Abdominal Manifestations of Situs Ambiguous abnormalities in adults. A study of four cases


1. Becado de Radiología, Universidad Mayor. Chile.
2. Radiólogo, Hospital Barros Luco Trudeau, Clínica Alemana de Santiago. Chile
3. Radiólogo, Facultad de Medicina. Clínica Alemana de Santiago - Universidad del Desarrollo. Chile

Abstract: Anomalies Situs (ASIT) has been described mainly in the pediatric population and there is little information on this in adults. These are very rare abnormalities; in the case of Situs Ambiguous (SAMB) in adults, there have only been a few published case series. SAMB, also called heterotaxy or heterotaxy Syndrome, is defined as the abnormal or ambiguous positioning of organs and vessels, that could also be associated with morphological alterations characteristic of each organ. There are two main groups within SAMB: polysplenia and asplenia. This publication presents a specific literature review, with emphasis on the classification of adult ASIT and abdominal findings in cases of SAMB. It will be complemented clinically and radiologically with four cases of abdominal tomographic imaging studies.

Keywords: Heterotaxy, Polysplenia, Situs ambiguous.

Resumen: Las Anomalías del Situs (ASIT) han sido descritas principalmente en la población pediátrica y existe escasa información sobre éstas en el adulto. Son anomalías muy raras; en el caso del Situs Ambiguous (SAMB) en el adulto sólo se han publicado pocas series de casos. SAMB, también llamado heterotaxia o Síndrome de heterotaxia, es definido como la posición anormal o ambigua de los órganos y vasos, que además pueden asociarse a alteraciones morfológicas características de cada órgano. Existen dos grandes grupos dentro de los SAMB: Poliesplenia y Asplenia. En la presente publicación se expone una revisión bibliográfica específica, la cual tiene énfasis en clasificación de las ASIT del adulto y los hallazgos abdominales en los casos de SAMB. Será complementada clínica e imaginológicamente con cuatro casos de estudios tomográficos abdominales.

Palabras clave: Heterotaxia, Poliesplenia, Situs ambiguous.


Correspondence: Dr. Andrés Labra W. / labraw@yahoo.com
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Introduction

Anomalies Situs (ASIT) has been described mainly in the pediatric population. In contrast, there is little information in literature about these entities in adults. This can be explained, at least in part, because there are pathologies strongly associated with these anomalies which prevent many patients reaching the adult age, such as congenital heart disease, immunodeficiency and intestinal obstruction. Furthermore, when adult patients present ASIT, they have it with mild symptoms and/or practically non-symptomatic, reasons for which they fail to require imaging studies for this cause. In fact, most ASIT adults are incidental findings in studies for unrelated pathologies such as appendicitis or cholecystitis.

These anomalies are very rare, Situs Inversus has been reported with a prevalence of approximately 0.01% of the U.S. population and in the case of Situs Ambiguous in adults (SAMB) only a few case series have been published.

Often ASIT and particularly SAMB seem confusing, since in the literature a variety of different terms have been used to classify or sub-classify them, such as heterotaxy syndrome or simply heterotaxy, asplenia and polysplenia syndrome or different isomerisms(1,2).

As will be explained further on, this text will introduce the use of the nomenclature used in the latest and most important publications related with the subject.

The aim of this publication is to present a literature review, with emphasis on the classification of adult
ASIT and the abdominal findings in cases of SAMB. It will be complemented by four cases.

A summary of the background, findings and diagnosis of the patients under study can be reviewed in Table I.

Terminology

As mentioned above, a variety of terms exist in the literature to refer to Anomalies Situs (ASIT) which added to its low frequency, often leads to confusion. This present article will develop based on the terminology and classification used by Fulcher and Turner in 2002 in their publication in Radiographics(2). This work presents a series of cases from a single center that includes SAMB, being the highest amount published to date with nine cases in almost thirty years. Similarly, it aims to promote the use in our area of this nomenclature and classification so as not to continue spreading the already extensive amount of synonyms for the same entities (Figure 1).

The term “Situs” refers to the position of the heart, viscera and great vessels relative to the midline. Thus, Situs Solitus refers to the normal position of the organs described above, whereby the cardiac apex is located to the left (Levocardia) the same as the spleen and stomach. On the other hand, the liver, gallbladder, and vena cava are found to the right. Also included in this concept is the correct positioning of the colon and the loops of the small intestine. In these conditions the presence of congenital heart disease (CHD) is 1%(2).

