PARTIAL AGENESIS OF THE DIAPHRAGM ASSOCIATED WITH PARTIAL AGENESIS OF THE PERICARDIUM IN AN ADULT BANTU MALE*

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An adult Bantu cadaver was sent in from the country after an autopsy had been done for an accidental death due to a car accident. There were multiple fractures of limbs and ribs and the immediate cause of death was probably rupture of the atria or an atrium. At autopsy a large diaphragmatic hernia of unusual appearance was found and the body was sent to the Department of Anatomy, University of Pretoria, with an interesting clinical history.

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During 1964 the deceased was treated in the Zeerust Hospital for 4 weeks after he had fallen off a wagon and had sustained severe injuries to his head. For 14 days he was unconscious and was nursed flat on his back. He was under medical supervision for 6 months and was then declared unfit due to permanent brain damage. No clinical signs were found to necessitate an X-ray examination of the thorax. There were never any complaints of dyspnoea or dyspepsia, or indications of cardiac incompetence.

The clinical history is most important in view of the anatomical findings submitted here.

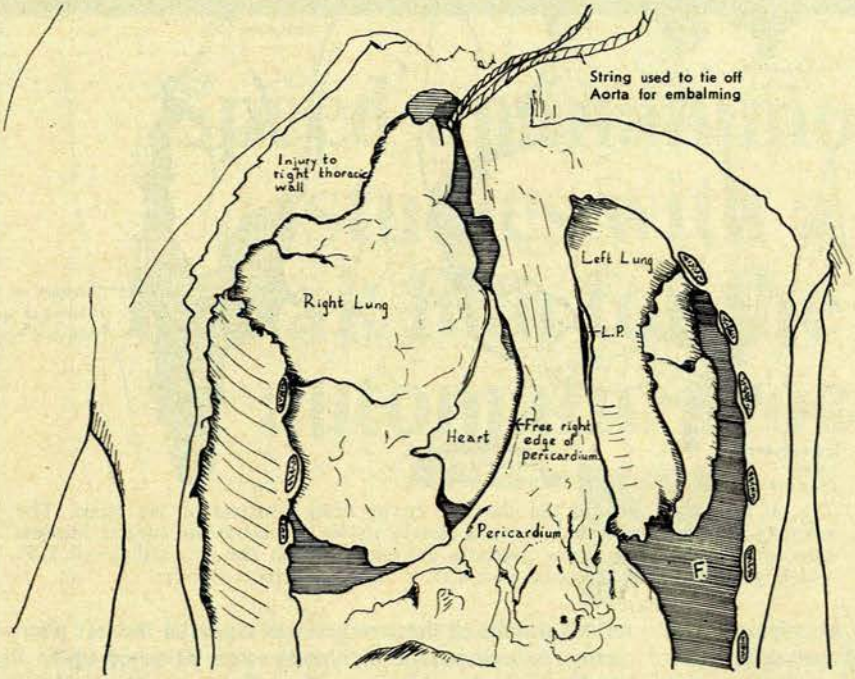


Fig. 1. Arrangement of organs in thorax after removal of the anterior wall. Note the displacement of heart to the right. The right lung, although hypoplastic, appears large due to its anterior position as compared with the left lung which was situated more posteriorly due to the distended fundus of the stomach in the left pleural cavity. L.P. = left phrenic nerve dissected out; F. = fundus of stomach was situated here.

ANATOMICAL FINDINGS

The abdominal contents, as far as the gastro-intestinal tract was concerned, consisted of duodenum, small intestine and large intestine, with the transverse colon and greater omentum sealing off the abdominal from the thoracic contents. The spleen was in the abdomen below the left posterolateral ledge of diaphragm. A small tongue-like projection of liver was wedged in below the smaller ledge of diaphragm on the right side. The kidneys and adrenals were in their usual position.

Inspection of the thoracic contents *in situ* after removal of the anterior thoracic wall showed that the mediastinum was displaced to the right. The pericardial sac had developed anteriorly and on the left side but was absent inferiorly (i.e. the diaphragmatic surface) and to the right (Figs. 1 - 3).

In the right pleural cavity was found the hypoplastic right lung which was tucked in between the liver and the heart anteriorly. Fig. 4 shows its actual appearance. The liver stretched up to a third rib and was entirely intrathoracic except for a small tongue of liver tissue caught below the right ledge of diaphragm (Figs. 5 - 7). The diaphragm showed a ragged edge after removal from the cleft in the liver (Fig. 8).

The visceral surface of the liver faced to the left and inferiorly. The stomach, with the exception of its fundus and the transverse colon, was related to it as part of the thoracic contents on the right side. The ligamentum teres of the liver was long and had a mesentery showing several varicosities. The lesser omentum was a continuous fold of peritoneum stretching from the gallbladder to the fissure of the ligamentum venosum, and was at the same time the elongated porta hepatis.

The heart was small and sharply displaced to the right. The ventricles were underdeveloped, especially the right which formed the right border of the heart. The pulmonary trunk and ascending aorta appeared to be of normal size (Figs. 2 and 9). Both the atria were situated posteriorly with the exception of their auricles. The superior and inferior caval veins were of smaller diameter than normal. The inferior vena cava, that is the prehepatic segment, showed considerable elongation and a distinct caval plica. The anatomy was distorted due to rupture posteriorly of a very thin atrial wall. There were, however, definite indications that there had perhaps been an incomplete separation of the embryonic atrium and associated maldevelopment. The autopsy findings confirmed this. The fact that the inferior vena cava opened in a recess where one of the right pulmonary veins also opened was a further indication of maldevelopment of the atrium. The atrial wall was extremely thin, closely resembling that of a vein, which could account for its rupture. The interatrial septum could not be identified but due to the trauma one could not come to a definite conclusion as to the correct state of affairs.

