

Situs Inversus Totalis - A Case Report

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Abstract: *SITUS INVERSUS TOTALIS* is a congenital positional anomaly characterized by transposition of abdominal viscera associated with a right sided heart (Dextrocardia). 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 when examined or visualized by tests such as x-ray filming.

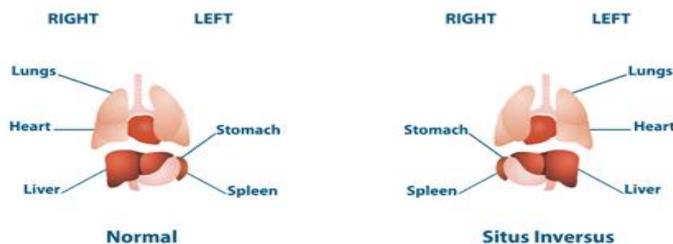
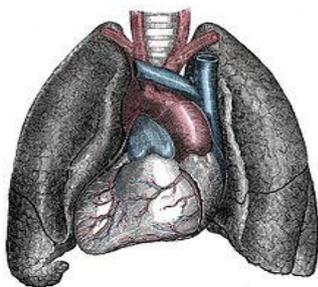
The term situs inversus is a short form of the Latin meaning inverted position of the internal organs. Generally individuals with situs inversus totalis are asymptomatic and have a normal life expectancy. Many people with situs inversus totalis are unaware of their unusual anatomy until they seek medical attention for an unrelated condition. The reversal of the organs may lead to some confusion as many signs and symptoms will be on the reverse side.

A 27 years old male patient reported to the Department of Nephrology with the c/o left sided flank pain since 1 week. The Chest X-ray, Ultrasonography, CT scan and MRI were done and he was diagnosed as **SITUS INVERSUS TOTALIS**.

The anatomic, pathologic, embryologic and etiology of complete Situs inversus and related abnormalities are presented in this case with special emphasis to genetic correlation.

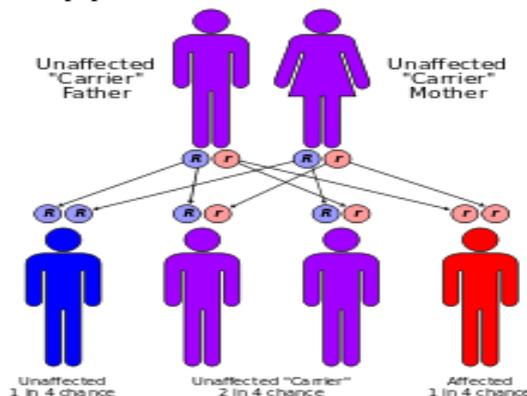
Key words: Situs inversus totalis, Dextrocardia, Congenital, Transposition, Thoracic and abdominal organs.

I. Introduction



Situs inversus causes the positions of the heart and lungs to be mirrored.

SITUS INVERSUS TOTALIS is also called Situs transversus or oppositus. It is a congenital condition in which the major visceral organs are reversed from their normal position. The normal arrangement is known as Situs Solitus. The other rare case is known as Situs ambiguus or heterotaxy, where in situs cannot be determined. Dextrocardia was first seen by **Leonardo da Vinci** in 1452-1519, and then recognized by **Marco Aurelio Severine** in 1643 and described more than a century later by **Matthew Baillie**. Matthew Baillie described the complete mirror-image reversal of the thoracic and abdominal organs in situs inversus. Situs inversus is present in 0.01% of the population.



Situs inversus is generally an autosomal recessive genetic condition, sometimes it can be X-linked and also found in identical twins.^[1, 2]

The condition affects all major structures within the thorax and abdomen; the organs are simply transposed through the sagittal plane. The heart is located on the right side of the thorax, the stomach and spleen on the right side of the abdomen and the liver and gallbladder on left side. The left lung is trilobed and right lung is bilobed. The blood vessels, nerves, lymphatics and intestines are also transposed. , The relationship between the organs is not changed, so functional problems rarely occur.

The **Situs inversus with Dextrocardia** or **Situs inversus totalis** has been estimated to occur once in about 6-8,000 live births. **Situs inversus with levocardia** or **situs inversus incompletes**.^[3] 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 thorax.

