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A Twins with Congenital Heart Disease: A Case Report

Mirza Syafaryuni[1]; Andi Alief Utama Armyn[1,2]; Yulius Patimang[1,2]; Muzakkir Amir[1,2]; Idar Mappangara[1,2]
[1]Department of Cardiology and Vascular Medicine, Faculty of Medicine, Universitas Hasanuddin, Makassar 90245, Indonesia
[2]Dr. Wahidin Sudirohusodo National General Hospital, Makassar, Indonesia

Abstract:-

- ***Background:***

Twin pregnancies have a higher risk of congenital heart defects compared to singleton pregnancies. The most commonly found congenital heart defect in both monochorionic and dichorionic twin gestations is ventricular septal defect (VSD). Approximately 10% of individuals with congenital heart defects will develop pulmonary arterial hypertension-congenital heart disease (PH-CHD).

- ***Case Presentation:***

We diagnosed an 11-year-old twin girl with ventricular septal defect. First Patient had a severe PH with mean Pulmonary Artery Pressure (mPAP) 68 and a defect size of 9-12 mm, while second patient had a mild PH with mPAP 30 and a defect size of 6-9 mm. The patient presented with failure to thrive, exercise intolerance during daily activities, and a history of recurrent respiratory tract infections. Echocardiography and right heart catheterization (RHC) results showed that the first patient with VSD Left to Right Shunt with low flow high resistance, non-reactive oxygen test and pulmonary hypertension. The second patient with VSD Let to Right shunt. Both of the patients were diagnosed with ventricular septal defect (VSD) and pulmonary hypertension (PH). However, they received different treatments based on the RHC results.

- ***Conclusion:***

Promptly addressing the long-term implications of CHD is essential to prevent pulmonary hypertension (PH) and systemic disorders. Advanced PH treatments are safe and can result in improved hemodynamic, exercise capacity, quality of life, growth and development and potentially survival.

Keywords:- *Congenital Heart Disease, Ventricle Septal Disease, Twins, Pulmonary Arterial Hypertension, Right Heart Catheterization.*

I. INTRODUCTION

In twin pregnancies, the risk of congenital heart defects is generally higher than in singleton pregnancies. The occurrence of congenital heart defects in singleton pregnancies is around 1.8% to 2%, whereas in twin pregnancies, it varies from 1.7% to 7.5%. Additionally, monochorionic twins have a higher prevalence of congenital heart defects compared to dichorionic twins. (Gijtenbeek & Haak, 2020; Nashat et al., 2017; Springer et al., 2014)

The most commonly observed congenital heart defect in both monochorionic and dichorionic twin gestations is ventricular septal defect (VSD) (Manning & Archer, 2006). The estimated global incidence of pulmonary arterial hypertension associated with congenital heart defects (PH-CHD) is that 10% of individuals with CHD will develop PH. The occurrence of PH in congenital heart disease (CHD) depends on the size and location of the defect, as well as the type of intervention performed. The therapeutic approach for PH-CHD not only focuses on treating vascular disease but also addresses the comorbidities associated with congenital defects. Cyanosis in Eisenmenger syndrome is a comorbidity that has multiorgan effects (Nashat et al., 2017). Early detection of congenital heart disease can prevent the development of systemic complications. Unfortunately, most patients with congenital heart disease (CHD) do not receive an early diagnosis unless they are already showing symptoms. (Jone et al., 2023).

II. CASE ILLUSTRATION

Two twins were brought to the outpatient clinic because their parents were concerned about their failure to thrive, which had been noticeable since they were 4 years old. The twins did not show signs of shortness of breath or cyanosis, but they both had a history of experiencing shortness of breath during daily activities and recurring respiratory tract infections. The twins were born prematurely at 7 months gestation, and there is no family history of congenital disease. Upon physical examination, both children had normal vital signs. The first child weighed 16 kg, while the second child weighed 24 kg.

The clinical examination showed that the twins had normal hemodynamic parameters and their oxygen saturation was 97-99% on room air. The first twins had clubbed fingers, and auscultation detected a grade 4/6 holosystolic harsh murmur was detected during auscultation at the left lower sternal border radiated to apex. The chest X-rays showed cardiomegaly and a left-to-right shunt (figure 1). The second twins no clubbing finger was found, and auscultation detected a grade 4/6 holosystolic harsh murmur was detected during auscultation at the left lower sternal border radiated to apex. The chest X-rays showed cardiomegaly and a left-to-right shunt (Figure 2). Echocardiography examination on first

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patient showed a Perimembran septal defect of ventricle with size 9.8 mm (Figure 3) and the second patient size of VSD 6.8 mm (figure 4).

