Unusual Variation of Asplenia Syndrome

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SUMMARY

The case of a 1-year-old cyanotic boy diagnosed with asplenia syndrome has been reported. By physical and laboratory examinations, levocardia, atrial inversion, primum ASD, single atrioventricular valve, single ventricle (left-hand morphology), rudimentary right ventricle (anterior, left-sided), pulmonary stenosis, left-sided vena cava, single vena cava superior were established and the case was diagnosed with asplenia syndrome. The patient has concordance between tracheo-bronchial situs and lung anatomy and inverted atrial and visceral situs, but without atrial isomerism that makes his case an unusual variation of asplenia syndrome.

Key Words:
Asplenia syndrome without atrial isomerism Tracheo-bronchial situs solitus

ASPLENIA syndrome is a disorder which is characterized by congenital absence of the spleen with malposition and malformation of other organs, especially those of the cardiovascular system. The spectrum of the syndrome, which was first defined at 1826, has continuously changed due to reports of cases that have different symptoms, and this has resulted in new characterizations of this syndrome.[1] In this report a case of asplenia syndrome with unusual thoracic and atrial situs discordance is presented.

CASE REPORT

A 1-year-old boy was admitted to the Department of Pediatric Cardiology, Gazi University Medical Faculty Hospital, because of cyanosis. He had

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Received for publication December 12, 1990.

Accepted May 13, 1991.

a history of recurrent respiratory system infections since he was 10 days old. His parents were first degree relatives, and the first child of the family died because of esophageal atresia. On physical examination body weight was 7 kg (below the 3rd percentile) and he was cyanotic. A third degree systolic murmur was heard at the left sternal border. He had a right inguinal hernia. Laboratory examination showed: hemoglobin=14 g/dl, MCV=74.3 μ, polymorphonuclear leucocytes=33,100/mm^3, red blood cells=4,970,000/mm^3 with aniso-poikilocytosis, hypochromia and normoblasts in his blood smear. ECG revealed negative P waves and a single-ventricle pattern in the precordial leads. His chest x ray showed moderate cardiomegaly and diminished pulmonary vascularity (Fig. 1). Bronchial situs solitus, a left-sided liver and a

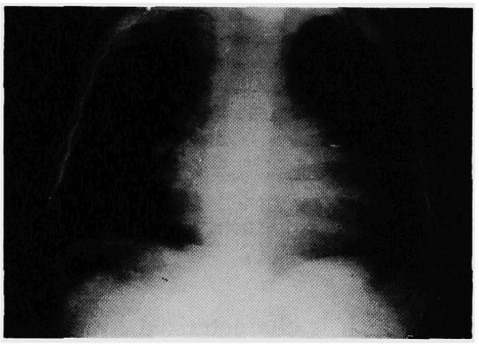


Fig. 1. Plain x ray showing moderate cardiomegaly and diminished pulmonary vascularity.

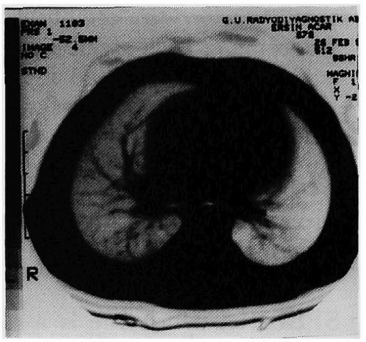


Fig. 2. Thoracic tomography showing bronchial situs solitus.

