Role of autopsy in diagnosing asplenia and right bilobed lung in a fetus with dextrocardia

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SUMMARY

We present a unique case of a fetus with dextrocardia, asplenia and a right bilobed lung in a primigravida woman in her 20s at 21 weeks' gestation. Prenatal ultrasound examination revealed dextrocardia and other anomalies such as atrioventricular septal defect and situs ambiguous with the gallbladder on the left, leading to termination of the pregnancy. Fetal autopsy confirmed the diagnosis, detected additional findings such as asplenia and right bilobed lung missed on ultrasound and highlighted the importance of autopsy in prenatal diagnosis. This rare case emphasises the value of a comprehensive prenatal assessment, fetal autopsy and a multidisciplinary approach in diagnosing, managing and counselling families affected by congenital anomalies. Timely detection and appropriate genetic counselling can guide affected families in making informed decisions regarding future pregnancies while providing closure and support in their grieving process.

BACKGROUND

Dextrocardia is a rare congenital anomaly in which the heart is located on the right side of the chest, with the apex pointing to the right. This condition occurs in approximately 1 in 12 000 live births and can occur in isolation or as part of more complex syndromes, such as heterotaxy.[1] Dextrocardia differs from dextroposition, where the heart is shifted to the right due to pathology of the diaphragm, lungs, pleura and other adjoining tissues. It has been seen in many studies that dextrocardia, along with other conditions such as asplenia or polysplenia, left or right isomerism and situs inversus, has resulted in atypical presentations in newborns with poorer outcomes.[2 3]

Heterotaxy syndrome refers to an anomaly in which the internal organs of the thoracoabdominal region show an atypical positioning along the left–right axis of the body. This condition encompasses individuals with various intricate cardiac anomalies. Patients with heterotaxy can be classified into subgroups, including those with asplenia and polysplenia syndrome or those with heterotaxy and isomerism of the right or left atrial appendages.[4]

Right atrial isomerism, asplenia syndrome or Ivemark syndrome is characterised by bilateral trilobed lungs with bilateral eparterial bronchi and superior vena cava draining to respective atria. Two atria are isomeric and the appendages of the right morphologic type. The coronary sinus is usually absent, and a persistent left superior vena cava drains directly into the left atrium. The atrial septum and relative position of great arteries are abnormal.[5]

Left isomerism is characterised by polysplenia, bilateral bilobed lungs with bilateral hyparterial bronchi and bilateral superior vena cava, which occurs in 50% of the cases. In the majority, there is atrial septal defect. Gallbladder is associated with the major lobe of the liver, which is the left lobe. Malrotation of the intestine is frequently seen. In some cases, partial anomalous pulmonary venous drainage is also present.[5]

Figure 1 Gross photograph of lungs showing bilateral bilobed lungs. Right lung measuring 3×2×1 cm and already sectioned left lung measuring 1.5×1.2×0.7 cm.

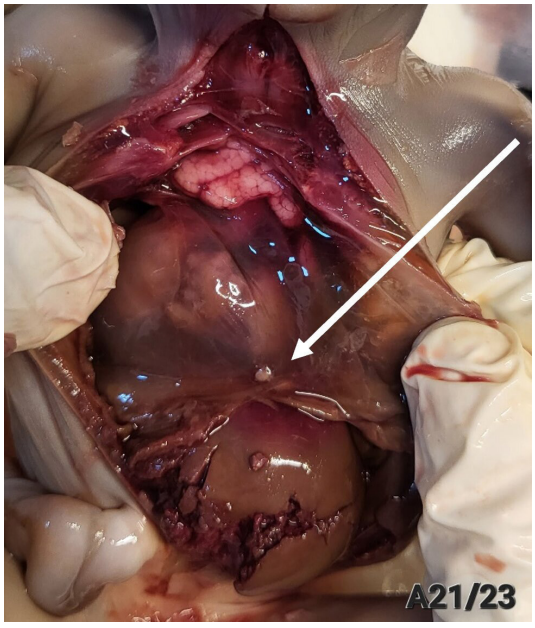

Figure 2 In situ view of fetus showing dextrocardia with the apex pointing towards the right (arrow).