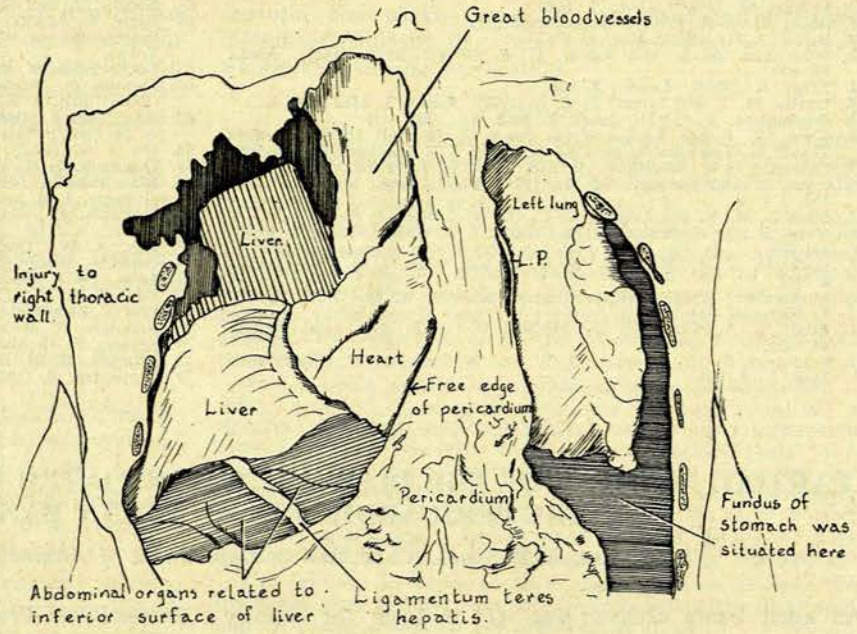


Fig. 2. The right lung was removed to show the liver and heart on the right side. L.P. = left phrenic nerve dissected out.

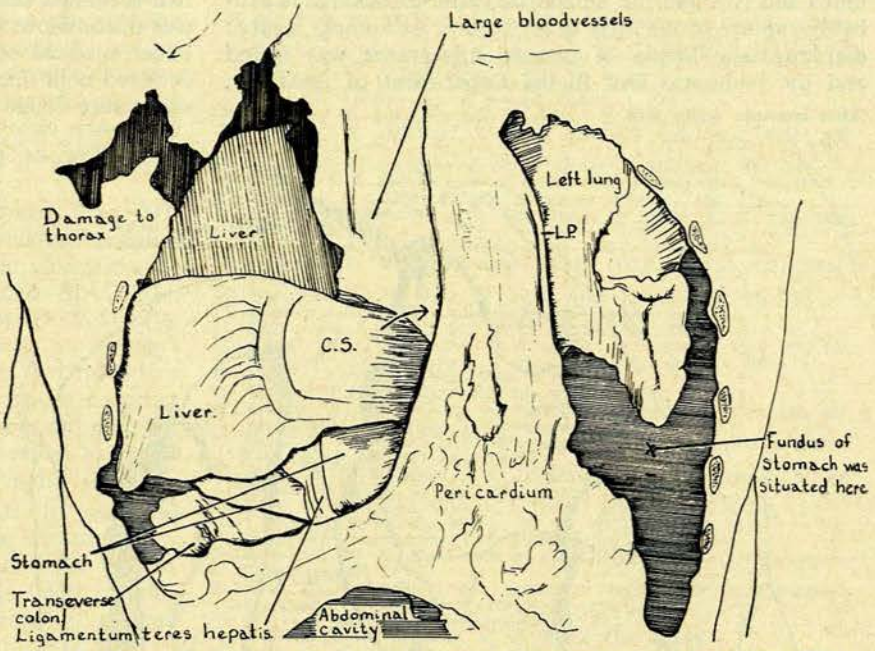


Fig. 3. The right side of the thoracic cavity after removal of the heart. The opening into the pericardial cavity is clearly visible as well as the cardiac impression on the liver. The arrow indicates the entrance into the pericardial sac. L.P. = left phrenic nerve dissected out; C.S. = cardiac surface of liver.

The left pleural cavity contained a fairly normal-looking lung which was somewhat smaller than usual but better developed than the right lung (Fig. 10). A grossly distended fundus of the stomach was found in the left pleural cavity. A long pedicle of blood-vessels stretched up to the fundus from the abdomen. The left gap in the diaphragm was overlain by transverse colon and greater omentum.

The diaphragmatic defect measured approximately 18 × 8 cm. in the midline. There was a total absence of diaphragm anteriorly (Figs. 8, 11 and 12). The existing

diaphragm resembled two posterolateral crescents, better developed on the left side. The larger gap was towards the right and communicated freely with the right pleural cavity and pericardial sac. The crura were present and the oesophagus pierced the diaphragm surrounded by fibres of the right crus. The stomach was, however, intrathoracic due to the fact that there was no diaphragm anterior to the hiatus. A strand of muscle fibres stretched from the left of the oesophagus to the plica of the inferior vena cava which was situated at the point where the diaphragm ended posteriorly as a clear-cut ridge (Fig. 13).