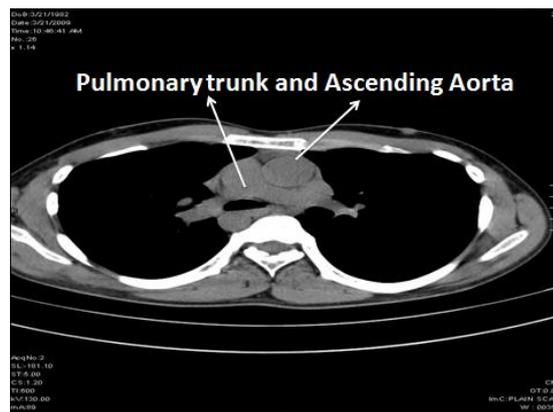
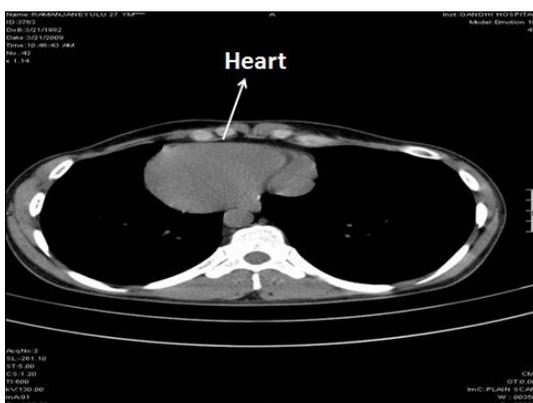
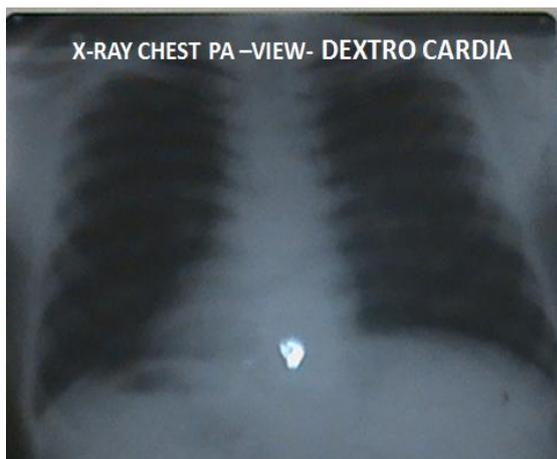
A Case Report

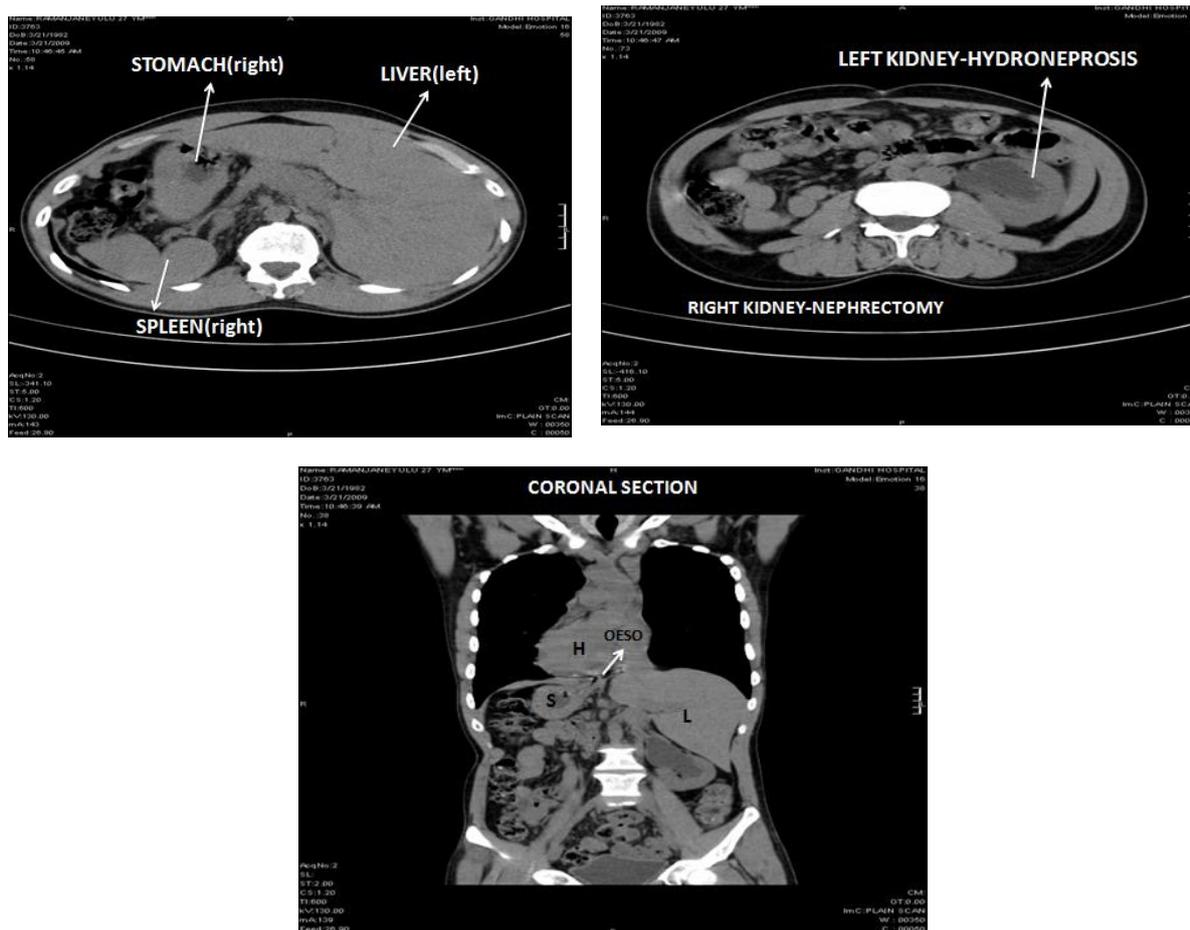
A 27 years old male patient reported to the Department of Nephrology with the c/o left sided flank pain since 1 week. He had no h/o of dysuria, oliguria, hematuria or UTI.

