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Incidentally Diagnosed Heterotaxy in a Child: A Case Report

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ABSTRACT: Heterotaxy syndrome (situs ambiguus) is a condition involving the abnormal arrangement of the internal organs of the thorax and abdomen. Complex congenital defects affect the heart, lungs, spleen, intestines, liver and other organs. This report describes a case of a 5 year old male child who presented with recent onset fatigue following exertion, difficulty breathing and one episode of cyanosis and had never been diagnosed prior to this. He was diagnosed incidentally on USG of the abdomen/pelvis. He was further evaluated with relevant investigations. As reported in prior research, early identification of anomalies is important for adequate and optimal management.

Keywords: heterotaxy syndrome, left isomerism, right isomerism, situs ambiguus, situs inversus

I. INTRODUCTION

Heterotaxy syndrome (Situs ambiguus) refers to the abnormal arrangement of the thoracic and abdominal internal organs and unlike situs inversus, it can cause serious complications. The basic structure of the heart can be altered and the number of lobes in each lung, as well as the length of the bronchi can also be affected. There can be asplenia or polysplenia, while the liver may not be in its normal position. The severity of the condition varies, depending on the specific abnormalities involved. The spectrum of heterotaxy syndrome can range from mild to life-threatening, inspite of treatment.[1]

Most patients with polysplenia syndrome die by the age of 5 years, most of which are due to cardiac causes. 5-10% of patients with polysplenia syndrome have minor cardiac abnormalities and may reach adulthood without complications.[2]

Heterotaxy is classified into two different types – bilateral left sidedness (polysplenia syndrome) or right sidedness (asplenia syndrome) – with some variations.[3] It usually occurs sporadically but some genetic abnormalities have been also reported.[4,5] The reported incidence of heterotaxy syndrome is 1–1.5/10 000 live births.[6] Patients with complex cardiac lesions and heterotaxy have a poor prognosis, with mortality of over 85% for patients with asplenia, and over 50% for patients with polysplenia. [7] 40–70% of patients with heterotaxy syndrome have involvement of various systems. Many of them have congenital gastrointestinal anomalies but a few present with other symptoms such as respiratory, genitourinary, central nervous system, and skeletal anomalies. [8]

CASE PRESENTATION

The patient is a 5 year old male child with no prior significant past history. He was not diagnosed previously and he presented to the emergency room with complaints of bluish discoloration of the lips while crying, breathlessness and a recent history of increased fatigue after exertion. He was born of a third degree consanguineous marriage, at term by a normal vaginal delivery. No similar conditions were reported in the family.

On admission, he was afebrile, tachypnoeic and had bradycardia at rest (50-60/minute). His resting oxygen saturation on room air varied between 88 to 92% and capillary refill time was 3 seconds with a blood pressure of 94/58 mmHg in the right upper limb in a supine position. He also had grade 2 clubbing and on further examination, pectus carinatum with a hyperdynamic precordium and hepatosplenomegaly. An ejection systolic murmur was heard in the second intercostal space at the upper left sternal border.

Further analysis of his anthropometry revealed his weight and height both to be below the 3rd percentile (10.9kg & 91cm respectively), head circumference was also below the 3rd percentile (46cm) and mid upper arm circumference was 11 cm. [9]

On admission, relevant laboratory findings included a low haemoglobin (8.9g/dL), a raised total leucocyte count of 23,900/cumm with a raised absolute eosinophilic count of 14,340/cumm

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(60%). Peripheral smear showed a microcytic hypochromic picture with adequate platelets and leucocytosis, with marked eosinophilia.

The patient was admitted in the PICU for stabilisation and started on Spirinolactone and Furosemide. ECG findings were unremarkable.

Radiograph of the chest showed cardiomegaly (figure 1).