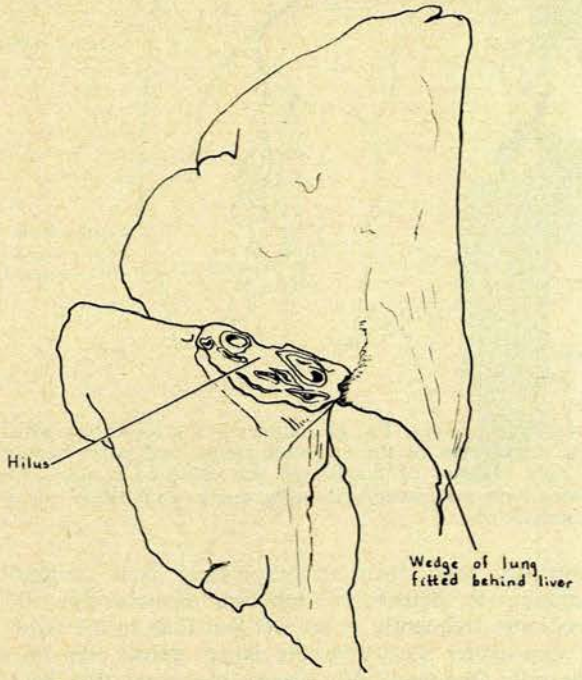


Fig. 4. Right lung, mediastinal view. Note the distortion of the lung.

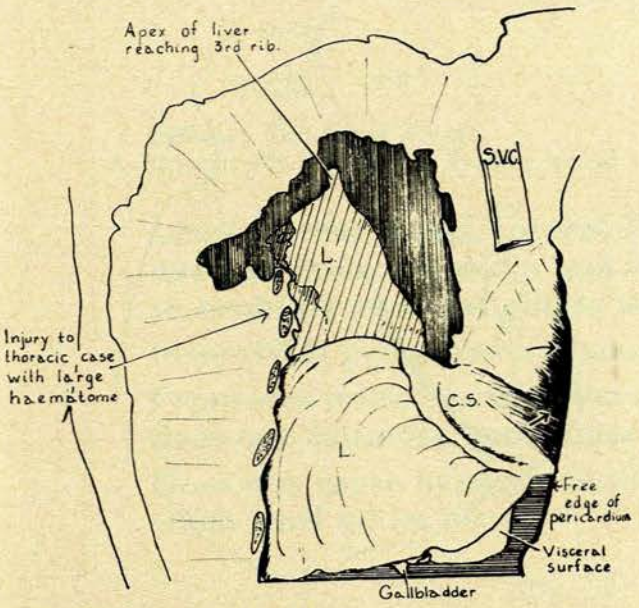


Fig. 5. The liver in the right thoracic cavity presenting a vertical part stretching up to the third rib, a flattened surface for the heart, a curved costal surface and a visceral surface facing inferiorly and to the left. The arrow indicates the entrance into the pericardial sac. C.S. = cardiac surface; S.V.C. = superior vena cava; L. = liver.

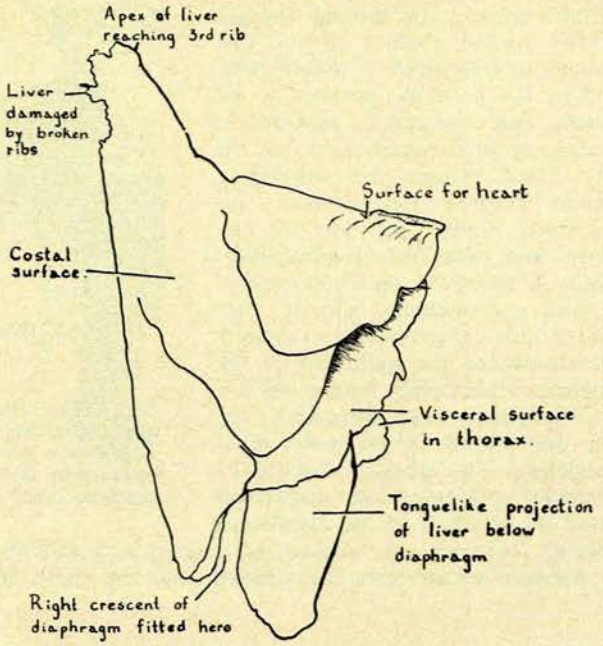


Fig. 6. The liver after removal, showing the same entities as in Fig. 5 as well as the tongue-like projection of the liver below the crescent of diaphragm which was adherent to the cleft in the liver.

The phrenic nerves were present on both sides and were well developed. The right phrenic nerve was found on an underdeveloped superior vena cava continuing down the posterior ridge of pericardium and then onto the inferior caval plica, and from there it entered and supplied the small posterolateral sickle-shaped ledge of diaphragm as well as the right crus (Figs. 8 and 13). On the left side the phrenic nerve had its usual relationship to the pericardium and entered and supplied the larger left posterolateral diaphragmatic crescent and crus (Figs. 1 - 3 and 12).

DISCUSSION

Various classifications are given for diaphragmatic hernia. Hume[1] refers to congenital and acquired hernia, of which the acquired can be either traumatic or non-traumatic. This case can definitely be regarded as congenital, representing a faulty subdivision of the embryonic coelom in view of the fact that the right and diaphragmatic surfaces of the pericardium were also absent, with concomitant anomalies of the heart. Four types of congenital diaphragmatic hernias are described by Hume:[2]

1. Hernia through the hiatus pleuroperitonealis.
2. Hernia through the dome.
3. Hernia through the oesophageal orifice.
4. Absence of the left half of the diaphragm.

