CASE REPORT

Unlikely culprit: congenital middle aortic syndrome diagnosed in the sixth decade of life

Muhammad Sajawal Ali,¹ Stefan Tchernodirski,² Divyanshu Mohananey,¹
Ahya Sajawal Ali³

¹Department of Medicine, John H Stroger Hospital of Cook County, Chicago, Illinois, USA

²Department of Medicine, University of Illinois at Chicago College of Medicine, Chicago, Illinois, USA

³CMH Lahore Medical and Dental College, Lahore, Pakistan

Correspondence to
Dr Muhammad Sajawal Ali,
sajawalhotiana@gmail.com

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SUMMARY

A 58-year-old woman was admitted with acute heart failure. She had a long history of resistant hypertension, with an unremarkable work up for secondary causes in the past. Her brachial blood pressure was 210/70 mm Hg, with ankle blood pressure of 100/70 mm Hg. CT angiogram revealed marked narrowing of the descending thoracic aorta between the left subclavian artery and the diaphragm, consistent with middle aortic syndrome (MAS). She was initially managed with diuretics and antihypertensives. Subsequently thoracotomy revealed a severely hypoplastic segment of the descending aorta. The diseased segment was resected and aortic reconstruction performed. Histopathology showed fragmentation of the medial elastic fibres and fibrosis of the medial and intimal layers. These findings along with gross aortic hypoplasia and absence of features of Takayasu's arteritis, suggest that our patient had congenital MAS. The patient has done well since her surgery. We believe this is the first case of congenital MAS reported in the sixth decade of life.

BACKGROUND

Middle aortic syndrome (MAS) refers to the segmental narrowing of the descending thoracic and abdominal aorta.¹ The disease can be either congenital or acquired. The congenital form typically presents in the first or second decade of life.² We describe a case of congenital MAS diagnosed in the sixth decade of life; to the best of our knowledge it is the first such case described this late in life.

CASE PRESENTATION

A 58-year-old Hispanic woman was admitted to the general medicine service with progressive dyspnoea on exertion, orthopnoea and paroxysmal nocturnal dyspnoea for the last 2 months. Her medical history was significant for resistant arterial hypertension, diagnosed 20 years earlier. Records showed that her blood pressure had been difficult to control despite her compliance with furosemide and maximal doses of hydralazine, carvedilol, enalapril and nifedipine. Recently, extensive work up for secondary causes of hypertension including serum aldosterone and renin levels, serum and urine metanephrines, thyroid function tests, 24 hour urine-free cortisol and renal artery Doppler ultrasound, had all yielded normal results.

On admission the patient's brachial blood pressure was 210/70 mm Hg, heart rate was 83/min and respiratory rate was 15/min. Her oxygen saturation by pulse oximetry was 95% while breathing

ambient air. Loud holosystolic murmur was present along the entire precordium and was also audible at the interscapular region on the posterior thorax. Lungs were resonant to percussion, with bilateral lower lobe crepitations on auscultation. Lower extremity pulses were diminished with a significant radiofemoral delay. Ankle blood pressure was only 100/70 mm Hg on both sides. Similar blood pressure differences were obtained and urgently reported to us by the concerned nursing staff as well.

INVESTIGATIONS

Blood work up was significant for an elevated serum creatinine of 1.8 mg/dL and blood urea nitrogen of 26 mg/dL. Her B-type natriuretic peptide was elevated at 172 pg/mL. Rest of the metabolic profile, complete blood count and urinalysis were unremarkable. ECG revealed normal sinus rhythm, left atrial enlargement and left ventricular hypertrophy. Transthoracic echocardiogram revealed an ejection fraction of 55–65% by visual assessment, grade 1 diastolic dysfunction and marked narrowing of the descending aorta distal to the take off of left subclavian artery (figure 1B).

