Case study

Eisenmenger Syndrome with Dextrocardia, Ectopic Left Kidney and Right sided Infantile Scoliosis: A Rare Case Report

Key words: Cyanosis, Dextrocardia, Eisenmenger, Heterotoxy, Hypoxemia

ABSTRACT:

Eisenmenger syndrome refers to any untreated congenital cardiac defect with intracardiac communication that leads to pulmonary hypertension, reversal of flow, and cyanosis. The previous left-to-right shunt is converted into a right-to-left shunt secondary to elevated pulmonary artery pressures and associated pulmonary vascular disease. Young male presented progressive dyspnoea, chest pain and anasarca and having cyanosis, dextrocardia, right scoliosis and ectopic kidney. A favourable prognosis for ES is achieved with early diagnosis and surgical intervention, whereas a poor prognosis is achieved with a late diagnosis and the onset of heart failure and pulmonary hypertension. Most patients die from heart failure, cardiac arrhythmia, and thromboembolic cerebrovascular disease. Patients with ventricular septal defects may benefit from taking medications like sildenafil and furosemide to improve their prognosis and quality of life. The longevity of these patients with functional limitations is still grim, despite therapy advancements. Further therapeutic interventions need to be made to reduce the symptoms.

INTRODUCTION

A rare birth disorder, heterotaxy syndrome, also called Isomerism, with a global incidence of 1 in 10,000, is associated with at least 3% of congenital cardiac abnormalities [1]. Visceral malposition and dismorphism are two signs of heterotaxy syndrome (within both the thorax and abdomen). Heterotaxy syndrome is defined by severe cardiovascular malformations. The majority of individuals with the heterotopia syndrome die naturally from difficulties as children. Visceral malposition and dismorphism are also characteristics of heterotaxy syndrome (within both the thorax and abdomen). Complex cardiovascular abnormalities are a feature of heterotaxy syndrome. Heart failure, abrupt cardiac death, and intrapulmonary bleeding are among the sequelae of extensive congenital heart abnormalities that most people with the heterotaxy syndrome experience before they reach adulthood [2].

Eisenmenger Syndrome (ES) is not a congenital heart disease. It is an entirely preventable, multisystem dis
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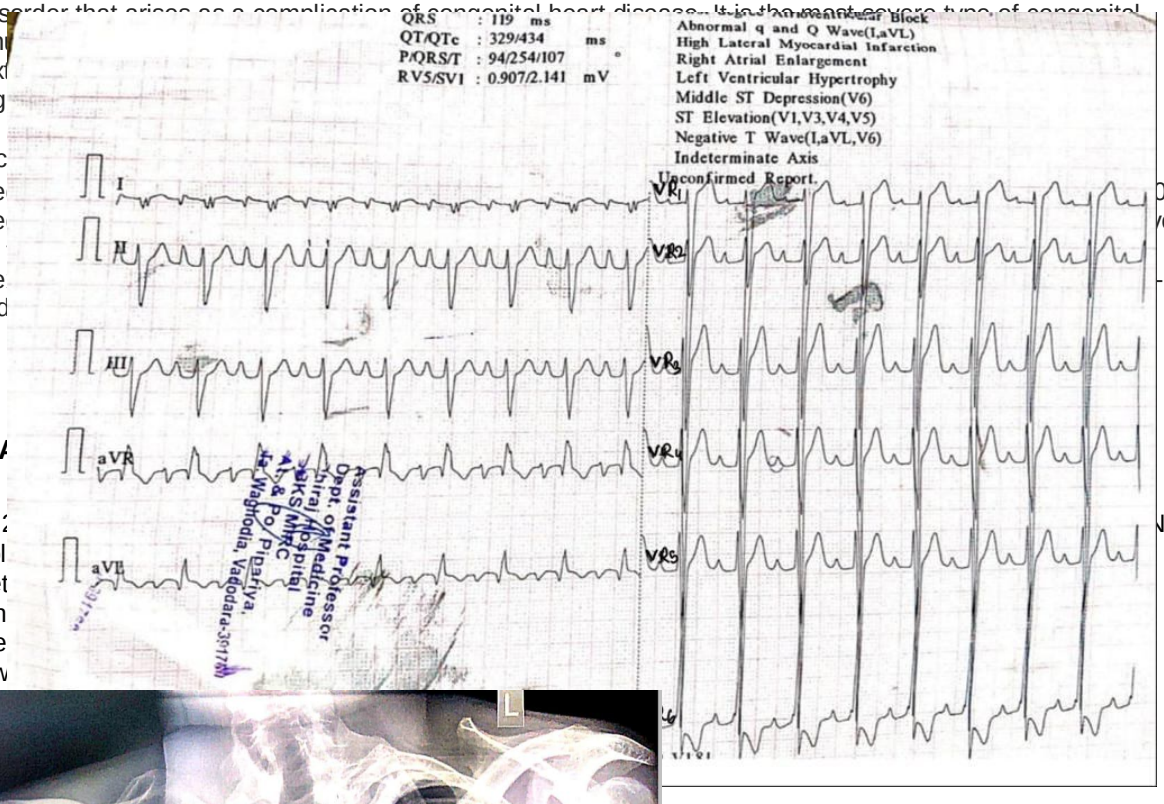