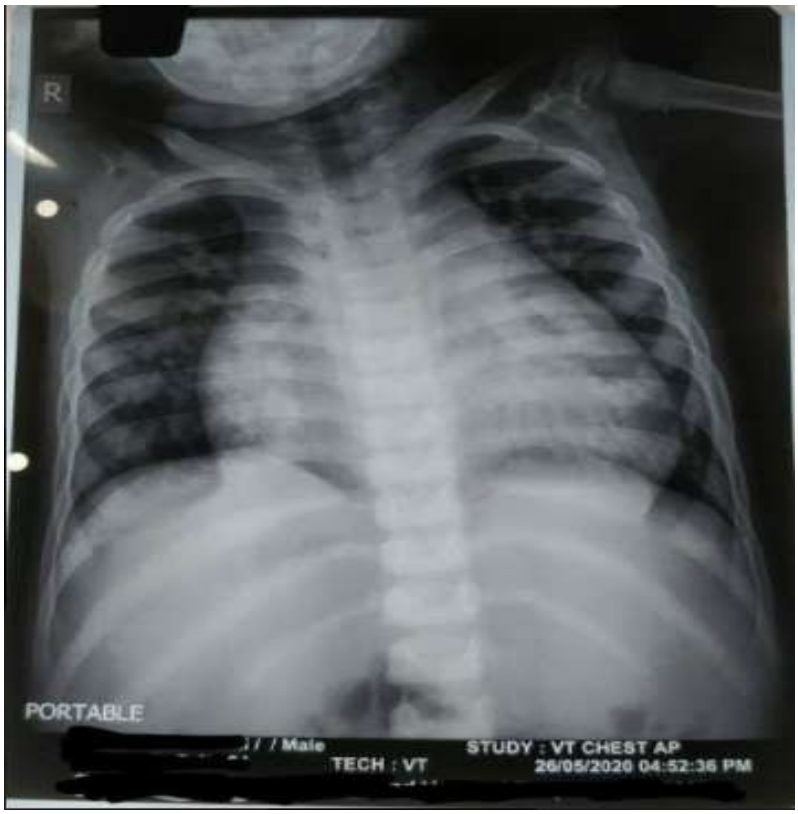

Figure 1. Chest radiograph shows cardiomegaly.

Ultrasound (US) of the abdomen and pelvis was obtained and revealed an ectopically located spleen, placed posteriorly to the liver on the right side, approximately 7.5cm, which was displacing the right kidney inferiorly. The liver measured 17cm and extended till the splenic fossa but had normal echogenicity and echotexture. Bilateral kidneys were unremarkable and no abnormal bladder masses or ureteral calculi were detected (figure 2).

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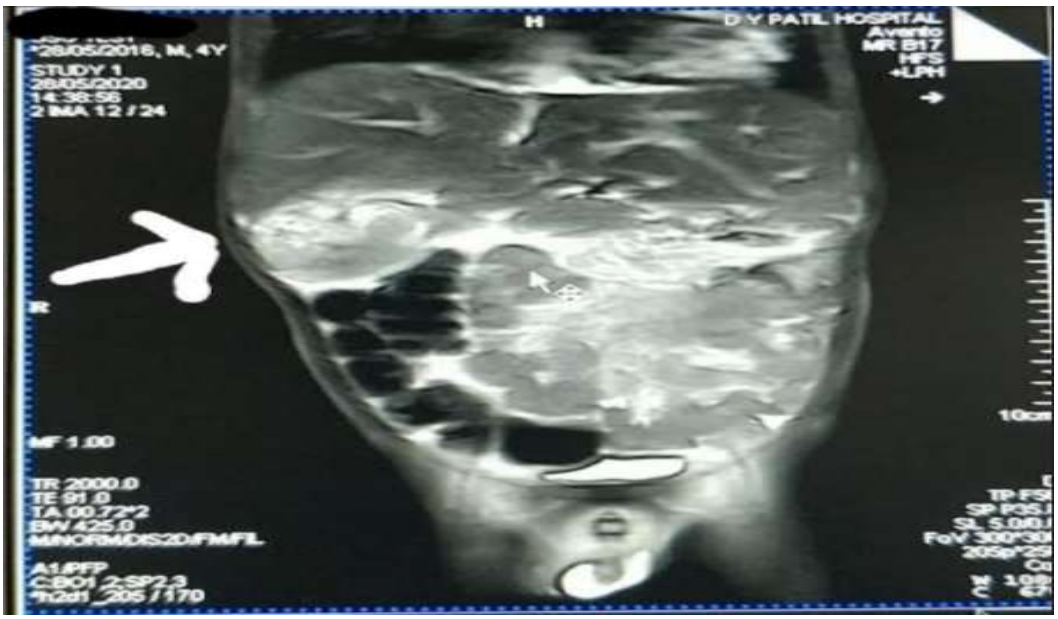

Figure 2. Ultrasonography of the abdomen and pelvis

A 2D echocardiogram showed severe tricuspid regurgitation with a peak pressure gradient of 70mmHg, severe pulmonary artery hypertension, severely dilated right sided chambers of the heart, a moderate secundum atrial septal defect with a bidirectional shunt. The interventricular septum was intact and the inferior vena cava could not be properly visualised but hepatic veins were dilated. It was likely to be Total Anomalous Pulmonary Venous Connection (TAPVC) or a Great Artery level shunt and hence a CT Cardiac Angiography was suggested.

