Case Report

Situs Inversus Totalis with Atrial Septal Defect : A Rare Association

Mahesh Dave[1], Puneet Patel[2], Saurabh Jain[2], Aniruddha Burli[2]

Situs inversus totalis is the complete inversion of position of the thoracic and abdominal viscera. It may be isolated or associated with malformations, especially cardiac and/or alimentary. Usually it remains asymptomatic and is discovered as an incidental finding in adulthood, but sometimes it may be discovered during infancy itself due to associated anomalies. We report a 19-year-old male found to have situs inversus totalis in association with Atrial Septal defect (ASD) while presenting to a medical OPD. This incidental finding situs inversus totalis in association with Atrial Septal defect (ASD) was detected by physical examination and was confirmed later by echocardiogram and other radiological studies. This report emphasizes the importance of complete physical examination with special reference to patients presenting with dextrocardia/situs inversus. [*J Indian Med Assoc* 2021; **119(6):** 60-1]

Key words **: Situs inversus totalis, Atrial septal defect, Congenital heart disease.**

Situs inversus totalis refers to a right-sided location of the heart within the thoracic cavity with complete mirror imaging of abdominal viscera. It is a primary manifestation of the abnormal lateralisation of the embryonic left-right axis during early development[1]. While its true incidence remains largely unknown, estimates range from 1 in 8,000 to 25,000 live births[2]. Chest radiograph, Ultrasonography and an electrocardiogram are enough to make a diagnosis of dextrocardia, while more recent imaging modalities like echocardiography and magnetic resonance imaging confirms the diagnosis[3]. Situs inversus totalis is usually associated with variable congenital anomalies which include primary ciliary dyskinesia (Kartageners syndrome) and Cardiac defects[4]. The common cardiac anomalies seen in situs inversus totalis include Transposition of great vessels (3-5%), Atrio-ventricular discordance and right sided aortic arch. Atrial Septal Defect (ASD) is very rare congenital anomaly associated with situs inversus totalis. Hence, we report this case of Situs inversus totalis with Atrial Septal Defect (ASD).

Editor's Comment :

- Situs inversus totalis is a rare condition characterized by completely reverse positioning of heart and abdominal viscera.
- Detection of prior cardiac anomalies is very important as invasive cardiac procedures, if needed, are very challenging in such patients.

Case Report

A 19 years old male came to medical OPD with complains of chest pain and shortness of breath on & off for past five months. Chest Pain was non-radiating, dull aching type, increased by exertion and was not associated with palpitations, diaphoresis, dizziness or loss of consciousness. He had no history of orthopnoea, Paroxysmal Nocturnal Dyspnea (PND) and bilateral swelling of lower limbs. Past history revealed recurrent episodes of upper respiratory tract infections since childhood. On general physical examination, Patient had pallor with no clubbing, cyanosis and pedal oedema. His vitals were normal. Cardio Vascular System (CVS) examination revealed apex beat on right side of chest, 10.5 cm lateral to mid sternal line in 5th Inter coastal space with normal character. On auscultation wide fixed splitting of second heart sound at right upper sternal border (no variation with respiration) was noted. Rest was normal. Abdominal examination showed no organomegaly, but on percussion, liver dullness was found on the left side upper abdomen. So clinically, situs inversus totalis with ASD diagnosis was suspected and the patient was subjected to further extensive workup. His all routine investigations which include Renal Function Tests (RFT), Liver Function Tests (LFT) and urine examination were within normal limits except for hemogram which showed Anaemia with haemoglobin 8.4gm%. X-ray Chest donewas showing heart on the right side with apex pointed towards the right side. The aortic arch was on the left side.The left hemidiaphragm was raised and the gastric bubble was on the right side.Abdominal ultrasound revealed that the liver and gallbladder were on the left side whereas the spleen was on the right side suggestive of situs inversus. His Echocardiography done and were suggestive of dextrocardia with Inferior venacava, superior venacava draining into left sided right atrium, pulmonary veins draining into right sided left atrium, AV/VA concordance, great arteries relation normal, ostium secundum type of atrial septal defect of 2.5 mm in size, with shunt from left atrium to right atrium, mild Tricuspid Regurgitation (TR) with dilated right atrium and right ventricle and intact interventricular septum.

Discussion

Situs inversus totalis is a rare congenital anomaly reported to occur in 1 in 8000 to 1 in 25,000 patients[5]. On review of literature the incidence approximately appears

Department of General Medicine, RNT Medical College, Udaipur, Rajasthan 313001

[1]MD, FICP, FRCP, Senior Professor & Ex-Head and Corresponding Author

[2]MD, Junior Resident

Received on : 18/11/2019
Accepted on : 31/07/2020

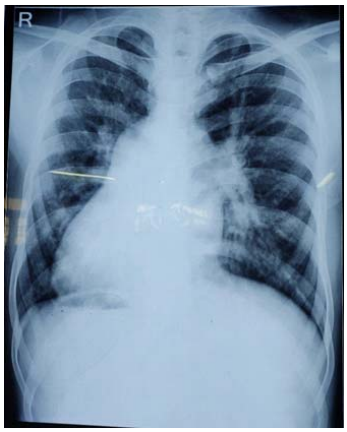

Fig 1 — X ray chest PA view showing dextrocardia with fundic gas shadow on right side