This particular case does not fit in with any of these types listed above. According to Harrington[2-5] the most

common sites of congenital hernia are probably found in the following order of frequency: Through the hiatus pleuroperitonealis (foramen of Bochdalik); through the oesophageal hiatus; through an anterior substernal opening (foramen of Morgagni or Larrey's space); and through the gap left by partial absence of the diaphragm, a gap which is usually situated in the posterior portion of the muscle. This case can be regarded as a large gap in the diaphragm, but the only place where the diaphragm showed normal development was posteriorly. Bowen[6] reviewed the literature and classified diaphragmatic hernia as traumatic and non-traumatic, and non-traumatic may be subdivided into congenital and acquired. The closure of the right leaf of the diaphragm takes place before the left leaf. The right leaf is protected by the liver and assists in its more rapid completion. The stomach reaches its permanent site below the diaphragm before the left half of the diaphragm is closed. Therefore any unusual increase in intra-abdominal pressure would force the stomach into the chest. In accordance with these embryological facts, congenital diaphragmatic defects are reported as occurring 10 - 18 times more frequently in the left leaf than in the right. In the case under discussion the larger defect was on the right side. Donovan[7] also stresses the point that because the liver lies over the right pleuroperitoneal hiatus, hernias occur more often on the left than the right side.

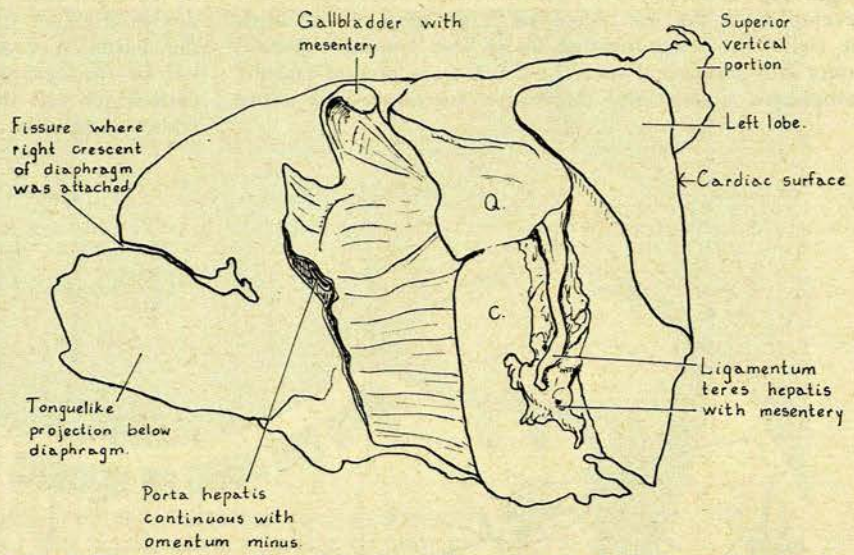


Fig. 7. The visceral surface of the liver. The gallbladder had a mesentery which was continuous with the peritoneum of the omentum minus and porta hepatis. The fissure where the right crescent of diaphragm was attached is seen. The ligamentum teres was long with a mesentery showing many varicosities. Q. = quadrate lobe; C. = caudate lobe.

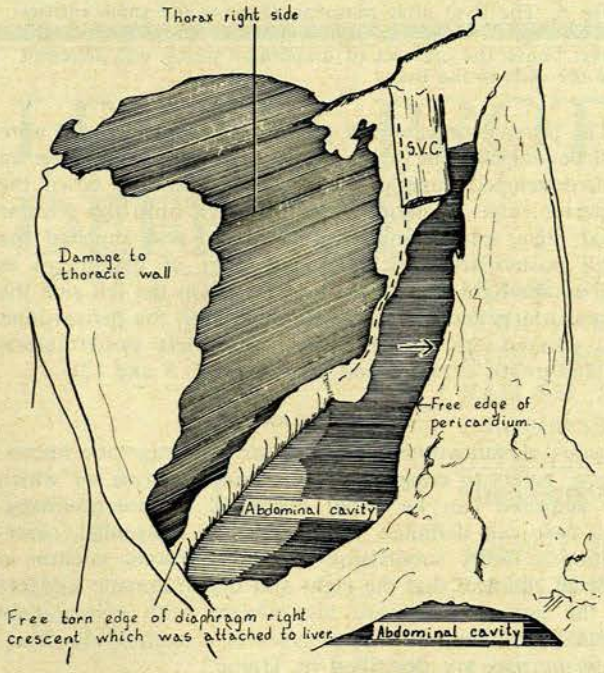


Fig. 8. The right thoracic cavity after the lung, heart and liver had been removed. The free torn edge of the diaphragm can be seen and below it the abdominal cavity. The pleural, pericardial and peritoneal cavities communicate freely. The course of the right phrenic nerve is indicated by the stippled line. Arrow indicates opening into pericardial cavity. S.V.C. = superior vena cava.

