

# Fetal Autopsy Showing Two Ventral Body Wall Defects: An Unusual Presentation

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## ABSTRACT

Thoracoschisis and gastroschisis are types of ventral body wall defects. Usually thoracoabdominoschisis presents as a continuous defect. Here the authors present the case of autopsy of 18 weeks old fetus with two well-separated ventral body wall defects. Thoracoschisis was a right lateral thoracic wall defect while gastroschisis, a midline infraumbilical abdominal wall defect. Through the right lateral thoracic wall parts of two organs appeared to be protruding. On dissection, 8<sup>th</sup> and 9<sup>th</sup> ribs with the liver and the lower lobe of the right lung were herniating outside the body. The right lung was haemorrhagic. Various embryological theories on ventral body wall defects have been discussed in view of the present case.

**Keywords:** Embryology, Fetus, Gastroschisis, Thoracoschisis

## CASE REPORT

An autopsy of 18-week-old fetus was performed. Mother was 19 years-old without history of any chronic illness or drug intake during pregnancy. Antenatal scan done at 18 weeks of pregnancy suggested a meningocele, after which the pregnancy was terminated surgically.

Fetal autopsy was performed after obtaining written consent. The fetus was male with anthropometric parameters corresponding to approximate 20-22 weeks. Gastroschisis with a small defect in the abdominal wall in midline adjacent and inferior to the umbilicus was noted. The umbilical cord was normally inserted and the abdomen was distinctly scaphoid. There was an approximately 3x2 cm defect in the right lateral thoracic wall through which parts of two organs appeared to be protruding. On dissection, this defect was found to be between the 8<sup>th</sup> and 9<sup>th</sup> ribs with the liver and the lower lobe of the right lung herniating outside the body [Table/Fig-1,2]. The round ligament was passing through the diaphragm connecting the liver above to the umbilical cord below. The stomach was notably horizontal in its orientation [Table/Fig-3].



**[Table/Fig-1]:** Fetal photograph showing two body wall defects- a midline infraumbilical gastroschisis and right lateral thoracoschisis. Note the traumatic appearance of the liver due to exteriorization. U-Umbilical cord. **[Table/Fig-2]:** Fetal photograph (lateral view) showing the extent of thoracic wall defect with liver and a tongue like protrusion of the lower lobe of the right lung herniating outside the body. Also seen is scaphoid abdomen and gastroschisis. R-Rib, SI-Small Intestine, U-Umbilical cord. **[Table/Fig-3]:** Fetal photograph after dissection showing a horizontally oriented stomach(S), diaphragm (D-outlined), haemorrhagic right lung and the absence of liver in abdominal cavity. (images from left to right).

The rest of the abdominal examination was normal. The right lung was haemorrhagic. An overlying area of haemorrhage was also found over the right scapula suggesting trauma to the fetus as was also suggested by the nibbled appearance of the liver. The heart and great vessels did not show any major defect. There were no constriction rings or limb reduction defect in the extremities. Grossly no neural tube defect was seen. The face was not dysmorphic and there were no facial clefts. The anal opening was normal and the external genitalia were of the normal male type. The placenta was not available for examination. The fetal radiograph showed 12 pair of ribs and a discontinuity in the right sided thoracic wall.

## DISCUSSION

Congenital thoracoschisis and gastroschisis are types of ventral body wall defects with evisceration of organs outside the body. Thoracoabdominoschisis implies a midline continuous defect in the thoracic and abdominal walls. A defect of only the ribs results in lateral thoracoschisis which is a very rare congenital anomaly. There are only nine published case reports on this condition. Karaman I et al., have presented their case and reviewed four earlier case reports [1]. Additionally two cases labeled as Congenital Intercostal Liver Herniation by Ertan G et al., one case each by Bhattacharya NC et al. and Seleim HM et al., were found to be published [Table/Fig-4] [1-8].

To summarise, all these cases had thoracic wall defect (left sided> right sided) with diaphragmatic defect such that the corresponding lobe of the liver (Left/Right) herniated outside the body and other intra-abdominal organs herniated in the thoracic cavity. All cases except three, had associated upper limb anomalies in the form of syndactyly, camptodactyly, digitalization of thumb, Poland anomaly, hypoplastic scapula and agenesis of forearm on the side of thoracoschisis. The case by Karaman I et al., however had only congenital heart defects which were Patent Ductus Arteriosus and Atrial Septal Defect [1]. Patent Ductus Arteriosus was also seen in the case by Seleim HM et al., [4].

