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Heterotaxy syndrome presenting as severe pulmonary artery hypertension in a young old female: case report

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Heterotaxy syndrome is a rare congenital disorder characterized by situs ambiguus, congenital heart defects and splenic malformations. We describe a case of 65 year young- old female who presented with sudden onset dyspnoea in emergency department. Her chest x-ray, 2 D echo and computerised tomography of chest was suggestive of sever pulmonary hypertension as a rare manifestation of Heterotaxy syndrome. To the best of our knowledge, pulmonary hypertension has not been previously reported as the main clinical feature in young – old patients with Heterotaxy syndrome.

Key words: Heterotaxy syndrome, Pulmonary hypertension, Young old, Congenital

INTRODUCTION

Heterotaxy syndrome (from the Greek heteros – different and taxis – arrangement) is a rare congenital disorder characterized by predominant malformations in the cardiovascular system, the lungs (symmetric lobulation), the spleen (polysplenia, asplenia, or hypoplastic spleen), and the gastrointestinal tract (situs ambiguus, other malrotations, liver and pancreatic malformation [1]. This case depicts two novel features: primary diagnosis of heterotaxy syndrome may be delayed until old age, and this syndrome may be associated with pulmonary hypertension, possibly on the basis of longstanding Porto systemic shunts. Few authors had reported this rare syndrome in adulthood as an incidental finding [2 3]. Here we describe a case of 65 year young – old female who presented with first time dyspnoea due to pulmonary hypertension as a manifestation of Heterotaxy syndrome.

CASE REPORT

A 65 year young – old previously healthy female was admitted to the emergency department of this hospital with breathlessness on exertion and palpitations since 1 month and aggravated since 3 days. Breathlessness Was NYHA Grade 4. Personal and family histories were otherwise insignificant. She had no history of hypertension, diabetes or any heart disease in the past.

Her physical examination results were normal, and there was no abdominal bruit on auscultation. The patient's blood pressure was normal at 130/70 mmHg, and her heart rate was 110 beats/minute. Her hemoglobin was 8.8 mg% and the total leukocyte count was 8400/mm. Her blood sugar, kidney function, and liver function were within normal limits. Her cardiovascular examination revealed signs of pulmonary hypertension in the form of diastolic shock and left parasternal heave. The electrocardiogram (ECG) showed signs of right ventricular hypertrophy with atrial fibrillation. Chest radiograph showed dilated central pulmonary arteries and cardiomegaly (Fig. 1).

High resolution computed tomography thorax revealed massively dilated pulmonary trunk measuring 63.5 mm in diameter with markedly dilated right and left pulmonary arteries up to the hilum showing abrupt narrowing

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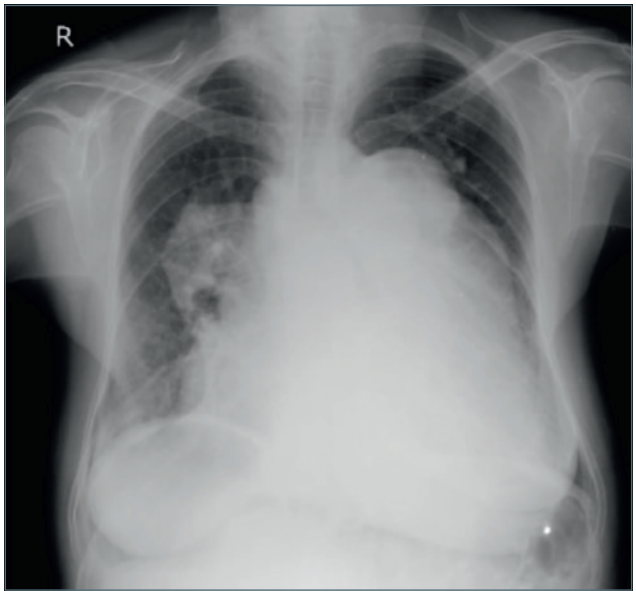

Figure 1. Showing massive cardiomegaly on chest x-ray PA view.

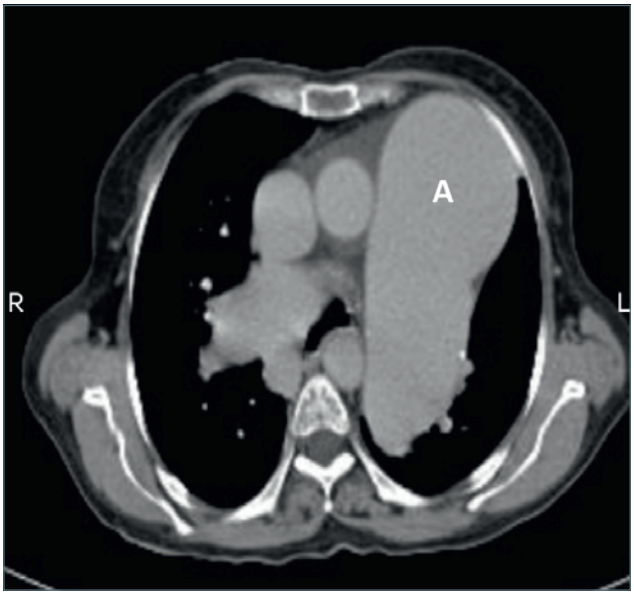

Figure 2. Showing huge pulmonary trunk (denoted by A) and markedly dilated left and right pulmonary arteries (denoted by B and C) on high resolution CT thorax.

of right and left pulmonary artery. Her liver was seen in midline, stomach on right side and spleen on right side with multiple splenenculas (Figs. 2-3). Her portal vein diameter was normal. 2d Echo was suggestive of severe pulmonary arterial hypertension with pulmonary arterial systolic pressure of 67 mm hg (Fig. 4).
The diagnosis of heterotaxy syndrome with severe pulmonary hypertension was, therefore, established. Symptomatic treatment with phosphodiaesterase-5 inhibitor Sildenafil led to gradual improvement of dyspnoea.