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Such cases of either right or left isomerism have an estimated prevalence of around 1.1/10 000 live births.[6]

While prenatal ultrasound is an invaluable tool in diagnosing congenital anomalies, it does have limitations. Its sensitivity and specificity can be influenced by gestational age, fetus's position, maternal obesity and the sonographer's expertise.[7–10] In complex cases, such as congenital heart anomalies, detection can be particularly challenging due to the small size and rapid motion of the fetal heart.[7 8 11] Moreover, visualising organs such as the spleen distinctly on prenatal ultrasound can be difficult, potentially leading to overlooked conditions such as asplenia.[12 13] This challenge was highlighted in a study by Fang *et al*, where they employed the use of alpha-fetoprotein levels as an alternative means for diagnosing asplenia, thereby bypassing the limitations of ultrasonography.[14] Similarly, identifying variations in lung lobulation can be challenging with ultrasound imaging due to the similar echogenicity of lung tissue and surrounding structures.[15] Furthermore, ultrasound imaging may encounter difficulties in accurately characterising and diagnosing cases of complex heterotaxy, as presented in this case report. This limitation is illustrated by a study by Xiuqing *et al*, which suggests that the diagnostic accuracy of ultrasonography is significantly higher for simple congenital heart diseases (CHDs) compared with complex CHDs.[16] Therefore, while prenatal ultrasound is a crucial tool for identifying congenital anomalies, its limitations warrant the need for additional methods such as fetal autopsy to confirm and complete the clinical picture.

CASE PRESENTATION

A primigravida woman in her 20s presented to the clinic at 21 weeks' gestation for a routine prenatal check-up. She reported no significant medical history and had not experienced any complications during the pregnancy. She had no history of diabetes, smoking or any known exposure to organic chemicals or pesticides. There was a history of consanguineous marriage. In addition, her family history was unremarkable, with no reported congenital anomalies or genetic disorders.

The radiologist performed a detailed ultrasound anomaly scan, which revealed several abnormal findings. The fetus was found to have dextrocardia, with the heart's apex pointing to the right side of the thoracic cavity. A complete atrioventricular septal defect (AVSD) was noted, with an unbalanced interatrial septum and dilated right atrium. The left ventricle appeared hypofunctional, and the cardiac axis showed deviation to the right.

The ultrasound also revealed situs ambiguous in the abdominal cavity, with the gallbladder on the left side and a persistent right umbilical vein.

After reviewing the prenatal ultrasound results and identifying the presence of dextrocardia and other anomalies, the fetus's condition was discussed with the parents. The parents were provided with detailed counselling, including the potential for adverse outcomes during the pregnancy, and various options were presented for terminating the pregnancy. Once the parents made the decision to terminate the pregnancy, informed and written consent was obtained. The pregnancy was terminated, and the fetus was sent to the histopathology section for autopsy with the parent's consent.

The fetus, weighing 300 g, was received in 10% formalin with an attached umbilical cord. Brain fixation was done by injecting 100% formalin through the anterior fontanelle. The fetus's measurements were: a crown-rump length of 16 cm, a crown-heel length of 26 cm, a head circumference of 16 cm, a chest circumference and abdominal circumference of 16 cm each, a biparietal diameter of 5 cm, a foot length of 3 cm and 10 digits present in both the upper and lower limbs.

A Y-shaped incision was made for a thorough examination, with the arms of the Y extending to the tops of the shoulders to free up the skin in the frontal neck. This Y-shaped cut met inferiorly in the midline at the xiphoid process. A midline vertical incision was made from the xiphoid process to the pubic symphysis and then passed around the left side of the umbilicus. On cutting the surface of the umbilical cord, three vessels were identified.

On internal examination, the thymus measured 2.5×1×0.5 cm and weighed 1 g, while the right lung was bilobed (figure 1), measuring 3×2×1 cm and weighing 3 g. The left lung was normal, the image shows an already cut specimen of the left lung measuring 1.5×1.2×0.7 cm (figure 1). It was also noted that the heart was right-sided with a right-sided apex, measuring 3×2.5×1 cm and weighing 3 g (figure 2). There was no evidence of an AVSD, atrial septal defect or ventricular septal defect. The liver was in a normal position, measuring 5×3.5×2 cm and weighing 5 g, with a normal position of the gall bladder. There was no evidence of situs ambiguous. However, the spleen was absent, which was not noticed on ultrasonography.

The kidneys and adrenal glands were normal in appearance. The right kidney measured 2.5×1×0.5 cm and weighed 1.5 g, while the left measured 1.5×0.7×0.5 cm and weighed 1 g. The right and left adrenal glands measured 1×0.5 cm and weighed 0.5 g each.

