Successful Live Saving Management Critical CHD By Early Diagnosed Fetal Echocardiography

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ABSTRACT:
Fetal echocardiography is a vital tool for identifying and assessing congenital heart disease (CHD), which accounts for 1% of live births globally. It offers significant prenatal and postnatal care benefits, potentially saving lives in severe cases, specially in critical a critical congenital heart disease (CCHD) cases. Early fetal echocardiography, conducted between 18-22 weeks of gestation, is recommended for fetuses with ductus venosus flow abnormality, increased nuchal translucency, or other CHD-like indicators. Early diagnosis allows for timely treatment and a better understanding of CHD effects. However, its use in developing countries remains limited due to high costs and lack of integrated health services. Prenatal diagnosis improves outcomes and survival rates of severe CHDs. However, there are limitations in implementing fetal echocardiography for early diagnosis, such as lower detection rates of cardiac abnormalities on screening ultrasound and difficulty obtaining complete fetal echocardiography views during early gestation. In this report, we present a case of a pregnancy with the result of fetal chocardiography were critical pulmonary stenosis, reduced flow from the right ventricle to the pulmonary artery, an open patent ductus arteriosus (PDA), and a patent foramen ovale (PFO) that was effectively managed in the infant after birth with prostaglandin infus and percutaneous transluminal balloon valvuloplasty (PTBV).

INTRODUCTION

Congenital heart disease (CHD) is are highly prevalent congenital malformations, constituting approximately 1% of live births globally.[1] The utilization of fetal echocardiography for the timely identification and assessment of congenital heart disease (CHD) offers significant benefits in terms of enhanced prenatal and postnatal care, hence potentially saving lives in instances of severe CHD.[1,2]

Fetal echocardiography has been used to identify CHD since the 1980s. Over time, improvements in ultrasound technology have facilitated enhanced precision and comprehensiveness in assessing fetal cardiac structure and function.[3] According to current guidelines, fetal echocardiography is advised for pregnancies deemed high-risk, such as those with a family history of CHD, extracardiac anomalies, monochorionic twins, or indications of CHD on a regular ultrasound examination.[4] Early fetal echocardiography, conducted during the gestational period of 18-22 weeks, is recommended explicitly for fetuses with ductus venosus flow abnormality, increased nuchal translucency, or other indicators that suggest an increased likelihood of CHD.[4,5]

The primary advantage of early fetal echocardiography is its capacity to identify critical CHD as early as 16 weeks into gestation. This facilitates the provision of scheduled treatment at a specialized medical facility equipped with pediatric cardiology and cardiac surgery resources, which is of utmost importance for newborns needing prompt postnatal medical intervention.[6,7] Additionally, early diagnosis gives families more time to comprehend the effects of CHD and decide on pregnancy management and newborn care.[8] However, the use of early fetal

echocardiography in developing countries is still limited due to the high cost and lack of integrated health services.[9] This case report presents successful life-saving management by utilizing fetal echocardiography in a developing country.

CASE DESCRIPTION

As an obstetrician and gynecologist advised, Mrs. S. is undergoing a fetal echocardiography test. The test revealed that the infant presented with right ventricular hypertrophy, suspected pulmonary atresia, diminished flow from the right ventricle to the pulmonary artery, critical valvular pulmonal stenosis, an open patent ductus arteriosus (PDA), and a patent foramen ovale (PFO) at week 34. The infant was later delivered by cesarean delivery with a birthweight of 3110 grams, a birth length of 49 cm, and a birth head circumference of 35 cm. The postnatal vital signs showed normal heart rate (150 bpm), respiratory rate (35 x/min), and axilla temperature (37ºC). The baby cried spontaneously and adequately, with no sign of cyanosis, but the spO2 level was 93% on the right arm (pre-ductal) and 89% on the right leg (post-ductal). After birth, the initial management of the infant was the initiation of prostaglandin E1 (PGE1/alprostadil) infusion to reopen the ductus arteriosus 10 nanograms/minute. On physical examination, a murmur was heard. Icterus neonatorum was found 2 days after birth and treated with phototherapy for 5 days. Trans thoracic echocardiography revealed critical pulmonal stenosis, pulmonary valve annulus diameter was 6 mm, severe tricuspid regurgitation, and small patent ductus arteriosus. When the baby was 1 week old, the oxygen saturation decreased to 65%. The pediatric cardiologist decided to do the percutaneous transluminal balloon valvuloplasty (PTBV). During the procedure, we could cross the right ventricular outflow tract (RVOT) by using the bottom end of the 0,014" coronary guide wire with the support of a microcatheter. Subsequently, we dilated the pulmonary valve with the 1,5x5 mm coronary balloon, inflated until up to 14 atm, then inflated mini this balloon size 6 mm until the waist disappeared. Supraventricular tachycardia (SVT) occurred during the procedure and was then converted by medication. The peripheral saturation increased from 65 to 85%.

