



A Rare Case of Heterotaxy Syndrome: Eisenmenger Syndrome with Dextrocardia in an Adult

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Authors' contributions

This work was carried out in collaboration by all authors. Author HL performed data collection and literature search, collaborated with author GT on the initial manuscript draft and wrote the subsequent revisions. Author GT assisted with literature search and assisted with the first draft of the manuscript. Authors AM, MN and TM assisted with manuscript revisions. All authors read and approved the final manuscript.

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Case Study

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ABSTRACT

Aims: Heterotaxy syndrome is a rare, complex, and confusing presentation in the realm of situs anomalies. The concurrence of heterotaxy syndrome with dextrocardia and Eisenmenger syndrome has never been reported in an adult. We described, for the first time, an adult patient who had heterotaxy syndrome with dextrocardia, complicated by Eisenmenger syndrome with large ventricular septal defect (VSD).

Presentation of Case: A 43 year-old female presented to the emergency room with worsening exertional dyspnea. She carried past medical history of cyanotic congenital heart disease since birth. Her physical examination findings were significant for hypoxia and clubbing with cyanotic fingers and toes, dextrocardia, and a 3/6-holosystolic murmur over the right lower sternal border. Laboratory investigation showed polycythemia and her chest x-ray showed dextrocardia. Echocardiography showed dextrocardia with a dilated left ventricle and a large VSD with left-to-right shunting. Computed tomography (CT) angiogram of the chest showed bilateral bi-lobed lungs with both main stem hyparterial bronchi, right-sided polysplenia, right-

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sided stomach with left-sided dominant liver.

Discussion and Conclusion: Heterotaxy syndrome has a complex variation in clinic presentation. We illustrate the occurrence of heterotaxy syndrome, polysplenia type with Eisenmenger syndrome and dextrocardia. Recognition of this rare anomaly is important for establishing a diagnosis in a patient with multiple organ displacement.

Keywords: Heterotaxy syndrome; Eisenmenger syndrome; dextrocardia; polysplenia.

1. INTRODUCTION

Heterotaxy syndrome is rare with an incidence of 1:10,000 worldwide and is associated with at least 3% of cases of congenital heart defects [1]. Patients with heterotaxy syndrome are subdivided into three subtypes according to the characteristic morphology of the spleen: asplenia, single-right-spleen, and polysplenia. The asplenia subtype of heterotaxy syndrome represents a right-sided isomerism where organs that are normally present on the right side of the body are present bilaterally coupled with a glaring absence of left-sided organs [2]. The single-right-sided spleen subtype of heterotaxy syndrome is very rare and therefore has not been studied adequately [3]. The polysplenia subtype of heterotaxy syndrome represents a left-sided isomerism where organs that are normally present on the left side of the body are present bilaterally with an absence of the right-sided organs. This subtype of heterotaxy syndrome also usually presents with bilateral bilobed lungs and hyparterial bronchi [4]. Heterotaxy syndrome presents with some degree of visceral malposition and dimorphism (within both thorax and abdomen). The cardiovascular malformations in heterotaxy syndrome are complex. Most patients with the heterotaxy syndrome die in childhood from complications of their complex congenital heart defects [3], including heart failure, sudden cardiac death, and intrapulmonary hemorrhage [5]. Approximately 10% of patients with polysplenia may reach adulthood without any complications [3].

Eisenmenger syndrome can be reversible (stages I-III) or irreversible (stages IV-VI), in which it is a fatal disease eventually causing decompensation and death despite vigilant treatment [6]. It has never been reported that heterotaxy syndrome, polysplenia subtype with dextrocardia and Eisenmenger syndrome are concurrent in an adult. The objective of this article is to describe, for the first time, an adult patient who had heterotaxy syndrome with dextrocardia, polysplenia subtype, complicated by Eisenmenger syndrome with large ventricular septal defect (VSD).

