

Thoracic ectopia cordis, sternal agenesis, partial ectopia hepatica and fissure abdominalis in a German Shepherd puppy with milder incomplete pentalogy of Cantrell. Clinical case

Ectopia cordis torácica, agenesia esternal, ectopia hepática parcial y fisura abdominal en un cachorro de pastor alemán con pentalogía incompleta de Cantrell más leve. Caso clínico

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ABSTRACT

A newborn German Shepherd breed male puppy was presented with the complaint that some organs were formed outside the chest and abdomen. The rhythmic movements of the swelling under the chest pointed out the heart and the swelling under the abdomen was covered with a membrane. On the lateral radiograph of body, sternal agenesis, extra-thoracic location of the heart, and normal diaphragmatic border were seen, while the swelling in the abdomen was thought to be liver. During the diagnostic steps, the puppy died, and necropsy was performed. At necropsy, it was observed that the heart wrapped with the pericardium was placed extra-thoracically (thoracic ectopia cordis) due to sternal agenesis, and the liver was partially located extra-abdominally (ectopia hepatica) due to fissura abdominalis, which was surrounded by the peritoneum. Both radiological and necropsy findings revealed that there was no rib structure after the costochondral joints. Diaphragm was present, and there was no abnormality in the other intrathoracic and intraabdominal organs. Here, it is aimed to report thoracic ectopia cordis, sternal agenesis, ectopia hepatica and fissura abdominalis in a German Shepherd puppy with milder incomplete pentalogy of Cantrell, and to present the radiological and necropsy findings of the anomaly.

Key words: Ectopia hepatica; fissura abdominalis; puppy; sternal agenesis; thoracic ectopia cordis

RESUMEN

A un cachorro macho recién nacido de raza pastor alemán se le presentó la novedad de que algunos órganos se habían formado fuera del pecho y el abdomen. Los movimientos rítmicos de la tumefacción debajo del pecho señalaban el corazón y la tumefacción debajo del abdomen estaba cubierta con una membrana. En la radiografía lateral del cuerpo se observó agenesia del esternón, ubicación extratorácica del corazón y borde diafragmático normal, mientras que se pensó que la tumefacción en el abdomen era hepática. Durante los pasos de diagnósticos, el cachorro falleció y se le realizó la necropsia. En la necropsia se observó que el corazón envuelto con el pericardio estaba ubicado extratorácicamente (ectopia cordis torácica) debido a la agenesia del esternón, y el hígado estaba parcialmente localizado extraabdominalmente (ectopia hepática) debido a la fisura abdominal, que estaba rodeada por el peritoneo. Tanto los hallazgos radiológicos como los de necropsia revelaron que no había estructura costal después de las articulaciones costochondrales. Un árbitro señala que el diafragma no presentaba alteraciones, al igual que el resto de los órganos intratorácico e intraabdominales. El presente trabajo tuvo como objetivo reportar ectopia cordis torácica, agenesia esternal, ectopia hepática y fisura abdominal en un cachorro de pastor alemán con pentalogía incompleta de Cantrell más leve, y presentar los hallazgos radiológicos y de necropsia de la anomalía.

Palabras clave: Ectopia hepática; fisura abdominal; cachorro; agenesia esternal; ectopia cordis torácica

INTRODUCTION

Multiple congenital anomalies are less common in domestic animals. Although the etiology of some congenital anomalies is not known, it is considered that anomalies are caused by genetic factors, mutations, chromosomal anomalies, infectious and environmental factors [1]. Ectopia is a congenital anomaly defined as an organ located anatomically in another part of the body or in another organ or tissue [2].

Ectopia cordis is the presence of the heart partially or totally outside the thoracic cavity [3, 4]. Similarly, ectopia hepatica is an anomaly characterized by different location of the hepatic parenchyma [5]. According to the position of the heart, ectopia cordis is classified as cervical, cervicothoracic, thoracic, thoracoabdominal and abdominal [3, 4]. In cases of ectopia hepatica, a small liver tissue may be attached to the gallbladder, intraabdominal organs and ligaments separately, or a small accessory liver lobe may be seen in different localization [5]. The reported sternum anomalies include branched xiphoid process, V-shaped bifurcation, sternum bifidum, synchondrosis sternii, wedge-shaped or asymmetric sternum, sternum gallinaceum and sternal foramen [6]. Fissura abdominalis is a defect caused by the absence of complete fusion of the abdominal wall in the median line [7].

