Case Report

A RARE CASE OF HETEROTAXIA: FETAL AUTOPSY FINDINGS

Daisy Dwivedi¹, Bal Chander¹

1. Department of Anatomy, Dr Rajendra Prasad Government medical College,
Kangra at Tanda, H.P., India

ABSTRACT

Visceral organs are asymmetrically distributed in thoracic and abdominal cavities but there is definite left right orientation along the midline. At times there may be disturbance of the same resulting from malrotation and/or movement during embryological development. We are presenting autopsy findings of a 20 weeks old male fetus wherein the stomach along with spleen and pancreas are on the right side. Rest of the thoracoabdominal viscera are in the anatomically correct locations. Such cases qualify for heterotaxia, denoting status somewhere between situs solitus which is the normal placement of organs and situs inversus which denotes mirror image of solitus. Heterotaxia can have a range of features. Such cases usually come to light within first year of birth but some cases remain asymptomatic till adulthood and are diagnosed incidentally. This article discusses the nomenclature with diagnostic criteria and points out the lacunae which make the present case difficult to classify.

Keywords: Heterotaxia, Fetal autopsy, Situs solitus

INTRODUCTION

Malrotation of intestinal loops with or without cardiac apex is referred to as situs inversus or situs ambiguus. Malrotation is a challenge for surgeons both diagnostically and therapeutically. Most of the cases come to clinical attention within first year of life. [1] However there are reports of such cases diagnosed in adult life. [2] The spectrum of features can range from complete mirror image of normal placement of viscera (situs inversus) to incomplete wherein a few organs are on the

opposite side and the others are on the normal side (situs ambiguus). [3] We are presenting a case that does not appear to fall under either of these two.

CASE REPORT

A 26 years old primigravida had presented for normal follow up. The mother had no adverse history and all routine investigations were normal. Ultrasound of the fetus did not reveal any abnormality. Following abdominal pains, the mother was brought to hospital where on

Address for Correspondence:

Dr. Daisy Dwivedi, Senior Resident, Department of Anatomy, Dr Rajendra Prasad Government medical College, Kangra at Tanda, H.P., India
Email - drdaisyd@gmail.com

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ultrasound, a diagnosis of intrauterine death was made. The fetus was sent for autopsy after informed consent of the parents.

RESULTS

A 20 weeks old male fetus weighing 390 grams and crown rump length of 17 cm was received for autopsy. There were no external malformations. A midline incision from neck to pubic region showed normal thoracic contents with no discernible gross abnormalities of the heart or lungs.

On autopsy, the stomach was seen to be lying opposite to its normal position, in the right hypogastrium and epigastric region. The head of pancreas was on the left side with its tail directed upwards towards right hypochondrium where spleen was also located. There were no signs of duodenal obstruction. Appendix, caecum and colon were in their normal position ruling out malrotation of midgut. There was no dextrocardia. (Fig. 1) All the organ weights were within normal range and were grossly normal. Umbilical cord stump showed three vessels. A diagnosis of situs ambiguous or heterotaxia was considered.

DISCUSSION

Situs solitus refers to the normal positions of cardiac apex, stomach, spleen, pancreas and intestinal loops and aorta on the left whereas liver and inferior vena cava is situated on the right of the midline. Situs inversus is the mirror opposite of solitus. It is of two types. Situs inversus with dextrocardia is more common and congenital heart disease occurs in 3 to 5% of the cases. Situs inversus with levocardia is extremely rare and congenital cardiac anomalies are seen in almost all the cases.

Situs ambiguous or heterotaxia is the third category which falls somewhere between solitus and inversus with congenital heart disease between 50 to 100% of the cases. [3, 4]. Heterotaxia is further classified as SA with polysplenia or with asplenia [5]. Incidence of heterotaxia is estimated to be between 1 in 10,000 to 40,000 live births [6]. In our centre this is the first case 1 in 10,000 pregnancies including abortions. The present case qualifies for heterotaxia since with the exception of stomach, spleen and pancreas all other thoracic and abdominal viscera are on the correct sides as in solitus. However, it fails to fit either in heterotaxia with polysplenia or asplenia since there is a right sided single spleen.

There are cases in literature similar to our case that do not fit into sub classification of heterotaxia. Isolated intestinal malrotation without other complications has been reported albeit without the benefit of complete autopsy and the authors have not commented on spleen or pancreas [7, 8]. Another case pertains to levocardia without defects with right sided stomach, spleen and colon and left liver. Information regarding pancreas is missing. [9]

Present case might come across as a first of its kind but from putative prognostic perspective it appears to be similar to other cases of heterotaxia. Vast majority of cases of malrotations come to clinical attention post birth within one year owing to pain abdomen and bilious vomiting. Management almost invariably involves major surgery. [7, 8, 9] But it should be noted that similar cases can also come to clinical attention during adult life, mostly as incidental findings [10].

Fig. 1. A: Right lobe, liver (right arrow); B: Heart (down arrow), Stomach (right arrow); C: Spleen (right arrow) and Pancreas (arrowhead)

CONCLUSION

The purpose of this case with extremely rare features is to highlight the importance of fetal autopsies. It is imperative that every intrauterine death be investigated and an attempt made to establish a diagnosis as per the established criteria or highlight the cases that do not conform to the same.

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