2. CASE PRESENTATION

A 43 year-old female presented to the emergency room with worsening exertional dyspnea. She was known to have significant cyanotic congenital heart diseases since birth. Due to limited access to health care in her native country, she was unable to have appropriate cardiology evaluation despite persistent exertional dyspnea. She denied tobacco, alcohol or illicit drug use. She denied any family history regarding congenital heart disease and was on no medication. On admission, the patient's pulse oxygen saturation was 76% on room air and improved to 80% with 100% Fraction of Inspired Oxygen (FiO₂) via a non-rebreather mask. The patient's blood pressure was 130/70 mmHg, pulse rate 92 per minute, respiratory rate 18 per minute and body temperature 98 degrees Fahrenheit. On physical examination, her extremities exhibited obvious clubbing and cyanosis (Figs. 1A and B). She was noted to have jugular venous distention and diffuse wheezes with bilateral basal rales on lung auscultation. The cardiac examination showed that the apex of the heart was on the right side of the chest, displaced 3 cm to the right of the midclavicular line; a 3/6-holosystolic murmur was noted on the right lower sternal border. Her standard electrocardiogram (ECG) (using left-sided chest leads) showed inverted P waves in limb leads and prominent R waves in V1-3 (Fig. 1C). Laboratory investigations were as follows: Hemoglobin – 20.9 g/dL, Hematocrit – 65.8%, WBC-count – 5.5 k/ μ L, Platelets – 70 k/ μ L, Sodium – 138 mmol/L, Potassium – 3.8 mmol/L, Chloride – 104 mmol/L, CO₂ – 23 mmol/L, BUN – 16 mg/dL, Creatinine – 0.7 mg/dL, Glucose – 84 mg/dL. BNP was 1306 pg/mL (normal range: <450 pg/mL in those <50 years of age).

Her chest x-ray showed dextrocardia with evidence of cardiomegaly and mild pulmonary vascular congestion (Fig. 2A). Her echocardiogram showed dextrocardia with a dilated left ventricle and a large VSD with left-to-right shunting (Fig. 2B). A computed