The Right Heart Catheterization (RHC) results of the first patient with mean Pulmonary Arterial Pressure (mPAP) 68, PARi 12.6 Wood unit, and PVR/SVR 0.8 underwent an oxygen test for 10 minutes with 10 liters of oxygen and RHC results of mPAP 68, PARi 10.3 Wood unit and PVR/SVR 0.82 (Figure 5). It was concluded that the patient had VSD Left to Right shunt with low flow, high resistance, pulmonary hypertension, and non-reactive oxygen test, then an oxygen test was carried out so that the first patient is advised to optimize PH therapy for 6-12 months and will be re-evaluated (Figure 6)

The second patient underwent RHC with results of mPAP 30, PARi 1.96 WU, PVR/SVR 0.25. It was concluded that the VSD findings in this patient had a VSD inlet left to right shunt with high flow low resistance and were planned for VSD device closure (figure 9 and figure 10). The results of LV-graphy showed a VSD with a size of LV side 9.6 mm and RV side 6.3 mm. The procedure was continued using the KONAR-MF Occluder LE-MFO 12-10 with good results (figure 9). Echocardiography was evaluated six hours after the procedure with an in-situ device and minimal residual shunt.

III. DISCUSSION

Congenital Heart Disease (CHD) refers to structural abnormalities of the heart or intrathoracic vessels that occur during fetal development and have functional implications. There has long been a suspicion that genetic defects play a role in the occurrence of these malformations (Springer et al., 2014). Abnormal heart structures that develop during embryogenesis lead to congenital heart defects. The heart, which is the first organ to develop in the embryo, plays a crucial role in supporting rapid growth and eventually forms four chambers. This intricate process of morphogenesis is crucial for the development of the heart and has an impact on blood flow. The presence of congenital heart defects during fetal development affects the circulation and hemodynamics of the fetus (Vijayalakshmi et al., 2013) (Kobayashi et al., 1998)

Cardiac catheterization can have a significant impact on the prognosis of individuals with congenital heart disease. Up to 28% of patients with congenital heart disease may develop pulmonary hypertension, and 12% may develop Eisenmenger syndrome. Echocardiography results in patients showed a 6.1% prevalence of atrial septal defect (ASD) or ventricular septal defect (VSD), which can cause pulmonary hypertension. PH-specific therapy has demonstrated long-term benefits in improving patient outcomes. Guideline-based therapy, including the use of sildenafil and bosentan, can be administered. (Chiu et al., 2022)

Base on the weight of the first patient, she was experiencing failure to thrive based on her age, and the first patient was more frequently affected by recurrent respiratory tract infection than her siblings. This in line with the clinical presentation of pulmonary hypertension symptom and sign: low cardiac output such as poor appetite, failure to thrive, lethargy, diaphoresis, tachypnoea, tachycardia, and irritability. After early childhood, children present with similar symptoms as adult. In older children, the most common symptom is exertional dyspnoea and occasionally chest pain. Frequently, RRTI in PH patients is caused by more reactive pulmonary blood vessels, which can later result in ventilation/perfusion mismatches due to alveolar hypoxia, worsening if not treated aggressively. (Widlitz & Barst, 2003).

Pulmonary hypertension in congenital heart defects occurs due to various pathophysiological factors that cause an increase in pulmonary vascular resistance (PVR), and ultimately malignant arrhythmias, right heart failure and premature death. As excessive pulmonary flow, due to a nonrestrictive left- to-right shunt, is often implicated in the development of PH in this group (Nashat et al., 2017). The World Symposium on Pulmonary Hypertension states that pulmonary hypertension (PH) is physiologically linked to congenital heart disease and biventricular heart disease. The results of cardiac catheterization showed that the mean pulmonary artery pressure was above 25 mmHg, and the pulmonary vascular resistance exceeded 3 Wood Units. According to the echocardiography criteria set by the European Society of Cardiology, if the peak tricuspid regurgitation velocity exceeds 3.5 m/s (with a pressure gradient above 46 mmHg) or if the tricuspid regurgitation velocity is above 2.8 m/s (with a pressure gradient above 31 mmHg), and there are indications of right heart pressure overload(Chiu et al., 2022).