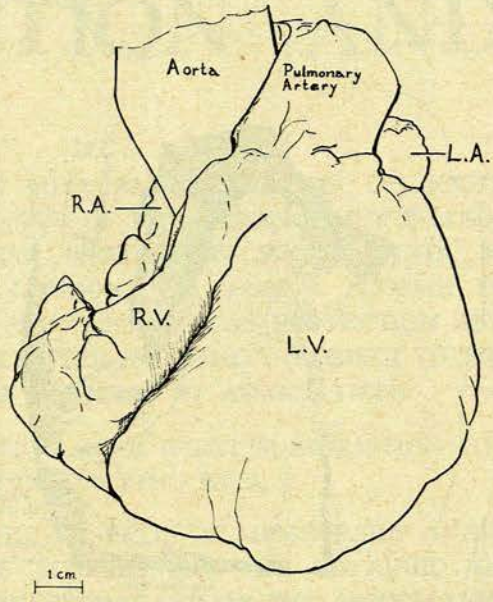


Fig. 9. An anterior view of the heart drawn to scale and enlarged × ·25. The heart shows the foetal arrangement with non-rotation to the left. The ventricles are anteriorly. The atria is posteriorly with only the right and left auricles visible. R.V. = right ventricle; L.V. = left ventricle; R.A. = right auricle; L.A. = left auricle.

The surgical problems as regards diaphragmatic hernias are discussed by Rives and Baker.[8] They mention that large marginal defects rarely occur in the anterior portion of the diaphragm and that very large defects in the diaphragm are much more common in its posterior than in its anterior portion. They mention in passing that they have never actually operated on a case with a large anterior defect. Schwalbe[9] referred to total agenesis of the diaphragm which was seen only in cases with associated gross congenital abnormalities incompatible with life. He mentions the publications of Von Gössnitz[10-12] on congenital diaphragmatic hernias, but this particular case does not fit in with any of the cases illustrated by him.

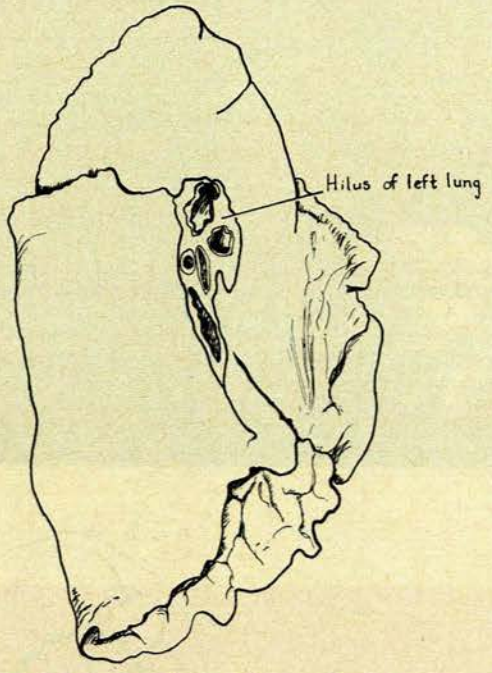


Fig. 10. Left lung, mediastinal view, relatively normal in appearance, although somewhat smaller than usual.

Embryology

As little information can be found in the literature about a large anterior defect of the diaphragm, one has to turn to the embryological development for an explanation. The primitive coelom arises by the splitting of the lateral mesoderm on either side of the body into splanchnic and somatic layers. The intra-embryonic portion of the primitive coelom gives rise to the body cavities. The heart develops in its most anterior portion. In its differentiation, as well as in its initial appearance, the pericardial part of the coelom is precocious (Fig. 14).[13] The first indication of the partitioning off of the pericardial region from the rest of the coelom is given by the formation of the septum transversum (Figs. 15 and 16).[14] At this stage it is essentially a shelf-like structure between the heart and the liver. Much of the superior surface of the septum transversum becomes the floor of the pericardial cavity. It seems logical that in this case the septum transversum did not develop and therefore the floor of the pericardial sac was absent and the heart rested directly on the liver. Absence of the septum transversum would

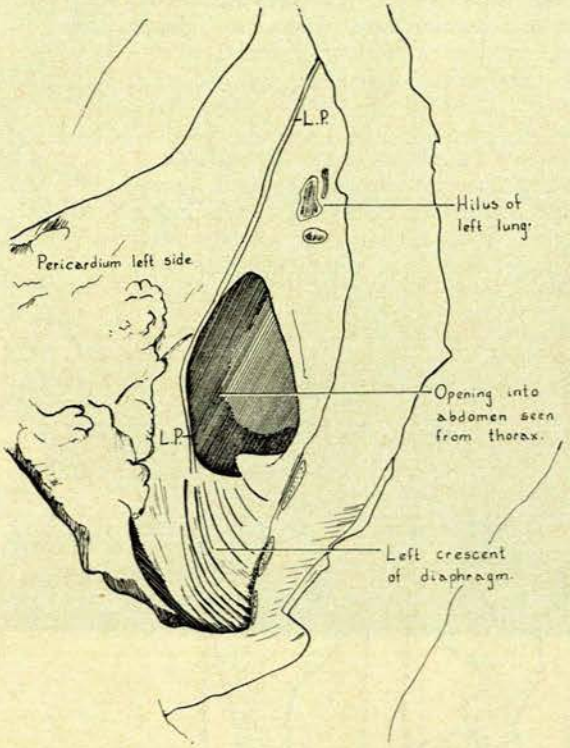


Fig. 12. Left thoracic cavity after removal of the lung. The oval-shaped opening into the peritoneal cavity is clearly seen and the posterolateral left crescent of the diaphragm is supplied by the left phrenic nerve. L.P. = left phrenic nerve.

