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Case Report

Late diagnosis of Ebstein anomaly after pregnancy: A rare case report

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ABSTRACT

Congenital heart disease is a concerning disease among those affected since it affects the patient's quality of life. One of which is Ebstein Anomaly (EA), with a prevalence of 1 in 200,0000 births with a clinical characteristic of displacement of the tricuspid valve that causes atrialization of the right-side heart. Women of childbearing age are at risk because they especially have asymptomatic EA, compromised to hemodynamic consequences, and risk of maternal complications. A 25-year-old woman visited the emergency department with worsening shortness of breath since last week, during rest, accompanied by decreased physical activity tolerance, bilateral ankle edema, cyanosis to the lips, and nail bed. She had echocardiography and cardiac multi-slice computed tomography to identify EA before and after her first child's birth. She was well controlled with a loop diuretic, phosphodiesterase type 5 inhibitor, and beta-blocker but could not care for her child due to her condition. The patient was discharged after symptoms subsided, awaiting further interventional evaluation. Although EA is rare, it has high morbidity and mortality, especially in women of childbearing age since it may be asymptomatic during childhood. Therefore, early recognition of EA is probably necessary for women who are planning pregnancy.

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INTRODUCTION

There is a risk of malformation during fetal development, and congenital heart disease (CHD) is the most common form (Jairo, 2018). From all prevalence of CHD, Ebstein anomaly (EA) is one of the rarest cases (Abramian et al., 2011). EA is cyanotic congenital heart disease of the tricuspid valve and is mainly caused by malformation of the Atrioventricular valve leading to a large atrial chamber in contrast to the ventricle of the right side. This anomaly is expected in 1 of every 200,000 births (Kanoh et al., 2018; Sharma et al., 2018; Sharma et al., 2020). Characterization of the disease is commonly recognized with clinical findings of tricuspid leaflet displacement, another additional finding such as atrial septal defect (ASD) or a patent foramen ovale (PFO), and abnormal conduction system presented with an accessory pathway (Kanoh et al., 2018; N. Sharma et al., 2018). Patient varies in clinical presentation depending on anatomic and hemodynamic factors depending on right heart function and severity of tricuspid regurgitation (TR) (Kanoh et al., 2018).

Patients with EA have a higher risk of mortality. Evidence-based European Surveillance of Congenital Anomalies (EUROCAT) acquired from 12 European countries for 29 years showed that 19 % of babies died before birth (Boyle et al., 2017). Although the number of mortality at birth and in neonates continues to rise, the survival rate of patients with congenital heart disease into adulthood is also increasing, and it is termed Adult with Congenital Heart Disease (ACHD) (Kwag et al., 2018). One of the main reasons for this to happen is that these patients did not feel any symptoms until adulthood and often been diagnosed during routine medical checkups or sometimes become symptomatic ranging from mild to severe, especially during pregnancy, as this case represents (Harris et al., 2020; Kwag et al., 2018). Due to late diagnosis during pregnancy, women with ACHD may experience severe maternal complications that may have a fatal outcome for the mother and the child (Kanoh et al., 2018).

Since this is a complex disease with a sinister outcome and rarely early diagnosis since it is rare to encounter in daily practice, we present this case of a young post-partum woman who sought medical attention at our health facility with congestive heart failure symptoms accompanied by cyanotic mucosal appearing and the history of EA that just been diagnosed since her third semester of pregnancy. This case presentation aims to review the diagnosis and prompt treatment of Ebstein anomaly with a late presenter.

CASE REPORT

A 25-year-old P1A0 female came to Siloam Diagram Heart Hospital and presented with a complaint of increasing dyspnea that was also felt during rest after delivering her first child. She was accompanied by decreased physical activity tolerance for the last week. She also noted bilateral pretibial edema and cyanotic appearance of the lips and nails. Her past medical record was unremarkable before pregnancy, but she remembered that she fainted during childhood during excessive exercise. She had delivered one child last year and was diagnosed with EA during pregnancy when she first complained of difficulty breathing when lying and bilateral ankle edema.