Sections were given from all the internal organs and the umbilical cord. Slides were stained with H&E. On microscopic examination, the umbilical cord showed three vessels. The umbilical cord, thymus, bilateral lungs, heart, liver, brain and kidneys showed normal histology. Adrenal glands showed normal histology with partial autolysis.

DISCUSSION

Dextrocardia with asplenia is a rare congenital disorder. Under normal circumstances, embryonic development involves situs determination, which ensures the correct positioning of internal organs within the body by establishing left–right asymmetry. A series of molecular events and physical forces guide this process.[5] However, in cases of dextrocardia, this process is disrupted, leading to the abnormal positioning of the heart. The precise cause of dextrocardia is not yet fully understood, but it is believed to result from a combination of genetic and environmental factors.

An early and accurate prognosis is essential for providing appropriate counselling and management options for affected families. Advanced imaging techniques, such as fetal echocardiography, can help confirm the diagnosis and identify associated cardiac and visceral abnormalities.[17]

In the indexed case, based on findings from prenatal ultrasound, the decision was made to terminate the pregnancy. However, this decision was shaped by the context in which it was made. The limited resources and access to expert perinatology consultation may have influenced the diagnostic and management approach.

The autopsy findings, however, did not substantiate all the initial prenatal diagnoses. The right lung was bilobed and the spleen was absent, neither noticed on ultrasonography. Dextrocardia was present, but there were no other cardiac anomalies which were mentioned in the ultrasonography report. Situs ambiguous was absent, and the stomach and gall bladder were in normal positions. The complexities of the case highlight the

limitations of ultrasound and the pivotal role of fetal autopsy in confirming and refining the prenatal diagnosis, particularly in settings with restricted resources or lack of specialised expertise.

Regarding additional diagnostic procedures, in this case, amniocentesis was considered after the anomaly scan. However, given the complexities and risks associated with amniocentesis, coupled with the family's decision not to proceed with any further testing, neither amniocentesis nor karyotyping was carried out. Additionally, the lack of infrastructure for DNA banking presented a further limitation.

While there are numerous case reports discussing individual anomalies related to the heart, lungs or spleen, few delve into the simultaneous occurrence of these anomalies, a condition known as heterotaxy syndrome. In a case report by Karki *et al*, they reported findings of dextrocardia with situs inversus and an absent left kidney in a 7-month-old baby.[18] Another study on fetal dextrocardia by Bernasconi *et al* compared diagnoses and outcomes in two tertiary centres, finding that the majority of fetuses with dextrocardia were associated with CHD.[19]

However, the complexity and rarity of heterotaxy syndrome are highlighted in a systematic review by Buca *et al*.[20] The study found that 83.4% of cases had major cardiac anomalies, with AVSD being the most common. It also emphasised the high rate of associated cardiac and extracardiac anomalies, the importance of the second trimester routine anomaly scan in detecting heterotaxy syndromes, and the need for prompt recognition and treatment of associated conditions such as biliary atresia and intestinal malrotation. These findings underscore the need for thorough prenatal diagnosis, follow-up and further research in prenatal diagnostic techniques.[18–20]

Cases involving dextrocardia, a right bilobed lung, and asplenia are rare. However, one unusual case reported by Dutta and Sarma[21] described dextrocardia in conjunction with bilateral bilobed lungs and a right-sided spleen. This emphasises the infrequency and intricacy of such cases, underscoring the need for accurate and comprehensive prenatal evaluations to identify these anomalies and offer appropriate counselling and management options for affected families.

Learning points

- Early and comprehensive prenatal ultrasound assessments are critical in detecting rare congenital anomalies, although with inherent limitations, particularly in assessing spleen and lung lobulation.
- Fetal autopsy plays a vital role in confirming prenatal diagnoses, providing comprehensive understanding of congenital anomalies and identifying potentially undetected conditions during prenatal imaging.
- Fetal autopsy findings guide medical teams in offering appropriate genetic counselling, allowing families to understand the potential risk of recurrence of congenital anomalies in future pregnancies.
- This case highlights the challenges of diagnosing complex congenital anomalies such as dextrocardia with asplenia using prenatal ultrasound imaging alone. Autopsy can provide crucial additional insights.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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