Ectopia cordis have been reported in cattle (*Bos taurus*), goat (*Capra hircus*), horses (*Equus caballus*) [3, 4] and human neonates [8, 9]; and ectopia hepatica is seen in cattle, dogs (*Canis lupus familiaris*) and cats (*Felis catus*) [5]. Although defects of the thorax are reported, sternal agenesis has not been reported together with the other congenital anomalies [3]. In addition, fissura abdominalis cases may be encountered in dogs with other anomalies [7].

Pentalogy of Cantrell defined as midline closure syndrome in humans and includes thoracoabdominal wall, sternal, diaphragmatic, pericardial and intracardiac defects [8, 10, 11, 12]. Depending on the size of the pathology, Cantrell's pentalogy is divided into different classes as class I, II and III. A complete pentalogy included all five defects, which named as complete pentalogy and classified as class I; an probable/incomplete pentalogy includes four defects including cardiac and ventral wall defects classified as class II and a milder incomplete pentalogy includes any combination of given pathologies with sternal defect classified as class III [8, 10, 11, 13]. Although some cases can be treated in human medicine, the majority of cases continue to have significant morbidity and mortality [10].

Taking these literatures into consideration, here, it is aimed to report radiological and necropsy findings of a multiple congenital anomaly (thoracic ectopia cordis, sternal agenesis, ectopia hepatica and fissura abdominalis) with a milder incomplete pentalogy of Cantrell in a German Shepherd puppy.

MATERIALS AND METHODS

Case history

A German Shepherd breed male puppy was presented with the complaint of abnormal thoracic and abdominal swellings. In the anamnesis, it is informed that puppy was one of the three puppies of the unrelated mother and father, and born with two different swellings that moved in the chest area and were motionless in the abdomen. One of the born puppies was normal, one was dead and the presented puppy had anomaly, as well. The mother was 4 years old and gave birth for the third time. No anomaly was observed in the offspring of the same mother and father born in the previous parturitions. The

pregnancy confirmed ultrasonographically at 30th day after mating and no medications were used during gestation.

Clinically, the puppy could not stand up and was restless, his body temperature was 36°C and the other vital parameters were within reference values. Due to the rhythmic movements of the swelling under the chest, it was thought to be the heart and the membrane around the heart could be a pericardium. The basis of the heart and the large vessels were in the thoracic cavity. A different swelling covered with a membrane was observed under the abdomen, which was directed about 1 cm in front of the umbilical cord towards the swelling in the chest. It was thought that this swelling might be a possible liver, because swelling had a solid-flexible consistency during palpation (FIG. 1).



FIGURE 1. Clinical view of the case, extra-thoracic located heart (H) and partial extra-abdominal located liver (L)

During radiological examination (Ajex Meditech Ltd., Ajex160H, Korea), spontaneous death occurred in the puppy. A necropsy was performed to detail the anomaly. However, advanced diagnostic techniques like CT and MRI could give valuable informations, these diagnostic aids were not performed considering the general condition of the puppy, technical reasons and the fact that they provide better information about cardiovascular system evaluations when the animal is alive.

Based on clinical, radiological and necropsy findings, the diagnosis of thoracic ectopia cordis, sternal agenesis, ectopia hepatica and fissura abdominalis was made in the puppy.

RESULTS AND DISCUSSION

Radiologically, the lateral radiograph of the body showed that there was no sternum, the heart was positioned extra-thoracically, and the swelling on the abdomen was at liver localization (FIG. 2). The diaphragmatic border was evident, and normal air radiolucent appearance was present in the trachea and lung area. Apart from these findings, no other anomaly reflected on the radiogram was found.

Diagnostic imaging methods such as CT and MRI provide detailed information about the identification, localization and extent of the pathology, but general anesthesia is required to ensure patient immobility while applying these imaging methods [14]. In the presented case, considering the general condition of the patient,