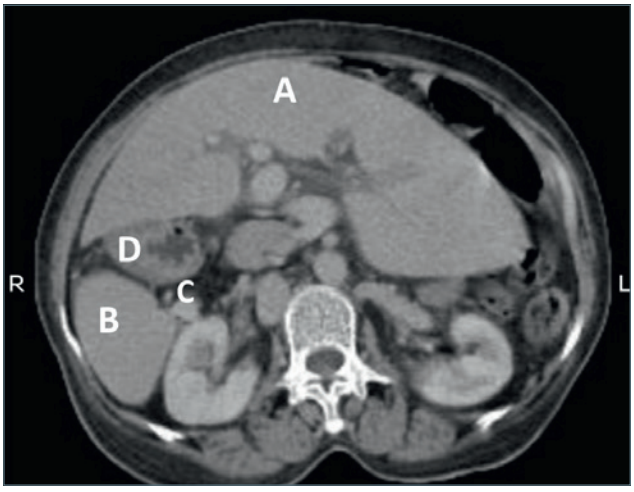

Figure 3. HRCT thorax with abdomen showing:
A) liver placed in midline
B) spleen on right side
C) splenenculi
D) right sided stomach

DISCUSSION

Heterotaxy syndrome also known as Ivemark syndrome or isomerism, includes a wide variety of clinical manifestations. Most cases are sporadic, but familial cases have been reported. The habitual and orderly arrangement of the organs in the human body is determined early in the embryonic development. The loss of such orderly arrangement may characterize situs inversus or a disordered and variable arrangement seen in heterotaxy syndrome) [1 2]. Heterotaxy syndrome presents an approximate incidence of 1:10,000 births and is slightly more prevalent in men, at a ratio of 2:1. As it is mostly manifested in childhood or infancy, their overall prognosis is reduced. Mortality and morbidity are usually related to the degree of Congenital Heart Disease [3]. Congenital Heart Disease may include abnormal localization of the cardiac apex, a common atrioventricular canal, anomalous systemic venous return (e.g., bilateral superior vena cava, interruption of the inferior vena cava with azygos continuation), atrial and ventricular malformations and septal defects, absent coronary sinus, malposition or transposition of the great arteries, pulmonic stenosis, pulmonary atresia, patent ductus arteriosus, and anomalous pulmonary venous connection [3].

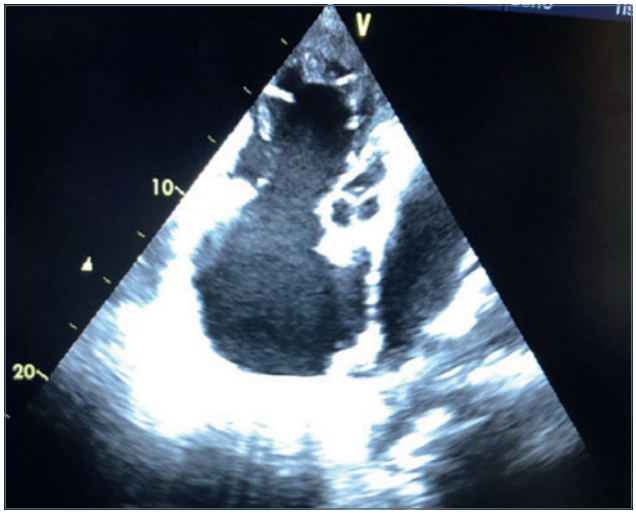

Figure 4. 2D Echo showing dilated right atrium and left atrium.

Our patient presented with symptoms of severe pulmonary hypertension. The most likely reason that our patient with heterotaxy syndrome was asymptomatic and felt well until 65 years of age was that she had no relevant structural or functional cardiac defect. The development of pulmonary hypertension may be due to left-right blood shunting; but in our patient significant cardiac shunt volume was ruled out by 2 D Echo. Pathophysiologically it may be explained by the presence of severe visceral malformations which happens in this syndrome.

Pulmonary hypertension may develop as a consequence of underlying splenic, hepatic, or vascular malformations. Chances are increased in patient with liver cirrhosis complicated with portal hypertension known as portopulmonary hypertension [4 5]. The exact mechanism of portopulmonary hypertension is unknown. Altered hemodynamics or incomplete metabolism of vasoactive substances in the liver is possible mechanisms for the development of pulmonary hypertension in these patients. These vasoactive substances include endothelin, vasoactive intestinal peptide, serotonin, and thromboxane A2. Endothelin is a well-known pulmonary vascular constrictor, and elevated levels of endothelin have been reported in patients with liver cirrhosis [5]. However parenchymal liver disease was excluded in our patient by Ultra Sonography of the abdomen and liver.

The number of asymptomatic patients with heterotaxy syndrome diagnosed in adulthood may rise with the increased utilization of CT and magnetic resonance imaging.

Declaration of Sources of Funding

This study was not funded.

Conflict of Interest

The authors declare no conflicts of interest.

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