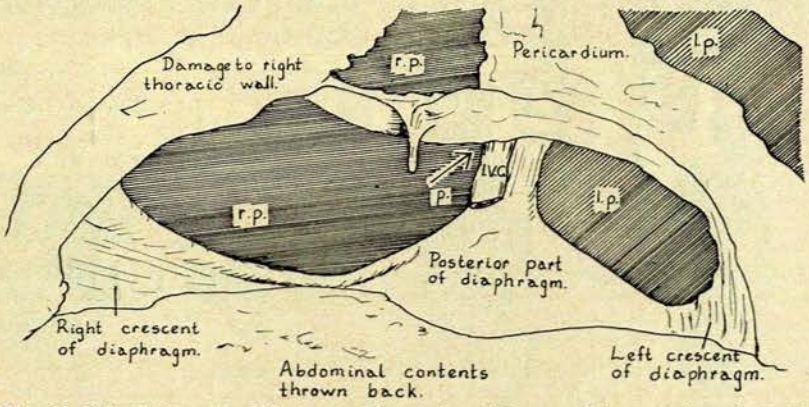


Fig. 11. Diaphragmatic defect seen from the abdomen. The openings into the right pleural cavity, pericardium and left pleural cavity are indicated by arrows. The smaller right and larger left posterolateral crescents of the diaphragm were the only parts which had developed and are clearly visible. r.p. = right pleural cavity; p. = pericardial sac; l.p. = left pleural cavity; I.V.C. = inferior vena cava.

also explain why the liver migrated up into the left thoracic cavity, causing underdevelopment of the left lung.

The complete isolation from one another of the pericardial, pleural and peritoneal portions of the coelom is brought about by the growth of additional folds which become associated with the septum transversum. On either side, the fold extending cephalad from the primary elevation about the duct of Cuvier is called the pleuropericardial fold, and the fold which extends caudad is called the pleuroperitoneal fold. The pleuropericardial folds complete the separation of the pericardial cavity and the rest of the coelom (Fig. 17).[15] In this particular case there was maldevelopment of the right pleuropericardial fold, which

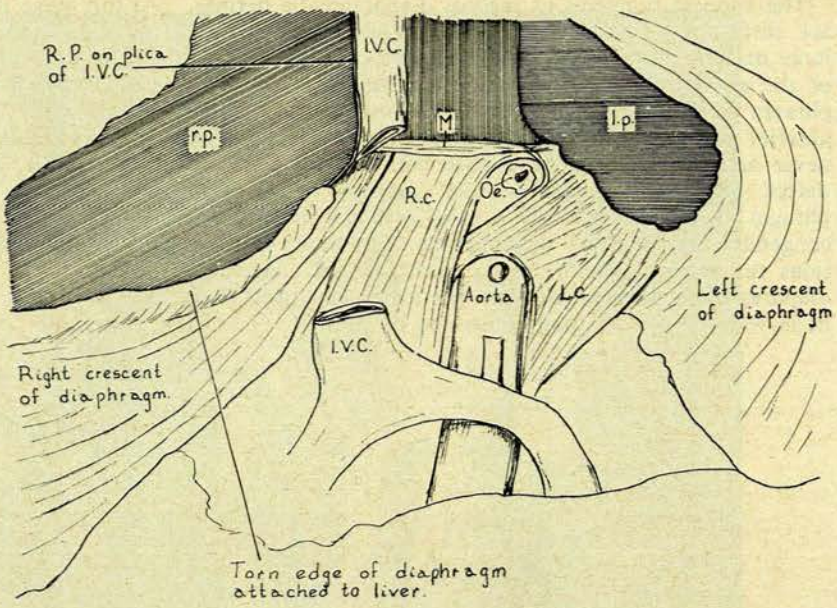


Fig. 13. Dissection of the crura of the diaphragm. The oesophagus and aorta are seen in their normal relationship to it. A strand of muscle fibres stretched from the left of the oesophagus to the inferior vena cava, which had a distinct plica. The right phrenic nerve was found on it and then entered the torn crescent of diaphragm. I.V.C. = inferior vena cava; r.p. = right pleural cavity; l.p. = left pleural cavity; R.P. = right phrenic; Oe. = oesophagus; R.c. = right crus; L.c. = left crus; M = muscle bundle.