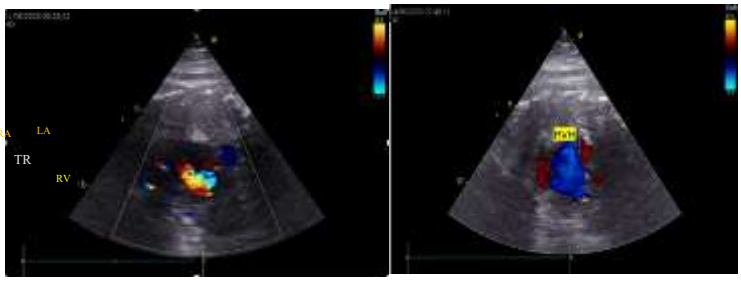

Figure 1. Fetal echocardiography of right ventricular hypertrophy, suspected pulmonary atresia, diminished flow from the right ventricle to the pulmonary artery, critical valvular pulmonary stenosis, an open patent ductus arteriosus (PDA), and a patent foramen ovale (PFO) at week 34.

The baby was then transported to the NICU, where the SVT reoccurred and was treated with adenosine but showed no improvement. Amiodarone was given later, and the sinus rhythm returned. Two days after PTBV attempt, second-degree AV block and atrial fibrillation occurred and were treated with bisoprolol 1x0,75mg, digoxin 2x0,015 mg, and prostaglandin 7,5 mcg/kg BW/min. Subsequent echocardiography imaging showed severe pulmonary stenosis, severe tricuspid regurgitation, mild PDA, and decreased systolic function of the left ventricle. The baby received bisoprolol 0.75 mg/24 jam, spironolactone 3.5 mg/24 jam, digoxin 2x0,015 mg), formula milk 12 x 31 ml, and the fluid intake was restricted to 20% (~130ml/kg BW/day). The conditions improved, and the baby was allowed to return home with the mother after 30 days of hospitalization.

Figure 1. Postnatal anterior-posterior x-ray of chest results showing right ventricular hypertrophy.

Figure 3 Trans thoracal echocardiography after birth. A. LA dilated, B & C: Colour doppler in 4 chambers view showed severe TR, PG 121mmHG, D & E. Short axis view showed critical pulmonal stenosis. F. Suprasternal view showed ductus arteriosus with pulmonic waist 0,5 mm.

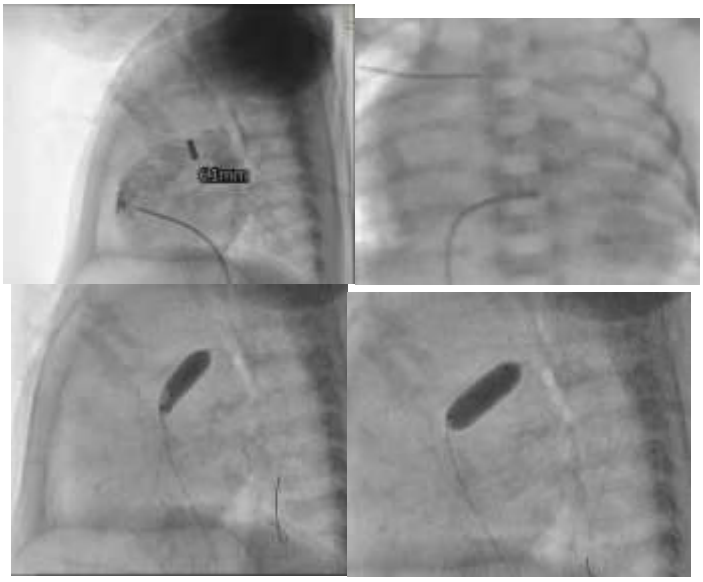

Figure 4. PTBV procedure. A. RVOT gram showed pulmonal stenosis with pulmonary valve annulus was 6 mm, B. cross the right ventricular outflow tract (RVOT) by using the bottom-end of 0,014" coronary guide wire with the support of microcatheter. C. Dilated

the pulmonary valve with the 1,5x5 mm coronary balloon, D. Inflated mini this balloon size 6 mm until the waist was disappeared.

