A Woman With Marfan Syndrome in Pregnancy: Managing High Vascular Risk With Multidisciplinary Care

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Abstract

Background: Women with connective tissue disorders are at risk for cardiovascular complications during pregnancy, but there are no guidelines for pregnant women with aortic root diameter > 45 mm or with rapid aortic widening. We describe the issues of practical significance in the management of pregnancy and delivery in a woman with Marfan syndrome (MFS).

Case: A pregnant woman with MFS presented for tertiary care at 26 weeks' gestation. Rapid aortic dilatation triggered a decision to undertake delivery preterm, with a resulting good neonatal outcome. A multidisciplinary approach aided in optimizing the monitoring and timing of delivery and subsequent aortic repair, and allowed planning for the management of a potential vascular catastrophe.

Conclusion: Having optimal maternal and neonatal outcomes for pregnant women with Marfan syndrome depends on a highly responsive and coordinated team effort, including meticulous planning for a vascular catastrophe.

Résumé

Contexte : Bien que les femmes qui présentent des troubles affectant les tissus conjonctifs soient exposées à des risques de complications cardiovasculaires pendant la grossesse, nous ne disposons d'aucune ligne directrice en ce qui concerne les femmes enceintes dont le diamètre de l'anneau aortique est supérieur à 45 mm ou qui connaissent un élargissement rapide de l'aorte. Nous décrivons les facteurs significatifs sur le plan pratique dans le cadre de la prise en charge de la grossesse et de l'accouchement chez une femme atteinte du syndrome de Marfan.

Cas : Une femme enceinte atteinte du syndrome de Marfan nous a consultés pour obtenir des soins tertiaires à 26 semaines de gestation. La dilatation rapide de l'aorte a mené à la décision de procéder à un accouchement préterme (lequel a donné lieu à une bonne issue néonatale). Le recours à une approche multidisciplinaire a contribué à l'optimisation du monitorage et de la chronologie de l'accouchement (et à la réparation subséquente de l'aorte), en plus de permettre la planification de la prise en charge d'une potentielle catastrophe vasculaire.

Conclusion : Dans le cas des femmes enceintes qui sont atteintes du syndrome de Marfan, l'obtention d'issues maternelles et néonatales optimales dépend de la mise en œuvre d'un effort d'équipe grandement attentif et coordonné (y compris celle d'une planification méticuleuse de la prise en charge d'une potentielle catastrophe vasculaire).

Key Words: Marfan syndrome, pregnancy, aortic root, vascular risk, multidisciplinary team, drills, high risk pregnancy

Competing Interests: None declared.

Received on August 28, 2014

Accepted on September 10, 2014

J Obstet Gynaecol Can 2015;37(8):724–727

INTRODUCTION

Marfan syndrome (MFS) is one of the most common inherited disorders affecting the heart and aorta during pregnancy.[1] The condition is diagnosed using the 2010 revised Ghent nosology with or without genetic testing.[1,2] MFS is associated with mutations of the FBN-1 gene, coding for fibrillin-1, a protein integral to the structure of connective tissue, and also regulates the TGF-beta pathway.[1]

The most common causes of death in pregnant women with MFS are aortic dissection or rupture.[3,4] Although a normal aortic root diameter does not exclude the possibility of dissection, the risk is increased in the presence of aortic root dilatation, particularly in pregnancy.[4] Aortic dissection in association with pregnancy is most likely to occur in the last trimester or early postpartum. This may be due to the altered hemodynamics of pregnancy and the postpartum period, causing arterial wall stress, or from hormonal effects on arterial wall structure.

In pregnant women, guidelines are in place to direct the management of those with a moderately dilated aortic root (< 45 mm)[5–8]; however, when the aortic root is > 45 mm in diameter, there is little guidance for managing pregnancies to prevent catastrophic events. We describe here the coordinated multidisciplinary approach to management of pregnancy and delivery, and subsequent aortic repair, in a woman with MFS who had a rapidly enlarging ascending aorta.

