CASE REPORT

Giant coronary artery aneurysm at autopsy

Alicia Blackman,¹ Wayne Mohammed²

¹Department of Anatomical Pathology, Eric Williams Medical Sciences Complex, Champs Fleurs, Trinidad and Tobago

²Department of ParaClinical, University of the West Indies at Saint Augustine, Champs Fleurs, Trinidad and Tobago

Correspondence to

Dr Wayne Mohammed,
wamed@live.com

All authors contributed equally to this paper.

Accepted 2 March 2018

SUMMARY

This case presents a woman in her early 20s who died after the sudden onset of chest pain. Five years earlier, she was investigated for a cardiac murmur during pregnancy and an echocardiogram revealed a 6.0×3.0 cm blood-filled sac compressing the left atrium and anterolateral aspect of the left ventricle with communication to the aortic root. She later had a CT scan of the chest with contrast, which showed aneurysmal dilatation of the left main coronary artery. She was placed on aspirin but defaulted from clinic 11 months post partum. At autopsy, a left coronary aneurysmal sac measuring 10.0×9.0 cm. was identified with a rupture measuring 7.0 cm in length and the pericardial sac contained 900 mL of blood with clots. The cause of death was cardiac tamponade secondary to rupture of the coronary artery aneurysm.

BACKGROUND

The most common aetiology of coronary artery aneurysm (CAA) is atherosclerosis. However, there are other important aetiologies such as congenital malformation. The pathogenesis, management and prognosis have not been well established due to the condition's rarity and a paucity of large studies on the subject. This case highlights its fatal outcome in a young woman with a possible congenital aetiology. The course of the disease illustrates the importance of thorough investigation and follow-up of patients with coronary artery aneurysm. This CAA is also one of the largest reported in the literature.

CASE PRESENTATION

The patient was a 22-year-old woman who was initially referred to the cardiology clinic at 17 years of age for evaluation of a cardiac murmur at 28 weeks of pregnancy. There was a history of cardiac murmur at a younger age, however it is unknown if she was formally investigated. There was no history of any serious childhood diseases including symptomatic cardiac disease or cardiac surgery. Antenatal clinic notes recorded that her VDRL test was negative. There was no history of trauma. At the initial evaluation, she was noted to have a blood pressure of 107/78 mm Hg and a pulse of 97/minute and normal. She was in no distress and had no pedal oedema. On cardiovascular examination, she had a palpable thrill at the left sternal border and a systolic murmur was auscultated in the left parasternal area. On examination of the respiratory system, she had good airway entry with no crackles.

INVESTIGATIONS

An echocardiogram revealed a mass described as a 6.0×3.0 cm blood filled sac compressing the left atrium and anterolateral aspect of the left ventricle with communication to the aortic root. Otherwise, structurally and functionally, the heart was normal. The differential diagnosis included an aortic root pseudoaneurysm, a left main coronary aneurysm and a left main coronary fistula. The patient later had a CT scan of the chest with contrast which showed aneurysmal dilatation of the left main coronary artery.

Serological investigation for the antinuclear antibody was negative.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis included an aortic root pseudoaneurysm, a left main coronary aneurysm and a left main coronary fistula on echocardiogram.

OUTCOME AND FOLLOW-UP

She remained asymptomatic during pregnancy and was awaiting coronary angiography after delivery to further investigate the aneurysm. She was placed on aspirin, but no anticoagulants were prescribed. She subsequently defaulted from clinic 11 months postdelivery.

Five years later, the patient presented to a local health facility with a sudden onset of chest pain and subsequent unresponsiveness. She died despite resuscitation and was referred for an autopsy.

At autopsy, notable examination findings included peripheral cyanosis with no obvious clubbing of the fingers. There was no chest wall deformity and no pedal oedema. The significant findings on dissection were confined to the cardiovascular system. The pericardial sac contained 900 mL of frank blood with clots. The heart weighed 360 g. A left coronary aneurysmal sac measuring 10.0×9.0 cm was identified. It communicated proximally with the ostium of the left coronary artery. Distally, it was continuous with the distal portion of the left anterior descending coronary artery (figure 1). The inner lining of the sac was thickened and there was a rupture measuring 7.0 cm in length. No thrombus was seen in the sac. The cardiac chambers appeared normal with no septal defects. The valves were also unremarkable.

