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CASE REPORT

Spontaneous ascending aortic rupture in a pregnant woman with neurofibromatosis type 1

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Abstract

Neurofibromatosis type 1 (NF-1) is an autosomal dominant disorder that affects 1 in 3000 individuals. Vascular involvement in NF-1 is a well-recognized, but rare, feature of this disease. In pregnant women, the risk of aortic dissection or rupture is elevated during pregnancy and the postpartum period. We report a pregnant woman who had a history of NF-1 with a spontaneous ascending aortic rupture. This rupture was successfully treated by emergent surgery. The mother and the 28-week-gestation newborn recovered uneventfully. During 7 years of follow-up, aorta of the patient shows no significant change. A review of the literature regarding the pathogenesis of this condition is also presented.

Keywords Thoracic aorta · Neurofibromatosis · Pregnancy

Introduction

NF-1 is an autosomal dominant disorder linked to chromosome 17. NF-1 affects approximately 1/3000 people and may primarily involve any tissue of the body, including connective tissue, nerve tissue, the vasculature, and others [1]. NF-1 vasculopathy is a rare clinical feature of NF-1. Arterial stenosis, arteriovenous malformations, and aneurysms have been reported in NF-1. Arterial lesions may involve the aorta and renal, mesenteric, carotid–vertebral, or intracranial arteries. Here, we report the rare case of spontaneous ascending aortic rupture in a pregnant patient with NF-1.

Case report

A 42-year-old pregnant woman with a positive family and personal history of neurofibromatosis type 1 (NF-1) visited the emergency department. She had sudden chest pain and left pleural effusion, which was identified at a nearby clinic. She was at the 30th week of gestation. She also had a history of hypertension, cesarean section, and subarachnoid hemorrhage, which was treated with clipping at 25 years. On examination, she had multiple cutaneous neurofibromata and café-au-lait spots. An emergent chest computed tomography scan showed rupture of the ascending aorta and left pleural effusion (Fig. 1a). A three-dimensional computed tomography scan showed a ruptured ascending aorta (Fig. 1b). The patient's vital signs were stable on admission to hospital.

The decision to perform cesarean section and hysterectomy was made to rescue the fetus and patient. Her blood pressure dropped to 60/40 mmHg during cesarean section and hysterectomy. Therefore, cardiopulmonary bypass was established with femoral venous drainage and femoral arterial return immediately after rapid closure of the abdomen. Great care was taken to achieve full hemostasis. We then rapidly opened her chest. There was no active bleeding or cardiac tamponade. Adventitial hematoma was detected on the ascending aorta to branches of neck vessels. An additional venous cannula was inserted via right atrium to obtain total cardiopulmonary bypass. The ascending aorta was replaced under deep hypothermic circulatory arrest with selective cerebral perfusion. An 8-mm small aortic tear with rupture of the ascending aorta was observed. There was no sign of aortic aneurysm, dissection, or other abnormalities. The patient was extubated on postoperative day 1. The patient's subsequent postoperative course was uneventful, and she was discharged on postoperative day 12. The newborn weighed 1482 g and was discharged on postoperative day 67 without any complications. During 7 years

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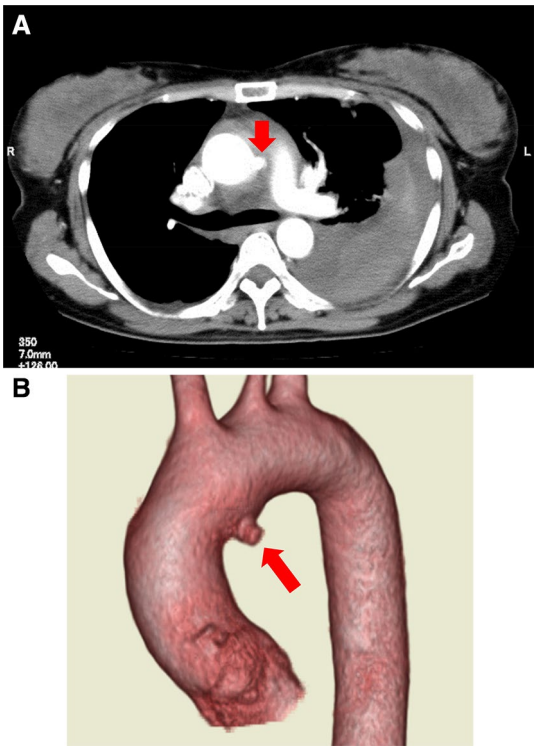

Fig. 1 **a** Preoperative computed tomography (axial view) shows a ruptured ascending aorta (red arrow). There was pleural effusion in the left thoracic cavity. **b** Three-dimensional computed tomography also shows a ruptured site of the lesser curvature side of the ascending aorta (red arrow)