DISCUSSION

Critical congenital heart disease is a particular type of CHD that requires early intervention within the first year of life to be treated successfully. About 25% of newborns with CHD have been found to have critical CHD.[10] Nearly all CHD, especially severe CHD, can now be treated to improve survival rates through advancements in medical science, surgery, and intervention procedures.[10,11] In significant congenital heart disease cases, intervention is often carried out within the first few weeks of life to improve hemodynamics and avoid end-organ dysfunction.[12]

Early fetal echocardiography, conducted between the ages of 18 and 22 weeks of pregnancy, supports the diagnosis of important congenital heart defects at the earliest possible stage. A large cohort study in Canada found that prenatal fetal echocardiography contributed to 249 (3.1%) near-miss diagnoses of severe CHD.[13] The study by Kondo et al. also highlighted that 5-day postnatal echocardiography screening is unhelpful for the diagnosis of CHD without prior prenatal diagnosis and instead leads to more false-positive cases.[14] Zhang et al.'s meta-analysis found that fetal echocardiography was 68.5% sensitive to the detection of CHD and 99.8% specific.[15]

In this case, the obstetric and gynecologist sent the patient to a pediatric cardiologist to perform the fetal echocardiography at 34 weeks of pregnancy because ultrasonography showed the right heart chamber dilatation and the mother with a history of recurrent premature ventricular tachycardia during pregnancy. However, early prenatal detection is still tricky because fetal echocardiography facilities are not widely available. Fetal echocardiography is indicated in high-risk pregnancies such as a family history of CHD or genetic disorders, the use of nonsteroidal anti-inflammatory drugs in the third trimester, exposure to cardiac teratogens (e.g., lithium, anticonvulsants), and TORCH infection during pregnancy.[16]

Although significant CHD can be accurately detected by early fetal echocardiography, certain anomalies might not be visible until later in pregnancy.[17] A small percentage of CHDs become apparent or become more evident as the pregnancy progresses. Therefore, an essential abnormality may not be discovered until late in the pregnancy or after delivery, while the heart evaluation may be normal at 18 weeks. This is accurate in some cardiomyopathies, cardiac tumors, and aortic or pulmonary stenosis. Fortunately, malformations that could be fatal rarely develop after 20 weeks of pregnancy. As a result, a typical mid-gestation transabdominal fetal echocardiogram should be used in conjunction with early fetal echocardiography, not in substitution for it.[1,7]

Prenatal diagnosis helps in a better understanding of CHD. It ensures that prenatal medical and interventional management, if possible, and delivery can be performed safely at a tertiary center. In a developing country, delay in admission could be due to delay in diagnosis, referral, financial, and social hindrances. The time needed for acceptance of the condition by the parents, the arrangement of funds for transport, and the time required for travel from a peripheral village to a major city further delay the presentation to a tertiary cardiac care center. In the absence of a strict newborn screening and limited echocardiogram availability, these children would present only after the closure of the ductus arteriosus. This delay results in hemodynamic instability leading to unfavorable outcomes.

Prenatal diagnosis, as opposed to postnatal diagnosis, has been shown in numerous trials to improve outcomes and survival rates of severe CHDs considerably. One study by Colaco et al. showed that 88.2% of patients with prenatal diagnosis of CHD, in contrast to 55.9% with postnatal diagnosis, get early medication for duct-related CHD.[2] When serious CHDs are identified during pregnancy rather than after birth, hospital costs are also reduced.[18] Early CHD implications counseling also helps families choose appropriate care. A prenatal diagnosis also makes scheduling delivery at a tertiary care facility with access to pediatric cardiology and cardiac surgery possible.[19]