Situs Inversus indicates the existence of a mirrored configuration in relation to Situs Solitus. Two major categories exist for Situs Inversus: Situs Inversus with Dextrocardia or with levocardia, where the variant with dextrocardia, also called Situs Inversus Totalis is vastly more frequent and appears with CHD in only 3-5% of the cases, whereas in the variant with levocardia practically all the cases appear with CHD(2).

Situs Ambiguous (SAMB), also known as heterotaxy or heterotaxy syndrome is defined as the abnormal position, ambiguous or in the midline of the organs and vessels, with a layout different to that found in Situs Solitus. CHD is present in 50 - 100% of the cases(2). This category is probably that which presents more problems when identifying abnormalities, and also therefore at the time of making a correct diagnosis and classification, because SAMB cases do not always come with the same group of abnormalities, but rather encompasses a spectrum of variants which may or may not be present, in addition to there being various degrees of severity. Furthermore, none of these constitutes a pathognomonic finding.

There are two major groups within SAMB: with polysplenia, sometimes also called polysplenia syndrome and left isomerism (for having only two bilateral lung lobes), and with asplenia, also called asplenia syndrome or right isomerism (for having three bilateral lung lobes). It is worth noting that in the category with asplenia CHD occurs in 99-100% of the cases and also tends to be more serious than in the other ASIT(1,2).

The classification used is summarized in Figure 1.

Abdominal findings of Situs Ambiguous abnormalities

SAMB may show a spectrum of abnormalities in relation both to the “Ambiguous” location of the organs, as well as the morphological characteristics thereof. We must remember it is not necessary that all organs are compromised.

Regarding the ambiguous position of the abdominal organs, in general, these can occur in a normal position, in the midline and continuing the spectrum until a mirror position. On the other hand, morphological alterations are characteristic of each organ.

Following will be described the principal findings in the literature and correlation with the cases presented in Table I.

Spleen: When present, may be either in normal, ambiguous or mirror position. This is of special importance, because based on the morphological findings the SAMB is subclassified: with polysplenia and with asplenia (Figure 1). Making this difference has importance because SAMB cases with asplenia are significantly less common in adults, given its high mortality rate during the first year of life because of its greater association with CHD both in frequency and in severity, and for decreased immunity. It is worth mentioning that polysplenia is defined as the presence of two or more spleens, however, the characteristic presentation is with multiple small rounded spleens of different sizes, also
**Table I. Situs Ambiguous Cases.**

<table>
<thead>
<tr>
<th>Patient</th>
<th>CASE 1</th>
<th>CASE 2</th>
<th>CASE 3</th>
<th>CASO 4</th>
</tr>
</thead>
<tbody>
<tr>
<td>Reason for consultation</td>
<td>Abdominal Pain, Obs. Diverticulitis</td>
<td>Asymptomatic</td>
<td>Abdominal pain, lipase (+)</td>
<td>Acute chest pain, Obs. SAA</td>
</tr>
<tr>
<td>Background</td>
<td>Male, 65 years</td>
<td>Female, 23 years, asthma</td>
<td>Female, 23 years</td>
<td>Female, 67 years, cholecystectomy</td>
</tr>
<tr>
<td>Tests conducted</td>
<td>CT of abdomen</td>
<td>MRI of abdomen</td>
<td>Cholangio-MRI, CT of abdomen and pelvis</td>
<td>Angio-CT thorax, CT of abdomen</td>
</tr>
<tr>
<td>Main Findings</td>
<td>Polysplenia</td>
<td>Polysplenia and mirror position</td>
<td>Polysplenia</td>
<td>Polysplenia</td>
</tr>
<tr>
<td></td>
<td>I IVC-C AV</td>
<td>I IVC-C AV</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>Short pancreas</td>
<td>Left IVC</td>
<td>Short pancreas</td>
<td>Short pancreas</td>
</tr>
<tr>
<td></td>
<td>Intestinal malrotation</td>
<td>I IVC-C AV Short pancreas</td>
<td>Vein passing through pancreas</td>
<td>IM</td>
</tr>
<tr>
<td></td>
<td>Polylobulated BV</td>
<td>IM Liver in midline Polylobulated BV</td>
<td>IM Polylobulated BV.</td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Pre-duodenal portal vein</td>
</tr>
<tr>
<td>Diagnosis</td>
<td>SAMB with polysplenia</td>
<td>SAMB with polysplenia</td>
<td>SAMB with polysplenia, Acute Pancreatitis</td>
<td>SAMB with polysplenia</td>
</tr>
</tbody>
</table>


called “splenules”. It is important to point out that in the case of presenting a study with typical characteristics of a SAMB, but only one spleen, it should be classified as SAMB with polysplenia, however, this form of presentation is uncommon(1-3).

