
Date of Peer Review: 06/09/2013.
Date of Acceptance: 13/09/2013.
Date of Publishing: 19/09/2013