Fetal echocardiography showed critical valvular pulmonal stenosis at 34 weeks. Critical pulmonary stenosis (CPS) is a severe pulmonary stenosis that occurs in neonates and causes obstruction of proper ventricular egress or flow to the pulmonary artery, requiring early intervention.[20] This obstruction occurs due to the fusion of the pulmonary valve leaflets, resulting in a conical valve with fibrous tissue in the center or maybe an orifice smaller than the pulmonary annulus. 3 Critical pulmonary stenosis is one of the congenital heart diseases (CHD) that causes cyanosis in newborns and has the potential to cause death. Pulmonary blood flow in neonates with CPS depends on the presence or absence of persistent ductus arteriosus (DAP).[21] Critical pulmonary stenosis is rare, with an estimated prevalence of 0.58 per 10,000 live births.[22]

In this case, fetal echocardiography also showed right ventricular dilatation and suspicion of pulmonary atresia. Postnatal management immediately gives prostaglandin infusion to ensure the ductus arteriosus remains open. One week later, the baby underwent a Percutaneous Transluminal Ballooning Valvulotomy (PBTV) procedure. These transcatheter-based techniques provide a suitable and effective alternative to open-heart operations.

Percutaneous balloon valvuloplasty is the primary intervention for neonates with critical or severe pulmonary stenosis. These patients undergo balloon valvuloplasty procedures in the cardiac catheterization laboratory. Typically, these procedures are performed while the patient is intubated and sedated or under general anesthesia. To better observe the pulmonary valve annulus, we performed lateral RV angiography and advanced the coronary wire and balloon before extending the Thysak balloon to open the valve.[23]

The likelihood of a successful balloon valvuloplasty procedure has increased and is often higher in institutions that handle many cases. Several notable studies involving less than 20 patients reported failure rates ranging from 20% to 45%. In major centers, the failure rate to accomplish an initial balloon valvuloplasty and cross the pulmonary valve is more like 5%–10%. The patient's condition demonstrates improvement following percutaneous transcatheter balloon valvuloplasty.[24]

However, there are still some limitations in implementing fetal echocardiography for the early diagnosis of CHD. The first one is that less experienced obstetric sonographers may have lower detection rates of cardiac abnormalities on screening ultrasound. More comprehensive fetal echocardiography performed by fetal or pediatric cardiac sonographers and interpreted by fetal or pediatric cardiologists provides more accurate prenatal cardiac diagnosis, with the highest reported detection rates of fetal cardiac abnormalities approaching 90% when performed by experienced fetal echocardiographers. This suggests that there is still room for improvement in detection rates. The second, obtaining complete fetal echocardiography views, can be more difficult during early gestation due to the small size of the fetal heart and surrounding structures.[25, 26]

CONCLUSION

In conclusion, the early prenatal diagnosis of critical CHD by fetal echocardiography, as presented in the case, was helpful for early detection, early diagnosis, early decision-making, and helping families comprehend the patient's conditions, which resulted in precise, appropriate treatment and overall better outcomes.

REFERENCES

1. Sun HY. Prenatal diagnosis of congenital heart defects: Echocardiography. Transl Pediatr. 2021;10(8):2210–24.
2. Colaco SM, Karande T, Bobhate PR, Jiyani R, Rao SG, Kulkarni S. Neonates with critical congenital heart defects: Impact of fetal diagnosis on immediate and short-term outcomes. Ann Pediatr Cardiol. 2017;10(2):126–30.
3. Ou Y, Bloom MS, Mai J, Francois M, Pan W, Xiao X, et al. Prenatal Detection of Congenital Heart Diseases Using Echocardiography: 12-Year Results of an Improving Program With 9782 Cases. Front Public Heal. 2022;10(May):1–10.
4. Donofrio MT, Moon-Grady AJ, Hornberger LK, Copel JA, Sklansky MS, Abuhamad A, et al. Diagnosis and treatment of fetal cardiac disease: A scientific statement from the american heart