The following findings were seen on CT Angiography (figures 3, 4, 5):

- Visceral situs inversus : multiple splenules were seen in the right hypochondrium and the largest portion of the liver was seen in the left hypochondrium. Stomach was seen on the right side.
- Thoracoabdominal situs-ambiguous : two lobes with two bronchi were seen in both lungs.
- Atrial situs-solitus: morphological right atrium was seen on the patients right side.
- Ventricular loop designation – D loop: morphological right ventricle was seen on the right side of the left ventricle.

-Position and relation of great vessels-dextromalposition : both great vessels are seen at the same coronal plane with aorta on the right side of the pulmonary artery.

The interpretation of the findings were as follows:

-Visceroatrial discoradance

-Atrial situs-solitus: SVC, IVC and coronary sinus drain into the morphological right atrium.

- Normal atrioventricular concordance: tricuspid and mitral valve form morphological right and left AV connection.
- Venticulo arterial concordance: both aorta and pulmonary artery arise from the morphological left and right ventricle.
- Venoarterial discordance: right superior and inferior pulmonary veins drain into the morphological right atrium and left superior and inferior pulmonary veins drain into the left atrium. (Partial Anomalous Pulmonary Venous Drainage – PAPVC)

-Large defect seen in the interatrial septum, measuring 1.4cm

- Pulmonary arterial hypertension.

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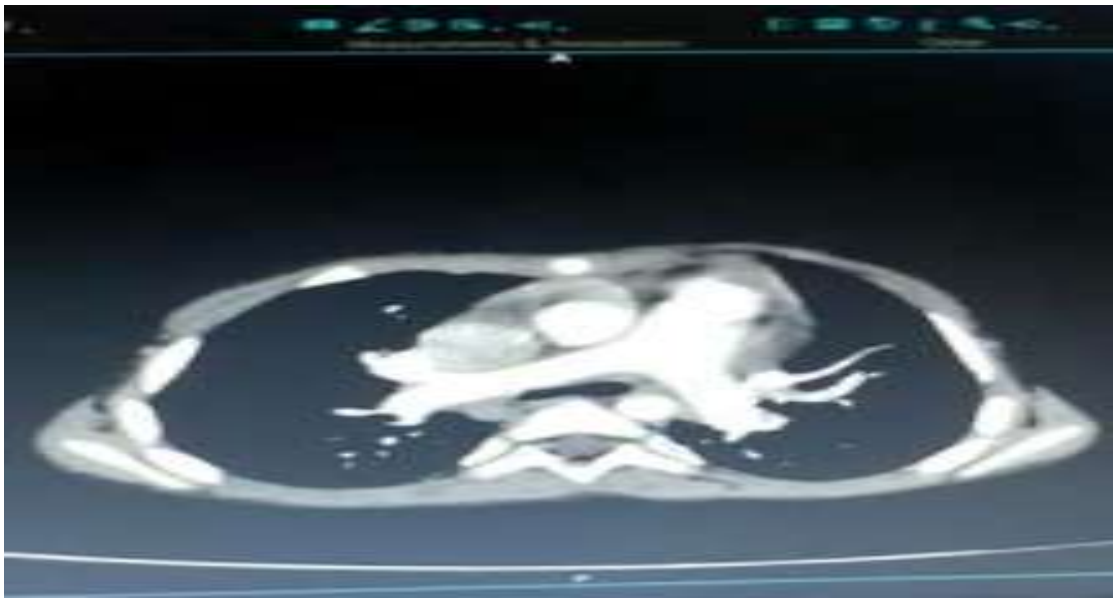

