SITUS INVERSUS TOTALIS WITH UNILATERAL UNDESCENDED TESTIS: A CASE REPORT

Qurat-ul-Ain, Talha Sohail, Noman Bashir✉, Somayya Siddiqa, Nazir Ahmed Malik

HITECH Institute of Medical Science, Taxila Cant - Pakistan

Address for correspondence:
Noman Bashir
HITECH Institute of Medical Science, Taxila Cant - Pakistan

E-mail:
nomangeoscientist92@gmail.com

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ABSTRACT

Situs Inversus Totalis (SIT) is a rare genetic anomaly that causes the heart and abdominal viscera to be positioned in a mirror image of their regular positions. It may present alone or in association with various associated anomalies. Most patients present with no symptoms and are often diagnosed incidentally; however, some have co-existent anomalies and must be ruled out.

A male neonate weighing 3.2 kg delivered at term via Em-LSCS presented with transient tachypnea of the newborn (TTN) and was on oxygen dependence in the neonatal intensive care unit (NICU). A General Physical Examination revealed heart sounds on the right side of the chest and empty left scrotal sac. Chest X-ray was done that showed Situs Inversus Totalis and congenital pneumonia. Ultrasonography of the scrotum revealed the absence of the left testis.

Situs Inversus Totalis should be evaluated and investigated to rule out associated anomalies, even when the finding is incidental. It is recommended that physicians promote regular medical examinations for their patients, since this practice can aid in the detection of anomalies, hence mitigating the risk of incorrect diagnoses and potential mortality resulting from delayed treatment.

Keywords: Siturs Inversus; Inguinal Cryptorchidism; Neonate

INTRODUCTION

A rare aberration known as Situs Inversus Totalis (SIT) is typified by the heart and abdominal organs being positioned in a mirror copy of their typical placements.[1] The abnormality has been widely acknowledged as a complex condition, with many genetic variables and maternal environmental modifiers being postulated as potential causes.[2] A few case reports from recent years have mentioned that certain diseases like skeletal dysplasia, retinitis pigmentosa, congenital heart defects, cystic kidney disease, and biliary atresia are often found in patients with SIT. However, the majority of patients are asymptomatic and show no obvious signs or symptoms. One possible hypothesis posits that the aetiology of SIT may also be implicated in congenital and acquired defects in several organs.[2] The assessment of the prevalence of SIT in newborns poses challenges due to its potential occurrence alongside other congenital abnormalities that may be incompatible with survival. However, the condition exhibits a prevalence rate of approximately 1 in 20,000 live births.[3]

Cryptorchidism, also known as undescended testis (UDT), is a prevalent genitourinary condition observed in male infants. In the majority of instances, it is quite likely for the testes to undergo spontaneous descent by the age of three months. However, if the testes fail to descend by the age of six months, the likelihood of spontaneous descent afterward is minimal.[4] The reported prevalence of this condition is observed to be elevated among infants who are born prematurely and have low birth weight, with reported rates reaching up to 45%. The prompt and appropriate referral for medical intervention, along with timely surgical procedures, have the potential to effectively mitigate the occurrence of infertility and lower the likelihood of developing malignancies.[5]

This case report and literature review describes a rare incidence of Situs Inverses Totalis in coexistence with unilateral undescended testis in a neonate.

CASE REPORT

A male neonate weighing 3.2 kg was delivered at 39±1 weeks through an Emergency C-section, due to pre-eclampsia and previous one scar. The mother was 29 years and a thalassemia trait.

Consanguinity was reported to be positive; mother had a history of a miscarriage, ectopic pregnancy and

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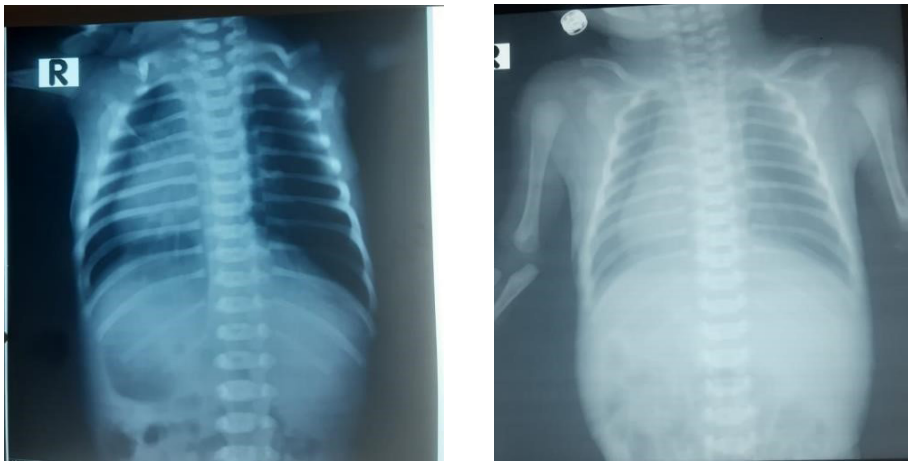
Intra-uterine death in previous pregnancies. The neonate had an APGAR score of 8/10 five minutes after birth. He was taken to Neonatal Intensive Care Unit (NICU) in Heavy Industries Taxila (HIT) Hospital, where he developed TTN with respiratory rate of 68. At room air, his oxygen saturation dropped to 75%, for which he was given oxygen via oxygen face mask at the rate of 1 liter per minute.