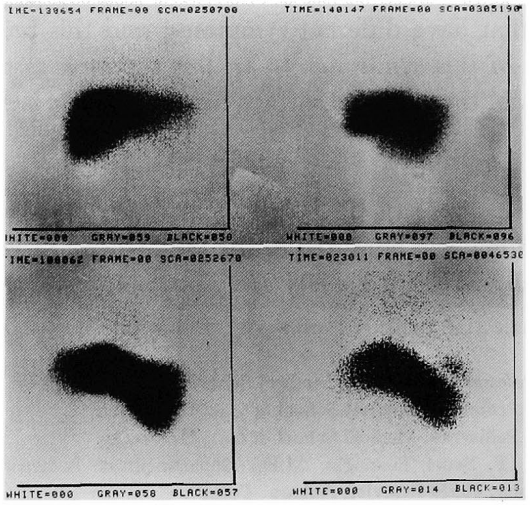


Fig. 3. Abdominal scintigraphy showing left-sided diffuse hepatomegaly and lack of spleen.

right-sided stomach were detected on thoraco-abdominal computerized tomography (Fig. 2). His abdominal scintigraphy findings were: left-sided diffuse hepatomegaly and lack of spleen (Fig. 3). His echo and angiocardiographic examinations showed levocardia, atrial inversion, primum atrial septal defect, single atrioventricular valve, single ventricle (left-hand morphology), rudimentary right ventricle (anterior, left-sided), pulmonary stenosis, left-sided vena cava inferior draining into the left-sided atrium, single vena cava superior

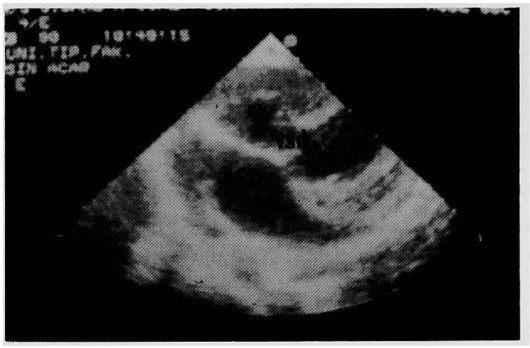


Fig. 4. His subcostal 4-chamber, 2-dimensional echocardiogram showing primum atrial septal defect (ASD), single atrioventricular valve, ventricular septal defect (vsd), rudimentary right ventricle.

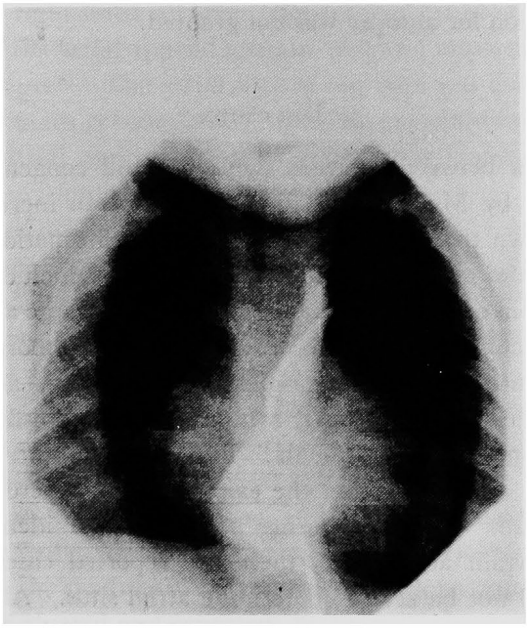


Fig. 5. His cineangiogram showing left-sided vena cava inferior connected to the left-sided right atrium.

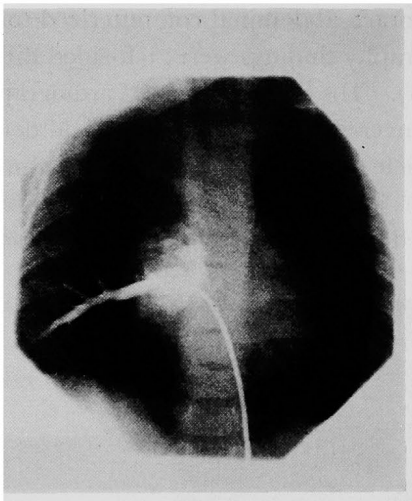


Fig. 6. His cineangiogram showing pulmonary veins connected to the right-sided left atrium.