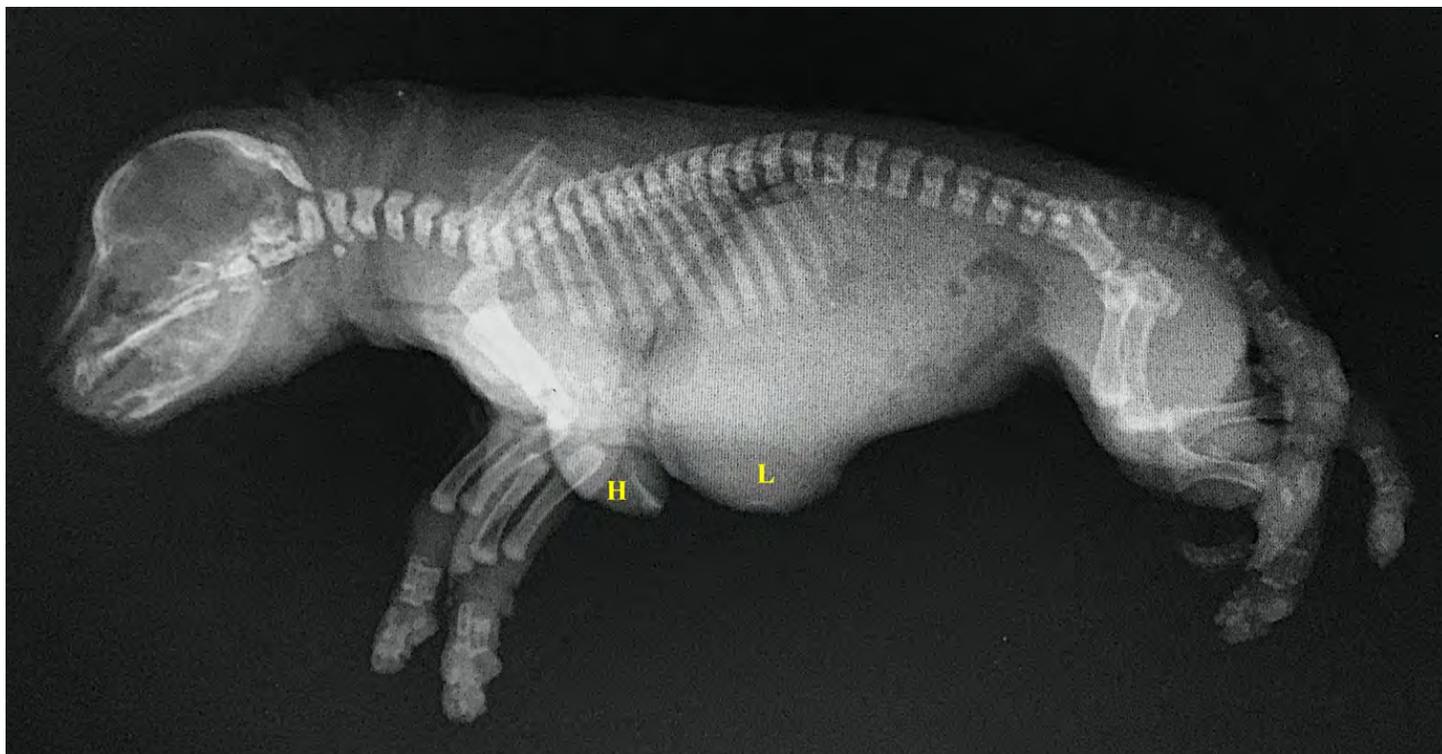


FIGURE 2. Lateral radiograph of the dog shows the absence of the sternum, and the heart (H) and liver (L) are partially located outside to body

the extent of the pathology and the technical conditions advanced diagnostic methods such as CT and MRI were not used. Furthermore, these imaging diagnostic methods are also used in postmortem examinations, but especially in postmortem cardiovascular system evaluations the methods have limitations [15].

At necropsy a longitudinal incision was made along the chest and abdominal median line, starting from the swelling under the chest to the abdominal swelling and caudal abdominal. Thoracic and abdominal cavities were exposed with the cutting procedure. First, the thorax was examined and the membrane on the extra-thoracic heart was found to be a pericardium (FIG. 3). No anomaly was detected in vascular and internal structures of the heart. The ventilation and lobular structures of the lung were normal, and there was no fluid in the peritoneal space. The diaphragm border was normal, but there was no sternum. Also, the parts of the ribs after the costochondral joints were absent (FIG. 4). The reason for the non-closure of the thoracic cavity was sternal agenesis. The cranial abdominal muscles were not closing the midline (fissura abdominalis); thus, the liver was found to be partially located in the abdominal swelling. The membrane covered the liver was the peritoneum (FIG. 3). No other pathology related to the other organs was detected in the examination of thoracic and abdominal cavities.

