

Case Report

Situs Inversus Totalis Detected Incidentally in a Young Patient

Lílian Sousa Santana¹, Laura Campos Modesto², Ana Amélia Damasceno Parreira³, Vitorino Modesto dos Santos⁴¹Master Institute of Education Presidente Antônio Carlos (IMEPAC), Araguari-MG, Brazil²Universitary Center (UNICEUB), Brasília-DF, Brazil³Department of General Surgery, Hospital of Base, Brasília-DF, Brazil⁴Department of Medicine, Armed Forces Hospital and Catholic University, Brasília-DF, Brazil

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Corresponding author's email:

vitorinomodesto@gmail.com

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ABSTRACT

Situs inversus totalis is a rare congenital anomaly characterized by the complete transposition of the thoracic and abdominal organs. We report the case of a previously healthy 21-year-old patient who was incidentally diagnosed with the condition at age 19, following an abdominal ultrasound requested on her own initiative due to a prior suspicion of an anatomical variation. The patient had a history of dextrocardia identified in childhood, without further investigation at the time. The diagnosis was confirmed by ultrasound, which showed complete inversion of the abdominal organs. This case reinforces the importance of early identification of congenital anatomical anomalies and their clinical relevance in diagnostic and surgical contexts.

Keywords: Dextrocardia; Diagnosis; Situs Inversus Totalis

Introduction

Situs inversus totalis (SIT) is a rare autosomal recessive entity characterized by the complete and specular inversion of the thoracic and abdominal organs in relation to their usual anatomical position; the incidence is estimated at between 1 in 8,000 and 25,000 newborns; is associated with multiple gene mutations affecting both sexes equally, and among all the congenital cardiovascular malformations, dextrocardia represents less than 3% of all cases (1-5). This condition can occur in isolation or associated with other anomalies, such as

primary ciliary dyskinesia, Kartagener syndrome, congenital heart disease, and significant vascular variations (1-5). Although some of the affected people can remain asymptomatic into old age, the early recognition is crucial, as SIT can alter the clinical presentation of common diseases, with misdiagnosis or adaptations in various surgical procedures and interventions (2,5). The objective of this case report is to enhance the awareness and suspicion index of general practitioners about this very rare condition.

Case Presentation

A 21-year-old female patient was previously healthy and without relevant comorbidities. At age 9, during a radiological investigation for an acute respiratory condition, dextrocardia was identified on a chest X-ray (Figure 1).



Figure 1. Plain chest X-ray: Classic appearance of dextrocardia.

At that time, no further investigation was performed, and she was not referred for a specialized follow-up. During adolescence, the patient became aware that dextrocardia could be associated with several other anatomical variations, but remained asymptomatic and without regular medical follow-up. At age 19, during an ultrasound exam of the right forearm to rule out carpal tunnel syndrome (confirmed by the classical imaging features), she also underwent a complete abdominal ultrasound. The routine anamnesis performed by the

radiologist informed her dextrocardia; then he recommended the abdominal exam to verify eventual SIT. The evaluation confirmed the complete transposition of abdominal organs, including the liver, spleen, stomach, and kidneys (Figure 2).



Figure 2. Abdominal echography: Abdominal organs that appear normal, but are in an inverted lateral position (situs inversus).

Both the results and images were provided by the patient, who reported some sporadic episodes of gastroesophageal reflux, without clinical or complementary evidence that could establish any direct causal relationship with the SIT. It is noteworthy that the specialized literature describes the majority of this group of patients with this condition presenting with preserved normal gastrointestinal functions. The patient remains asymptomatic, without any recurrent respiratory

symptoms or repeated infections, nor cardiac abnormalities; nevertheless, she has missed the scheduled

specialized follow-up appointments as was clearly directed by her generalist physician.

Discussion

Cases of SIT result from alterations in the embryonic lateralization process responsible for defining the left-right asymmetry of the thoracic and abdominal organs. Although the mechanisms involved are quite complex, failures in this process can occur, resulting in either a partial or complete inversion of the viscera, as was described in this case report. Most individuals with SIT remain asymptomatic throughout their lives, and their diagnoses are usually incidental, as was the example of the young patient here described. Nevertheless, early diagnosis of this condition is essential, as it can mimic the common conditions such as appendicitis, cholecystitis, or chest pains arising in unusual sites (5). Furthermore, SIT may be associated with conditions such as primary ciliary dyskinesia, which presents with recurrent respiratory infections and alterations in the ciliary motility; as these associations occur due to failures in the ciliary function during embryonic development, they may also result in conditions such as the Kartagener syndrome (1-4). However, in the present case, the patient did not have either repeated infectious episodes or respiratory manifestations that could be suggestive of such hypothetical condition. From a gas-

trointestinal standpoint, the digestive function is generally preserved; however, symptoms such as gastroesophageal reflux may occur, although there is no evidence of a direct causal relationship with the situs inversus (5). In this case study, the patient presented with mild symptoms sporadically, without significant clinical repercussions. However, in surgical contexts, the SIT may represent a technical challenge, requiring adaptations in the surgical approach, as well as in the composition of the surgical team and equipment. The laparoscopic procedures may require a "mirrored configuration" of the operating room, as has been described in those case reports involving the Nissen fundoplication (2,5). The early recognition of this anatomical inversion is an essential tool for risk reduction and to avoid errors related to the spatial orientation during eventual invasive procedure. In the present case, the patient did not present with heart disease, recurrent respiratory infections, or manifestations consistent with primary ciliary dyskinesia, conditions frequently associated with SIT (1-4). The patient's current clinical evolution remains stable, and at this time, there is no need for ongoing specialized close monitoring.

Conclusion

SIT is a rare congenital anomaly that, despite its unusual anatomical presentation, generally does not cause more significant clinical repercussions. The case described demonstrates that the diagnosis can occur incidentally, and that many individuals remain asymptomatic throughout their lives. The early recognition of this condition is crucial, mainly in emergency assistance or surgical management, because anatomical inversion can alter the interpretation of clinical signs and require diverse technical procedures. Proper documentation and increased awareness among healthcare professionals regarding these anatomical variations can contribute

to greater safety and technical accuracy during the clinical and surgical management of this very uncommon group of patients. In this specific case, the patient and her parents did not have the necessary orientations about the clinical significance of the dextrocardia incidentally found. Therefore, she passed over more than a decade before the diagnosis of SIT, when the radiologist's good history-taking practice sparked her interest in an abdominal US examination.

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