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Polysplenia Syndrome: A Case Report

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Abstract **Case Report**

Polysplenia syndrome is a rare congenital disorder belonging to the large spectrum of situs ambigus syndromes. It is characterized by the association of multiple spleens with other vascular, cardiac, intestinal and biliary anomalies. We report the case of an 8 year-old-girl suffering of chronic abdominal pain resistant to symptomatic treatment. Radiological investigations showed right-sided stomach midline liver, two spleens in the retro-gastric region, absence of retrohepatic inferior vena cava and direct drainage of the hepatic veins to the right atrium. No cardiac anomaly was detected. This disorder is often diagnosed incidentally during imaging studies for unrelated medical conditions or when patients present with symptoms of associated anomalies. The aim of this case report is to increase the awareness of this rare entity in pediatric groups presenting common digestive symptoms but with functional impact on daily life.

Keywords: Polysplenia, Spleen, Heterotaxy, Left Atrial Isomerism, Intestinal Inversion, Cardiac Anomalies.

INTRODUCTION

Heterotaxy syndrome is defined as an abnormal morphology and position of the thoracoabdominal organs with usual left-right asymmetry (situs solitus) or reversed/mirrored arrangement of the abdominal and thoracic organs (situs inversus). Polysplenia syndrome is a type of left atrial isomerism characterized by multiple small spleens, often associated with cardiac malformations and with situs ambiguus of the abdominal organs [1]. There is a high association of congenital gastrointestinal abnormalities, such as malrotation of the bowel, biliary atresia, volvulus, and splenic anomalies that can significantly affect the long-term survival. This article presents a case of chronic abdominal pain as an atypical symptom of polysplenia syndrome.

CASE PRESENTATION

An 8-year-old girl with no previous medical history, was admitted to the pedriatric departement of military hospital of Rabat. She suffered from a chronic abdominal pain in the epigatric area since the age of 2 years old. Symptomatic treatments were initated after several medical visits without an outcome. The abdominal pain was associated to other symptoms of reflux such as burning sensation and regurgitations. Physical examination revealed a tenderness over the epigastric quadrant. The weight and height were within standards P: 32kg (+1DS) T: 135 cm (+2DS).

Biological tests resultas were normal. The blood count cells showed hemoglobin (13.2 g/dL), leukocytosis (9600/µL) with neutrophilia (4400); lymphocytes (3300/µL) and platelet count (45600/mm3). The liver and pancreatic tests were in the norms (ASAT= 23 ALAT= 13 Lipase= 10).

Chest X-ray showed levocardia and stomach gas on the right side (Fig 1). Abdominal ultrasound visualized a 40 mm an isoechogenic image in hepatorenal zone. Abdominal CT scan (with angioscan) revealed an incomplete situs inversus, median liver and the presence of two spleens in the retro-gastric region, interruption of inferior vena cava (IVC) in the retrohepatic region. (Fig 2)

Transthoracic echocardiography revealed levocardia, direct drainage of the hepatic veins into the right atrium, no transposition of the great vessels, nor interauricular communication (CIA) or interventricular communication (CIV).

Oesogatro-duodenal fibroscopy showed a discret sliding hiatal hernia and congestive pangastritis. The histological resulats of gastric biopsies revealed a moderate antrofundial chronic gastritis and the presence of Helicobacter Pylori (HP).

The patient was treated for HP+ gastritis. Since épigastric pain persisted, a second gastrointestinal

endoscopy was performed with normal resulats. We concluded that the chronic abdominal pain was related to situs inversus of polysplenia syndrom. A follow up was managed for this patient to detect further complications.

