doi: 10.31674/mjmr.2020.v05i03.001

MJMR

A CASE REPORT: PRENATAL DIAGNOSIS OF FETAL CARDIAC RHABDOMYOMA

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ABSTRACT

Fetal cardiac rhabdomyoma is a rare disease, and most of the cases are detected incidentally during the antenatal ultrasound. This is a case discovered during antenatal ultrasonography at 29 weeks of gestation. Multiple hyperechoic round shape masses were seen in the cardiac. After birth, the baby had been under a paediatric cardiologist follow-up with regular echocardiography. Management was mainly conservative at the time being as there was no sign of obstruction.

Keywords: Cardiac Rhabdomyoma; Prenatal Diagnosis; Fetal Echocardiography

INTRODUCTION

Primary congenital cardiac tumour is a rare condition. The incidence is around 1-2/10000 (Suwardewa *et al.*, 2016). The primary congenital cardiac tumour can be divided into two groups which are benign and malignant. Benign tumours are rhabdomyoma, teratoma, fibromas, hemangiomas, and myxomas. The malignant tumours are extremely rare, which included rhabdomyosarcomas and fibrosarcoma (Yuan, 2017). Among the primary cardiac tumours, cardiac rhabdomyoma is the most common type, with a 60% incidence.

The aetiologies of cardiac rhabdomyoma remain unclear, but it is highly associated with tuberous sclerosis complex. Tuberous sclerosis is diagnosed in most cases of multiple fetal cardiac rhabdomyoma (Ekmekci *et al.*, 2018). Tuberous sclerosis is a neurocutaneous syndrome inherited in an autosomal dominant pattern with an incidence of about 1/6000 to 1/10000 live births (Bejiqi, Retkoceri & Bejiqi, 2017). It is a rare genetic disease with multiple non-cancerous tumours grows in the brain or other organs.

Cardiac rhabdomyoma is commonly presented as multiple large lesions than single or small lesions. It may grow in utero and subsequently regress without treatment. Therefore, the prevalence is higher in children if compared to adults. Generally, the management for cardiac rhabdomyoma is conservative if no signs of obstruction. Surgical resection might be indicated in the case of cardiac function being compromised. Most of the patients have a good prognosis if cardiac function is not affected. The neonatal mortality rate is about 4 to 6%(Ekmekci *et al.*, 2018). We reported a case of fetal cardiac rhabdomyoma was found incidentally during a routine antenatal check-up in the primary care setting.

CASE STUDY

39 years old gravida 6 Para 1+4 presented at 29 weeks gestation came for routine antenatal follow-up and was found to have multiple homogenous cardiac lesions. She was referred for a detailed scan, and fetal echocardiography of a cardiac malformation was detected. The pregnancy was spontaneous and there was no consanguineous marriage.

The patient was diagnosed with type 2 diabetes mellitus five years ago and was treated with two oral hypoglycaemic agents before pregnancy. Her HbA1c was 6.8 during the first trimester. Her metformin was continued, and recombinant synthetic human intermediate-acting insulin was started at 19 weeks because of suboptimal blood sugar profile. Subsequently, her blood sugar profile was optimized, and her HbA1c was reduced to 5.8% during the third trimester. Aspirin was started at 12 weeks as prevention of preeclampsia as the patient was diabetic.

When concern about her past obstetric history, she had four abortions from years of 2008 to 2014 without dilation and curettage. She delivered her first son four

years ago via emergency lower segment caesarean section because of foetal distress. This is her second marriage and her second child with her current husband.

For her current pregnancy, a detailed scan was done in an obstetric clinic by a maternal-fetal medicine specialist at 31 weeks. The result revealed multiple cardiac rhabdomyoma without other abnormalities. She was referred to a paediatric cardiologist to have fetal echocardiography at 32 weeks. The fetal echocardiography demonstrated the presence of multiple globular hyperechoic mass at the right atrium, the right ventricle, and the left ventricle. There were normal cardiac chambers and good biventricular function. The masses were located at the right atrium measured 10x10mm, intraventricular septum on right ventricular site measured 13mmx13mm, tricuspid annulus measured 8x10mm, and left ventricular near mitral valve annulus measured 4mmx5mm. There was a mild turbulent flow at the superior vena cava right junction, possibly due to the mass. Otherwise, there was no obvious inflow obstruction to both right and left ventricular. Hence, the diagnosis was multiple rhabdomyoma with normal function and the possibility of mild obstruction at the right atrial superior vena cava junction.

