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FIRST-TRIMESTER AORTIC ROOT REPLACEMENT IN A PREGNANT WOMAN WITH PREVIOUSLY UNDIAGNOSED MARFAN SYNDROME

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ABSTRACT

Aortopathy encompasses a spectrum of aortic aneurysms and dissections and represents a major cause of cardiovascular morbidity during pregnancy, particularly in women with heritable connective tissue disorders such as Marfan syndrome. Evidence guiding the management of severe aortopathy diagnosed in early pregnancy remains limited. We report the case of a pregnant woman with previously undiagnosed Marfan syndrome who presented at 10 weeks' gestation with decompensated heart failure due to severe aortic regurgitation and a massive aortic root aneurysm. Transthoracic echocardiography revealed marked dilatation of the aortic root and ascending aorta, accompanied by left ventricular dilatation and systolic dysfunction. Following multidisciplinary evaluation, urgent surgical intervention was deemed necessary because of prohibitive maternal risk. The patient successfully underwent aortic root and valve replacement during the first trimester. Postoperatively, cardiac function improved, and the pregnancy was continued without major cardiovascular complications, ultimately resulting in the delivery of a healthy neonate. This case highlights the complexity of managing severe Marfan-associated aortopathy diagnosed during early pregnancy and demonstrates that life-saving aortic surgery may be considered in carefully selected patients when maternal risk is extreme, emphasizing the importance of individualized, multidisciplinary decision-making.

KEYWORDS

Marfan syndrome, pregnancy, aortopathy, aortic root replacement, multidisciplinary management

LEARNING POINTS

- Pregnancy may unmask previously undiagnosed Marfan syndrome and carries a high risk of life-threatening aortic complications.
- Current guidelines recommend pre-pregnancy surgery when the aortic root diameter reaches ≥4.5 cm; however, management of severe aortopathy detected during pregnancy remains challenging and individualized.
- In carefully selected high-risk patients, severe aortic root aneurysm complicated by heart failure may necessitate life-saving aortic surgery even during the first trimester, guided by multidisciplinary decision-making and European Society of Cardiology recommendations.

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INTRODUCTION

Aortopathy encompasses a spectrum of aortic aneurysms and dissections. Although definitions vary by anatomic location, a localized dilation exceeding 1.5 times the expected diameter is generally considered diagnostic[1]. Aortic aneurysms are often clinically silent until complicated by dissection, which carries high mortality, particularly during pregnancy[2,3].

Pregnancy-associated aortic complications are frequently linked to heritable connective tissue disorders, most commonly Marfan syndrome[4]. Hemodynamic and hormonal changes during pregnancy may accelerate aortic dilatation and precipitate clinical presentation of previously unrecognized disease[5]. According to the modified World Health Organization (WHO) classification, an aortic root diameter >45 mm in women with Marfan syndrome represents WHO class IV risk, in which pregnancy is contraindicated because of extreme maternal danger[6].

Evidence guiding the management of severe aortopathy first diagnosed during early pregnancy remains limited. We report a case of previously undiagnosed Marfan syndrome presenting in the first trimester with massive aortic root aneurysm and heart failure, successfully managed with urgent aortic surgery.

CASE DESCRIPTION

A 29-year-old primigravida at 10 weeks' gestation was referred to a tertiary centre for suspected Marfan syndrome and severe aortic root aneurysm. She reported progressive exertional dyspnoea and orthopnoea. Physical examination revealed marfanoid habitus, arachnodactyly, bounding carotid pulses, and peripheral oedema.

Transthoracic echocardiography demonstrated massive dilatation of the aortic root and ascending aorta with severe aortic regurgitation and left ventricular dilatation with moderate systolic dysfunction *(Table 1)*. The parasternal long-axis view demonstrated marked dilatation of the sinus of Valsalva *(Fig. 1)*. Severe left ventricular dilatation was also evident on parasternal long-axis imaging *(Fig. 2)*. Colour Doppler imaging confirmed severe central aortic regurgitation *(Fig. 3)*. The apical four-chamber view demonstrated markedly increased left ventricular end-diastolic volume *(Fig. 4)*. Based on systemic features and imaging findings, Marfan syndrome was considered the most likely diagnosis. Genetic testing was unavailable.