association. Circulation. 2014;129(21):2183–242.
5. Xu H, Zhong L, Deng J, Peng J, Dan H, Zeng X, et al. High expression of ACE2 receptor of 2019-nCoV on the epithelial cells of oral mucosa. Int J Oral Sci. 2020;12(1):1–5.
6. Islam N, Saha S, Islam T, Mukherjee J, Roy M. Clinical Significance of Fetal Echocardiography in Diagnosing Congenital Cardiac Anomalies: An Experience From Eastern India. J Radiol Clin Imaging. 2023;06(02):107–17.
7. Irshaid LW, Elfky N, Ahmed B. Prenatal detection of critical congenital heart disease. Donald Sch J Ultrasound Obstet Gynecol. 2016;10(2):131–5.
8. Donofrio MT. Predicting the Future: Delivery Room Planning of Congenital Heart Disease Diagnosed by Fetal Echocardiography. Am J Perinatol. 2018;35(6):549–52.
9. Hunter LE, Simpson JM. Prenatal screening for structural congenital heart disease. Nat Rev Cardiol [Internet]. 2014;11(6):323–34. Available from: http://dx.doi.org/10.1038/nrcardio.2014.34
10. Oster ME, Lee KA, Honein MA, Riehle-Colarusso T, Shin M, Correa A. Temporal trends in survival among infants with critical congenital heart defects. Pediatrics. 2013 May;131(5):e1502-8.
11. Olney RS, Ailes EC, Sontag MK. Detection of critical congenital heart defects: Review of contributions from prenatal and newborn screening. Semin Perinatol. 2015;39(3):230–7.
12. McClain MR, Hokanson JS, Grazel R, Van Naarden Braun K, Garg LF, Morris MR, et al. Critical Congenital Heart Disease Newborn Screening Implementation: Lessons Learned. Matern Child Health J. 2017;21(6):1240–9.
13. Cardinal MP, Gagnon MH, Têtu C, Beauchamp FO, Roy LO, Noël C, et al. Incremental Detection of Severe Congenital Heart Disease by Fetal Echocardiography Following a Normal Second Trimester Ultrasound Scan in Québec, Canada. Circ Cardiovasc Imaging. 2022;15(4):E013796.
14. Shibata N, Kondo T, Kazama S, Kimura Y, Oishi H, Arao Y, et al. Impact of predictive value of Fibrosis-4 index in patients hospitalized for acute heart failure. Int J Cardiol [Internet]. 2021;324:90–5. Available from: https://doi.org/10.1016/j.ijcard.2020.09.056
15. Zhang YF, Zeng XL, Zhao EF, Lu HW. Diagnostic value of fetal echocardiography for congenital heart disease. Med (United States). 2015;94(42):e1759.
16. Willim HA, Cristianto, Alice Inda Supit. Critical Congenital Heart Disease in Newborn: Early Detection, Diagnosis, and Management. Biosci Med J Biomed Transl Res. 2020;5(1):107–16.
17. Nayak K, Naveen Chandra GS, Shetty R, Narayan PK. Evaluation of fetal echocardiography as a routine antenatal screening tool for detection of congenital heart disease. Cardiovasc Diagn Ther. 2016;6(1):44–9.
18. Jegatheeswaran A, Oliveira C, Batsos C, Moon-Grady AJ, Silverman NH, Hornberger LK, et al. Costs of prenatal detection of congenital heart disease. Am J Cardiol. 2011;108(12):1808–14.
19. Rychik J, Donaghue DD, Levy S, Fajardo C, Combs J, Zhang X, et al. Maternal psychological stress after prenatal diagnosis of congenital heart disease. J Pediatr [Internet]. 2013;162(2):302-307.e1. Available from: http://dx.doi.org/10.1016/j.jpeds.2012.07.023
20. Soeroso S. Penyakit jantung bawaan non sianotik. Buku ajar Kardiol anak Jakarta Ikat Dr Anak Indones. 1994;203.
21. Fiore AC, Jureidini S, Keenan W, Johnson RG. Cardiac surgery in the newborn: improved results in the current era. Mo Med. 2004;101(6):603–7.
22. Peterson RE, Levi DS, Williams RJ, Lai WW, Sklansky MS, Drant S. Echocardiographic predictors of outcome in fetuses with pulmonary atresia with intact ventricular septum. J Am Soc Echocardiogr. 2006;19(11):1393–400.
23. Rao PS. Percutaneous balloon pulmonary valvuloplasty: state of the art. Catheter Cardiovasc Interv. 2007;69(5):747–63.
24. Koentartiwi D. English Case report: Succesfully Management of Critical Pulmonary Stenosis in a 12-Month-Old Boy By percutaneous transluminal balloon valvuloplasty. WMJ (Warmadewa Med Journal). 2021;6(2):67–74.
25. Schneider PD, Groner A. Understanding the limitations of fetal cardiac imaging. Neoreviews. 2018;19(3):e134–42.
26. Co-Vu J, Ivsic T. Fetal echocardiography to diagnose fetal heart disease. Neoreviews. 2012;13(10).