# Case Report: Polysplenia Syndrome, a Wide Range of Congenital Heart Malformations in an Adult

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**ETHICS STATEMENT**

Written informed consent was obtained from the individual(s) for the publication of any potentially identifiable images or data included in this article.

**AUTHOR CONTRIBUTIONS**

All authors listed have made a substantial, direct and intellectual contribution to the work and approved it for publication.

**ABSTRACT**

**Background:** Polysplenia syndrome is a rare disease where are observed two or more spleens associated with left-sided isomerism. In this condition there are many visceral and vascular congenital malformations, including cardiac anomalies.

**Case summary:** We present a case of a 60-year-old male with polysplenia syndrome and multiple cardiovascular abnormalities, but despite these malformations, the patient survived to adulthood with minor symptoms.

**Discussion:** This case report highlights the importance of a multimodality imaging approach in patients with complex congenital heart disease, leading to an unsuspected diagnosis at the adult age.



# CASE REPORT

## INTRODUCTION

Polysplenia syndrome, also known as left isomerism or Ivemark syndrome, is a subtype of heterotaxy syndrome whose main feature is the presence of two or more spleens as part of a left-sided isomerism [1]. This condition is associated to a wider spectrum of visceral and vascular congenital malformations, including cardiac and biliary ones [2]. Although the presence of multiple spleens is a common feature, some cases show a single bilobed spleen; in any case these multiple spleens will be located along the greater curve of the stomach due to their embryonal development [3].

The main features of this syndrome include cardiac malformations such as ventricular or atrial septal defects or outflow abnormalities [4]. Additionally, some patients can present bilobed lungs bilaterally, wrong positioned stomach, middle line liver and an interruption of the inferior vena cava, determining a different venous flow through a proeminent azygos vein or an hemiazygos vein, both running parallel to the descending aorta [5-7].

The advance of the imaging made the diagnostics of this syndrome more common in adults [8]; the presence of unfused spleens should raise to the radiologist the suspicion that other features of this syndrome could be present, helping him to make the right diagnosis [9].

## CASE PRESENTATION

A 60-year-old male patient, without limitations for daily activities, presented to our Hospital University Center with a diagnosis of a complex congenital heart disease (CHD) and situs inversus totalis during childhood. At the age of 6-8, he was offered cardiac surgery, but his father declined the procedure due to its high risk **(**Table 1**)**.

On physical examination a systolic murmur was heard throughout the precordium and marked cyanosis with drumstick fingers was noted.

The electrocardiogram revealed an ectopic atrial rhythm 54 bpm and right ventricular hypertrophy criteria.

The transthoracic echocardiogram documented dextrocardia

with dextroapex, a double outlet right ventricle with a large ventricular septal defect and preserved biventricular systolic function.

A cardiac magnetic resonance was performed for further evaluation of the patient's cardiovascular anatomy. Abdominal images showed situs inversus with multiple spleens, suggesting the diagnosis of a heterotaxy syndrome (left isomerism) (Figure 1C). Dextrocardia with dextroapex (Figure 2) and a double outlet right ventricle was confirmed. Malposition of the great arteries and a right aortic arch was identified with mirror-image branching (Figure 1A). A atrioventricular canal defect was present (Figure 3) along with a supravalvular pulmonary stenosis. Bilateral superior vena cava were found draining to the "functionally" right atrium, on the right through the coronary sinus. There was interruption of the inferior vena cava with continuation into the hemiazygous vein (Figure 1B), draining in the left superior vena cava. The hepatic veins also drained directly to the "functionally" right atrium and the pulmonary veins drained into the "functionally" left atrium.

Despite these cardiovascular malformations, the patient had preserved biventricular function. However in the late gadolinium enhancement (LGE) images, intra-myocardial LGE in the mid-basal morphologically right ventricle and interventricular septum was found, compatible with fibrosis (Figure 4).

A cardiac CT confirmed the presence of symmetric bilobed lungs with bilateral hyparterial bronchi.

Given the stability of the patient's condition, he was considered for clinical follow-up.

## DISCUSSION

Polysplenia syndrome is a rare variant of heterotaxy syndrome, a congenital disorder characterized by an anomalous position of thoracic and abdominal organs [1]. In relation to the typical organs distribution (situs solitus) or the mirror distribution of them (situs inversus), heterotaxy determines a a wrong position of these organs, that can include some vascular malformations [10,11].

The patients with asplenia have a per year mortality rate of 85%, while the ones with polysplenia have a mortality rate of around 50% [12]. Early surgery is generally crucial, particularly for those with cardiac and gastrointestinal abnormalities, even though the late complications and the mortality rates remain high [13].

The exact cause of polysplenia syndrome remains unclear but is probable that is determined by a combination of genetic and embryonal factors [14]. Among the potential causes there are left-right axis modifications during the early embryologic development and mutations in some of the 80 genes responsible for the regular axial development of organs [15].

