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Heterotaxy Syndrome In A Middle-Aged Pakistani Male

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Abstract

Background: Heterotaxy syndrome (or situs ambiguous) is an extremely rare disorder in which the viscera are arranged in an abnormally asymmetrical pattern around the midline along with cardiac and spleen abnormalities. The management of this disorder depends upon the extent and variability of the organ involvement and a multi-disciplinary approach is often required.
Case presentation: A middle-aged male presented in pulmonology OPD with with complaint of hemoptysis and shortness of breath for 3 weeks. He had a history of cyanotic spells since childhood. His HRCT chest revealed dextrocardia, generalized mild centrilobular emphysema of the lungs with fibrotic bands and mild cylindrical bronchiectasis and his echocardiography showed dextrocardia along with ventricular septal defect and pulmonary hypertension. He was found to have abnormally positioned abdominal viscera along with multiple spleens and he was diagnosed as a case of heterotaxy syndrome with left isomerism. His respiratory symptoms were treated conservatively and the patient was referred to the cardiology unit for management of cardiac defects.
Conclusion: This manuscript describes a case of heterotaxy syndrome which is a rare disorder with significant mortality and morbidity. The patient may present with vague symptoms. Early involvement of all the relevant specialities might help in a prompt diagnosis and timely management, which may improve the disease outcome.

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1. Introduction

Heterotaxy syndrome (or situs ambiguous) is an extremely rare disorder in which the viscera are arranged in an abnormally asymmetrical pattern around the midline along with cardiac and spleen abnormalities [1]. While cardiac anomalies are most frequently described, there is a wide variability in the pattern of organ asymmetry reported in published literature[2]. Most of the cases are diagnosed in childhood and it is important to distinguish this disorder from situs inversus in which all the organs are displaced in a mirror-image pattern but the diagnosis is based on cardiac abnormalities and spleen defects[3]. The management of this disorder depends upon the extent and variability of the organ involvement and a multi-disciplinary approach is often required.

This manuscript describes a rare case of a middle-aged male with a history of cyanotic spells since childhood, who was diagnosed with a case of heterotaxy syndrome at our centre. This might be the first case of heterotaxy syndrome reported from the twin cities of Islamabad and Rawalpindi.

2. Case Presentation

A 41-year-old male presented in pulmonology OPD with with complaint of hemoptysis and shortness of breath (MRC grade-3) for 3 weeks. He was married with two children (no history of assisted fertilization) and was a non-smoker. He gave no history of fever or weight loss but he had been prescribed antituberculous drug treatment twice for the same complaints in the past based on history and chest X-ray findings. The patient had a history of cyanotic spells since childhood but no proper investigation was done at that time.

On examination, the patient had grade 4 clubbing of fingernails, peripheral cyanosis and coarse crepitations on chest examination. The apex beat was located on the right 5th intercostal space with palpable thrill and loud P2. His chest X-ray revealed an increased cardiothoracic ratio with an opacity in the right middle and lower zone (figure 1). His initial laboratory investigations revealed polycythemia with hematocrit of 68% and type 1 respiratory failure on arterial blood gas analysis. His sputum for gene Xpert was negative and his HRCT chest revealed dextrocardia, generalized mild centrilobular emphysema of the lungs with fibrotic bands mild cylindrical bronchiectasis and features of pulmonary arterial hypertension. CT scan was also indicative of abnormal liver positioning and polysplenia (figure 2).

His 2D-ECHO showed dextrocardia, situs inversus, large inlet ventricular septal defect and severe pulmonary hypertension with Eisenmenger syndrome. His ultrasound abdomen was done which revealed an abnormal arrangement of the liver and spleen around the midline as well as polysplenia. Based on his clinical history, examination and radiological findings, he was diagnosed as a case of heterotaxy syndrome with left isomerism. His respiratory symptoms were treated conservatively, two sessions of venesection were done and the patient was referred to the cardiology unit for further management of cardiac defects.

5. Discussion

The word heterotaxy is derived from the Greek word heteros meaning different and taxis meaning arrangement. It is a rare birth defect in which the chest and abdominal organs are abnormally arranged around the central axis. It is distinct from situs inversus in which the arrangement of organs in the chest and abdomen is mirror imaged[1].

Heterotaxy syndrome was first reported by the Swedish paediatrician Ivermark in 1955. Its prevalence is estimated to be 1 in 10,000 people worldwide accounting for 3 % of all congenital heart defects[4]. Many genes are implicated in the pathogenesis of this disorder including Pitx2, Lefty, Cited2, and Sonic Hedgehog genes but the exact cause of the syndrome still eludes the scientists[5].

Heterotaxy syndrome can be divided into left isomerism and right isomerism based on defects observed in organs. Right, isomerism presents in infancy and represents a poorer prognosis due to the presence of a severe form of cyanotic heart disease6. Left isomerism usually presents with multiple spleens and bilaterally bilobed lungs while spleen may be absent in patients with right isomerism. Commonly involved organs are the heart, stomach, spleen, intestines, bronchial tree and lungs. Cardiac anomalies include tetralogy of Fallot, transposition of great vessels and atrial or ventricular septal defects. Asplenia and polysplenia may occur but the spleen is usually hypo-functional with an increased risk of infections from capsulated organisms[6,7].

The syndrome is diagnosed in childhood if severe cardiac defects are present. It is typically diagnosed by imaging of internal organs and echocardiography is used for the detection of cardiac defects[8]. Treatment depends upon specific organs affected in each individual. For those diagnosed in infancy, surgery may be needed to correct cardiac defects and in cases of intestinal malrotation. Regular vaccinations against encapsulated microorganisms may be carried out in patients with splenic problems to prevent recurrent infections. Hence, a multidisciplinary approach is required for timely diagnosis and interventions to reduce the significant morbidity and mortality associated with this disorder.

5. Conclusion

Heterotaxy syndrome is a rare disorder with significant mortality and morbidity. The patient may present with vague symptoms and clinical findings may be confusing. Early involvement of all the relevant specialities might help in making a prompt diagnosis and timely management of the disease-related issues may improve the outcome.

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M.S - Experimentation/Study Conduction
A.T.D - Analysis/Interpretation/Discussion
S.K, A.T.D - Manuscript Writing
M.S, F.K, A.T.D - Critical Review
F.K, A.T.D - Facilitation and Material analysis

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