CAROTID ARTERY DISSECTION IN A CASE OF TURNER'S SYNDROME

DR. INDERPREET SINGH, SCF,
DR. JOY SINGHAL, IMT-1,
DR ASOKE DATTA, CONSULTANT
STROKE MEDICINE

NHS Stockport NHS Foundation Trust

INTRODUCTION

Turner's syndrome (TS) is a genetic disorder resulting from complete or partial absence of one of the X chromosomes. It occurs in approximately 1 in 2,500 live-born females and is associated with short stature, gonadal dysgenesis, and a range of other phenotypic features. Cardiovascular manifestations are particularly common, with up to 50% of patients experiencing congenital heart defects, aortic dilation, and aortic dissection. However, the presentation of stroke due to carotid/vertebral artery dissection in patients with TS is rare.

CASE PRESENTATION

- A known Turner's syndrome mosaic in her early forty's presented to the emergency department due to signs and symptoms suggestive of a cerebral vascular event. She was a non-smoker and consumed 9 units of alcohol per week. She was a Year 3 School teacher. She had two successive visits to
- the emergency department. She presented the first time due to worsening of her asthma, palpitations and tingling sensations affecting hands. She was tachycardic, HR 140. Chest Xray was suggestive of a vascular abnormality of the aortic root. As seen in Figure 1, Aortic root dilatation was suspected which warranted further investigations.
- CT Angiogram [Figure 2] showed the right sided aortic arch having retro-tracheal component with aortic aneurysm involving the proximal descending aorta. Patient was discussed with Cardiothoracic surgery and was deemed to be a candidate for non-urgent surgery and to be followed up later. Patient was discharged home.

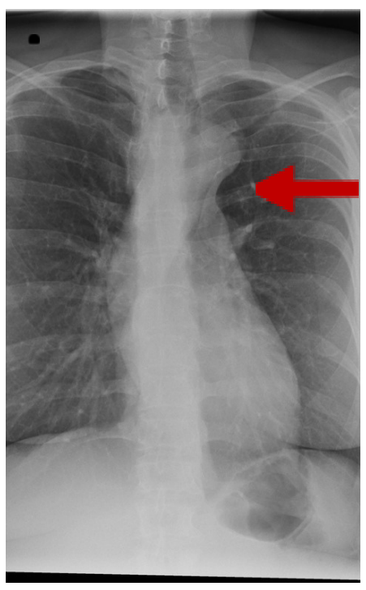


Figure 3.CT scan showing loss of grey-white matter differentiation in post central gyrus

Figure 1. PA Chest X-Ray showing Aortic root dilatation

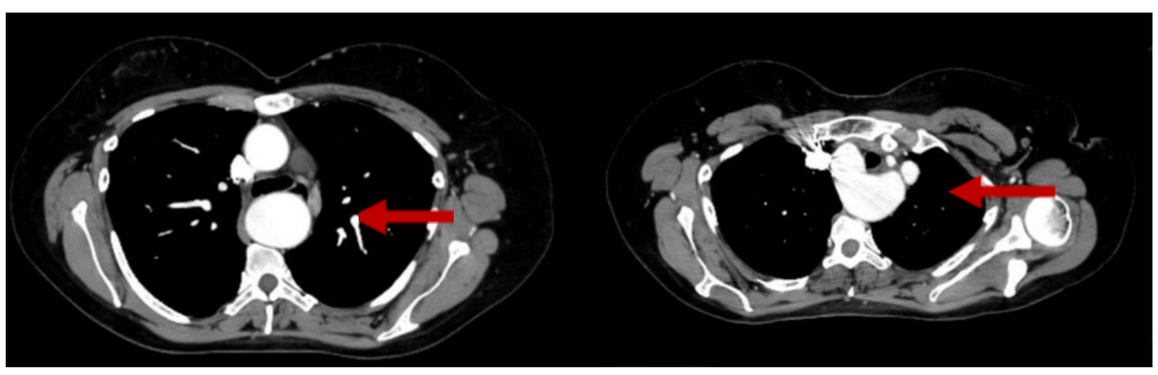


Figure 2. CT Angiogram showing aortic aneurysm of 45 mm

Three days later, she presented in the hyper acute stroke unit complaining of sensations of pins and needles in her right arm along with weakness in right upper and lower limb. On examination, her GCS was 15/15 and NIHSS was 6. An urgent plain CT head revealed loss of grey-white matter differentiation in left post central gyrus.