DISCUSSION

A CAA is defined as a localised dilatation of the coronary artery exceeding 1.5–2.0 times the diameter of the adjacent segments.¹ The size at which it is called a giant CAA has not been established

To cite: Blackman A, Mohammed W. *BMJ Case Rep* Published Online First: [please include Day Month Year]. doi:10.1136/bcr-2017-222816

Figure 1 Heart with the aneurysmal sac opened at upper left. The upper probe enters the sac through the left coronary ostium and the lower probe shows the communication with the left anterior descending artery.

with suggestions of sizes exceeding 2.0 and 5.0 cm being used.² The shape can be described as saccular or fusiform and it can be multiple or single.^{1 3 4}

CAAs have a reported incidence of 0.02%–0.04% in the general population⁵ with giant CAAs having a prevalence of 0.02% in a large surgical population.² The artery most commonly involved is the right main coronary artery followed by the left circumflex, the left anterior descending artery and the left main coronary artery.^{1 3 4} It is noted that 57 of the 79 cases reported in the medical literature between 1812 and 1960 were men. In one study, the average age at autopsy diagnosis was 54.4 years.⁶ The presence of CAAs in patients <33 years is considered as congenital.¹ Congenital aneurysms have less sex predilection.⁷

There are multiple aetiologies implicated in the formation of CAAs. The most common aetiology documented in adults is atherosclerosis with other causes including congenital, vasculitis, mucocutaneous lymph node syndrome, connective tissue disorders, infectious, iatrogenic and trauma and illicit drug use.^{1–3 5 6 8}

In this case, congenital malformation is the aetiology of interest given the lack of history and evidence to support vasculitis, connective tissue disease and iatrogenic causes. There is also a lack of evidence of atherosclerosis at autopsy to support the most common aetiology.

Congenital malformations of the coronary arteries include absence, abnormal distribution, aneurysm formation, fistulous communication, aberrant origin, stenosis and occlusion. In young patients, the underlying aetiology is assumed to be congenital but it is uncertain as to whether the aneurysm is present at birth or develops later.⁷

The pathogenesis for CAAs is not known; but there are a range of hypotheses generated based on the aetiology. It is well recognised in connective tissue diseases such as Marfan syndrome that aneurysmal formation can occur. Marfan syndrome is an

autosomal-dominant disorder in which there is an association with mutations in the gene that encodes fibrillin which is structurally related to and interacts with a family of latent transforming growth factor beta (TGF- β) and holds it in an inactive complex.⁶ It is also been suggested that abnormalities in the TGF- β signalling pathway can cause development of Marfan syndrome or that abnormalities in the receptors of TGF- β can cause phenotypes similar to Marfan syndrome.⁹ Loeys-Dietz syndrome is a recently described autosomal-dominant syndrome in which there is aortic aneurysmal formation and the underlying pathogenesis is thought to be mutations in the TGF- β receptors.¹⁰ Given the similarities between these syndromes, it has been suggested that an excess of active TGF- β could be responsible for the aneurysmal formation. Excessive active TGF- β is thought to lead to cystic medial degeneration commonly seen in Marfan syndrome.⁶ Unfortunately, the histological features of the aneurysm wall in our case showed predominantly fibrosis with no evidence of cystic medial degeneration.

The presentation of CAAs is variable but is mostly asymptomatic. Angina pectoris, myocardial infarction, sudden death, fistula formation, haemopericardium, tamponade, compression of surrounding structures and congestive heart failure are some of the known presentations. In patients with giant CAAs, the presentation can also be one of superior vena cava syndromes or a mediastinal mass being misdiagnosed as a thymoma or cardiac tumour. In one case report, the patient presented with a murmur.¹¹ This is similar to the patient in our case whose first presentation was a murmur but was otherwise asymptomatic until demise due to cardiac tamponade secondary to rupture of the CAA.

The gold-standard investigation is coronary angiography. However, in lieu of this, multislice CT is a feasible alternative as it assists in the delineation of the anatomy of the aneurysm and the adjacent structures.¹

It is important to note that due to the rarity of CAAs, standardised treatment measures have not been established and there are no guidelines supportive of medical versus surgical management in elective settings.³ Surgical methods include ligation and/or resection of the CAA with a distal bypass and also percutaneous stenting. Conservative management is inclusive of antiplatelet and anticoagulant therapy to prevent thrombus formation and embolisation.^{1–3} In our case, the patient received antiplatelet therapy in the form of aspirin. The patient defaulted from clinic after delays in obtaining coronary angiography and therefore there was no opportunity for surgical management.