During hospitalization, the patient developed recurrent chest pain concerning for aortic complications. Advanced imaging was unavailable. A multidisciplinary team recommended pregnancy termination because of extreme maternal risk, but the patient elected to continue the pregnancy. Given the critical aortic dimensions and worsening heart failure, urgent surgical intervention was undertaken.

At 10 weeks' gestation, she underwent aortic root and valve replacement using a modified Bentall–de Bono procedure under cardiopulmonary bypass. Postoperative recovery

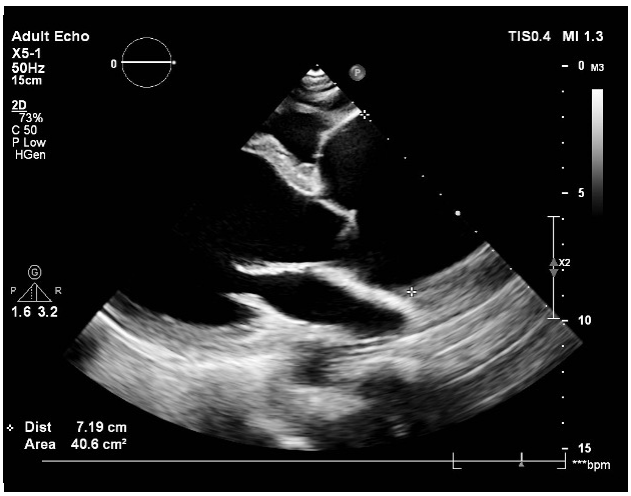


Figure 1. Transthoracic echocardiogram (parasternal long-axis view) demonstrating marked dilatation of the sinus of Valsalva (7.19 cm).

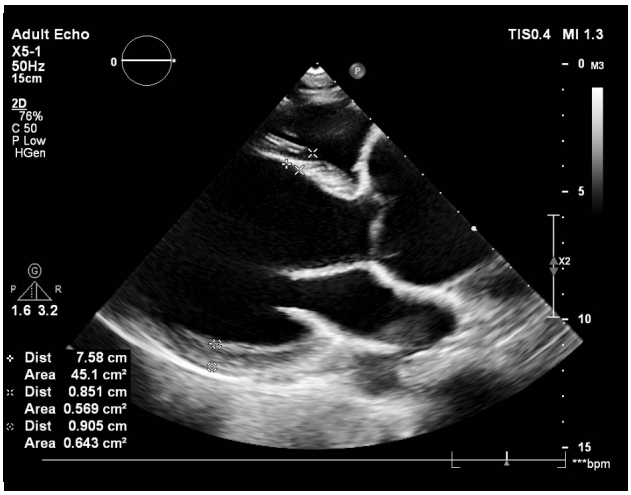


Figure 2. Parasternal long-axis view showing severe left ventricular dilatation with an end-diastolic diameter of 7.58 cm.

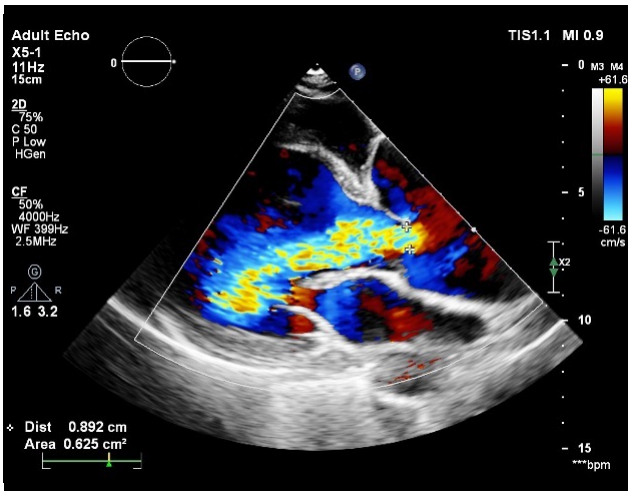


Figure 3. Color Doppler imaging demonstrating severe central aortic regurgitation.

was uncomplicated apart from a conservatively managed pneumothorax. Follow-up echocardiography showed a significant reduction in left ventricular dimensions and improved systolic function.