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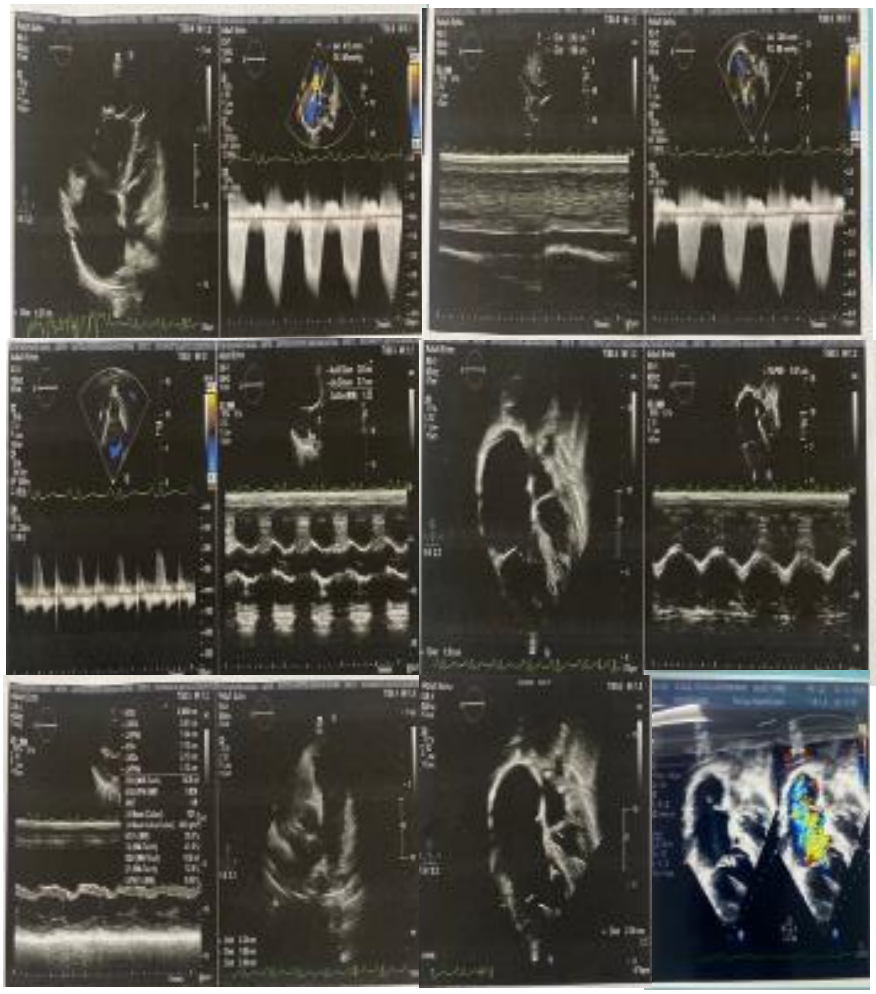

Figure 1. TE image showing large right atrial chamber with the displacement of the tricuspid valve, severe tricuspid regurgitation, normal RV function, and high probability of pulmonary hypertension. The Doppler showed a severe tricuspid regurgitation.