It is recognised that giant CAAs carry a higher mortality.⁶ Possible sequelae of giant CAAs include thrombus formation with distal embolisation, fistula formation and rupture.¹¹ In our patient, the massive size of the aneurysm made rupture a more likely outcome.

Giant CAAs are rare entities and this case highlights its fatal outcome in a young female with a possible congenital aetiology. This CAA is one of the largest reported in the literature. Possible

Learning points

- ▶ Availability of investigative tools to adequately assess suspected coronary artery aneurysm (CAA) is critical for diagnosis.
- ▶ Timely intervention—surgical or conservative as appropriate—is important to avoid fatal complications.
- ▶ Where surgical intervention is delayed, active follow-up of patients with CAA may be necessary to prevent default from medical care.

factors contributing to the fatal outcome include delays in management and the eventual default from clinic.

Contributors AB was responsible for performing the autopsy, interpreting and reporting on the findings and preparing the final autopsy report; authored the draft of the case report and made necessary amendments. WM was the supervising pathologist at the autopsy who examined and confirmed the findings and determined the cause of death; assisted in the writing of the case report and suggested amendments to the case report, approving the final draft.

Funding This research received no specific grant from any funding agency in the public, commercial or not-for-profit sectors.

Competing interests None declared.

Patient consent Detail has been removed from this case description/these case descriptions to ensure anonymity. The editors and reviewers have seen the detailed information available and are satisfied that the information backs up the case the authors are making.

Provenance and peer review Not commissioned; externally peer reviewed.

© BMJ Publishing Group Ltd (unless otherwise stated in the text of the article) 2018. All rights reserved. No commercial use is permitted unless otherwise expressly granted.

REFERENCES

- 1 Dunder C, Tigen K, Pala S, *et al.* Congenital left main coronary artery aneurysm. *Cardiol J* 2011;18:430–3.
- 2 Lazar JF, Compton M, Li F, *et al.* Excising a giant: report of a 7-cm coronary artery aneurysm. *Tex Heart Inst J* 2013;40:173–5.
- 3 Tuncer E, Onsel Turk U, Alioglu E. Giant saccular aneurysm of the left main coronary artery. *J Geriatr Cardiol* 2013;10:110–2.
- 4 Chen YT, Hwang CL, Kan MN. Large, isolated, congenital aneurysm of the anterior descending coronary artery. *Br Heart J* 1993;70:274–5.
- 5 Keyser A, Hilker MK, Husser O, *et al.* Giant coronary aneurysms exceeding 5 cm in size. *Interact Cardiovasc Thorac Surg* 2012;15:33–6.
- 6 Nichols L, Lagana S, Parwani A. Coronary artery aneurysm: a review and hypothesis regarding etiology. *Arch Pathol Lab Med* 2008;132:823–8.
- 7 Seabra-Gomes R, Somerville J, Ross DN, *et al.* Congenital coronary artery aneurysms. *Br Heart J* 1974;36:329–35.
- 8 Ebina T, Ishikawa Y, Uchida K, *et al.* A case of giant coronary artery aneurysm and literature review. *J Cardiol* 2009;53:293–300.
- 9 Defendi GL. Genetics of Marfan Syndrome. *Medscape* 2017 <http://emedicine.medscape.com/article/946315-overview#a5> (cited 2017 Mar 23).
- 10 Loeys BL, Schwarze U, Holm T, *et al.* Aneurysm syndromes caused by mutations in the TGF- β receptor. *N Engl J Med Overseas Ed* 2006;355:788–98.
- 11 Crawley PD, Mahlow WJ, Huntsinger DR, *et al.* Giant coronary artery aneurysms: review and update. *Tex Heart Inst J* 2014;41:603–8.

Copyright 2018 BMJ Publishing Group. All rights reserved. For permission to reuse any of this content visit <http://group.bmj.com/group/rights-licensing/permissions>.
BMJ Case Report Fellows may re-use this article for personal use and teaching without any further permission.

Become a Fellow of BMJ Case Reports today and you can:

- ▶ Submit as many cases as you like
- ▶ Enjoy fast sympathetic peer review and rapid publication of accepted articles
- ▶ Access all the published articles
- ▶ Re-use any of the published material for personal use and teaching without further permission

For information on Institutional Fellowships contact consortiasales@bmjgroup.com

Visit casereports.bmj.com for more articles like this and to become a Fellow