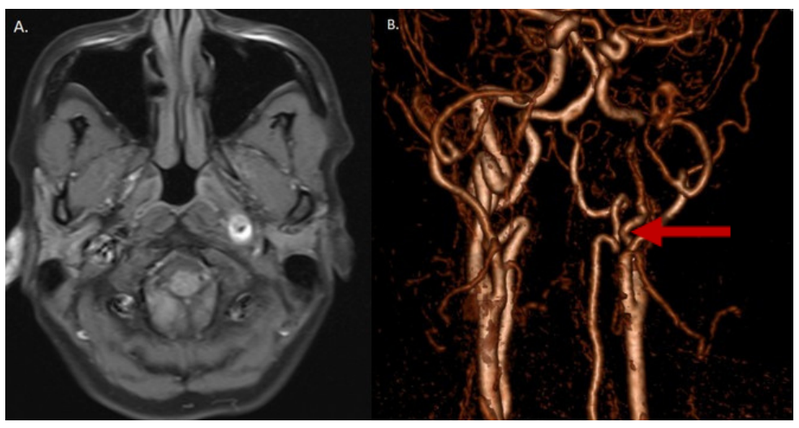
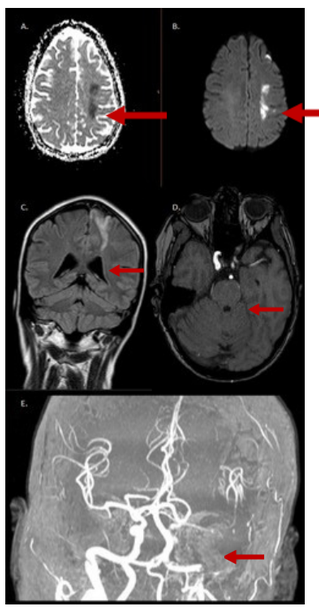


Figure 3 MR Angiography of Carotids. A.Left ICA dissection. B.3D reconstruction of the vessels



- ECHO showed dilated descending aortic root (4.4cm), normal bi-ventricular function with no significant valvular abnormalities. USG Carotids didn't show any
- abnormality. Initial laboratory investigations, including complete blood count, electrolytes, lipid profile, renal and liver function tests, coagulation profile, and lipid panel were within normal limits apart from haemoglobin of 119 g/L and slight but persistently raised CRP within the range of 18- 30 mg/L. Thyroid function test, Ferritin levels, Vitamin B12 levels, Bone profile and Endocrine panel unremarkable. Luteinising Hormone < 0.1, FSH 0.8 and oestradiol 43.
- After initial assessment, patient was admitted in stroke ward. She was started on Aspirin 300mg and betablocker due to tachycardia. She underwent comprehensive physical and occupational rehabilitation and was discharged with minimal residual weakness.

Figure 4 .MRI scan of brain confirming stroke. Red arrows for area of interest. A & B:Diffusion weighted and ADC images showing acute stroke. C. FLAIR-LONGTR sequence showing the extent of infarction. D.T2W-TSE showing partially occluded MCA, possibly due to thrombus/emboli. E.S3Di-MC showing 3d visualisation of the vessels.

DISCUSSION

This case report describes a rare presentation of stroke in a patient with Turner's syndrome. While aortic dissection and other cardiovascular complications are well recognized in patients with TS, the association between TS and cerebral infarction due to vessel dissection has been infrequently reported in the literature. Dissection has been associated with dilation (in the relevant vessel), along with other risk factors such as hypertension, bicuspid aortic valve and coarctation of aorta, none of which were present in the patient. This leans towards a possibility of a systemic vasculopathy presenting with dissection of ICA and contributing to the dilatation of the aorta. Widespread vasculopathy has been proposed, leading to dilatation of arteries beyond the aorta—carotids and brachial artery. This could have been the case in our patient, in which undetected carotid artery dilatation contributed to the dissection. Reasons for this "vasculopathy" are not clearly understood; composition and remodelling defects of connective tissue, abnormal smooth muscle contractility or inflammatory processes (mildly raised CRP in our patient without any obvious infectious process) have been theorised. These defects can be exacerbated by other features of TS like oestrogen deficiency (normal in our patient), hypertension and atherosclerosis. Females with TS are also found to have increased intimal media thickness, an indirect indicator of atherosclerosis

CONCLUSION

The pathophysiology underlying the increased risk of vascular complications in patients with TS remains incompletely understood. However, structural abnormalities of the aortic media, impaired vascular smooth muscle function and endothelial dysfunction have been implicated. In addition, the absence of a second X chromosome may lead to haploinsufficiency of genes critical for vascular development and maintenance, predisposing these patients to vascular complications.

In managing patients with TS who develop stroke, it is important to consider potential underlying aetiologies, such as genetic connective tissue disorders and vasculopathies. Early detection and aggressive management of cardiovascular risk factors are essential to minimize future events