Figure 3. CT Angiography

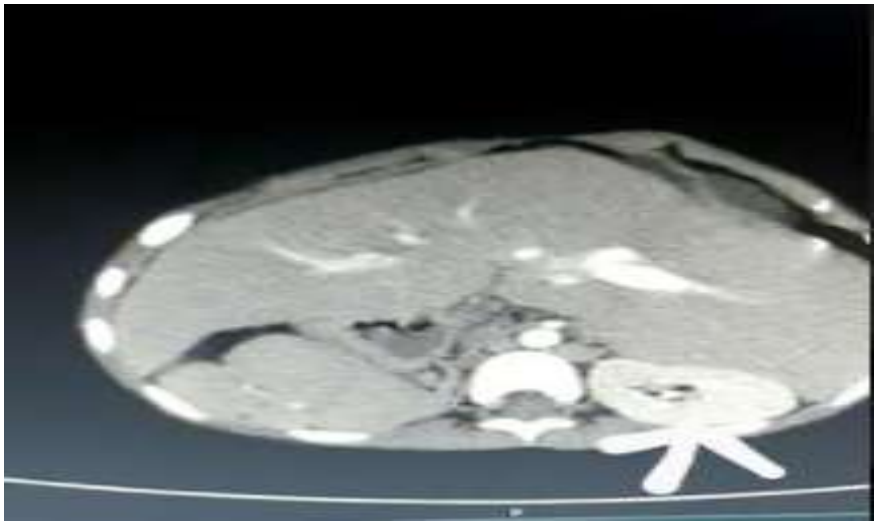

Figure 4. CT of the abdomen showing left kidney but right kidney cannot be visualised in this cut due to inferior displacement.

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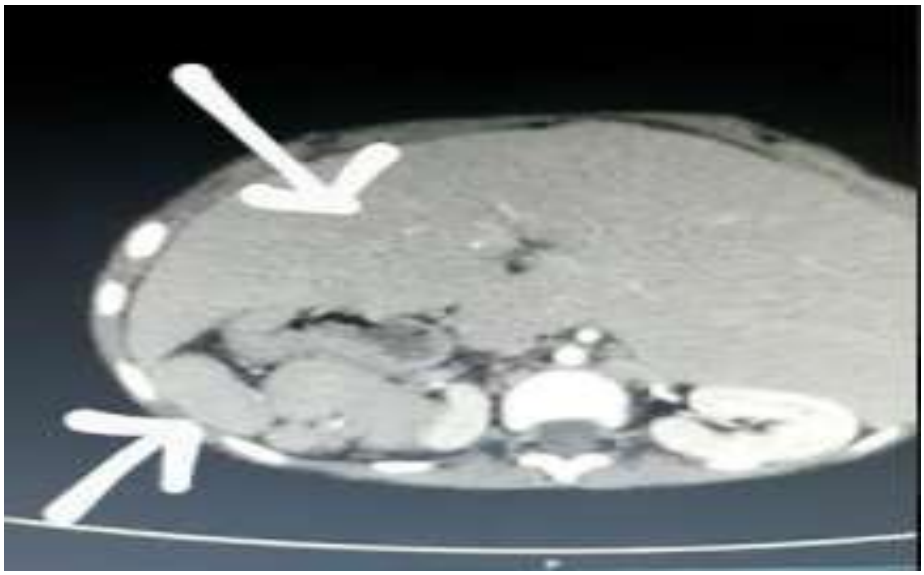

Figure 5. CT of the abdomen showing the liver spanning across the abdomen.

II. DISCUSSION

Based on genetic information, the organs of the human body are arranged in an orderly manner during early embryonic development.[10] The failure of this arrangement may characterize situs inversus or a disordered and variable arrangement (heterotaxy syndrome). Heterotaxy syndrome has an incidence of 1:10,000 births and is seen more often in males, with a ratio of 2:1.[11]

Almost 50% to 100% of patients have a co-exsiting cardiac anomaly and, these are responsible for the severity and the mortality of the syndrome. Asplenia presents as duplication of structures located on the right side of the body, with trilobed lungs, left atrium with morphology corresponding to the right atrium, liver positioned in the middle of the body, left-sided aorta and inferior vena cava, and intestinal malrotation. In most such cases, death is an early event, usually due to complications from congenital heart defects, such as a single atrioventricular chamber. [12] Other reported complications are immunological changes and intestinal volvulus.[13]

Polysplenia includes duplication of the structures in the left side of the body, bilobed lungs, right atrium anatomically identical to the left atrium, liver centrally positioned, absence of the hepatic segment of the inferior vena cava with continuity through the azygos or hemiazygos vein, along with malrotation of the intestines. Cardiac anomalies are less frequently and milder, which might explain detection of such findings in individuals of more advanced ages. [12]

The spectrum of findings in such a syndrome make the individualization of cases important, as most do not adhere to a specific classification. In conclusion, as heterotaxy syndrome has an unusual presentation, there is requirement of additional information to optimize medical care for these patients in order to reduce the morbidity and mortality.