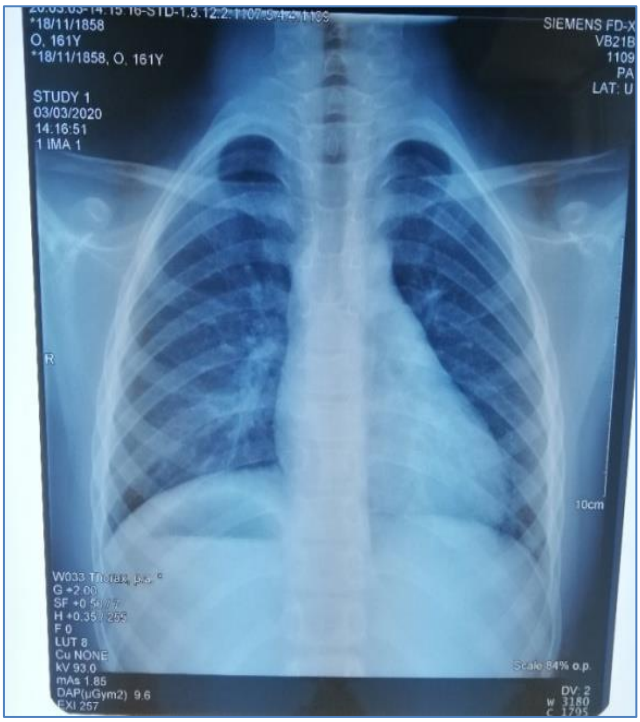

Fig. 1: X ray chest (frontal incidence) showing levocardia and right-sided stomach gas

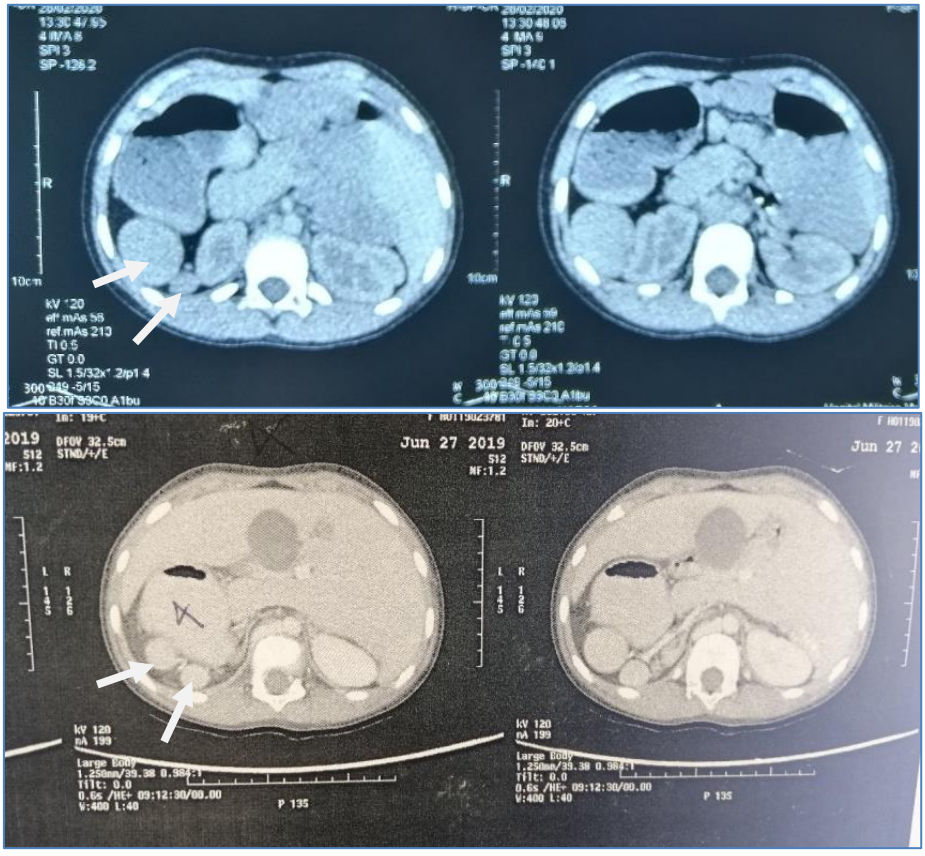

Fig. 2: Axial CT images of abdomen showing midline liver, two spleens in the retro-gastric region (showed with arrows) and interruption of inferior vena cava with a continuation of azygos vein.

DISCUSSION

Heterotaxy is defined as abnormal arrangements of the abdominal and thoracic organs, due to a change in the orientation of the left–right axis in early embryonic development. Heterotaxy is classified into two categories: right isomerism (asplenia syndrome) and left isomerism (polysplenia syndrome). Heterotaxy syndrome has an estimated incidence of 1 in 6000 to 20,000 live births, with a female predominance [1].

