

Case Report

Situs Inversus Totalis

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Abstract

A 2 months old girl weighing 3.2kg, 1st issue of a non-consanguineous parents immunized as per EPI schedule was born at term at home by NVD presented to the emergency department of NBMCH with severe respiratory distress and fever for 5 days. Her mother also complained of unable to feed for 1 day. For routine examination, she was referred to the Medical Imaging Center for X-ray chest & abdomen, USG of whole abdomen, echocardiography and some radiographic studies like CT scan etc. But unfortunately, we could not done CT scan due to unavailability. However, by the USG and X-ray we observed a situs inversus totalis. We found right sided heart, stomach and spleen as well as left sided liver with gall bladder. Her parents were unaware about her unusual anatomy.

Key words: *Situs inversus totalis, Dextrocardia, Congenital abnormality, Transposition of viscerae.*

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Introduction

Situs inversus totalis is a congenital positional anomaly characterized by transposition of abdominal viscera associated with a right sided heart (Dextrocardia). Situs inversus totalis is a condition in which the organs of the chest and abdomen are arranged in perfect mirror image of normal.³ Both male & female ratio is 3:2.¹³

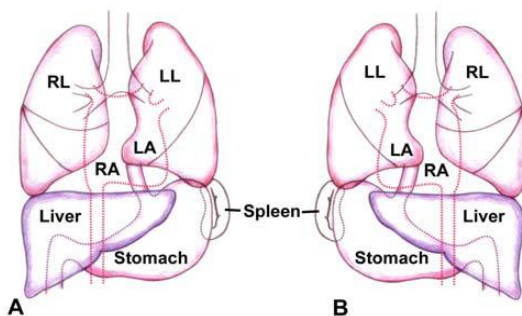


Figure 1: Schematic Diagram of Mirror Image of Situs Inversus

(Adapted from Wilhelm A, 2011)³

It was Mathew Baillie who first described situs inversus totalis in early twentieth century. A transposed thoracic and abdominal organ is a mirror image of the normal anatomy confirmed by radio imaging such as x-ray film and USG of abdomen. Generally individuals with situs inversus totalis are asymptomatic and have a normal life expectancy. Many people with situs inversus totalis are unaware of their unusual anatomy until they seek medical attention for an unrelated condition. The reversal of the organs may lead to some confusion as

many signs and symptoms are found on the reverse side.

The prevalence of situs inversus varies among different populations which is less than 1 in 10,000 population. Situs inversus is a rare congenital anomaly characterized by transposition of the abdominal organs, viscera& vasculature. When associated with dextrocardia, it is known as situsinversus-totalis.¹ This condition is generally an autosomal recessive genetic condition. It may or may not be associated with dextrocardia.^{1,2} Generally genetic anomaly is discovered incidentally when radiographic assessment of patient is performed. The heart is located on the right side of the thorax, the stomach and spleen are also located on the right side of the abdomen and the liver and gall bladder on left.

The situs inversus with dextrocardia or situs inversus totalis has been occurred once in about 6000-8000 live births. Situs inversus with levocardiaorsitus inversus incompletes is an another rare condition (1 in 22,000 of general population) in which the heart is found on the normal left side of the thorax⁶. Situs inversus with levocardia or dextrocardia without situs inversus present with much higher rates in congenital defects than situs inversus with dextrocardia. We report this case of situs inversus discovered in the medical imaging center following image studies compared with normal anatomy in relation to severe pneumonia.

Discussion

Situs inversus is generally an autosomal recessive genetic condition, sometimes¹² it can be X-linked and also found in identical twins^{10,16} and there is no difference between races.