THE CASE

A 28-year-old primigravida with MFS, who had recently moved to our province, was referred urgently to our maternal and fetal medicine unit. She had a singleton pregnancy at 26 weeks' gestation. She had had identification of the FBN-1 gene mutation one year before her pregnancy, and she had had genetic counselling at that time. However, she had not had pre-conception counselling with a maternal–fetal medicine subspecialist, and had not been made fully aware of the potential complications of pregnancy associated with MFS.

She had had no complications arising from MFS. Her father, two paternal uncles, and two cousins had MFS, and her father had died at age 52 from thoracic aortic dissection. She denied smoking, alcohol consumption, and illicit drug use, and was on no medication. Her pre-pregnancy BMI had been 18.2 kg/m^2. At her first visit, blood pressure was 100/60 mmHg. Her prenatal course had been unremarkable to the time of presentation.

On arrival in the province at 23 weeks' gestation, the patient had undergone review by a cardiologist. She had begun treatment with metoprolol 12.5 mg twice daily. Echocardiography performed at 23 weeks' gestation demonstrated an aortic root diameter of 43 mm; however, because of technical issues, it was uncertain whether the maximum diameter had been measured. Accordingly, she had undergone thoracic and abdominal MRI at 25 weeks, and this showed a dilated aortic root with a diameter of 47 mm at the mid-sinuses. The ascending aorta had a normal measurement of 30 mm, and the remainder of the thoracic aorta had normal dimensions. The main pulmonary artery was mildly dilated at 32 mm. The remainder of the echocardiogram was normal, with no mitral regurgitation or left ventricular dysfunction. There were no abnormalities of the abdominal aorta on MRI, but dural ectasia was identified.

Fetal ultrasound at 26 weeks confirmed a single intrauterine pregnancy with appropriate estimated fetal weight. Fetal anatomy and cervical length were within normal limits. Prenatal investigations including routine serology, hematology, liver function, renal function, and aneuploidy screening showed unremarkable results.

After counselling from the maternal and fetal medicine unit at 26 weeks, the patient confirmed her commitment to continue with the pregnancy. The Halifax protocol for multidisciplinary management of connective tissue disorders with cardiovascular involvement in pregnancy[9] was used to guide prenatal care. The first formal multidisciplinary meeting regarding this patient's management was held at 29 weeks' gestation. The meeting included representatives of maternal–fetal medicine, cardiology, cardiovascular surgery, obstetric and cardiovascular anaesthesia, neonatology, operating room leaders, and cardiovascular intensive care coordinators, as well as clinical nurse specialists in cardiology, neonatology, and obstetrics. Details of the case, the results of imaging, and relevant medical literature were reviewed and plans made for delivery. The group was led by the representatives of maternal–fetal medicine and cardiology.

With an aortic root diameter of 47 mm, the patient was advised that her risk of maternal aortic dissection or rupture was at least 10%, on the basis of available evidence.[7] Delivery was planned for 33 to 34 weeks, taking the maternal and neonatal risks into account. Plans were also made for emergency Caesarean section and aortic repair or perimortem Caesarean section in the event of aortic dissection occurring in the interim. Caesarean section and vaginal delivery instruments were stocked in the local emergency room, as were intrapartum and postpartum medications. Emergency department staff underwent specific drills for the management of aortic dissection in a pregnant woman, including the potential need for perimortem Caesarean section in the event of hemodynamic collapse. The patient and her partner were informed of the early symptoms of aortic dissection, and were instructed to carry a detailed plan of care at all times, listing contact information for all those involved in her care to ensure continuous availability of emergency care. Details of the patient's condition and of her delivery plans were provided to the provincial emergency health services, and collaborative plans made in anticipation of the need for emergency transportation. The patient relocated to within one kilometre of both the general and the women's hospitals; she had continuous outpatient supervision and access to emergency services. Outpatient management, with relocation of the patient to be close to both hospitals, allowed rapid access to specialized prenatal care while still having expedited access to emergency cardiovascular and critical care. A callout list of attending physicians and staff