On Examination, heart sounds were audible at the right side of chest, liver was palpable at left upper quadrant and left scrotal sac was empty. Chest indrawing and flaring of ala nasi was positive. Posteroanterior chest radiograph was done that revealed Situs Inversus Totalis. Right-sided anatomical features include the stomach, descending colon, aortic arch, and cardiac apex. The upper left quadrant is where the liver is situated. The abdominal viscera are positioned mirror-imagery in relation to Situs Solitus, as confirmed by abdominal sonography. The inferior vena cava is left-sided, while the abdominal aorta is right-sided. Left testis, measuring (12×05 mm) was seen in left inguinal canal near deep ring. 2D Echocardiography revealed Isolated mirror image dextrocardia and no evidence of pulmonary hypertension was identified. Ultrasonography of scrotum revealed absence of left testis. Other investigations done included CBC (Normal), C-reactive protein (Negative), ABGs (Normal) and Serum electrolytes (Normal).



The patient was admitted for observation and treatment for pneumonia done. The patient recovered gradually, saturation was maintained at room air and he was discharged on twelfth day of life, in good health with instructions to consult a pediatric surgeon to treat for the undescended testis after six months of age.

DISCUSSION

Situs anomalies refer to a wide array of congenital abnormalities affecting the visceral and vascular systems, which present a diverse variety of radiographic symptoms. The term Situs Solitus is used to describe the typical arrangement of asymmetrical anatomical structures, in which the heart's apex, left atrium, left bilobed lung, stomach, spleen, and aorta are all located to the left of the midline and the liver, gallbladder, right bilobed lung, and right inferior vena cava are located to the right.[6]

Situs Inversus has been classified into two distinct subtypes: situs inversus with levocardia, which is often referred to as Situs Inversus Incompletus, and situs inversus with dextrocardia, commonly known as Situs Inversus Totalis. The categorization is established based on the alignment of the heart's base-to-apex axis. In cases of levocardia, this axis is directed towards the left, whereas in dextrocardia, it is directed towards the right. The prevalence of situs inversus is higher in individuals with dextrocardia compared to those with levocardia.[7]

Situs Inversus Totalis was seen in the patient under consideration. The orientation of the heart's base-to-apex axis was observed to be directed towards the right side, whereas the stomach was on the right side of the abdomen and the liver was on the left. The absence of any accompanying symptoms in the patient can be attributed to the presence of a "mirror-image" arrangement of the heart and abdomen viscera. The presence of cardiac anomalies is considerably low in association with SIT, only 3%-5% of patients have co-existing congenital heart disease, compared to a very high prevalence in patients with situs inversus with levocardia. The most common anomaly reported is transposition of great vessels,[8] hence, an echocardiography must be done to eliminate any such anomaly. SIT is highly associated in co-existence with other anomalies. Several pulmonary and vascular abnormalities[9] and other anomalies, including duodenal atresia, asplenism, multiple spleens, ectopic kidney, and horseshoe kidney, have been associated with this disorder. Multiple visceral abnormalities, both congenital and acquired, have been linked to the underlying cause of SIT.[2] In our case, the patient has co-existence of unilateral undescended testis, it suggests a correlated developmental abnormality of organs that develop from mesoderm (heart and testis) and endoderm (alimentary canal).

Kartagener syndrome is the medical term for the occurrence of both Situs Inversus Totalis and primary ciliary dyskinesia. Approximately 20% of instances of Situs Inversus Totalis (SIT) are documented as being associated with Kartagener Syndrome. Coughing, choking, and other respiratory distress signs are present from birth in patients with this illness, and they progress to situs inversus, chronic sinusitis, and bronchiectasis during the course of their lives. Impairment of ciliary motility is observed, leading to an increased

susceptibility to recurrent sinopulmonary infections and infertility.[10] Typically, the fall of the testes to the scrotum occurs during the gestational period between 25 and 35 weeks. The prevalence of undescended testis is significantly higher in preterm newborns compared to term infants. While the diagnosis is made at birth in 1% to 4% of term infants, the incidence climbs to 45% in preterm infants.[4] In around 45% of instances, the testis experiences natural descend by the age of 3 months. However, the likelihood of descent diminishes beyond the age of 6 months, necessitating surgical intervention. This intervention is necessary due to the elevated risk of testicular cancer and impaired fertility.

CONCLUSIONS

Situs Inversus Totalis (SIT) is a rare congenital anomaly that warrants comprehensive evaluation and investigation upon detection, due to its correlation with infrequent yet potentially lethal comorbidities. A comprehensive evaluation is necessary, utilizing diverse imaging modalities, in order to exclude any concurrent problems. During preoperative and surgical care of their patients, pediatricians, surgeons, and radiologists must be attentive for the presence of this abnormality. Additionally, it is crucial to engage in patient and attendant counselling in order to provide comprehensive care.

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Author's Contribution

NB designed and wrote initial manuscript. QUA, TS and SS collected patient data and helped in literature review. NAM helped in finalizing the manuscript. Authors agree to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflict of Interest

Authors declared no conflict of interest

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None

Data Sharing Statement

The data that support the findings of this study are available from the corresponding author upon reasonable request.