On general examination the patient was pale, no icterus or pedal edema. BP recorded was 130/ 90 mm Hg, Heart: S1, S2 are heard in **right** 5th intercostal space. Past h/o right sided nephrectomy .Known diabetic and hypertensive.

Investigations done

- Blood Sugar – 70 mg%
- Blood Urea- 150 mg%
- Serum Creatinine – 7.1 mg%
- X-ray- chest PA view – showed Dextrocardia
- X-ray KUB-Normal
- U/S Abdomen- showed left hydronephrosis ,Abdominal situs inversus
- CT SCAN Chest and Abdomen revealed Situs inversus totalis
- 2D Echo - Dextrocardia with normal sized cardiac chambers and normal LV function
- HIV – Non reactive
- HBS Ag – Negative





H: Heart; S: Stomach; L: Liver ; OESO: Oesophagus

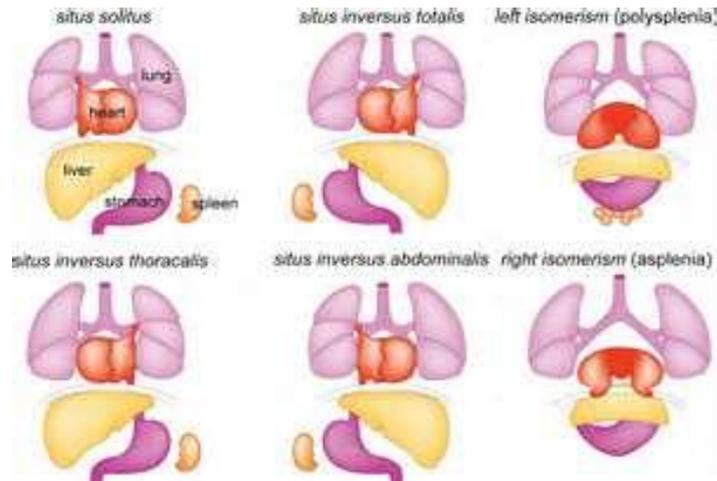
Diagnosis: Situs inversus with Dextrocardia with Chronic Kidney Disease, Chronic Renal Failure and on Dialysis once a week.

II. Discussion

Situs inversus totalis is a condition in which the organs of the chest and abdomen are arranged in perfect mirror image of normal. Complete situs inversus is a rare syndrome, of autosomal recessive inheritance. Distribution is the 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 the 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 the asymmetric viscera is conserved; it is known as Situs Solitus and when there is complete inversion of the lateralization of the organs (mirror image), it is known as Situs inversus totalis^(18,19,20,21)

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.**^(18,19,20,21) The individuals with situs inversus are phenotypically unimpaired, and can lead normal healthy lives, without any complications related to their medical condition. Many people with situs inversus totalis are unaware of their unusual anatomy until they seek medical attention for an unrelated condition.