At week 38 of pregnancy, the patient was admitted for induction of labour using a prostaglandin pessary. However, an emergency lower segment caesarean section was performed because of impending scar dehiscence. Although the intraoperative findings revealed that there was no scar dehiscence. The baby was delivered with a good Apgar score of eight at the first minute and nine at the fifth minute. The baby was born with a weight of 3.2kg and 50cm in length. The baby was intubated at 25 minutes of life due to worsening respiratory distress. She was discharged well on day 10 of life.

Neonatal echocardiography performed on day one of life confirmed the prenatal finding of multiple rhabdomyomas with multiple small rhabdomyomas over the left ventricular wall. There was no plan for surgical intervention because of no obstructions. The child was thriving well and under a paediatric cardiologist for regular follow- up. She had no signs or symptoms suggestive of tuberous sclerosis. She is currently six months old with weight and length of 8kg and 66cm, respectively.

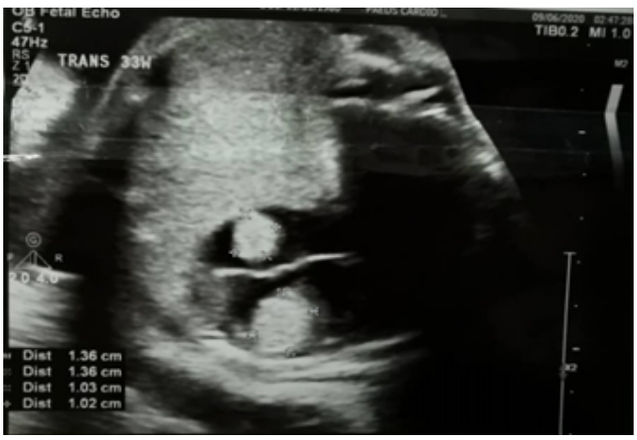

Figure 1: Globular Hyperechoic Mass in the Right Ventricle Measured 13mmx13mm and the Right Atrium 10mmx10mm.

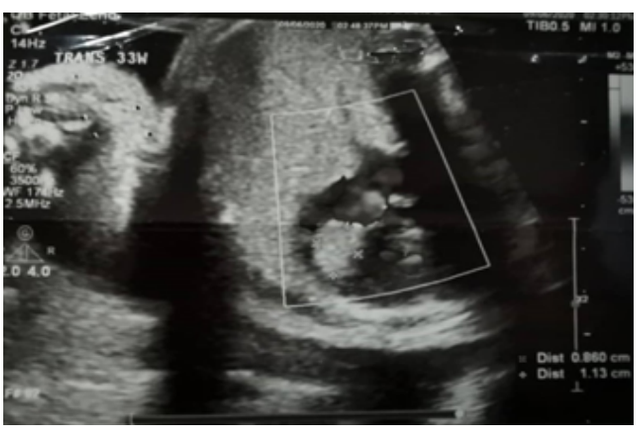

Figure 2: The Mass Located at Tricuspid Annulus Measured 8mmx11mm

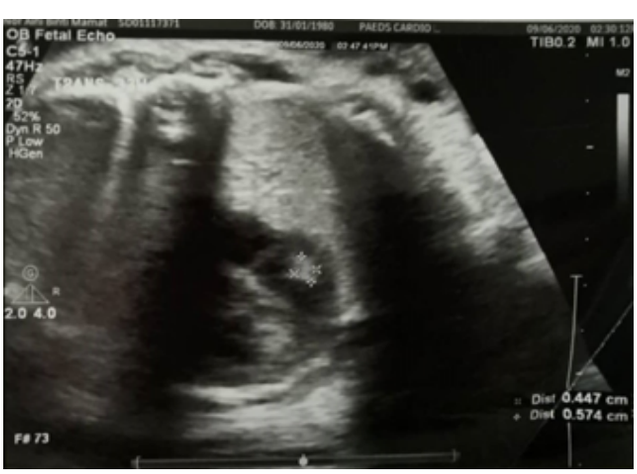

Figure 3: A Small Rhabdomyoma Located at Left Ventricle Near Mitral Valve Annulus Measured 4mmx5mm