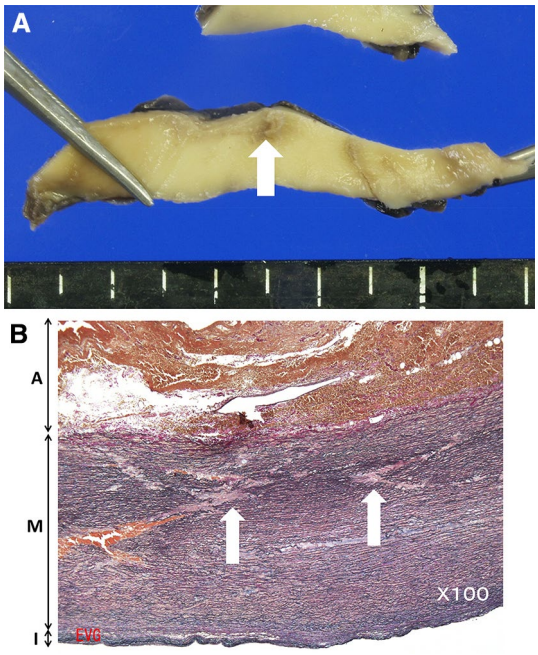

Fig. 2 **a** Macroscopic finding of a pathological specimen shows a tear in the ascending aorta (white arrow). **b** Microscopic finding of a pathological specimen (elastic van Gieson stain) shows an irregular break in medial elastic fibers (white arrows). *A* adventitia, *M* media, *I* intima

of follow-up, a chest computed tomography scan showed no major changes at the systemic aorta, including repaired aortic lesions.

A pathological specimen showed no findings of aneurysmal change and dissection (Fig. 2a). An irregular breakage site of medial elastic fibers was observed only around the ruptured aortic wall by microscopic findings of the specimen (Fig. 2b). This break in medial elastic fibers is not a typical finding for the aorta of pregnant women. Some scattered S100 protein positive cells were detected only in the media on immunohistochemical staining. There was no definitive histological evidence of neurofibromal invasion of the aortic wall and mesodermal dysplasia.

Discussion

Vascular involvement in NF-1 is a known feature of this disease. Hence, spontaneous rupture of a major artery is extremely uncommon in this patient population. Cases of spontaneous aortic rupture are even more uncommon. A case of recurrent aortic pseudoaneurysm after endovascular aneurysm repair at the distal infrarenal aorta has been reported [2]. A case of rupture of a dissecting aneurysm of the renal artery in a pregnant woman with neurofibromatosis was also reported [3]. However, these cases showed pseudoaneurysm or dissecting aneurysm. Our case showed no sign of aneurysmal change or dissection in the aorta. We experienced a rare case of spontaneous rupture of thoracic aorta without aneurysm or dissection in a pregnant woman with NF-1.

The previous reports showed that underlying causes of aortic rupture in patients with NF-1 were due to associated connective tissue anomalies. Two pathogenic mechanisms have been suggested. One mechanism is smooth muscle (mesodermal) dysplasia and direct vascular invasion by neurofibromatous tissue. The second mechanism of aneurysm formation is often found in larger vessels where neurofibromatous or ganglioneuromatous tissue invades and weakens the arterial wall [4]. In the current case, there was no cause of the aneurysm like mesodermal dysplasia or neurofibromatous tissue involvement adjacent to the vessels in our specimens. For the cause of rupture in our case, we cannot exclude the possibility of complete blow-out rupture of small neurofibromatous tissue directly invading the aortic wall. Even histology did not show neurofibromatous tissue invasion around the aortic tear.

In pregnancy, women with aortic disease, such as arteritis and aortitis, are at considerable risk of aneurysmal formation and dissection with potential for catastrophic outcomes.

Pregnancy results in an increased risk of dissection because of physiological and hormonal changes that occur, particularly in those with connective tissue disorders, genetic syndromes, congenital heart disease, and other heritable and acquired conditions involving the aorta [5]. Pregnancy introduces a progressive, estrogen-mediated, structural weakening of the aortic media due to reticulin fragmentation and elastin fiber disorganization that peaks in the third trimester and postpartum period [6]. However, a break in medial elastic fibers in our case is not compatible with this ordinary structural weakening of the aortic media during pregnancy. This breakage could be a cause of spontaneous rupture of the aorta in our case. During 7 years of follow-up, computed tomography showed no considerable change in our patient's aorta or major arteries.

Conclusion

We recommend paying careful attention to spontaneous aortic or arterial rupture when encountering sudden onset of pain with signs of blood loss in pregnant patients with NF-1. Once a diagnosis is made, appropriate surgical treatment without delay should be performed to rescue the mother and child.

Compliance with ethical standards

Conflict of interest None of authors had a conflict of interest concerning this study, and none received outside support for this research.

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