Ectopias can be seen in one organ or may be observed with other multiple anomalies [2]. These abnormalities affect an organ, system, or even some systems (cardiovascular or gastrointestinal) [4]. The prevalence of thoracic ectopia cordis varies among species [3] and there are a few case reports of ectopic liver in Veterinary Medicine [5]. And among these developmental anomalies a Cantrell's pentalogy, a rare syndrome of human neonates, defined as a collection of

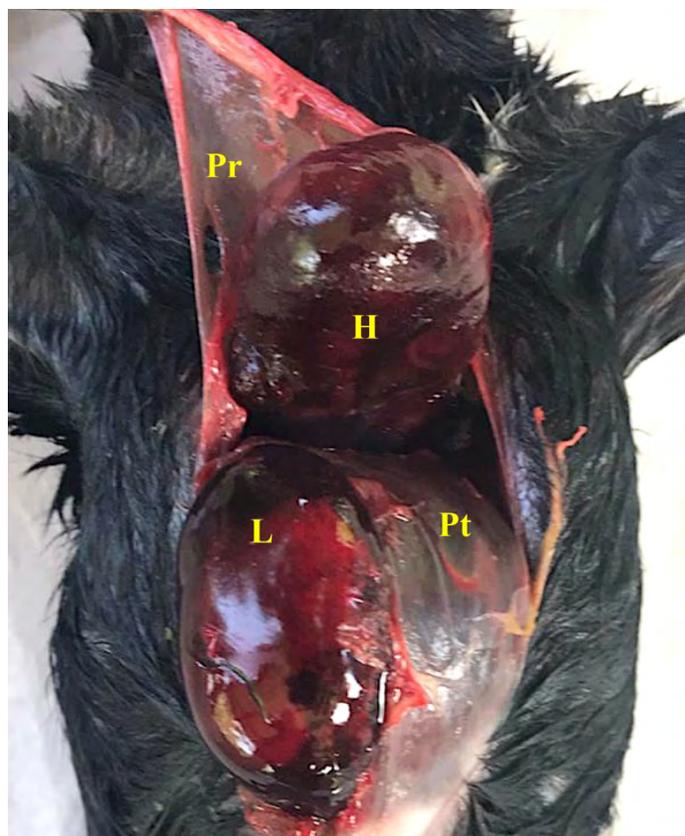


FIGURE 3. At necropsy, the appearance of the heart (H), pericardium (Pr), liver (L) and peritoneum (Pt)

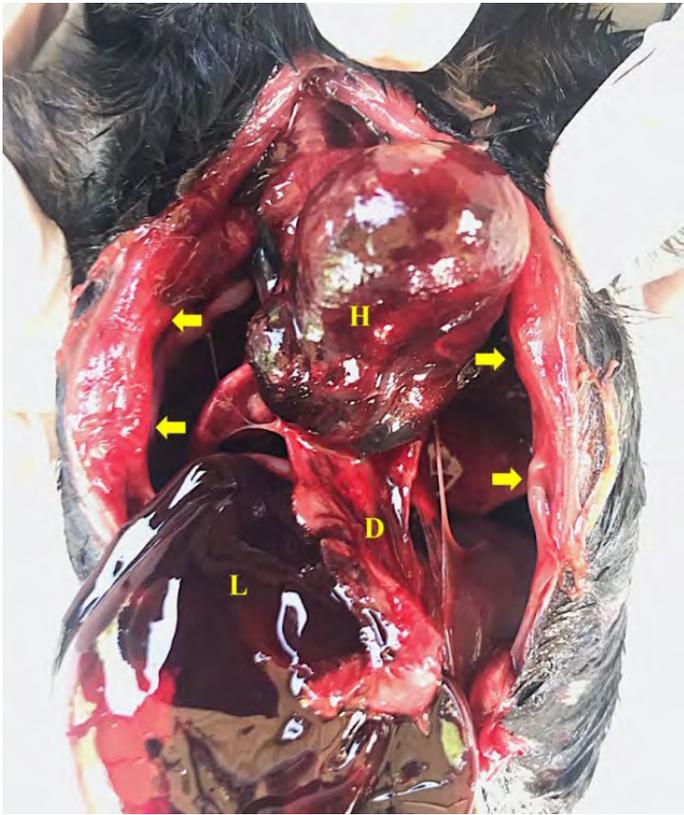


FIGURE 4. In the examination of the thoracic and abdominal cavities, absence of the sternum and partial parts of the ribs after the costochondral joints (arrows), the appearance of the heart (H), diaphragm (D) and liver (L)