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DECLARATIONS
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Conflict of interest: None

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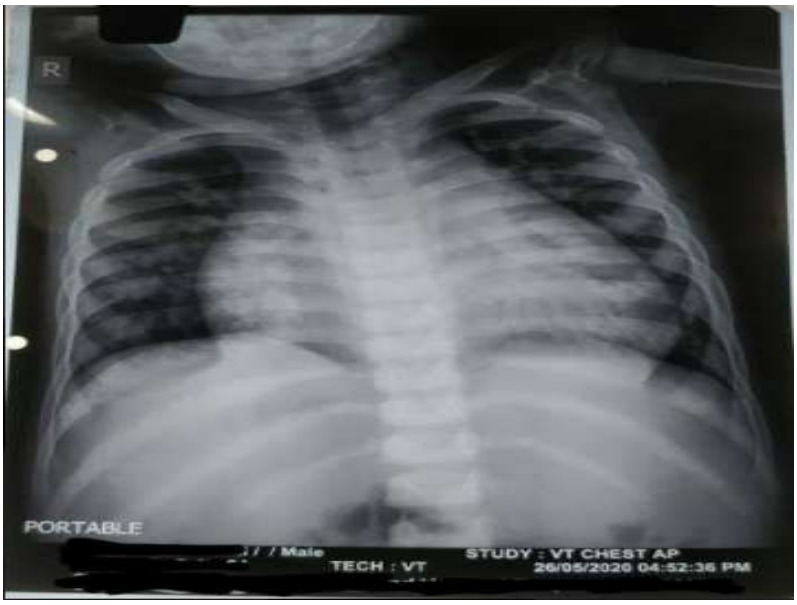

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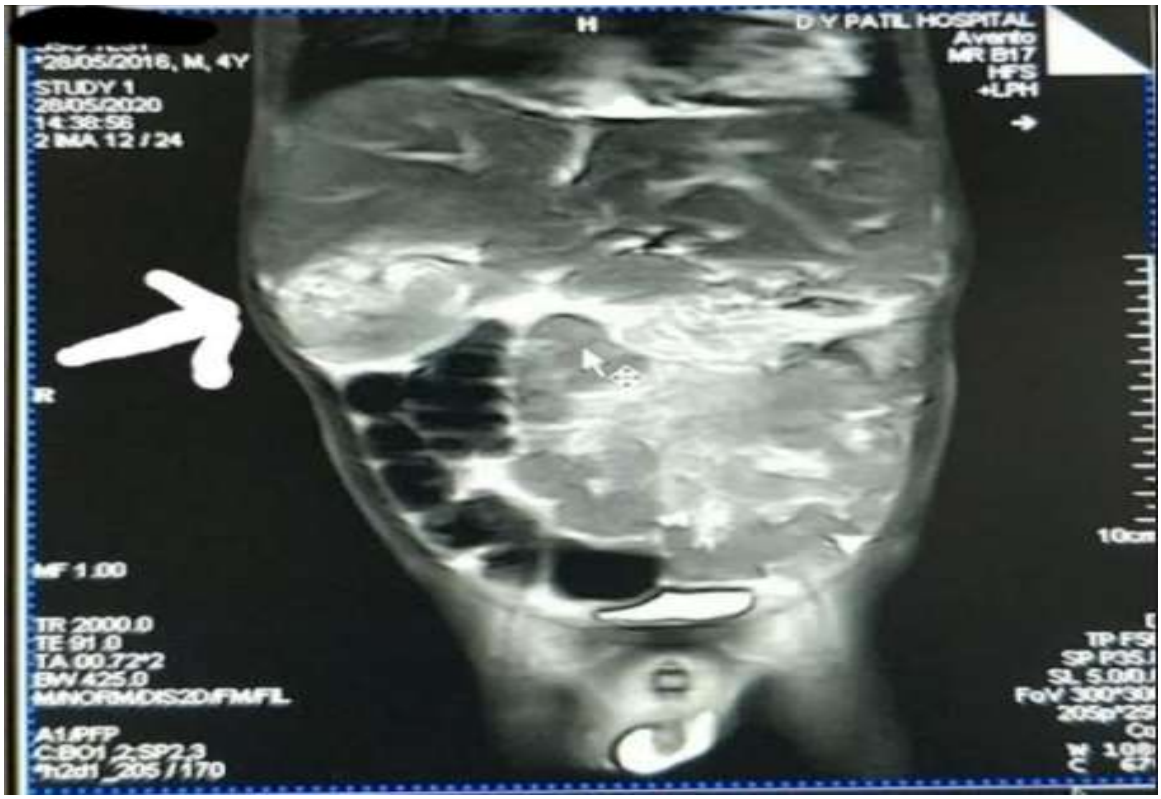