The four cases presented show polysplenia (Table I). The variation between them was a spleen composed of few splenules (Image 1), morphology described as characteristic of polysplenia with multiple splenules (Image 2) and one case where this organ was found in a mirror position in the upper right quadrant (Image 3).

Liver, gallbladder and biliary tract: The most frequent are the positional abnormalities, characteristically the liver and gallbladder can be found in the midline (Image 4) or mirrored.

A polylobulated aspect of the gallbladder has not been described in other studies, however, this finding was present in three of our cases (Image 5). Pancreas: As an abnormal position the head of the pancreas is described to be found to the right of the midline (Image 6). Morphologically the SAMB is associated with short pancreas (no tail and/or body), also that a vessel/vein can traverse the gland (Image 7)(2,4).
Image 1. Axial CT of the abdomen in portal venous-phase which demonstrates polysplenia with few splenules (Case 1).

Image 2. Coronal T2 weighted sequence MRI of the abdomen. Shows characteristic morphology of the polysplenia composed of multiple splenules in the upper left quadrant (Case 2).

Image 3. Coronal T2 weighted sequence MRI of the abdomen. Polysplenia can be seen in mirrored position in the upper right quadrant (Case 2).

Image 4. Axial GRE T1 weighted out-of-phase MRI of the abdomen. It shows the position of liver in the midline and the stomach to the right (Case 2).

Image 5. Axial T2 weighted MRI of the abdomen. Gallbladder can be seen with a polylobulated aspect (Case 3).

Image 6. Axial CT of the abdomen in portal venous-phase which shows the location of the head of the pancreas to the right side of the midline (Case 1).
Image 7. Axial CT of the abdomen in portal venous-phase. Note the short pancreas (no tail) and a vessel/vein which passes through the gland (Case 3).

Interruption of the inferior vena cava (IVC) with azygos-hemiazygos vein continuity: the intrahepatic portion of the IVC has been described in normal position, to the left (Image 8) or also found duplicated. Often the intrahepatic portion of the IVC in the caudate lobe is not observed and the intrahepatic portion continues with the azygos or hemiazygos system (Image 9); this type of abnormality is rare in patients with SAMB with asplenia[1-4].

Image 8. Axial GRE T1 weighted in-phase MRI of the abdomen. It shows the position of intrahepatic IVC (VCI) to the left of the midline and the aorta (AA) (Case 2).

All of our reported cases had IVC interruption with azygos vein continuity.

Aorta: this is found in its normal position in the majority of the described cases. It has been reported to the right of the midline when it coexists with duplicated IVC, also cases have been described where the IVC and the aorta are found on the same side[1,2,3].

Gastrointestinal tract: the stomach may present a normal or mirror position (Image 10). In regard to the small intestine and colon, varying degrees of intestinal malrotation can be observed, the most often described being that the small intestine is found to the right of the midline and the colon to the left (Image 11). Other degrees of malrotation can be observed, for example, when the cecum is not completely fixed to the retroperitoneum[2]. Also described in the literature is the position of the portal venous in front of the duodenum, which possibly may cause symptoms of partial intestinal obstruction (Image 12)[4].

Image 9. Sagittal CT of the abdomen and pelvis in portal venous-phase, reconstruction with average intensity projection (MIP). Interruption of the inferior vena cava (VCI) with Azygos vein (Az) continuity, can be observed (Case 3).

Image 10. Coronal T2 weighted MRI of the abdomen. Stomach is observed in mirror position (Case 3).
Conclusion

The diagnosis and classification of SAMB using abdominal studies can be difficult because they do not always present the same group or degree of abnormalities. On the other hand, given that historically a variety of terms have been used to refer to different types of ASIT, we proposed consensus on the terminology used in the present publication and which is summarized in Figure 1, leaving aside other terms to avoid confusion.

Currently, the greater availability of imaging methods have shown that ASIT, especially the SAMB type, are not exclusive to the pediatric age and are not always associated to morbidity, on the contrary, they can be present in adults and as an incidental finding.

It is important to know the different variants and incorporate the concept that it is about a broad spectrum of possibilities, in order to arrive more easily to a correct diagnosis and classification of the ASIT with which we are confronted.

Bibliography


Image 11. Axial CT of the abdomen in portal venous-phase. Notice how the colonic loops are almost exclusively to the left of the midline and those of the small intestine to the right (Case 4).

Image 12. Axial CT of the abdomen in portal venous-phase. Observe how the portal vein (solid arrow) is found in position pre-duodenal locating in front of the duodenum that is collapsed (area above the dotted arrow) (Case 3).