Мак Мед Преглед 2023; 78(1): 28-30

MMP

Original article

SITUS INVERSUS TOTALIS IN A NEWBORN WITH CONGENITAL HEART DISEASE

SITUS INVERSUS TOTALIS КАЈ НОВРОДЕНЧЕ СО КОНГЕНИТАЛНА СРЦЕВА МАЛФОРМАЦИЈА

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Abstract

Introduction. Situs inversus totalis (SIT) is rarely reported in newborns. Isolated or associated with other congenital abnormalities, most often with congenital heart malformations (3-9%), SIT can often be an accidental finding.
Case report. We report a term-newborn with SIT and complex congenital heart defect, diagnosed prenatally by fetal ultrasound. SIT was confirmed with plane film X-ray (liver on a left side, spleen on right side). Heart ultrasound was done and revealed a complex cardiac malformation (CHM) - dextrocardia, single ventricle, tricuspidal valvular atresia, hypoplastic aortae, ASD II, PDA. The baby was transferred to a heart surgery center where the neonate was treated but unfortunately passed away.
Conclusion. SIT with CHM is a rare condition in neonatal period. Although a myriad of congenital malformations can be accompanying, isolated SIT is the most common. SIT and CHM is a condition challenging for surgical treatment.

Keywords: situs inversus totalis, newborn, congenital heart malformation

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Апстракт

Вовед. Situs inversus totalis (SIT) е ретка кондиција кај новороденчиња. Може да биде изолиран или асоциран со вродени абнормалности, најчесто со вродени срцеви малформации (3-9%), но често може да биде и случаен наод.
Приказ на случај. Доносено новороденче со SIT и комплексна вродена срцева мана, пренатално дијагностицирана со ултразвук. Со бебиграм се потврди SIT и се детктираше: црн дроб на левата страна, слезината на десната страна. Ехокардиографски се потврди комплексна срцева малформација-декстрокардија, single ventricle, трикуспидална валвуларна атрезија, хипопластична аорта, ASD II, PDA. Новороденчето се префрли во кардиохируршки центар, се изврши хирушка интервенцеја, но поради комлексноста на срцевата мана за жал почина.
Заклучок. SIT во комбинација со конгенитална срцева мана е ретка состојба во неонаталниот период. Иако мноштво вродени малформации можат да бидат придружни, но изолираниот SIT е најчест. SIT и конгенталните кардиопатии се состојби кои се вистински предизвик за изведување на хируршки третман.

Клучни зборови: situs inversus totalis, новороденче, вродени срцеви малформации

Introduction

Situs inversus totalis (SIT) is a rare congenital abnormality. The abdominal and thoracic organs are positioned as a mirror-image transposition [1,2]. SIT is categorized as solitus, inversus, and ambiguous; solitus is the normal arrangement of the organs, situs inversus totalis is a mirror image of the normal position of internal organs [2], while situs ambiguous or heterotaxy is the random arrangement of internal organs [3].
There is a lengthy history of description of SIT, from Aristotle (BC. 384-322) who described this condition in animals [4] to Fabricius (1600 a.d.) who reported SIT in humans [5]. The first X-ray report was the transposition of the viscera in 1897 by Vehsemeyer [6].
It has been found that some conditions are risk factors for the development of SIT. Among them, the most important are: family history of heart defects, family history of noncardiac anomalies, maternal diabetes, paternal smoking, antitussive use, and low socioeconomic status [7].
Dextrocardia occurs when the heart fails to migrate to the left chest. In situs inversus totalis the heart tubes are rotated to the left and the placement of the heart and other internal organs is a mirror image of the normal arrangement. A chain of signaling molecules has been implicated in influencing organ rotation and migration. „Sonic hedgehog" (Shh) is a protein that affects the expression of two transforming growth fac-

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tors, called Nodal and Lefty [1,8]. A number of genes, more than 100, are associated with laterality, including genes for primary ciliary dyskinesia (PCD) [1].
We present a term-newborn with a congenital heart defect.

Case report

A 27-year-old patient with first regularly controlled pregnancy was hospitalized at the University Clinic for Gynecology and Obstetrics in Skopje in February 2024. A prenatally verified complex cardiac anomaly (AVSD-atrioventricular septal defect, single ventricle, stenosis valvulae mitralis) was US diagnosed as well as situs inversus totalis. Family history was uneventful. A term boy in 38+3 week of gestation, with birth weight of 3470 g, Apgar score 2/6/7 in the first, fifth and tenth minute was born by elective caesarean section. After primary resuscitation with ambu ventilation with 100% oxygen, the baby was transferred to the NICU. A plain film X-ray was done and situs inversus totalis (dextrocardia, liver-left positioned, spleen on the right) was confirmed (Figure 1). Due to acidosis (pH=7.09, pCO_2=69.1, pO_2=41.6,