Based on the above findings, MAS was suspected. CT angiogram revealed a 7 cm long segment of aortic narrowing, starting distal to the left subclavian take off (figure 1A). At its narrowest the aortic diameter was 6 mm and established a normal calibre distal to the diaphragm (figure 2). Prominent internal mammary artery collaterals were noted. Cardiac catheterisation revealed

Figure 1 (A) Sagittal reconstruction of the CT angiogram shows the point of maximal aortic narrowing (arrow). (B) Two-dimensional echocardiography redemonstrates narrowing of the descending aorta (arrow).

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normal coronary arteries and a peak-to-peak pressure gradient of 55 mm Hg across the descending thoracic aorta. Erythrocyte sedimentation rate and C reactive protein were within normal limits.

DIFFERENTIAL DIAGNOSIS

Our diagnosis was heart failure with preserved ejection fraction due to uncontrolled hypertension, caused by MAS. However, at this point we were still not sure whether our patient's MAS was congenital or acquired.

TREATMENT

Initial treatment consisted of intravenous furosemide to control volume overload and pulmonary oedema, and blood pressure control with hydralazine, carvedilol, extended-release nifedipine and later, enalapril. This resulted in subjective and objective improvement.

Cardiothoracic surgical consultation was obtained; given the long stenotic aortic segment and the fact that our patient was having organ dysfunction from uncontrolled hypertension, surgical intervention was recommended. The patient underwent thoracotomy; intraoperatively, severe hypoplasia of the descending thoracic aorta, extending from the left subclavian artery to just above the diaphragm was found. Intercostal arteries T4-T11 were divided and aortic reconstruction was performed with 20 mm Hemashield Dacron graft. Somatosensory evoked

potentials were monitored to look for possible spinal cord ischaemia; they did not change during the procedure. The patient recovered from the surgery without complications. Histopathology of the resected segment revealed thinning of the arterial wall. There was marked fibrosis of medial and intimal layers (figure 3A). The medial fibres were fragmented and in disarray (figure 3B).

OUTCOME AND FOLLOW-UP

It has been 3 years since the patient underwent surgery and she continues to do well. She has excellent blood pressure control and is currently requiring only one antihypertensive drug. Of note she is off diuretics and has not developed any heart failure symptoms since. The patient's renal function has also remained within normal limits.

DISCUSSION

The term MAS was first used by Sen *et al*³ in 1963 to describe subisthmal blocks in the descending thoracic and abdominal aorta. Other authors have also used the terms midaortic syndrome or midaortic dysplastic syndrome to describe such lesions.^{4 5} Ninety-eight per cent of all aortic blockages are at the level of ductus arteriosus, MAS is different from the usual aortic coarctations since it usually affects more distal aortic segments and can also potentially affect long segments, up to several centimetres at a stretch.⁶ Principal clinical features of MAS are refractory hypertension, heart failure and lower extremity claudication.⁷ Interestingly our patient did not report any claudication symptoms.

MAS can either be congenital or acquired. 61–90% of the cases are congenital and these can be associated with other disorders such as fetal alcohol syndrome, neurofibromatosis, Williams syndrome, Alagille syndrome, congenital rubella syndrome, tuberous sclerosis and Schimmelpenning-Feuerstein-Mims syndrome.^{6 8–10} In its acquired form the most common aetiology of MAS is Takayasu's arteritis.¹¹ For the congenital form of the disease, different series have reported a mean age presentation ranging from 2.7 to 19.5 years.^{2 12} These patients have mean life expectancy of 35 years and have a very high mortality by the time they reach fifth decade of their lives.^{13–15} This reduced life expectancy is usually because of complications from uncontrolled hypertension such as coronary artery disease, heart failure, cerebrovascular accidents, renal failure, etc.¹⁶ The acquired form of the disease, on the other hand can

Figure 2 Axial view of the CT angiogram with the narrowest segment of the descending aorta (arrow).

Figure 3 (A) (elastic stain, $\times 4$) Arrow points to the intimal fibrosis. (B) (elastic stain, $\times 20$) Fragmentation and disarray of the medial elastic fibres can be seen.

potentially present later in life. Kim *et al*⁴ described five patients in the sixth decade of their lives, with MAS secondary to Takayasu's arteritis.