DISCUSSION

Cardiac tumours are very uncommon in a neonate. Among the causes of primary cardiac tumours, rhabdomyoma is the most common to be seen in up to 60% of cases. About 90% of the cardiac rhabdomyoma is multiple tumours (Yuan, 2017). Rhabdomyomas are usually demonstrated as homogenous, hyperechogenic round masses in the ventricles. It might appear as multiple foci in the ventricles and septal wall (Chao *et al.*, 2008). The ultrasound findings of this case were consistent with common locations of rhabdomyoma which were where located in the bilateral ventricles.

Rhabdomyoma can be asymptomatic, but it might cause heart failure in the neonatal periods on another end of the spectrum. The outflow obstruction, arrhythmia, or valve dysfunctions, and thromboembolic stroke are the complications of rhabdomyoma that may happen during the neonatal period (Bassirou *et al.*, 2020). Besides, rhabdomyoma will be presented as biphasic evolution in which they grow in fetus up to 32 weeks of gestation and slowly regress during the first year of life(Bassirou *et al.*, 2020). The exact mechanism of regression still incomplete; there may be due to reduced estrogen level (Ekmekci *et al.*, 2018). However, the rapid growth of cardiac rhabdomyoma in utero was uncommon. The fetus had a higher risk of mortality if the tumour size was greater than 20mm in diameter (Chao *et al.*, 2008). The larger size of the mass carries a higher risk of hemodynamic instability or outflow obstruction and subsequently resulted in a poor outcome for the fetus. Fetal arrhythmia was associated with the hydrops fetalis, and postnatal fetal arrhythmia will increase the risk of mortality. Surgical intervention is mostly required if the hemodynamic instability component is present.

The incidence rate of tuberous sclerosis is 50 to 86% in patients with cardiac rhabdomyoma (Suwardewa *et al.*, 2016). Clinical presentation of tuberous sclerosis included dermatological manifestations (hypopigmented macule, sebaceous adenoma, and shagreen patches), cerebral abnormalities of calcification, periventricular nodule, seizure, and cerebral atrophy. Among the signs and symptoms of tuberous sclerosis, cardiac rhabdomyoma might be the earliest signs that could be detected antenatally. A detailed scan should be performed with specific attention to brain ventricle dilation and kidney dysplasia. In this case, a detailed scan was performed at 31 weeks of gestation, and there was no abnormality was found except the cardiac rhabdomyoma.

Furthermore, investigating family history also plays an important role in detecting tuberous sclerosis, especially the family history of mental retardation and epilepsy or any cutaneous signs of tuberous sclerosis among the family members (Suwardewa *et al.*, 2016). However, in this case, the child and family members had no other signs and symptoms of tuberous sclerosis seen and genetic testing was not done for tuberous sclerosis. There were no other genetic abnormalities that associate with cardiac rhabdomyoma.

Nonetheless, postpartum echocardiographic monitoring is necessary. In this case, the child was followed up with a paediatric cardiologist for regular echocardiography and showed no signs of heart failure.

CONCLUSION

In summary, this case highlights the importance of prenatal detection of the cardiac tumour. Cardiac rhabdomyoma is the most common primary cardiac tumour which can be detected during the prenatal care with the antenatal ultrasonography. Further Investigation with foetal echocardiography is essential in the presence of cardiac mass. The prognosis of the foetus is good if there are no signs of obstruction.

Conflict of Interest

The authors declare that there are no conflicts of interest relevant to this article.

ACKNOWLEDGEMENT

The authors would like to thank the patient for her permission and cooperation in writing this case report. The informed consent of the parents of the case was obtained before the preparation of the case report.

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