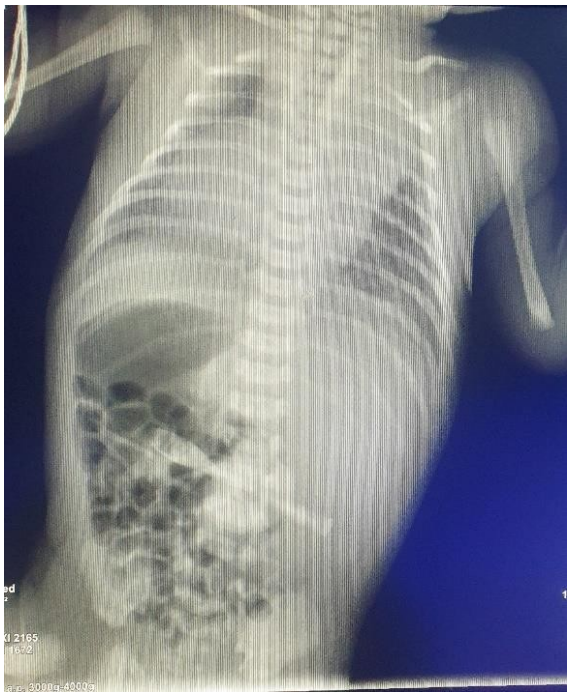

Fig. 1. Situs inverus totalis with complex congenital heart anomaly

HCO_3=20.6, BE=-10.4, sO_2=73.2%) the baby was intubated and put on conventional mechanical ventilation (SIPPV+VG). A pediatric cardiologist was consulted and a heart ultrasound was performed. A complex heart malformation was diagnosed (dextrocardia, single ventricle, tricuspidal valvular atresia, hypoplastic aorta, ASD II, PDA). The newborn was transferred to a cardiac surgery center. Surgery was performed, but unfortunately, due to the complexity of the heart anomalies, the newborn died at the age of 6 days.

Discusion

The incidence of situs inversus totalis is estimated to be 1:6,500 to 1:25,000 [1], with a male-to-female ratio of 3:2 [8].
The first suspicion of situs inversus totalis can be raised after a careful physical examination, ECG, as well as with imaging techniques. Typical findings are dextrocardia, left-sided liver and right-sided spleen [9-11], confirmed by electrocardiography inversion of electrical waves [12]. Computed tomography (CT) or magnetic resonance imaging (MRI) are useful imaging techniques that provide a detailed description of situs anomalies well before delivery [13]. SPECT/CT labeled with 99mTc is used to differentiate between polysplenia and abdominal masses [14].
The diagnosis of SIT is rare in the neonatal period. SIT is most often incidental during radiographic evaluation [15]. In our patient, the diagnosis of SIT and complex congenital heart malformation were detected in the uterus. We confirmed SIT with plain film X-ray (dextrocardia, left-placed liver and right-placed spleen) and heart ultrasound (dextrocardia with complex congenital heart disease).
The fact that the patient has a diagnosis of situs inversus totalis is an important finding [16]. The diagnosis of SIT is especially important in emergency surgical situations. Acute abdominal emergencies, such as cholecystitis, acute appendicitis or spleen injury, the manifestation of symptoms and signs is unusual [17,18]. Sometimes technical modifications are needed in surgical interventions. Patients with SIT can also develop malignant or benign neoplasms [19,20], and accurate diagnosis is required for localization and treatment. In patients with SIT, organ transplantation is more complicated [21].
Most people with situs inversus live normal lives [12]. SIT can be isolated or associated with other congenital abnormalities. The most common is intestinal malrotation, which affects 40-90% of patients [22]. Other malformations are duodenal atresia, biliary atresia, gastrochisis, congenital coronary abnormalities, ventricular septal defect, congenital heart disease [23,24]. SIT is also a part of some syndromes, such as Kartagener's syndrome (situs inversus totalis, abnormal paranasal sinuses and bronchiectasis) [25] and Ivemark's syndrome (SIT and asplenia) [26]. SIT has been shown in 20% of patients [27]. The rate of congenital heart disease is estimated to be ~0.6% in situs solitus (normal anatomy), 3-9% in situs inversus totalis, and almost 80% in situs ambiguous (1). SIT with dextrocardia and congenital heart defect has been observed in 3-5%. The most common congenital heart

defect in SIT is transposition of the great vessels [28]. Situs inversus totalis with left heart is a rare condition [29] and is mostly associated with congenital heart disease [30]. Our patient was the newborn with a rare congenital heart defect associated with SIT-single ventricle, tricuspid valvar atresia, hypoplastic aortae. Due to the complexity of the heart defect, despite cardiosurgical interventions, the newborn died.

Conclusion

Situs inversus totalis is a complex disorder in embryological morphogenesis. Efforts should be done to discover associated congenital anomalies and the possibility of life-threatening complications should always be kept in mind. A multidisciplinary approach is important in all stages of diagnosis and monitoring the patients. It is mandatory to inform clinicians of the anatomical mirroring to prevent complications during some interventions.

Conflict of interest statement. None declared.

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