A Rare Case of Situs Ambiguous Anomaly with Right Sided Isomerism and Ventricular Septal Defect

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Abstract: Situs ambiguous anomaly describes an abnormal arrangement of visceral organs in the thoracic and abdominal cavities across the normal left–right axis of the body. This syndrome is mostly diagnosed as a serious cyanotic cardiac disease in the infancy. In this report, we present a case of situs ambiguous anomaly with acyanotic heart disease which was diagnosed during evaluation for recurrent chest infections. The patient had centrally located liver, asplenia, ventricular septal defect, endocardial cushion defect, left sided inferior vena cava and right sided aorta. These congenital anomalies were found to be compatible with situs ambiguous anomaly with right isomerism.

Key words: Heterotaxy syndrome; situs ambiguous; isomerism; congenital heart disease.

INTRODUCTION

Position of the viscera (heart, lungs, liver, spleen and bowels) is defined very early in embryogenesis. Defects in asymmetry may occur during embryogenesis and there is a spectrum of malformations ranging from reverse asymmetry (situs inversus) to a complete lack of physiologic asymmetry (situs ambiguous)[1]. Cyanotic heart diseases are the most often encountered manifestations of the situs anomalies in the infancy[2,3]. In this case report, we present a atypical case of situs ambiguous anomaly with right sided isomerism with ventricular septal defect and endocardial cushion effect.

CASE REPORT

A 5 month-old girl was referred to the pediatric outdoor as a case of recurrent chest infections for further evaluation and management. She was born to a non-consanguinous parents after a full-term pregnancy with no significant antenatal/neonatal history and no significant family history. With an apparent normal development, the patient was hospitalized at the age of 3 months in a pediatric department with respiratory distress. There was no history of cyanosis. On physical examination, a 3rd degree cardiac murmur was identified. Her chest x-ray was normal. Abdominal ultrasonography showed stomach on the right side, aorta on the right side, inferior vena cava on the left side and spleen is not visualized (figure 1).

Figure 1. Abdominal ultrasonography showing right sided aorta with left sided inferior vena cava and asplenia

Liver was in midline with normal echo texture. There was no abnormality found in portal vein, pancreas, both kidneys and ureters. Echocardiography showed a gap of 0.79 cm in interventricular septum in perimembranous region with left to right shunt (figure 2).

Figure 2. Echocardiography showing ventricular septal defect with left to right shunt
Echocardiography also showed the endocardial cushion defect. Both left and right ventricular outflow tract were normal. Computed tomography of abdomen was not done due to lack of resources. As per aforementioned features a diagnosis of situs ambiguous anomaly with right sided isomerism with complex congenital heart disease was made.

**DISCUSSION**

Situs inversus is a mirror image of the normal situs where all internal organs occupy the corresponding contralateral side and are otherwise normal. Situs ambiguous or Heterotaxy syndrome are the intermediate presentations between situs solitus and situs inversus[2]. Traditionally Heterotaxy is divided into two general categories: left isomerism and right isomerism.

Left isomerism, or situs ambiguous with polysplenia, has a female predominance and mostly present as complex cardiac defects like partial anomalous pulmonary venous return, atrial septal defect, and a common atrioventricular canal[3,4].

Right isomerism, or situs ambiguous with asplenia, has a male predominance and has a high incidence of cyanotic congenital heart diseases such as a common AV canal, univentricular heart, transposition of the great arteries, or total anomalous pulmonary venous return[3,4,5]. In contrast to left isomerism, the spleen is absent and the IVC typically runs ipsilateral to the aorta[3].

In our literature research, we came across three case presentations related to heterotaxy syndrome. Himanshu et al, reported heterotaxy syndrome with complete right atrial isomerism[8]. Hrusca et al, reported heterotaxy syndrome with complete right atrial isomerism[8].

Most patients with right isomerism succumb within the first year of life due to cardiovascular compromise, sepsis due to asplenia, sudden death and cardiac arrhythmias [9]. The prognosis remains poor despite modern surgical techniques. It is crucial to reveal the anatomical features in such patients by using imaging modalities prior to surgery and invasive intervention to prevent the possible risks and complications.

**References**