Recent studies suggest that left-right asymmetry defects is to be due to genetic abnormalities in lefty genes, nodal genes, and ZIC 3, ACVR2B and Pitx2 genes. Mutation of genes present on chromosome 12.^{12,15} In the absence of congenital heart defects, individuals with situs inversus are phenotypically unimpaired and can live normal healthy life without complications (Figure 2). About 25% of individuals with situs inversus have an underlying condition known as primary ciliary dyskinesia (PCD). Situs inversus with PCD together known as "Kantagener Syndrom".¹⁴

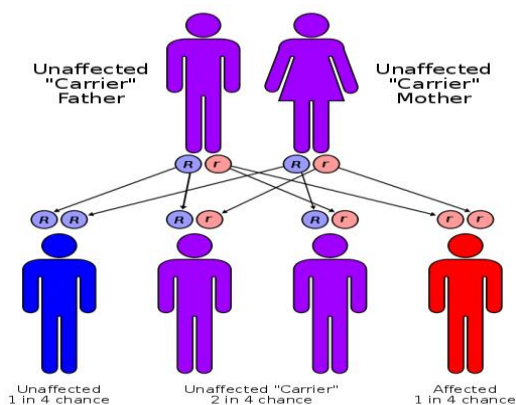


Figure 2. Situs Inversus has an Autosomal Recessive pattern of inheritance

(Adapted from Kosaki et al, 1999)¹²

Situs inversus occurs more commonly with dextrocardia.³ A 3%-5% incidence of conge-

nital heart disease is observed in situs inversus with dextrocardia, usually with transposition of the great vessels, 80% of these patients have a right-side aortic arch.

Situs inversus with levocardia rare⁵ and it is almost always associated with congenital heart disease.^{6,7,8,9} Many people with situs inversus totalis are unaware of their unusual anatomy until they seek medical attention for unrelated condition. The reversal of the organs may then lead to some confusion, as many signs and symptoms would be on the wrong side.

For example, if an individual with situs inversus develops appendicitis, they will present to the physician with lower abdominal pain in the left side instead of right where the appendix generally lies. Physician may confuse in such situation.

Abdominal and chest X-ray ultrasonography and CT scan also facilitate reliable and early diagnosis if patient is unaware of his unusual anatomy. In patient with situs inversus totalis, who present with an acute abdominal pain having appendicitis that involving left ileac region, the diagnosis become more complex and difficult. Until 2008 fewer than 10 cases of appendicitis associated with situs inversus were reported in the literature.¹¹ Thus, in the event of a medical problem, the knowledge of situs inversus can expedite diagnosis.

The occurrence of situs inversus is very rare. A few cases have been reported in the literature which causes diagnostic problem that can be solved by the medical image particularly X-ray chest and abdomen, USG

of the abdomen, echocardiography and CT scan. These procedures allows for early management of the disease and guidance for the best approaches.

The case

A 2 month old girl weighing 3.2 kg, 1st issue of non-consanguineous parents immunized as per EPI schedule was born at term at home by NVD presented to the emergency department of NBMCH with severe respiratory distress and fever for 5 days. Her mother also complains of unable to feed the baby for last 1 day. With above these complaints she was admitted in the department of Pediatrics.



Figure-3: X-ray of Chest and Abdomen

On clinical examination, we found the patient is dysphonic, HR-120/min, Temp-102⁰F, RR-66 breath/min, chest in drawing and fast breathing present, mildly anemic, non-icteric, mild-cyanotic, Heart- s_1+s_2 audible at right side of chest, lungs-rhonchi present, other clinical signs revealed normal.

We have done only chest X-ray and USG. But when we proceed to listen the heart sound it was not found in left side .On

curiosity, further checked in the right side it was clearly audible with shifted apex beat. She was provisionally diagnosed as a severe pneumonia with consolidation. As the size of the chest of baby was small, we were confused about the position of the heart of the baby. Then we referred him to medical imaging center for chest X-ray, USG of whole abdomen, echocardiography and CT scan etc. But patient party could not be done CT scan and echocardiography due to unavailability and financial incapability.

We observed dextrocardia with situs inversus which is known as situs inversus totalis. Here we found right sided heart, stomach and spleen as well as left sided liver with gall bladder. All the thoracic and abdominal organ and viscera were completely inverted.

Conclusion

Patient with situs inversus who present to the junior doctor at emergency department may have diagnostic problems in physical examination because of their unusual anatomy. It is important to inform senior and expert medical personnel to diagnose a case like situs inversus totalis in order to decrease errors and prevent complications that arise during patient assessment and care, particularly in case of appendicitis and other abdominal organ anomalies. Both practical experience and theoretical knowledge play a vital role to diagnose such abnormality and always needs interdepartmental co-operation to manage the complications effectively and efficiently.

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