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c QRS complexes, LV strain pattern

On examination, the patient was poorly built and malnourished with BMI-14.67 kg/$m^{2.}$ cyanosis of tongue, lips and all the digits and grade 2 clubbing. His pulse was 104/minute, regular but low volume and blood pressure was 84/62 mmHg. His respiratory rate was 26/ minute with oxygen saturation of 76% on room air. JVP was elevated. His cardiovascular examination revealed that he had Right sided bulging asymmetric chest wall with apical impulse present in Right 6th intercostal space in mid-axillary line, 11 cms away from sternal border. Parasternal heave was present . On auscultation Loud S2 was heard in pulmonary area with no murmurs appreciated. His lungs had bilateral fine basal crepitations. His abdominal examination revealed hepatomegaly with tenderness hypochondriac and epigastric region. Back showed drooping of shoulder of right side with scoliosis. Routine investigation showed erythrocytosis with hemoglobin 18.9 g/dl (12-15 g/dl), packed cell volume was 57.5%, platelets 1,50,000 cells/cu.mm, Mean Corpuscular Volume (MCV) 88.8 fl , Mean
Corpuscular Hemoglobin (MCH) 29.2 pg/cell, creatinine was 1.2 mg/dl and urea 91 mg/dl, Bleeding time 1 minute 10 seconds and activated clotting time 3 minutes 25 seconds. Uric Acid 13.8 mg/dl. Peripheral smear examination showed Normocytic Normochromic blood picture with Reticulocyte count of 3%. ECG revealed signs of both Left Ventricular Hypertrophy (LVH) and Right Ventricular Hypertrophy (RVH), Large Biphasic QRS complexes in V2V5, with LV strain pattern in V6, Right Atrial Enlargement

Echocardiography report showed SitusSolitus and Dextrocardia, with Criss-cross AV connection and L-looping of ventricle. Large upper muscular Ventricular Septal Defect extending upto sub-aosric region with Severe Pulmonary Arterial Hypertension and Bi-ventricular dysfunction with LVEF 20-25% and Dilated IVC. USG Abdomen showed Rt kidney appears malrotated 9.8x4.4cm the hila is seen to be facing supero-medially with unremarkable hila/focal lesion or collection, left kidney is not visualized in left renal fossa?absent?ectopic however it's location /position in abdomen can't be ruled out due to excessive bowel gas.

Further evauqtion with CT KUB is recommended.CT-KUB (IMAGE 3) revealed left kidney was small in size measuring 6.5x 4.5cm visualised in left pelvic cavity suggestive of ectopic left kidney. HRCT- thorax report showed that Dextrocardia, Cardiomegaly, Prominent mean pulmonary artery measuring 30mm and Ground Glass Opacities with peribronchial and interlobular septal thickening noted in right middle and lower and left lower lobes along with Scoliotic deformity.

With evidence obtained from above investigations, patient is known to have Dextrocardia (IMAGE 1), Congenital heart disease- ES, Ventricular septal defect with PAH and ectopic left kidney in left pelvic cavity. Patient in hospital for 10 days and during his stay he was given Inj
Amoxicillin+ clavulinic acid (1.2gm) TDS for 5 days, Tab Digoxin (0.25) once daily,
Tab furosemide (10) 1/2 tab once daily, Tab Ecosprin (75) once daily, Tab Folic Acid
(5) once daily, Tab Febuxostat (40) once daily, Tab Calcium and Tab Vitamin D3 Patient had undergone phlebotomy 4 times after which improvement of cyanosis as well as well as orthopnea and PND and significant fall in hemoglobin levels from 18.9 g/dl to 15.6 g/dl.

DISCUSSION

heterotaxy syndrome might result from
gene mutations. Clinically, there are twotypes:
unique as patient presented with dextrocardia
sized kidney and right sided scoliosis. Variable
ss multiple organ systems. Dysrhythmia, heart
ne main cardiac complications. Extra-cardiac
rhosis/ hepatocellular carcinoma, renal
ertility [6].

n disease that develops as a result of a significant
onary vasculature and causes supra-systemic
en PVR exceeds SVR, which causes cyanosis.
s and young children who have extensive, un-
DA. If left untreated, Eisenmenger syndrome is
can also occur in people with complex intra-
rs may play a role in Eisenmengersyndrome,