congenital midline defects. A Cantrell's pentalogy is characterized by thoracoabdominal wall, sternal, diaphragmatic, pericardial defect and intracardiac anomaly. And it could be classified according to the number of included pathologies as complete or incomplete [10, 11, 13]. Considering this information, it is aimed to contribute to the literature reporting thoracic ectopia cordis, sternal agenesis, ectopia hepatica and fissura abdominalis in a German Shepherd puppy with milder incomplete pentalogy of Cantrell.

Ectopia cordis results from incomplete fusion of the body cavity, especially in the abdominal wall, during development of the fetus [3, 9]. In ectopia cordis cases, the heart appears partially or completely outside the thoracic cavity and may be complicated with other malformations either cardiac or non-cardiac. It is possible to encounter ectopia cordis especially with abdominal wall, sternum, diaphragm, pericardium and heart defects [4, 8, 9]. The thoracic ectopia cordis anomaly encountered in the presented case was together with ectopia hepatica, sternal agenesis and fissura abdominalis anomalies. The sternum has different variations and anomalies [6], and some sternal pathologies such as a shortened and widened manubrium, sternal cleft, sternal foramen, xyphoid cartilage hypoplasia, double appearance sternabrae and presence of multiple sternabrae can be seen in cases with cervical ectopia cordis [1, 4, 6]. In this German Shepherd puppy, radiological and necropsy findings revealed that there was no sternum (sternal agenesis) and rib structure after the costochondral joints.

As reported in the literature, diagnostic imaging techniques present details in ectopia cases [3, 4, 5]. Radiological evaluation of the cases with ectopia cordis is not so much affected due to a fast heartbeat. Radiographs shows the apical sites of the ribs and their sternal connections, if there is [3]. In the presented case, *premortem* radiological examination helped the diagnosis of sternal agenesis. On the lateral radiograph, the heart was outside, the diaphragmatic border was normal and the swelling on the abdomen was in the localization of the liver.

In an ectopia cordis, surgically, after the heart is placed in the chest cavity, death may occur in the early postoperative period and the survival time of the cases varies from 3 minutes to several years. In addition, neonatal death usually occurs if the heart is displaced through the sternum or rib [4]. On the other hand, surgical intervention has been reported to be successful in patients with ectopia hepatica [2]. This means that patients with ectopia hepatica have a better chance of life than ectopia cordis. However, as in this presented case, death was inevitable due to ectopia cordis and ectopia hepatica, the case was died following to the radiological examination.

As it is understood from the literature [1, 3, 6], necropsies performed in cases with anomalies provide information about the organs and systems where pathologies are seen, and also helps the specific diagnosis of the pathology. Ectopia cases can be noticed immediately after birth. While death is inevitable in ectopias related to the heart, lung, and chest wall, the patient can survive in other ectopia, which are not observed from the outside, but where organs and tissues enter another organ or tissue. These ectopias can mostly be determined in *post mortem* necropsies [2]. In the presented case, necropsy findings provided more explanatory information about the details and specific diagnosis of the anomaly. With the necropsy, it was observed that ectopia cordis was formed due to sternal agenesis, and ectopia hepatica was formed as a result of fissura abdominalis. As reported previously [3], ectopia cordis in this case raised the suspicion that there may be any other intracardiac pathologies, but no pathology was found in the anatomical structures of the heart. Differently from other reported cases [10, 11], which reported as incomplete Cantrell's pentalogy but even the intracardiac evaluations was not performed due to owner's permission or postmortem changes and excluded, in our case we had to chance to perform a cardiac evaluation immediately after death of the puppy. And based on the findings of no cardiac and diaphragmatic anomalies, but on the numbers of included pathologies combination of sternal and thoracoabdominal defects with ectopia cordis and ectopia hepatica the case classified the case as class III-a mild incomplete form of Cantrell's pentalogy.

CONCLUSION

In conclusion, developmental anomalies such as thoracic ectopia cordis, sternal agenesis, ectopia hepatic and fissura abdominalis may have a chance to be treated, but it should be known that when these anomalies are seen together in the same case (as in the presented case), death is inevitable in the early postnatal period.

Conflict of interest

The authors declare no conflict of interest.

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