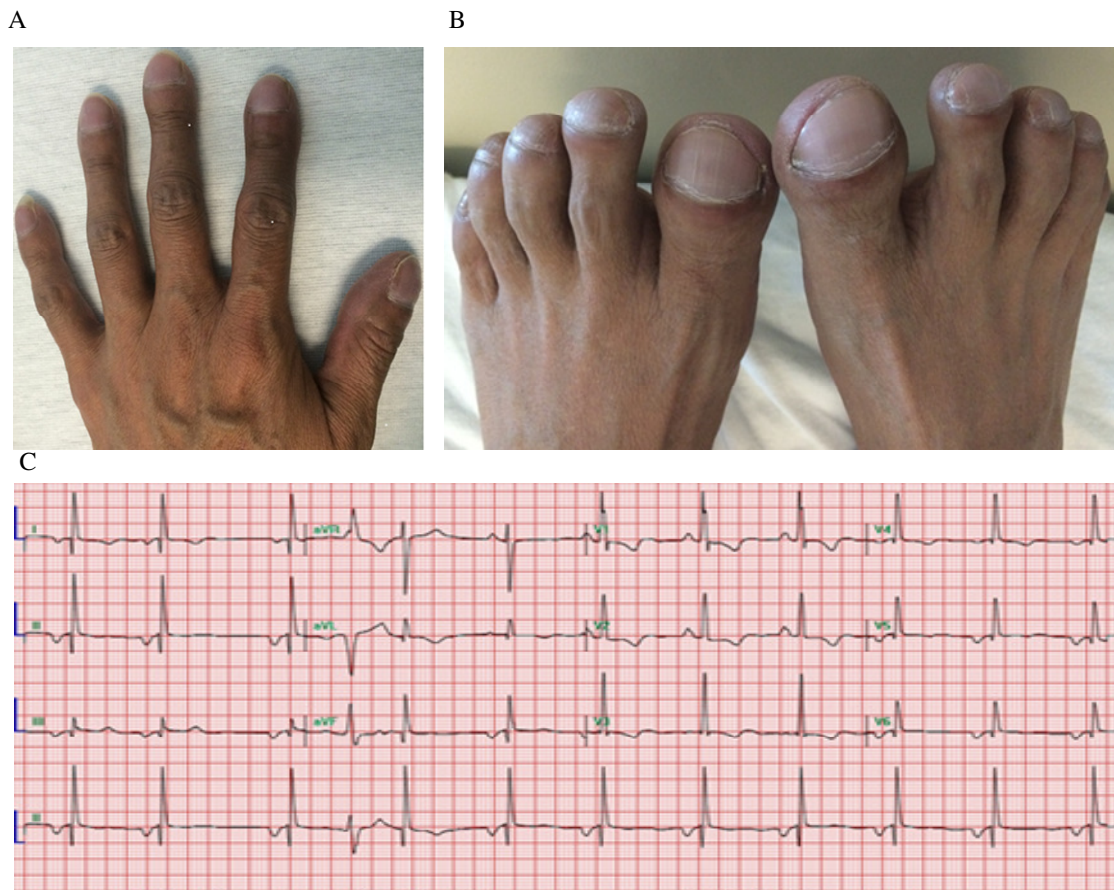


Fig. 1. View of clubbing in the patient's fingers (A) and toes (B), as well as illustration of dextrocardia in her standard electrocardiogram (ECG) (C) (using left-sided chest leads) with inverted P waves in limb leads and prominent R waves in V1-3

tomography (CT) angiogram of the chest showed both lungs to be bi-lobed with both main stem hyperarterial bronchi. It also showed dextrocardia, cardiomegaly with marked dilatation of right and left coronary arteries, left-sided dominant liver, and right-sided gastric bubble with evidence of right-sided polysplenia (Figs. 2C and D). The patient was subsequently diagnosed with heterotaxy syndrome, polysplenia-subtype, dextrocardia and Eisenmenger syndrome. She was treated with supportive care, remained stable and was discharged home awaiting surgical correction of her large VSD.

3. DISCUSSION

Heterotaxy syndrome, a complex variation in clinical presentation, occurs as result of genetic

mutations or mechanical malrotation during lateralization. It clinically presents as three subgroups as described above. Our case is unique as the patient presented with bilateral bi-lobed lungs, dextrocardia with VSD, right-sided polysplenia, left-sided stomach and left-sided dominant liver.

While such mutations are responsible for the isolated versus the combined events, it is the development of the underlying congenital abnormalities that decide disease presentation and provide each patient with their own unique variation [7]. In an autopsy study of 146 patients with polysplenia, 56% of the subjects had abdominal heterotaxia, 55% had bilateral bi-lobed lungs and at least half of the patients had cardiac anomalies [7].