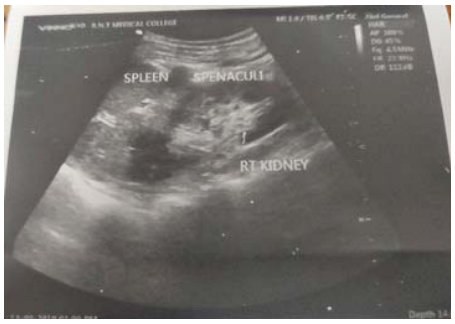

Fig 2 — USG Abdomen showing situs inversus

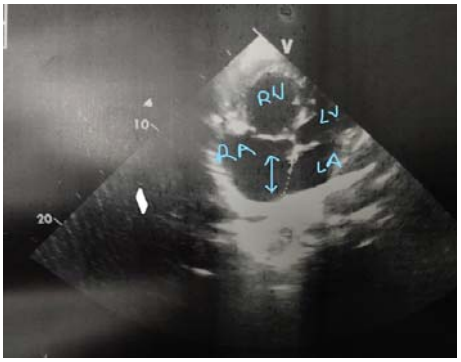

Fig 3 — Echocardiography (4 chamber view) showing Ostium secundum type of ASD. The defect is showed with dotted line measuring 2.5mm

to be 1:10,000 adults and although it appears to be genetically determined the exact mode of inheritance is not clear[6]. The male-to-female incidence is 1:1. The arrangements of the position of the abdominal viscera in dextrocardia may be normal (situs solitus), reversed (situs inversus totalis), or indeterminate (situs ambiguous or isomerism) in 32 to 35%, 35 to 39%, and 26 to 28% of cases, respectively.The interatrial septum plane is oblique in cases of laevocardia, with the left atrium more posterior than the right atrium. In dextrocardia, the interatrial septum is directed anteriorly and to the right, with the morphologic right atrium situated to the right and slightly posteriorly, and the morphologic left atrium to the left and slightly anteriorly[7].

The diagnosis of dextrocardia by foetal echocardiogram has estimated an overall incidence of between 0.22% and0.84% in pregnant women referred for pre-natal cardiac ultrasound. A large proportion of these foetuses have associated Coronary Heart Disease (CHD). The prevalence of dextrocardia in an adult population is unknown. With the advancement of new surgical techniques to correct previously fatal cardiac abnormalities, patients with dextrocardia and CHD surviving to birth are increasingly surviving into adulthood[8].

Dextrocardia with a normal abdominal situs is often associated with congenital cardiac anomalies take transposition of the great vessels and Atrial Septal Defects (ASDs) 6 and Ventricular Septal Defects (VSDs) 7 in 90 to 95% of cases, However, dextrocardia with situs inversus is associated with a lower incidence of congenital heart disease[9]. Associated Cardiac anomalies described in patients of dextrocardia with situs inversus are: VSDs, ASDs, complete AV canal defect, pulmonary atresia, TOF and double outlet right ventricle. Kulkarni and Inamdar reported a case of large peri membranous VSD associated with dextrocardia and situs inversus.[10]Situs inversus totalis may be associated with other congenital anomalies such as duodenal atresia, asplenium, multiple spleens, ectopic kidney, horseshoe kidney, and various pulmonary and vascular abnormalities[11]. Situs inversus totalis without other congenital anomalies have normal life expectancy. The recognition of Situs inversus totalis is also important for preventing surgical errors that result from the failure to recognize reversed anatomy. Cardiac interventions like transcatheter closure of Patent Ductus Arteriosus (PDA), ASD, VSD and procedures like Balloon Mitral Valvotomy (BMV) also pose orientation problems for the operator.

Conclusion

Situs inversus totalis remains a rare finding in adults, even in a highly select group of patients with known congenital heart disease. Complete diagnostic work up of suspected cases by various imaging modalities is required. Doctors should encourage routine physical examination which could help identify this anomaly, preventing wrong diagnosis and possibly death due to delay in management.

References

1 Perloff JK, Marelli AJ — Perloff's Clinical Recognition of Congenital Heart Disease, 6th Edition, Philadelphia, PA: Saunders: Elsevier; 2012, 15-30.
2 Casey B — Two rights make a wrong: human left–right malformations. *Human Molecular Genetics* 1998; 1565-71.
3 Maldjian PD, Saric M — Approach to Dextrocardia in Adults: Review. *AJR* 2007; 188: S39-49.
4 Olbrich H, Haffner K, Kispert A, Volkel A, Volz A, Sasmez G, *et al* — Mutation in DNAH5 causes primary dyskinesia and randomisation of left-right asymmetry. *Nat Gente* 2002; **30:** 143-4.
5 Layton WM Jr — Random determination of developmental process. *J Herred* 1976; **67(6):** 336-8.
6 Madan Kumar K, Babu C, Chander S, Kumar A, Balchander J, Nachipaan M — Complete A-V canal defect with dextrocardia with CCTGA – A case Report. IJTCVS. 2003; 19:55.
7 Lev M, Liberthson RR, Eckner FA — Pathologic anatomy of dextrocardia and its clinical implications.Circulation 1968; 37: 975–999. Ho SY, Mccarthy KP and Faletra FF. Anatomy of left atrium for interventional echocardiography. *Eur J Echocardiogr* 2011; **12:** i11-i15.
8 Bernasconi A, Azancot A, Simpson JM, Jones A, Sharland GK — Fetal dextrocardia: diagnosis and outcome in two tertiary centres. *Heart* 2005; **91(12):** 1590-4.
9 Piryani RM, Shukla A, Prasad DN, Kohli SC, Shrestha G, Singh D — Situs inversus with dextrocardia with multiple cardiac lesions in adult. *Kathmandu Univ Med J* 2007; **5(2):** 247-9.
10 Kulkarni PR, Inamdar VV — Situs inversus with dextrocardia associated with ventricular septal defect. Government medical college, Nanded. *J Anatom Soc India* 2005; **54(1):** 50(abstract)
11 Iino K, Watanabe G, Ishikawa N, Tomita S — Total endoscopic robotic atrial septal defect repair in a patient with dextrocardia and situs inversus totalis. *Interact CardioVasc Thorac Surg* 2012; **14(4):** 476-7.