The remainder of the pregnancy was uneventful under

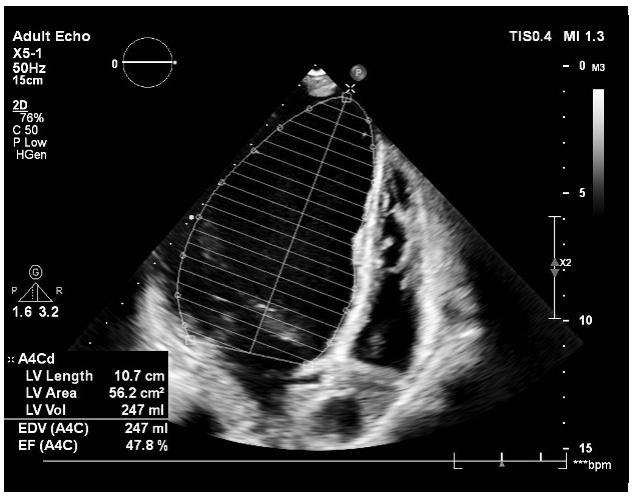


Figure 4. Apical four-chamber view showing marked left ventricular dilatation with an end-diastolic volume of 247 ml.

close surveillance. Caesarean delivery was performed at 35 weeks' gestation, resulting in a healthy neonate. At 5 months postpartum, echocardiography demonstrated normalization of left ventricular size and function.

DISCUSSION

Aortic complications are a leading cause of maternal mortality in women with heritable thoracic aortic disease, with Marfan syndrome representing the most frequent underlying aetiology[4,7]. The risk of dissection increases substantially when aortic root diameter exceeds 45 mm, particularly during pregnancy, prompting guideline recommendations for prophylactic surgery before conception[6,8].

This case is notable for the early gestational presentation of previously undiagnosed Marfan syndrome complicated by massive aortic root aneurysm, severe aortic regurgitation, and heart failure. In such scenarios, management is challenging because surgical intervention during pregnancy—particularly in the first trimester—carries significant foetal risk, while conservative management exposes the mother to potentially fatal complications.

Current European Society of Cardiology (ESC) and American College of Cardiology/American Heart Association guidelines discourage pregnancy continuation in women with Marfan syndrome and an aortic root diameter >45 mm but provide limited guidance for patients diagnosed during pregnancy without dissection, yet with progressive dilatation or heart failure [6,8]. Although aortic surgery during pregnancy is generally deferred to the second trimester when feasible, isolated reports demonstrate that first-trimester surgery may be lifesaving in carefully selected cases managed at experienced centres[9,10].

Our case illustrates that individualized, multidisciplinary decision-making can result in favourable maternal and foetal outcomes even in extremely high-risk situations, when maternal risk is prohibitive and alternative options are limited.

Measured structures	10-weeks pregnancy	12 weeks pregnancy (2-week post-operative)	5-months post-partum	Normal range by ASE
Ascending aorta diameter (cm) Indexed (cm/m²)	6 3.55	-	-	2.7 ± 0.4
Sino-tubular junction diameter (cm) Indexed (cm/m²)	7.4 4.38	-	-	2.6 ± 0.3
Aortic valve annulus (cm) Indexed (cm/m²)	3.4 2	-	-	2.3 ± 0.3
Valsalva sinuses diameter (cm) Indexed (cm/m²)	7.19 4.25	2.5 1.48	-	3.0 ± 0.3
LV End-diastolic diameter (cm) Indexed (cm/m²)	7.58 4.48	6 3.55	5.44 3.34	3.8-5.2 2.3-3.5
LV End-systolic diameter (cm) Indexed (cm/m²)	5.79 3.43	4.9 2.9	4.25 2.61	2.2-3.5 1.3-2.1
LV end diastolic volume (ml) Indexed (ml/m²)	247 146	146 86.39	124 76.1	46-106 29-51
LV end systolic volume (ml) Indexed (ml/m²)	129 76.3	90 53.25	65.9	14-42 8-24
Stroke volume (ml)	118	38.36	46.9	>55
LV ejection fraction (%)	47.8	56	58.1	60-100

Table 1. Preoperative and postoperative transthoracic echocardiographic measurements.

Abbreviations: LV, left ventricle; ASE, American Society of Echocardiography, –, indicates aortic root and ascending aorta replaced with a composite graft and bioprosthetic valve; postoperative diameters not applicable. Early postoperative measurements reflect altered loading conditions following cardiopulmonary bypass.

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