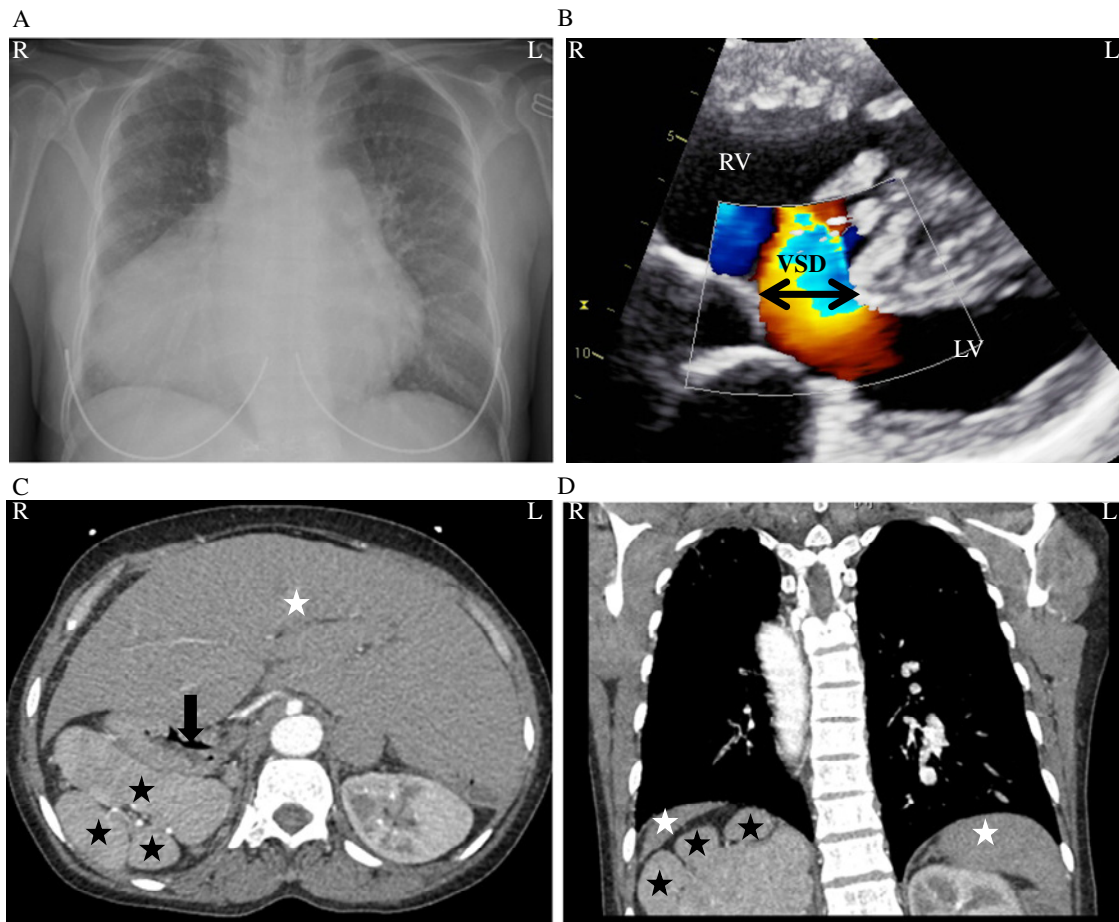


Fig. 2. Illustrations of the patient's chest x-ray (A), echocardiogram (B) and CT-angiogram (C and D)

Two-head black arrow indicates a large VSD with left-to-right shunting. White star and black star indicate the liver and spleen, respectively. The black arrow indicates agastric bubble. R: right; L: left.

These malrotations can be isolated as dextrocardia or situs inversus; they can also present in conjunction with each other as dextrocardia with situs inversus[8]. In addition, at least 12% of patients with Primary Ciliary Dyskinesia present with heterotaxy syndrome [1]. However, it is quite rare that a patient presents with a combination of situs ambiguous (heterotaxy syndrome), right-sided polysplenia type and dextrocardia with a large VSD ultimately leading to Eisenmenger syndrome. In our case, the untreated VSD induced left-to-right shunt in dextrocardia, which is responsible for the hypoxia, clubbing fingers and polycythemia vera.

Heterotaxy syndrome causes variable complications in multiple organ systems. Main cardiac complications include dysrhythmia, heart failure, thromboembolism and infectious

endocarditis; extra-cardiac complications include plastic bronchitis, liver dysfunction/ cirrhosis/ hepatocellular carcinoma, renal dysfunction, diabetic mellitus, mental disease and subfertility in female [4]. The prognosis of heterotaxy syndrome largely depends on the severity of the associated cardiac anomalies. Although medical and surgical treatments are advanced, the long-term prognosis of these patients is still unsatisfactory. Right isomerism has an overall 5-year survival ranging from 30% to 74% due to the worst forms of congenital heart disease; left isomerism, on the other hand, is improved with 5-year survival rates ranging between 65% and 84% [4].

4. CONCLUSION

This case illustrates the very rare adult occurrence of heterotaxy syndrome, polysplenia

type, with Eisenmenger syndrome and dextrocardia. It is crucial for clinicians to recognize this complex anomaly and conduct a thorough investigation for associated multiple organ abnormalities in the heterotaxy syndrome.

CONSENT

Not applicable.

ETHICAL APPROVAL

The authors hereby also declare that all examinations and interventions have been examined and approved by the ethics committee of Raritan Bay Medical Center and have therefore been performed in accordance with the ethical standards laid down in the 1964 Declaration of Helsinki.

COMPETING INTERESTS

Authors have declared that no competing interests exist.

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