Currently, we are looking at hemodynamics based on RHC data and vasoreactivity tests to predict patient outcomes after closure. Further management of VSD patients with pulmonary hypertension is based on hemodynamic criteria obtained from RHC results based on PVR and pulmonary to systemic resistance ratio, with the following criteria: a. Baseline PVR index < 6 Woods units/m2 is associated with a resistance ratio < 0.3 without vasoreactivity testing, which is interpreted to indicate a good outcome after surgery resulting in biventricular circulation; b. An acute vasoreactivity test using oxygen/nitric oxide is recommended if a baseline PVR index is found between 6 and 9 Wood units/m2 with a resistance ratio of around 0.3-0.5. Although there is no absolute consensus, the results of surgery are considered successful if an evaluation results in a decrease in the PVR index of 20%, a decrease of approximately 20% in the ratio of pulmonary and systemic vascular resistance, a final PVR index < 6 Wood units/m2, a final resistance ratio <0.3. (Beghetti & Tissot, 2010; Dimopoulos et al., 2014)

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In this case, both patients were diagnosed with the same condition but required different treatment approaches. Both patients had recurring respiratory tract infections, but one of them was underweight. Echocardiography and RHC examinations confirmed the presence of pulmonary hypertension. In First patients with negative AVT, this requires optimization of pulmonary hypertension therapy. In contrast to the second patient, from the RHC results, it was decided to close the VSD with a device.

The management algorithm PH in patients with PH and congenital heart disease (CHD) incorporates the findings from right heart catheterization (figure 10). If the results of acute vasoreactivity testing (AVT) were positive, a device was used to perform VSD closure. For patients with negative AVT results, PH therapy was administered for the next 6 months, followed by a repeated RHC. Patients with a left-to-right shunt may experience an increase in pulmonary arterial pressure (PAP) and pulmonary blood flow (Qp). However, if the mean pulmonary arterial pressure (mPAP) or QP remains within the normal range, or if there is only a slight increase in pulmonary vascular resistance (PVR), closure of the shunt may be an option (Nashat et al., 2017). The 5th World Health Organization (WHO) and ESC meeting decided to take a conservative approach to CHD. These factors, in addition to guidelines, include defect type, age, the pulmonary-to-systemic vascular resistance (PVR: SVR) ratio, and the total pulmonary-to-systemic flow (Qp: Qs) ratio.

Based on the PH therapy algorithm in congenital heart disease, patients who have been catheterized are evaluated to see PARi and PVR / SVR. In the first patient, PARi values > 8 and PVR / SVR > 0.5, and vasoreactivity tests were carried out with negative results. Based on the chart above, patients are carried out to optimize PH therapy. In contrast to the second patient, PARi values were obtained < 6 and PVR / SVR < 0.3 so that it continued for VSD closure. (Nashat et al., 2017; Pan et al., 2011).