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 with Dextrocardia is situs inversus totalis because of the cardiac position, as well as the atrial chambers and reversed abdominal viscera. [1, 2, 3, 4, 5, 6.]

When situs cannot be determined, the patient has situs ambiguus or heterotaxy. Which occurs in one in 20,000 births [Fuster, et al] (7,8,9). In these patients, with situs ambiguus, both stomach and liver move towards the center (midline), and the stomach develops behind the liver. In these patients the spleen may be absent or multiple and the bowel is malrotated.

There are two subtypes of **situs ambiguus** (10, 11)

1. Right isomerism or asplenia syndrome.
2. Left isomerism or polysplenia syndrome.

In Asplenia or right isomerism syndrome or bilateral right sidedness is associated with right atrial morphology i.e. both atria, liver are centrally located and both lungs have trilobed morphology. In case of Polysplenia or left isomerism syndrome or bilateral left sidedness is associated with bilateral spleen, left atrial morphology and both lungs are bilobed. In both the cases the apex of the heart can be located in either hemi thorax and also may be related to congenital cardiac anomalies.

Having no spleen can lead to risk of infections and polysplenia have the chance of rupture of spleen during accident causing internal bleeding.

Cardiac situs is determined by the location of the atria. In both Situs Solitus and Situs inversus the ventricles may present in two positions:

D-Loop or right sidedness i.e. right ventricle is anterior and to the right of left ventricle (normal). **L-Loop** or left sidedness, right ventricle is posterior and to the left of the 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 the right (17).

3% of people with situs inversus totalis have some form of congenital heart disease [Fuster, et al] (7,17)

About 25% of individuals with situs inversus have an underlying condition known as **primary ciliary dyskinesia (PCD)**. PCD is a dysfunction of the cilia that manifests itself during the development of fetus. Situs inversus with PCD together known as Kartagener syndrome characterized by the triad of situs inversus, chronic sinusitis, and bronchiectasis. (Wilhelm) (9,12,13,14,15,16)

The Situs inversus partialis involves a single viscus. Dextrocardia is the most common example of partial situs inversus. The abdominal organs, mainly the intestinal tract which develops from the midgut is chiefly involved in malposition, but for the foregut and hindgut is considered to be more stable and fixed in their positions. The error in location of the foregut situated within the abdomen i.e. the stomach and duodenum down to the biliary papilla is excessively rare.

The Max.A.Almy et.al reported a case of inversion of the stomach alone. The right-sided stomach behind the left lobe of liver was discovered during a cholecystectomy and later confirmed radiographically as Situs Inversus of Stomach. The majority of cases of right-sided stomach are associated with eventration of the diaphragm. (22)

The Structural and functional brain asymmetries are seen in human situs inversus totalis. The two major anatomic asymmetries of the cerebral hemispheres, the frontal and occipital petalia, are reversed in individuals with Situs Inversus.⁽²³⁾

Our patient was 27 years old male had Situs inversus totalis associated with chronic renal failure, right kidney nephrectomy and left kidney hydronephrosis. (Diagnosed by radiography). He was on dialysis once in a week. The prognostic factors could not be assessed in the present case, since the patient was referred to Higher Centre for management and was lost for further follow up.

III. Conclusion

The prevalence of situs inversus varies among different populations but is less than 1 in 10,000 people. Situs inversus totalis has number etiologic theories and it is thought that many factors may be involved in causing situs inversus. In human embryo of few millimeters, in median sagittal plane presents two sides which are mirror image of each other. As a result of an altered relationship between the embryo and chorion or environmental influences on the zygote, it can lead to asymmetrical development. Imaging studies such as MRI, CT or ultrasound may be advised in suspected diagnosis of situs inversus. Imaging studies will also rule out the possibility of situs ambiguous. Situs inversus also complicates **organ transplantation** operations as donor organs will almost certainly come from Situs Solitus donors.