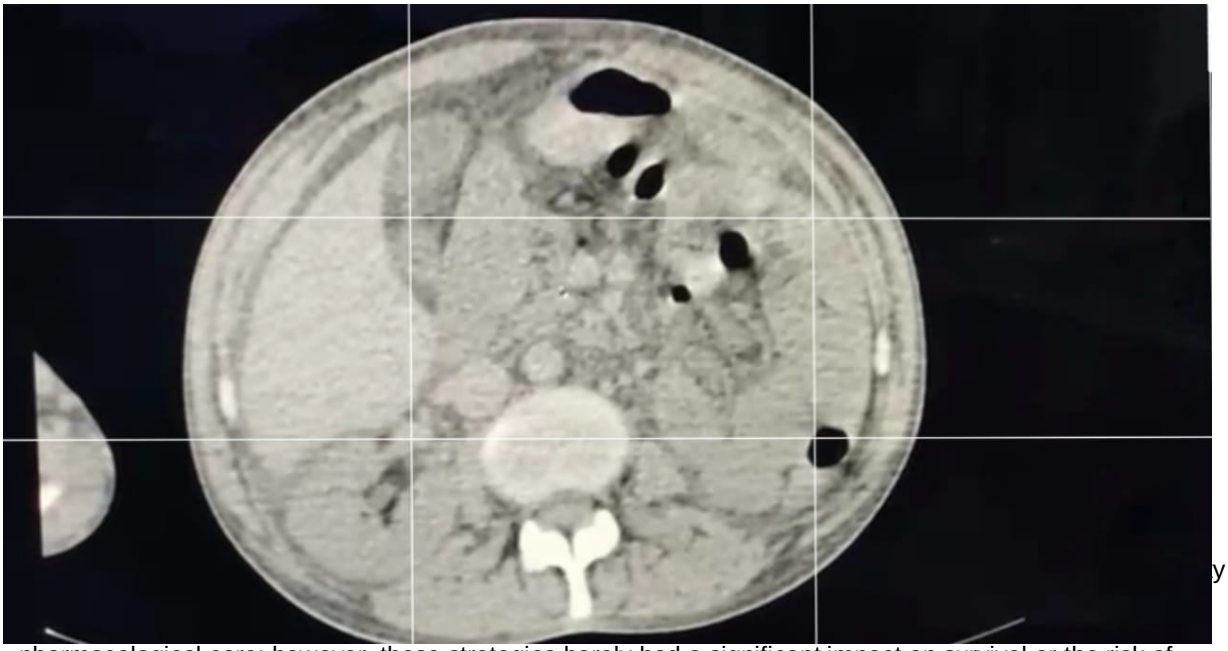

IMAGE 4: CT-KUB suggestive of Right kidney in Normal shape and position while Left Kidney appearing small in size and in Left Pelvic cavity suggestive of Ectopic Left Kidney.

pharmacological care; however, these strategies barely had a significant impact on survival or the risk of disability. Cardiopulmonary transplantation, which is impractical in most contexts, can treat ES. Eisenmenger Syndrome has historically been treated with warfarin to prevent clotting. In older patients, surgical correction of the underlying heart defect is primarily unsuitable. [12] When sildenafil and furosemide are administered together, cGMP elevation-mediated cochlear toxicity may result in hearing loss with sensorineural origin. The reason that the index patient experienced hearing improvement after stopping sildenafil is indicative that this hearing loss is reversible. Hence, if at all co - administration of ototoxic drugs such loop diuretics/CYP3A4 inhibitors, and PDE5 inhibitors is required, careful prudence and monitoring are indicated.

Individuals with advanced symptoms of ES who are resistant to medical treatment may be eligible for lung or heart transplantation along with correction of the cardiac defect. In terms of outcomes, ES is comparable to other cases where combined heart-lung transplantation is being used. 63 patients who underwent heart-lung or lung transplantation for ES were identified in a recent international survey. Early mortality was 11%, and 15 years after transplantation, survival was 41%. [13]

The severity of the underlying cardiac anomalies significantly affects the heterodoxy syndrome prognosis. Although these patients are undergoing reducing surgical and medical treatment, whose long-term outlook is still not favourable. [6] Eisenmenger patients have a variable prognosis, but it is better when compared to individuals with much more severe forms of PAH. Mortality in childhood is unusual, but it becomes significantly more prevalent in the fourth or later decades of life. [14]

CONCLUSION

A favourable prognosis for ES is achieved with early diagnosis and surgical intervention, whereas a poor prognosis is achieved with a late diagnosis and the onset of heart failure and pulmonary hypertension. Most patients die from heart failure, cardiac arrhythmia, and thromboembolic cerebrovascular disease. Patients with ventricular septal defects may benefit from taking medications like sildenafil and furosemide to improve their prognosis and quality of life. The longevity of these patients with functional limitations is still grim, despite therapy advancements. Further therapeutic interventions need to be made in order to reduce the symptoms. This case illustrates Heterodoxy syndrome with Dextrocardia and ES with right sided scoliosis with Left ectopic kidney. This case is very crucial among physicians for complex and multi organ involvement. Hence, through investigations and management is required for treating such complex case.

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