Others associated malformations are inferior vena cava agenesis, a continuous azygos system, and the confluence of the hepatic veins directly into the right atrium [16]; even if the cardiac abnormalities are less common in polysplenia than asplenia, are still observed in a high percentage of cases, with only 10% of probability to reach the adulthood [17]. Besides the many cardiac malformations already described, there is as well the possibility to have the "bilateral pulmonary atria", when the right pulmonary veins end into the right atrium instead of the left [18,19].

The syndrome is more frequent in women, with an incidence of 1\250,000 newborns [20]. It can be diagnosed both in childhood and adulthood, and the diagnosis is typically made with a prenatal ultrasound recognizing the lateralization abnormalities and thanks to a thoraco-abdominal computed tomography for detailed assessment [21].

Treatment strategies depend on type and severity of cardiovascular malformations, on patients age and clinical conditions [22]. Surgery is recommended to solve cardiac defects in childhood because of a worse prognosis in most cases [23]. In adulthood is preferred a symptomatic therapy, specifically focusing on the cases that involve the portal veins, intestinal malrotations or vascular abnormalities, to prevent surgical complications [24]. There are no specific guidelines for long term follow-up and for surveillance of these patients [25] .

Summarizing, polysplenia syndrome is a rare and complex condition associated to severe cardiovascular malformations. Cardiac abnormalities are quite common, and often an early surgical management can be crucial to improve the outcomes; in any case, in this case report the aim is to highlight that even with multiple malformations some patients can live with minor symptoms and have a good prognosis with an individualized management, without surgery.

## CONCLUSIONS

The importance of this article lies in presenting and describing a case of left isomerism with multiple cardiovascular abnormalities (double outlet right ventricle, atrioventricular canal defect, supravalvular pulmonary stenosis, bilateral superior vena cava, uninterrupted inferior vena cava) and a long survival.

Furthermore, this article highlights the critical role of multimodality imaging in diagnosing and evaluating the associated abnormalities, specifically for a right management and prognosis of these patients.

**Data availability statement**

The original contributions presented in the study are included in the article, further inquiries can be directed to the corresponding author.

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## FIGURES

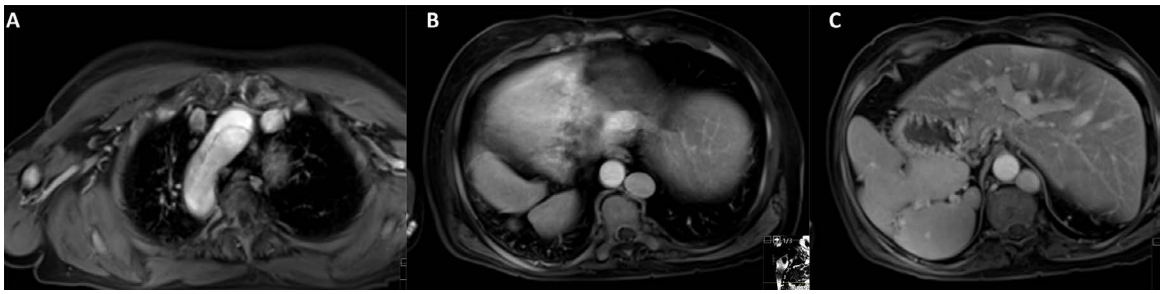


**Figure 1:** Axial T1 fat-saturated (T1FS) image after intravenous gadolinium showing right-sided aortic arch (A); hemi-azygos continuation of the inferior vena cava (B) and abdominal situs inversus with left-sided liver and polysplenia on the right upper abdominal quadrant (C).

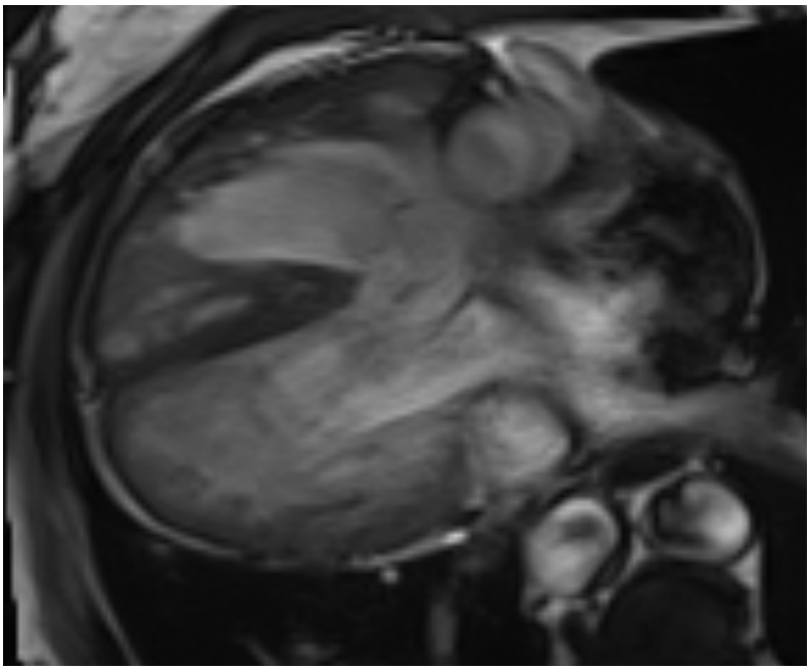

**Figure 2:** Balanced Steady State Free Precession (SSFP) cine image in the horizontal long-axis of the left ventricle shows dextrocardia and dextroapex. Right ventricular hypertrohpy is also seen.