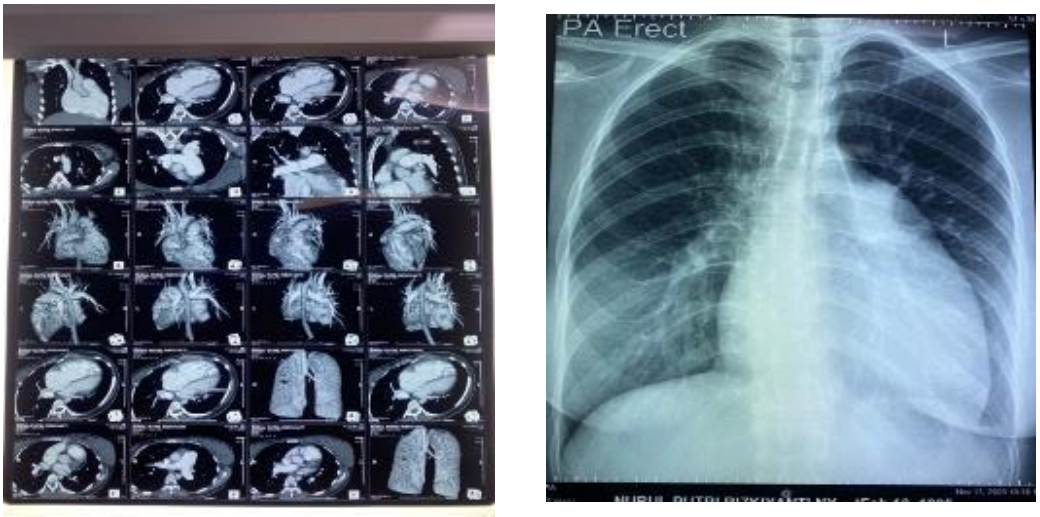

Figure 2. Cardiac Magnetic Resonance (left), Rontgen Thorax Posterior-Anterior (right)

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One year ago, she was evaluated with a non-invasive imaging modality, such as echocardiography, and further evaluated with cardiac multi-slice computed tomography (MSCT) after delivery two months ago (**Figure 1**). She gave birth by a cesarian due to her condition, and her symptoms are controlled by diuretics, beta-blockers, and PDE-5 inhibitors after delivery. Despite good medical compliance, she is limited to childcare and relies on her spouse. On admission, the patient had difficulty breathing and was pale and weak. On examination, obtained as follows: blood pressure 160/121 mmHg, heart rate 105 bpm, respiratory rate 24 rpm, SpO_2: 82% without oxygen supplement. JVP was elevated, bilateral crackles at both lung bases, 5/6 holosystolic murmur at LLSB with S3 gallop, and bilateral pretibial edema. The blood test was within the standard limit, and routine MSCT Thorax showed mild cardiomegaly with bilateral lung congestion (**Figure 2**). The patient was optimized with IV diuretics and MRA in addition to routine medication during hospitalization and discharged three days later. The clinical approach in EA diagnosis was done with echocardiography and CT-Cardiac before and after delivery.

The patient was hospitalized until decongestion was sufficient to alleviate symptoms and was discharged three days later after symptoms were relieved. She is also advised to consult for cardiac intervention for right heart pressure evaluation and more advanced treatment options.

DISCUSSION

The epidemiology of congenital heart disease in Asia has recently increased compared to other continents (van der Linde et al., 2011). Based on global data on CHD prevalence in metanalyses, the study found that there were 1,161,030 CHD cases between 1970 and 2017. This evidence also shows that Asia has the most significant number of CHD at birth, with 9,342 cases per 1000 population, which is higher than in other regions, such as Europe and the United States of America (Liu et al., 2019). Nevertheless, the exact cause of this high incidence of CHD is still unknown, but previous data showed that only 20% of the CHD incidence is related to genetic syndromes, teratogen exposure, or maternal diabetes (Blue et al., 2012). From all the data regarding CHD, EA is categorized as a rare case compared to other types (Sainathan et al., 2020). Supporting the claim, the prevalence of EA to date recorded up to 0.534% and an incidence of up to 0.044 cases per thousand births with CHD (Liu et al., 2019). EA patients are more prevalent in females than males from those births (Wu et al., 2018). This may lead to cases where females who had EA and survived until adulthood (ACHD) will be exposed to morbidity and mortality risk, especially during pregnancy, which may burden the heart workload. Pregnancy physiologic adaptation, including hemodynamic and hormonal changes, may worsen this disease outcome (Elkayam et al., 2016b; Elkayam et al., 2016a). These changes also increase the risk of arrhythmias, heart failure, and thromboembolism, which puts ACHD patients at a higher mortality risk than pregnant females without EA (Adam, 2017; Lammers et al., 2021; Ramage et al., 2019).

EA is categorized as a cyanotic congenital heart disease that features an abnormal displacement of the tricuspid valve toward the apex and an abnormally formed tricuspid valve causing regurgitation. In our case, the patient had severe tricuspid regurgitation. These anomalies are also associated with a shunt at the atrium septal wall (ASD Secundum or PFO) and a concealed accessory pathway; hence, the latter will make the patient prone to medical conditions, such as arrhythmias. The consequence of various

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degrees of hypertrophy and thinning walls during embryogenic development (Kanoh et al., 2018; Sharma et al., 2018; Susilowati, 2020). A prior study showed that EA could be diagnosed by ultrasound as early as 14 weeks of gestation but may not be detected until adulthood since echocardiography is not a routine check-up in a healthy adult (Boyle et al., 2017). In our case, the patient did not check with a cardiologist because she did not know.