References

- [1]. Yokoyama T, Copeland, Jenkins NA, Montgomery CA, Elder FF, Overbeek PA (1993). "Reversal of left- right asymmetry: a situs inversus mutation". *Science* 260 (5108): 679-682.
- [2]. Gedda L, Sciacca A, Brenci G, et al. (1984). "Situs viscerum specularis in monozygotic twins." *Acta Genet Med Gemellol (Roma)* 33 (1): 81-5. PMID 6540028.
- [3]. Gindes L, Hegesh J, Barkai G, Jacobson JM, Achiron R. Isolated levocardia: prenatal diagnosis, clinical importance, and literature review. *J Ultrasound Med.* Mar 2007;26(3):361-5.
- [4]. Fung TY, Chan DL, Leung TN, Leung TY, Lau TK. Dextrocardia in pregnancy: 20 years' experience. *J Reprod Med.* Jul 2006;51(7):573-7.
- [5]. Sirin BH, Kurdal AT, Iskesen I. Congenitally corrected transposition of the great arteries plus dextrocardia operated with an unusual operative technique. *Thorac Cardiovasc Surg.* Sep 2008;56(6):367-9.
- [6]. Hagler DJ, et al. Cardiac malpositions and abnormalities of atrial and visceral situs. In: Adams F.A. et al. eds. *Heart Disease in Infants, Children and Adolescents*. 5th ed. Baltimore:Williams and Wilkins 1995;1307-36
- [7]. Fuster, Valentin, Alexander, R. Wayne, O'Rourke, Robert, et. Al. *The Heart*. Chapter 81: Cardiovascular Diseases Caused by Genetic Abnormalities
- [8]. Wilhelm, Annamaria. "Situs Inversus." eMedicine. Dec. 5, 2007. <http://www.emedicine.com/radio/topic639.htm>
- [9]. "Dextrocardia with Situs Inversus." WebMD. Aug. 8, 2007. <http://www.webmd.com/heart-disease/dextrocardia-with-situs-inversus>
- [10]. Winer-Muram HT. Adult presentation of heterotaxic syndromes and related complexes. *J Thorac Imaging.* 1995;10(1):43-57. [Medline]
- [11]. Applegate KE, Goske MJ, Pierce G, Murphy D. Situs revisited: imaging of the heterotaxy syndrome. *Radiographics.* Jul-Aug 1999;19(4):837-52; discussion 853-4.
- [12]. Ferrero G.B. et al. A submicroscopic deletion in Xq26 associated with familial situs ambiguus. *Am J Hum Genet* 1997;61:395-401.
- [13]. Kinney TB, DeLuca SA. Kartagener's syndrome. *Am Fam Physician.* Jul 1991;44(1):133-4.
- [14]. Schidlow DV. Primary ciliary dyskinesia (the immotile cilia syndrome). *Ann Allergy.* Dec 1994;73(6):457-68; quiz 468-70.
- [15]. Yarnal JR, Golish JA, Ahmad M, Tomashefski JF. The immotile cilia syndrome: explanation for many a clinical mystery. *Postgrad Med.* Feb 1982;71(2):195-7, 200-2, 209-11 passim.
- [16]. Gierich J, et al. Primary ciliary dyskinesia in situs inversus without bronchiectasis. *Pneumologie* 1997;51:1127-32.
- [17]. Silverman NH. An ultrasonic approach to the diagnosis of cardiac situs, connections, and malpositions. *Cardiol Clin.* Aug 1983;1(3):473-86.
- [18]. Kosaki R. et al. Left-right axis malformations associated with mutations in ACVR2B, the gene for human activin receptor type IIB. *Am J Med Genet* 1999;82:70-6.
- [19]. Ryan A.K. et al. Pitx2 determines left-right asymmetry of internal organs in vertebrates. *Nature* 1998;394:545-51.
- [20]. Piedra M.E. et al. Pitx2 participates in the late phase of the pathway controlling left-right asymmetry. *Cell* 1998;94: 319-24.5043-8.
- [21]. Heymer J. et al. The expression pattern of nodal and lefty in the mouse mutant Ft suggests a function in the establishment of handedness. *Mech Dev* 1997;66:5-11
- [22]. Max A. Almy, M.D., Fred H. Volk, M.D. and Charles M. Graney, M.D. *Genesee Memorial Hospital Batavia, N. Y.* (Copy right 2013 by Radiological Society of North America.)
- [23]. Kennedy DN, O'Craven KM, Ticho BS, Goldstein AM, Makris N, Henson JW. *Neurology.* 1999 Oct 12; 53(6):1260-5.