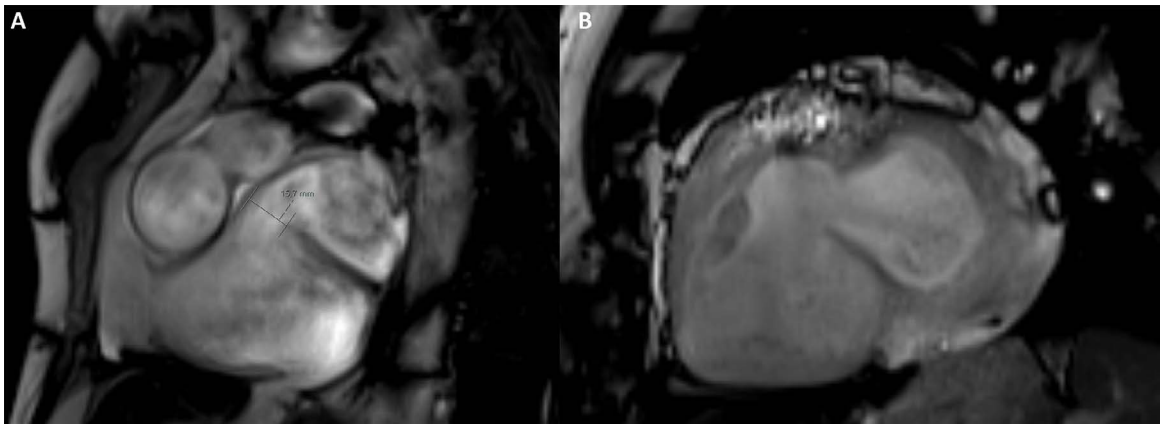


**Figure 3:** Balanced SSFP cine image in the short-axis of the left ventricle showing atrial septal defect (A) and sub-pulmonary ventricular septal defect (B).

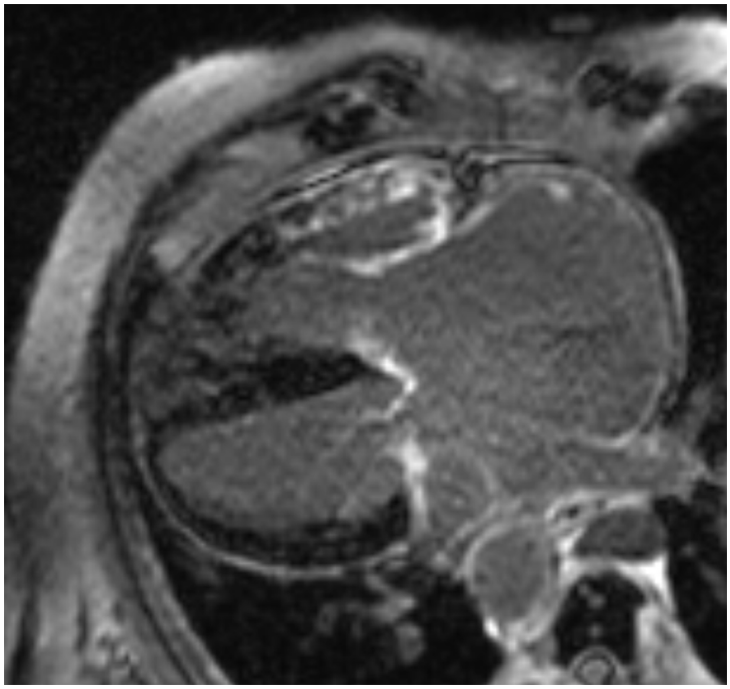

**Figure 4:** Late gadolinium enhancement (LGE) Phase-Sensitive Inversion Recovery (PSIR) image in the horizontal long-axis of the left ventricle shows intra-myocardial LGE on the basal portion of the right ventricle and extensive LGE involving both atrioventricular valves. Mild enhancement of the atrial walls can also be seen.

TABLE

| **DATE** | **EVENT** |
|---|---|
| Age of 5 | Diagnosis of congenital heart disease and *situs inversus totalis* |
| Age of 6-9 | He was offered cardiac surgery (refused) |
| Age of 12 | Started the follow up at our center |
| 2008 (unspecified) | Hip surgery for septic arthritis |
| 2012 (unspecified) | Followed by the nephrology clinic for proteinuria |
| 2013 | Diagnosis of type 2 diabetes |
| Currently | Stage 3 chronic kidney disease |
| Currently | Periodic follow up for the congenital heart disease |

KEYWORDS

*MRI, Polysplenia, Congenital, Heart, Cardiovascular*