➢ *Evidence Attachment*

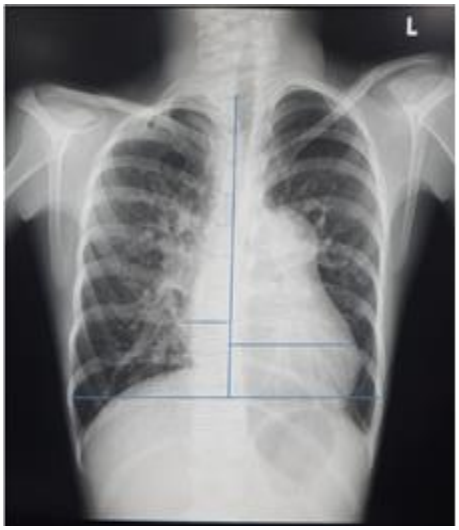

Fig 1 Chest X-ray 1st Patient

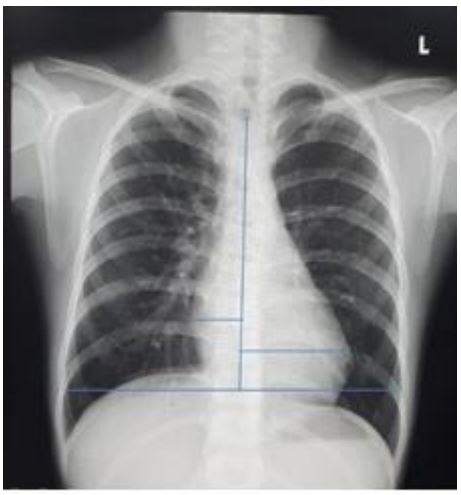

Fig 2 Chest X-ray 2nd Patient

Fig 3 Echocardiography 1st Patient

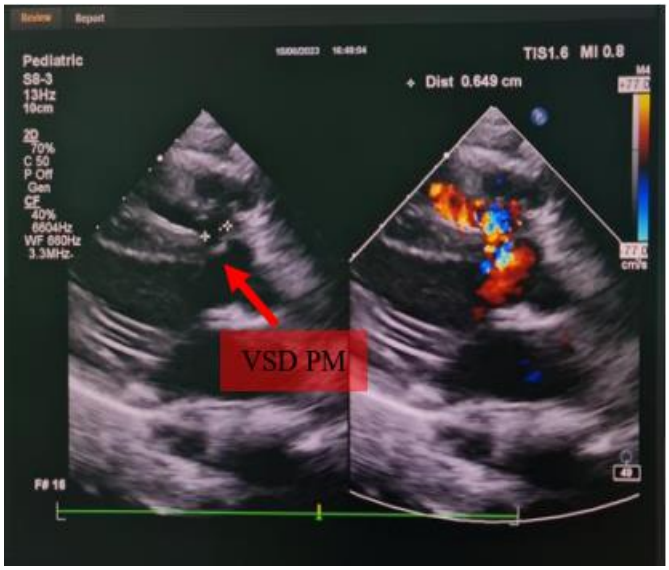

Fig 4 Echocardiography 2nd Patient

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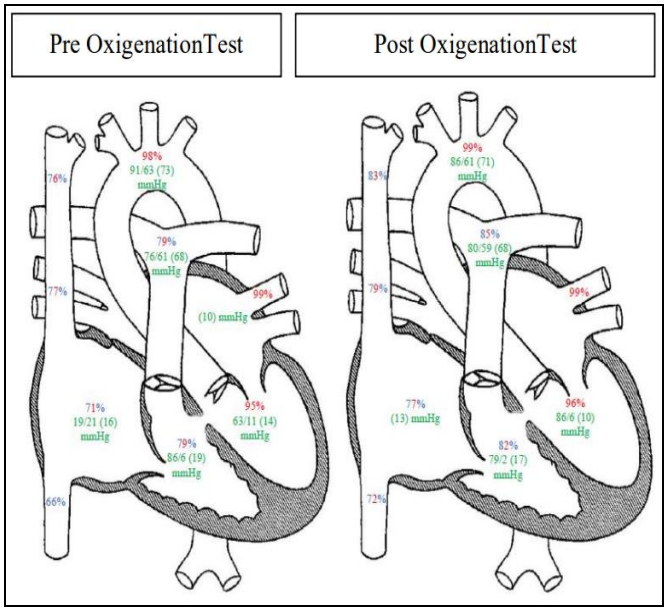

Fig 5 Right Heart Catheterization 1st Patient

Table 1 Right Heart Catheterization 1st Patient 's Report

Right Heart Catheterization:

	Pre Oxygen Test		Post Oxygen Test	
	Tekanan (mmHg)	**Saturasi (%)**	**Tekanan (mmHg)**	**Saturasi (%)**
AoD	91/63 (73)	98	86/61 (71)	99
RA	19/21 (16)	71	20/19 (15)	77
IVC		66		72
HSVC		76		83
LSVC		77		79
PV		99		99
RV	86/6 (19)	79	79/2 (17)	82
PA	76/61 (68)	79	80/59 (68)	85
LV	90/11 (37)	95	86/6 (10)	96

Conclusion:
VSD Perimembran Bidirectional Shunt
Low Flow, High Resistance
Pulmonary Hypertension
Non Reactive oxygen Test

Suggestion:
Optimalization of PH treatment

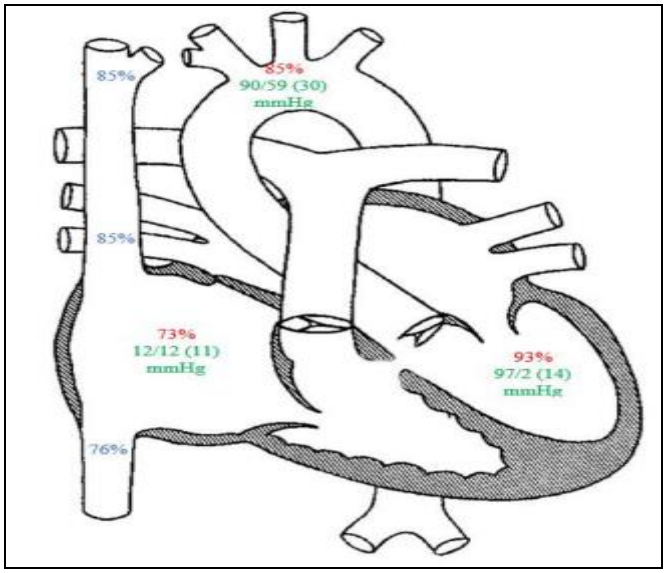

Fig 6 Right Heart Catheterization 2nd Patient

Table 2 Right Heart Catheterization 2nd Patient's Report

Right Heart Catheterization:

	Pressure (mmHg)	**Saturation (%)**
RA	12/12 (11)	73
IVC		76
HSVC		85
LSVC		85
PA	40/20 (30)	85
AoD	90/59 (72)	85
LV	97/2 (14)	93

Conclusion:
Ventricle Septal Defect Inlet Left to Right Shunt
High Flow, Low Resistent
Pulmonary Hypertension
Plan:
Ventricular Septal Defect Device Closure

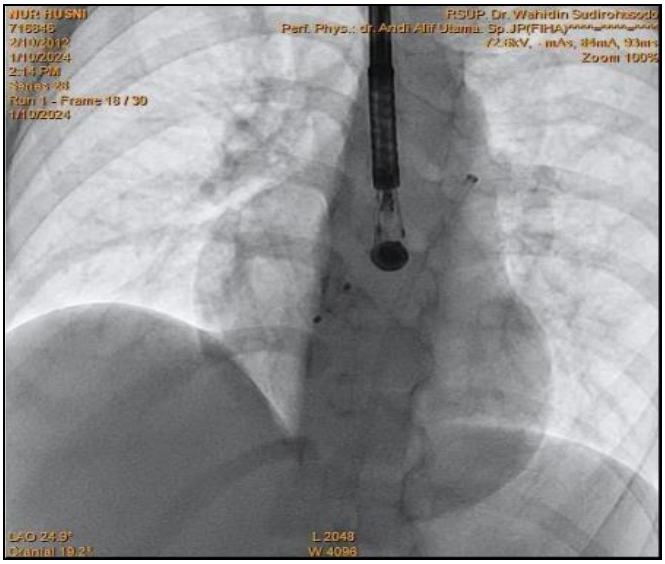

Fig 7 Post VSD Device Closure

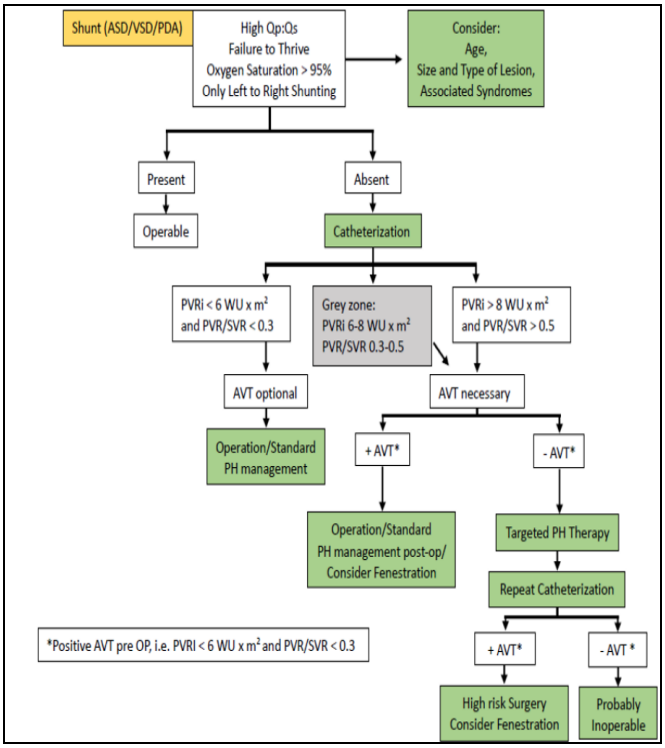

Fig 8 Algorithm Management of PH in PH-CHD Patient

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IV. CONCLUSION

Congenital heart disease (CHD) is more common in twin pregnancies than in singleton pregnancies. It is essential to promptly address the long-term implications of CHD in order to prevent pulmonary hypertension (PH) and systemic disorders. Moderately-sized VSD is associated with PH that may progress to severe irreversible pulmonary vascular disease (Eisenmenger syndrome). The Children with monochorionic twins had an incidental of congenital heart disease, and in this case the patients have the same congenital heart disease but different treatment. There is evidence suggesting that advanced PH treatments are safe and result in improved hemodynamics, exercise capacity, quality of life, and potentially survival.

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