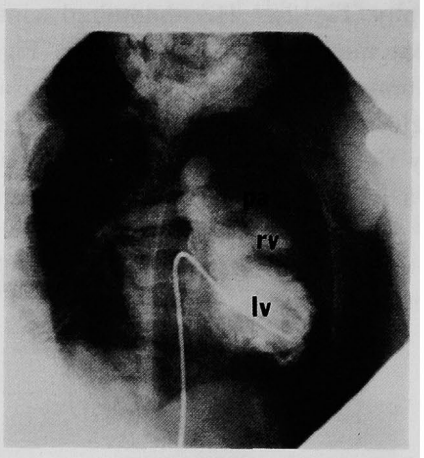


Fig. 7. His cineangiogram showing left-sided rudimentary right ventricle and infundibular pulmonary stenosis.

draining into the left-sided atrium, hepatic veins draining into the vena cava inferior (Figs. 4–7). A Blalock-Taussig shunt was made for palliation. On the second day following surgery the patient died because of respiratory failure. Permission for autopsy was not granted.

Discussion

The relation between asplenia syndrome and congenital heart disease was first defined by Martin in 1826. Since then an increasing number of reports have shown that cardiovascular system malformations are one of the main characteristics of the syndrome.[2)–5)] This case includes the most common malformations of the asplenia syndrome, namely large atrial septal defect, single atrioventricular valve, single ventricle (left-hand morphology), visceral situa inversus, but does not have right atrial isomerism as expected. The asplenia syndrome was first defined as a lateralization disturbance and later as right atrial isomerism and asplenia. But, as the number of reported cases increased, it was understood that the existing criteria were not sufficient for defining the situs.[6)–8)] Though using parameters for identifying the atrial situs brought certain advantages, there are reported cases in which these parameters could not be used to determine atrial situs. Although the trend is to identify atrial situs according to the morphology of the atrial appendages, there are cases where it is not possible to identify left and right atria even

by use of some other anatomical criteria like pectinate muscle or atrial septum. This paradoxical situation caused the growth of new definitions such as situs ambigious and situs undeterminate, but there is still a controversy regarding identification and definition.[9),10)] Identification of atrial situs was shown to be the first step in sequential analysis and categorization of congenital cardiac malformations.[11)-13)]

According to this definition Putschar and Mannion defined the position of the organs as right atrial isomerism in asplenic patients.[6)] The identification of atrial situs depends on some morphological criteria which are: connection of the systemic and pulmonary veins; the anatomical characteristics of the atrial septum; the external and internal morphology of the atrial appendages. Recently, transesophageal echocardiography has been used to determine the atrial situs according to the atrial appendages.[14),15)] Ultrasonographic and tomographic examination of the abdominal great vessels according to the criteria of Huhta et al is a guide to definition.[16)-18)] The atrium to which the inferior cava drains is identified as the right atrium.[19)] In this case, the inferior caval vein ascended to the left and anterior of the aorta in echocardiographic examination. Angiocardiographic results supported this finding and showed that the left-sided inferior caval vein drains to the left-sided atrium into which the left-sided superior caval vein drains as well, and hepatic veins drain directly into this atrium. Pulmonary veins drain into the right-sided atrium. Besides the external shape, the internal architecture of the atrial appendages also plays an important role in determining the atrial situs.[8)] The atrial situs of our case was defined as atrial inversion according to his echoangiocardiographic examinations and direct visualization of the atrial appendages during surgery. Unfortunately we did not have the necropsy findings for further confirmation.

In addition to the difficulties in determining the atrial situs, there are also discordances between atrial situs and visceral situs, tracheo-bronchial and atrial situs, and bronchial and lung morphologies.[4),17),20),21)] Besides, these splenic anomalies are not always accompanied by symmetry of the thoracic organs.[22)] The nosology of asplenia syndrome has a very wide spectrum. Each newly defined case brings in a new concept. We believe the case mentioned above (concordance between tracheo-bronchial anatomy and the lungs, tracheo-bronchial situs solitus, inverted atrial and visceral situs, without atrial isomerism) is an unusual variation and a unique example in the literature as far as we have surveyed. In order to settle the discussion about the naming and defining of the syndrome, every new case with its unique properties should be throughly evaluated and reported.

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