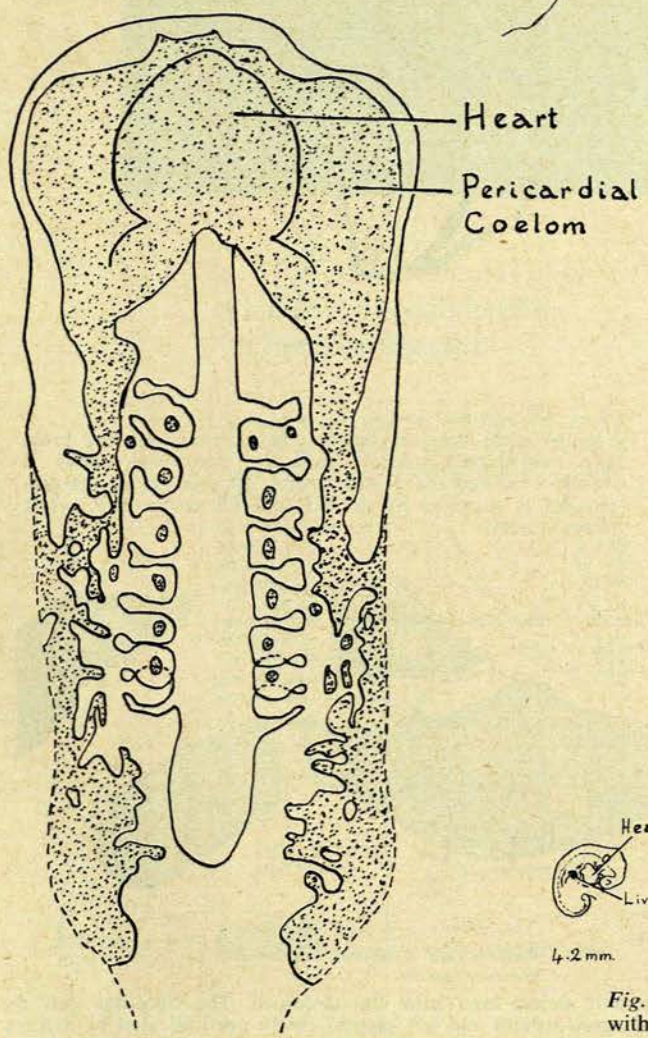


Fig. 14. Coelom human embryo, 8 somites, to show that the pericardial coelom is precocious in its development (redrawn and adapted from Starck).[13]

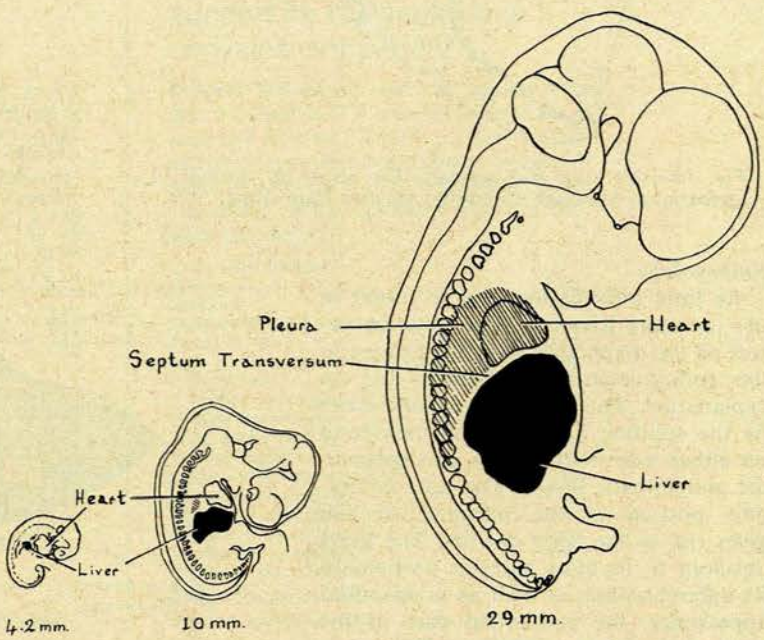


Fig. 15. The relationship and size of the heart as compared with the liver is shown at 4·2 mm., 10 mm. and 29 mm. The expansion of the liver has an important relationship to the development of the diaphragm (redrawn and adapted from Blechschmidt).[14]

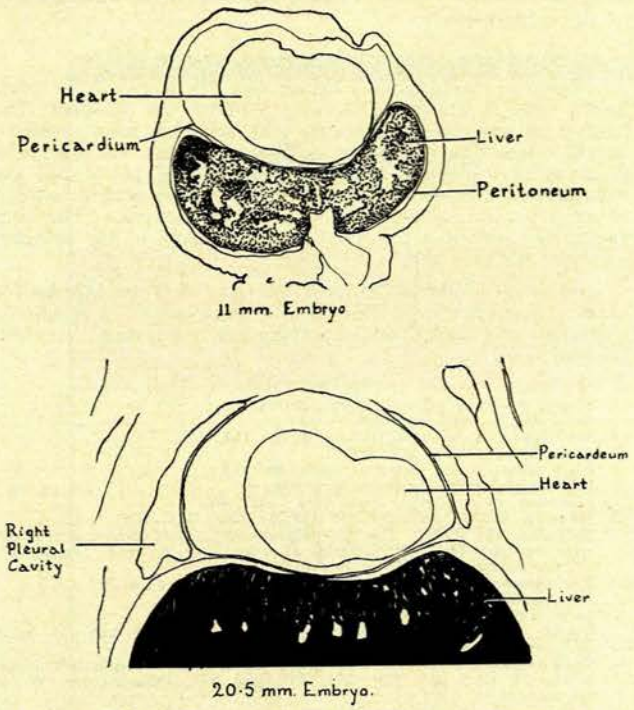


Fig. 16. The liver was originally small as compared with the heart. In the 11-mm. embryo the upper edge of the liver curves convexly upwards along the diaphragm until it partially covers the heart like a saddle. Later in the 20·5-mm. embryo the liver becomes flatter and at the same time the pleura protrude into the enlarging space on either side of the heart as the pleural sacs (redrawn and adapted from Blechschmidt).[34]