Polysplenia syndrome affects approximately one in 10,000 live births [1]. It associates multiple spleens with other malformations usually cardiac, vascular, visceral and biliary.

This syndrom is underdiagnosed in pediatric groups, only hundred cases have been reported so far in several studies [2]. In literature, familial cases of polysplenia syndrome are observed and the transmission is autosomal recessive. The malformations appear between the 4th and 6th week of embryonic life. Therefore a prenatal diagnosis by early fetal ultrasound (17th week of gestation) is necessary in familial cases [3].

The pathogenesis of this syndrom remains incompletely understood. Embryological, teratogenic and genetic factors have been suggested [4]. The exact genetic mechanisms that triggers polysplenia syndrome are still unknown, requiring more investigations of ante- or post-mortem cases.

Polysplenia syndrome is often discovered during routine radiological examination or abdominal surgery. There is no specific clinical symptom of polysplenia syndrome. The clinical findings are variable and range from asymptomatic to serious cardiac complications. [2]. In our patient's case, the only symptom presented was abdominal pain.

Clinical diagnosis can be a challenge for physicians. The polymorphism of polyspenia syndrome leads to a large screening of associated malformations using a complete radiological work-up: thoraco-abdomino-pelvic CT scan with thoracic and abdominal angioscanner. It is difficult to detect the number and position of spleens by only abdominal ultrasound. The CT scan of the abdomen is an additional important tool for further exploration of other vascular and digestive anomalies [5].

The visceral anomalies include situs ambigus type; stomach located on the right in 50% to 87% of cases and liver in medianline [6]. Bilateral bilobed lungs are observed in 48.9%. The common mesentery with intestinal malrotation are noted in over 75% of cases. Duodenal and jejunal strictures, small or annular pancreas are found in 40% of cases [7, 8]. Biliary atresia can be present in 50% of cases [9]. Several severe complications can occur such an intestinal occlusion or a splenic torsion revealed by acute abdominal pain or vomiting in children and adolescents [10].

The most frequent vascular anomaly, occurring in up to 80% of cases is the agenesis of suprarenal inferior veina cava (IVC) and continuation of azygos vein [7]. Other vascular malformations include direct drainage of supra hepatic veinas in right atrium and preduodenal portal vein (37%).

Cardiac anomalies are frequent and severe (85-90% of cases): transposition of the great vessels, atrioventricular and/or interventricular septal defects, common atrioventricular canal, pulmonary artery stenosis, single ventricle [11].

Cardiac malformations associated to polysplenia syndrome are less frequent and viable into adulthood, unlike asplenia, where cardiac anomalies are lethal. Escobar *et al.*, [12] showed that the 5-year survival rate was 86% for polysplenia syndrome compared to 53% for asplenia syndrome. Non cardiac anomalies and pulmonary vein stenosis are predictors for death while in the polysplenia syndrome. The presence of univentricular circulation and left ventricular circulation are predictors for the poor outcome.

Polysplenia can be associated with complex heart manifestations and significant cardiac anomalies from the first day of life. It was reported in a new born boy a rare combination of polysplenia, jejunal atresia, and malrotation, complicated by hypoplastic heart syndrome responsable of high mortality rate [13]. Another rare case of polysplenia syndrome with complex anomalies was published, concerning a one-month-old Infant with severe cardiac malformations, congenital alveolar dysplasia and hepatic fibrosis [14].

Polysplenia syndrome requires no specific medical treatment [7]. The digestive malrotations, biliary anomalies and also cardiac complications may need surgical interventions in emergency cases. The recognition of this pathology can help avoid many diagnostic errors, and also guide any surgical procedure performed on the abdominal level.

CONCLUSION

The diagnosis of polysplenia syndrome is challenging due to the variable presentation and associated anomalies. In this case, imaging exams were crucial in confirming polysplenia syndrome. Early diagnosis and appropriate management with close monitoring and follow-up are essential to prevent cardiac and digestive complications and ensure favorable outcomes.

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