This unclear etiology and unclear symptoms make it hard to diagnose this disease early. Until today, most modalities used in diagnosing are still considered essential in diagnosing EA after it is severe enough to cause deterioration (Yuan, 2017). Chest X-ray, Echocardiography, and cardiac imaging using CT or MRI are the main modalities for assessing this disease (Qureshi, 2018). The gold standard among these modalities is echocardiography since it is non-invasive, relatively quick, and does not need a stable patient. The main finding in echocardiography is an apical displacement of the TV septal leaflet or posterior of $\geq$ 8 mm/m^2 with an elongated anterior leaflet (Qureshi, 2018). Cardiac CT-scan or CMR is also essential to supplement the findings in echocardiography to detect PFO, which is usually hard to find in the previously mentioned modality (Baumgartner et al., 2021). In our case, the patient had CXR, TEE, and CMR so that we could conclude the diagnosis.

Physiologic hemodynamic changes affect the cardiovascular system workload, for instance, increasing plasma volume by up to 30-50% (Kanoh et al., 2018; Sharma et al., 2018; Katsuragi et al., 2013; Sanghavi & Rutherford, 2014). Therefore, these changes increase preload, worsening tricuspid regurgitation, and increasing right atrium pressure. Increased preload will cause the right ventricle to compress the left ventricle, impairing left ventricular diastolic and systolic function. As a result, both right and left side hearts are affected. On the other hand, there is also a gradual increase in catecholamine levels during late pregnancy that causes an increase in stroke volume and heart rate up to approximately 30-50% due to increased maternal and fetal demand. The catecholamine increase is related to cardiac arrhythmias (Kanoh et al., 2018; Sharma et al., 2018; Christ et al., 2014). In our patient, congestive heart failure is caused by severe regurgitation of the tricuspid valve and marked cardiomegaly, worsening the right heart failure condition after delivery.

Despite complications in pregnancy, these patients also face hemodynamic changes during delivery and the postpartum period. P asma volume increases during labor due to uterine contractions and Valsalva during vaginal delivery. T e Valsalva also predisposes to right-to-left shunts in patients with EA who have ASD or PFO previously (Chopra et al., 2010). The latter complication of atrial transient reversal shunt has a 14% risk of paradoxical embolus (Hakman & Cowling, 2022). In the postpartum, the increased plasma volume may persist due to autotransfusion from uterine involution and edema resorption (Bishop et al., 2018). She was one year after delivery, and the symptoms of heart failure worsened.

The treatment for EA is surgical or conservative. R pair of the TV lesion is preferred whenever indication criteria are fulfilled and feasible. However, the timing of surgery remains challenging and should be done by a congenital surgeon with specific experience. Patients with EA who are not surgery candidates but also have developed heart failure are treated with standard heart failure treatment, including diuretics to decrease preload and digoxin (used to enhance muscle contractility and anti-arrhythmia). Symptomatic rhythm disorder can be treated conservatively; however, it is preferred by

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electrophysiologic interventions. The use of oral anticoagulant is recommended if there is a history of paradoxical embolism or atrial fibrillation (AF) and may be considered if there is an increase in thromboembolic risk or right to left (R-L) shunt through PFO (Baumgartner et al., 2021). We decided to manage the heart failure condition first; then, we recommended the right heart catheterization and surgery.

CONCLUSION

EA is a cyanotic congenital cardiac condition affecting the tricuspid valve that is mostly brought on by a malformed atrioventricular valve, which results in a large atrial chamber as opposed to a right-side ventricle. In particular, women of reproductive age with asymptomatic EA are at risk due to hemodynamic implications and the possibility of maternal and after-delivery problems. Surgery or conservative measures are used to treat EA. When possible and the indication conditions are met, repair of the TV lesion is preferable. Even though EA is uncommon, it has a significant morbidity and death rate, particularly in women of reproductive age, since it may not show symptoms until later in life. Therefore, it is likely vital for women hoping to become pregnant to recognize EA early.

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