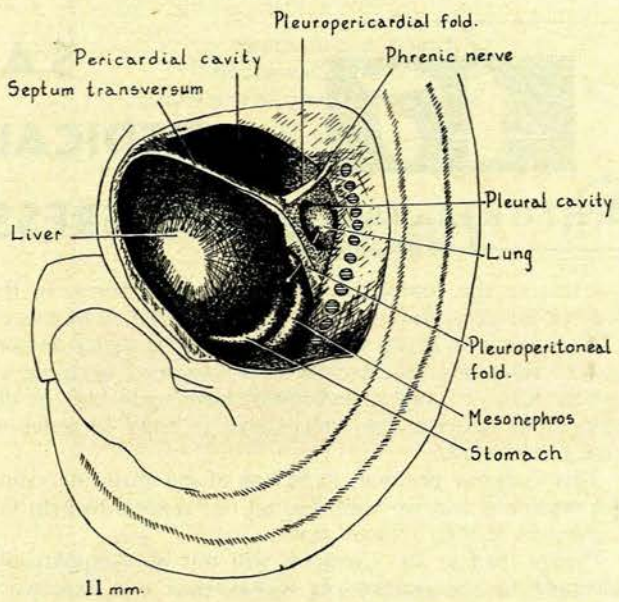
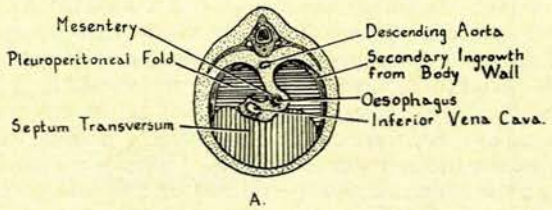


Fig. 17. Human coelomic cavity 11 mm., embryo viewed from the left side after removal of the body wall. Septum transversum and pleuropericardial and pleuroperitoneal folds are shown. Pleuroperitoneal canal indicated by arrow (redrawn and adapted from Arey).[35]

explains the direct relationship of the right lung to the heart. For a considerable time pleural canals posterior to the septum transversum form a communication between the thoracic and abdominal cavities. Gradually these canals are obliterated by the outgrowth from the posterolateral body wall of the pleuroperitoneal folds. From the beginning these folds are continuous with the dorsal margin of the septum transversum, and when their posterior borders meet and fuse with each other and with a small persisting part of the primary dorsal mesentery the diaphragm is completed. This takes place at about the eighth week. Considerably later a secondary bilateral ingrowth of muscle from the body wall forms a narrow ridge around the posterolateral margins of the diaphragm. On the right side the secondary ingrowth of muscle was probably responsible for the narrow crescent of diaphragm attached to the liver.



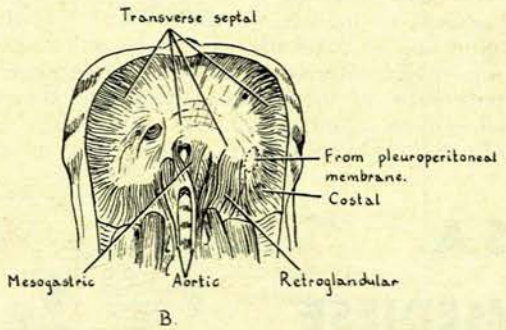


Fig. 18. A: Diagram indicating the embryological origin of various regions of diaphragm (according to Broman,[16] redrawn from Patten[17]). B: General areas of the adult diaphragm contributed by the primitive components (adapted from Wells,[19] redrawn from Hollinshead[20]).

Pericardial, pleural and peritoneal cavities thus freely communicated in this case. On the left side the crescent of diaphragm was better developed and served as a shelf which delimited the spleen to the abdomen. On this side the pleuroperitoneal fold probably did develop, as was also the case with the pleuropericardial fold. The embryology reviewed here refers to the work of Broman[16] and is referred to by Patten,[17] Potter[18] and Starck[22] (Fig. 18A). Wells,[19] however, came to the conclusion that the development of the human diaphragm is more complicated and that it develops by the fusion of not 4 but 6 elements, some of these paired. There are transverse septal membranes; right and left pleuroperitoneal, which contribute a small rather than a large area; right and left costal; right and left retroglandular; aortic; and mesogastric membranes (Fig. 18B).[20] It is difficult to explain

embryological development of the diaphragm described by Wells.

The prehepatic portion of the inferior vena cava was much longer than usual but smaller in diameter, probably due to the displacement of the liver and anomalies of the heart. The heart was small, with displacement to the right, and no rotation had occurred; hence it was still in the foetal position (Fig. 9). There was a strong indication of an anomalous pulmonary vein and incomplete separation of the primitive atrium and underdevelopment of its walls, which was probably the reason why the atrium ruptured, the thickness of its wall being comparable with that of a vein.

Respiration

Finally, the most interesting point which arises is the mechanism of respiration in this case and survival into adult life. Potter points out that in her experience infants with a large diaphragmatic hernia had associated pulmonary hypoplasia of such a severe nature that the lungs were incapable of supporting extra-uterine existence, and death occurred immediately after birth or within a few hours. Furthermore, such small portions of the diaphragm have usually been present that to effect a closure would have been extremely difficult had the infants survived long enough for operation to be performed. In this case the liver apparently served to seal off the right side of the thorax and the liver supported the diaphragmatic surface of the heart. The opening in the diaphragm as a whole, and especially on the left, was further sealed by the transverse colon and in particular by a very well-developed omentum majus. Morrison[21] called the omentum majus the 'policeman' of the abdomen, and the extremely well-developed omentum majus was probably the main reason why this subject survived into adulthood in spite of gross congenital defects. One is again amazed at the adaptability of the human body.

SUMMARY

A congenital diaphragmatic hernia was discovered in an adult Bantu male at autopsy following a motor car accident. The diaphragm was absent anteriorly with relatively small posterolateral ridges but was better developed on the left side. The diaphragmatic surface and right side of the pericardial sac were absent and there was consequently free communication between peritoneal, pleural and pericardial cavities. The congenital abnormalities are discussed in the light of the embryology and the literature is reviewed.

I am greatly indebted to Dr C. B. Otto of Great Mariko for sending this interesting case to the Department of Anatomy, University of Pretoria, and supplying the case history and clinical findings.

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