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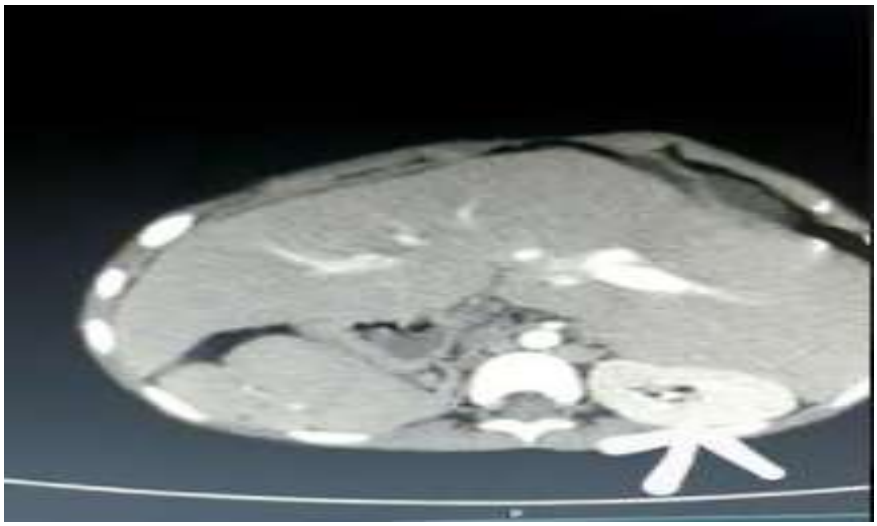

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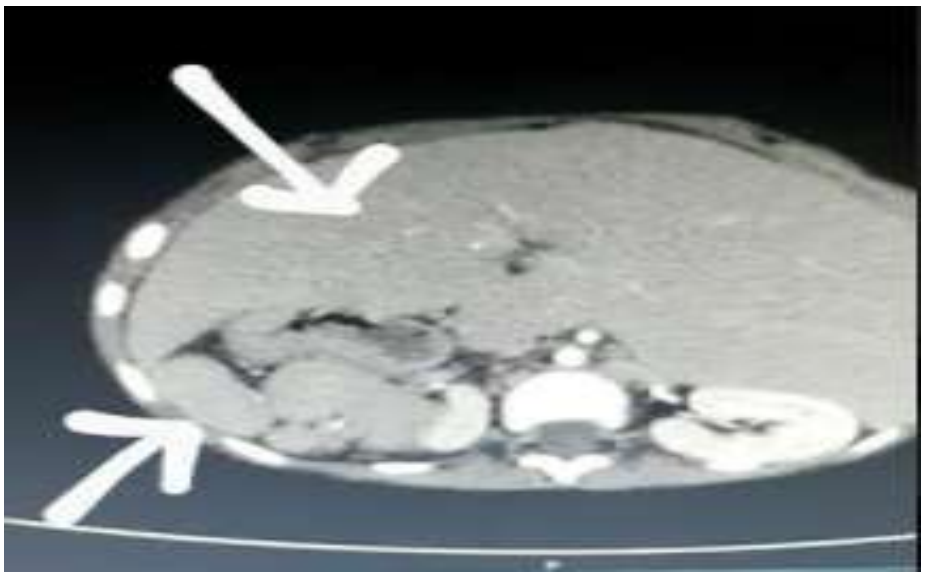

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