Our case is unique since to the best of our knowledge, this is the first time that congenital form of MAS is being described this late in life. There are several factors that point towards our patient's disease process being congenital. Firstly, our patient had no history of systemic symptoms such as fever, malaise, arthralgias, weight loss, rash, etc. These symptoms are often present especially in early phase of Takayasu's arteritis.¹⁷ Secondly, Takayasu's arteritis typically affects the aortic arch and other large vessels taking off from the arch.¹⁸ In our patient there was no clinical or radiological evidence of aortic arch involvement. Thirdly, the disorganised medial elastic fibres, medial and intimal layer fibrosis and absence of inflammatory cells on histopathology as seen in our patient are in line with the previous descriptions of congenital aortic coarctations.^{19–21} Fourthly, our patient's diseased aortic segment was grossly hypoplastic which means that the insult must have occurred at a very early stage of development before the aorta had a chance to mature. On the other hand, acquired forms of MAS have been described as having normal external diameter, with luminal narrowing resulting from adventitial and intimal thickening.³

We hypothesise that our patient had some aortic narrowing since childhood but likely survived into adulthood by developing collaterals, which for most part permitted sufficient blood supply to the lower half of her body. However, with ageing two overlaying processes, led to further luminal narrowing of an already hypoplastic aortic segment. Firstly, the turbulent blood flow at the level of the aortic lesion caused accelerated atherosclerosis. Secondly, the abnormal intima in congenital coarctations causes increased fibrin deposition. This process has already been well described by Kennedy *et al*.²² Therefore, the superimposed atherosclerosis and fibrin deposition in what was already a congenitally narrow aortic segment, diminished the blood flow to a point whereby the collaterals proved inadequate. At this stage our patient's hypertension worsened, which in turn precipitated heart failure.

MAS is a complex disease and the treatment strategies have to be individualised. Most authors have however, favoured surgical intervention.²³ The surgical intervention typically involves aorto-aortic bypass of the affected segment.²⁴ This can be accomplished using either a woven Dacron graft or a Gore-tex, anastomosed to the descending aorta distal to the obstruction in an end-to-side fashion using a 4–0 polypropylene continuous suture. The graft is then passed proximally and anastomosed to the ascending aorta in a similar fashion.²⁵ The ascending aorta is the preferred site for proximal anastomosis because of technical ease and excellent exposure. If the patient is young an oversized graft must be used to allow for the linear growth without kinking. In addition to aorto-aortic bypass, other surgical interventions include patch aortoplasty and primary aortic repair after aortic lengthening.²⁶ Short-term complications of surgical intervention include bleeding and thrombosis while long-term complications include graft infection, restenosis, degradation of the material, enteric fistulas, etc.²⁷ For the patients who have small isolated diseased aortic segments or those who are not surgical candidates, endovascular intervention is an option. Unfortunately, with endovascular interventions long-term success is low and about half of these patients may end up needing a second intervention in a year's time.²⁶

Finally, our case also demonstrates the importance of a good physical examination. Our patient had resistant

hypertension for 20 years and a pretty exhaustive work up had been carried for secondary causes. Perhaps a thorough examination of lower extremity pulses earlier on could have alerted clinicians to the possibility of an obstructive aortic lesion. This would have expedited the diagnosis and institution of appropriate treatment.

Learning points

- Congenital forms of middle aortic syndrome (MAS) and other obstructive lesions of the aorta can rarely go undiagnosed late in to adult life.
- MAS and obstructive lesions of the aorta should always be considered as differentials for resistant hypertension in the absence of alternative explanations.
- Simple physical examination manoeuvres such as checking lower extremity pulses and blood pressure can yield valuable diagnostic clues.
- Surgery is the treatment of choice for MAS, and results in good blood pressure control and preservation of organ function.

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