The case of this study differs from the cases of thoracoabdominoschisis in not being a continuous midline body wall defect but two well-separated defects i.e., a right lateral thoracic wall and a midline infraumbilical abdominal wall defect. It also differs from the cases of thoracoschisis by neither having any associated limb or heart defects. Also, in the index case there is associated gastroschisis along with thoracoschisis. The case by Ertan G et al., had omphalocele along with thoracoschisis [2]. However, omphalocele is considered to have an altogether different aetiology i.e., failure of the bowel loops to return into the abdominal cavity after physiological herniation rather than defect in ventral abdominal wall closure leading to gastroschisis [9].

A number of theories on the embryological basis of ventral body wall defects have been proposed. We ruled out some of these as:

1. Early embryonic maldevelopment due to defect in the germ disc leading to blastogenic anomalies[10]: This presents with multiple congenital anomalies which were not there in this case.
2. Amniotic band disruption theory wherein a primary rupture of amnion causes direct mechanical disruption of organs. This would present with body wall defects together with, constriction rings with limb deficiencies, cranio-facial anomalies lacking in the present case [10].

Author	Thoracic defect	Abdominal defect	Additional abnormalities
Karaman I et al., [1]	Left eighth intercostal space	None	ASD, PDA
Ertan G et al., [2]	Case 1-Right sided thoracic wall defect Case 2- Right lateral chest wall defect	Case 1- Diaphragmatic eventration Case 2-Omphalocele with eventrated diaphragm	Case 1-B/L short arms with clinodactyly of the right 5 <sup>th</sup> finger, a digitalised thumb and camptodactyly of the left index finger
Bhattacharya NC et al., [3]	Right thoracic wall defect	Right supraumbilical abdominal wall defect	Agenesis of the right upper limb with a rudimentary finger-like structure attached to the right shoulder
Seleim HM et al., [4]	Left anterior thoracic wall widely spaced ribs	-	Dextrocardia, PDA
Davies MRQ and Cywes S [5]	Left third intercostal	Left anterolateral diaphragmatic defect	No left forearm, syndactyly, dextrocardia
Bamforth JS et al., [6]	Left sixth rib	Left posterior diaphragmatic defect	Left Poland anomaly, left scapula hypoplastic, no humerus-radius-ulna, dextrocardia, ectopic pancreas in the duodenum, intestines in the left hemithorax
Derbent M and Balci S [7]	Right second to fourth ribs	Right anterolateral diaphragmatic defect	-
Biri A et al., [8]	Left (location not specified)	Hiatus hernia	Left forearm agenesis, right arm-hand agenesis, spleen, stomach, colon, liver herniated into the thorax, VSD
Present case	Right eighth and ninth ribs	Gastroschisis (adjacent and inferior to the umbilicus)	None

**[Table/Fig-4]:** Cases of thoracoschisis reported in literature. ASD: Anterior septal defect; PDA: Patent ductus arteriosus; VSD: Ventral septal defect; B/L: Bilateral

- Vascular disruption theory: Here altered vascular blood flow leads to disruption and incomplete embryonic development. This is less likely in this case as there were two distant body wall defects and not a localised one [10].
- Body wall hypothesis proposed by Duhamel B in 1963 [11] says that a primary effect on the mesenchyme (lateral plate mesoderm) of the lateral body folds by a teratogen is responsible for ventral body wall defects. The teratogenic effect is on cell proliferation, migration, production of extracellular matrix resulting in failure of proper movement and/or fusion of lateral body folds. According to Sadler TW and Rasmussen SA [12], this is the most unifying theory to explain ventral body wall defects such as ectopia cordis, gastroschisis and bladder exstrophy.

In this case, authors speculated that a teratogen acted additionally on paraxial mesoderm, the cells which migrate into the lateral body folds to form rib cartilages and muscles of the limbs and body wall

thus explaining the associated lateral thoracic wall defect in the form of thoracoschisis. However, it is intimidating to know why a teratogen act on two different developmental blocks and why only on the right thoracic paraxial mesodermal cells.

Therefore, this case was presented due to unusual type of body wall defect in two different regions of the body i.e., thorax and abdomen-one in midline and the other lateral.

There are a few more points in this case that we would like to highlight. The antenatal diagnosis of meningomyelocele was incorrect highlighting the importance of doing fetal autopsies. The maternal age was 19 years. Notably the only epidemiologic feature found consistent with cases of gastroschisis is young maternal age [13]. The horizontal position of the stomach noted in this case has been proposed as a marker for diaphragmatic hernia on antenatal USG scan [14]. In the absence of any report of the familial occurrence of such case, we considered the malformation to be a sporadic event and counseled the parents for a low probability of recurrence.

## CONCLUSION(S)

This case of fetal autopsy had two well-separated ventral body wall defects which help in understanding of the embryological basis of body wall defects. The Body Wall hypothesis proposing a primary teratogenic effect on the mesenchyme of the lateral plate mesoderm is considered to be the most satisfactory explanation for majority of the ventral body wall defects.

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