SITUS INVERSUS TOTALIS - A RARE CASE REPORT

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ABSTRACT

BACKGROUND

A 60 years old previously asymptomatic female was admitted with accelerated hypertension and she was incidentally found to have mirror images of normal anatomical organs in chest and abdomen called situs inversus totalis. Situs inversus totalis is a congenital anomaly characterized by transposition of abdominal viscera associated with dextrocardia. Complete situs inversus is a rare syndrome of autosomal recessive inheritance. The anatomic, pathologic, embryologic and aetiology of complete situs inversus and related abnormalities are presented in this case with special emphasis to genetic correlation.

KEYWORDS

Accelerated Hypertension, Complete Situs Inversus, Congenital Anomaly, Dextrocardia, Transposition of Abdominal Viscera.

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BACKGROUND

Situs inversus totalis is a congenital condition, in which the major visceral organs are reversed from their normal position. The situs inversus is present in 0.01% of the population. It is an autosomal recessive genetic condition, sometimes it can be X-linked and also found in identical twins.^[1,2] The persons with situs inversus totalis are usually asymptomatic and have a normal life expectancy. The situs inversus totalis has been estimated to occur once in about 6000 - 8000 live births. Situs inversus with levocardia^[3] is another rare condition occurring in 1 in 22,000 of general population, in which the heart is found on normal side of thorax.

CASE REPORT

A 60 years old female patient was admitted with a history of headache and giddiness of 2 days' duration. She did not give any history of significant illness in the past. There was no significant illness in the family. On examination, she was conscious, oriented, her pulse rate was 80 beats per minute, BP was 180/100 mmHg in supine and upright position. She did not have any signs of focal neurodeficit.

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Her investigations revealed a haemoglobin of 12.4 gm%, total leukocyte count of 10100/mm³ with polymorphs of 86%, lymphocytes 12%, PCV 40.6%. Blood sugar, renal function tests, liver function tests and sr. electrolytes were within normal limits. She was not immunosuppressed. Chest radiogram showed Dextrocardia. Ultrasound abdomen revealed liver in left hypochondrium, spleen in right hypochondrium. Inferior vena cava noted left to the aorta, that is features of situs inversus totalis. Her ECG revealed inversion of P waves in leads I and aVL, dominantly negative QRS waves in leads I, V1-V6, extreme QRS axis, flattened T waves in aVR and inverted T waves in lead I and aVL. The features suggesting dextrocardia. Her 2D Echo revealed Dextrocardia with normal sized cardiac chambers and normal LV function. With the clinical and investigation results, a diagnosis of situs inversus totalis was made. She was started on calcium channel blockers for her hypertension. She was completely asymptomatic till date.

Left Sided Leads

Right Sided Leads

Chest Radiogram **USG Abdomen and Pelvis**

- Liver in left hypochondrium.
- Spleen in right hypochondrium.
- Features s/o situs inversus totalis.

CT Abdomen and Pelvis

2D Echo

DISCUSSION

Mathew Baillie first described situs inversus totalis in early 20th century. Distribution is same for both genders (1:1) and there is no difference between races. Rarely situs inversus can run in families, but most often it is an isolated and accidental event occurring in an individual for the first time in family.

In human and in vertebrates, anatomic asymmetry is established during embryogenesis. The left-right axis is determined at the beginning of the embryonic development with the formation of the dorso-ventral and cephalocaudal axes. The cardiac tube curve to the right is the first sign of asymmetry. The left-right gradient has been established at cellular level. The left-right relation of asymmetric viscera is conserved; it is known as situs solitus and when there is complete inversion of the lateralisation of the organs (mirror image); it is known as situs inversus totalis.^[4,5,6,7]

Recent studies suggest that left-right asymmetry defects 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.^[4,5,6,7] The individuals with situs inversus and phenotypically unimpaired and can lead normal healthy lives without any complications related to their medical conditions.

Situs inversus can be classified further into situs inversus with levocardia or situs inversus with dextrocardia. The classification of situs is independent of the cardiac apical position. The terms levocardia and dextrocardia indicate the direction of the cardiac apex at birth, but not the orientation of the cardiac chambers. In levocardia, the base to apex axis is towards the left and reverses in case of dextrocardia. Isolated dextrocardia is situs solitus with dextrocardia. The cardiac apex points to the right, but the viscera are otherwise in their usual positions. Situs inversus dextrocardia is situs inversus totalis because of the cardiac position as well as the atrial chambers and reversed abdominal viscera.^[1,2,3,8,9,10] Cardiac situs is determined by the location of the atria. In both situs

solitus and situs inversus, the ventricles may present in two positions.

1. D-loop or right sidedness, that is right ventricle is anterior and to the right of the left ventricle (normal).
2. L-loop or left sidedness, right ventricle is posterior and to the left of left ventricle (inversion of ventricles). Dextrocardia is cardiac malposition in which the heart is in the right hemithorax with the base to apex axis pointing to right^[11]; 3% of people with situs inversus totalis have some form of congenital heart disease.^[12,11] About 25% of individuals with situs inversus totalis have an underlying condition known as primary ciliary dyskinesia.

PCD is the dysfunction of the cilia that manifests itself during the development of foetus. Situs inversus with PCD together known as Kartagener syndrome characterised by triad of situs inversus, chronic sinusitis and bronchiectasis.^[13,14,15,16,17]

CONCLUSION

Situs inversus totalis is mostly an incidental finding. Most individuals are unaware of having situs inversus totalis in them and they come to know during screening investigations for unrelated illness. Though Kartagener syndrome can be associated with situs inversus totalis, so physician must be vigilant in suspected cases and should take help of radiological investigations to confirm the diagnosis. Situs inversus also complicates organ transplantations as donor organs will almost certainly come from situs solitus donors.

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