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IMAGES IN CARDIOLOGY

Cardiology Journal
2011, Vol. 18, No. 5, pp. 568–569
10.5603/CJ.2011.0016

ISSN 1897–5593

Interrupted aorta in mosaic Turner syndrome

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A 28 year-old female with a four year history of primary infertility, oligomenorrhoea and menorrhagia presented to the sub-fertility clinic at our institution for further investigations. Her short stature (141 cm) and obesity (73 kg; body mass index 36.7 kg/m^2) prompted karyotype testing which confirmed mosaic Turner syndrome.

Otherwise, she had had poorly controlled hypertension for many years and was currently being prescribed labetalol for presumed primary hypertension. Her blood pressure remained elevated at 158/ /104 mm Hg, but attempts to add other antihypertensive medications had not been tolerated due to troublesome side effects. Echocardiography had previously shown a bicuspid aortic valve, but no left ventricular hypertrophy or other structural abnormality.

Due to concerns about potential secondary causes for hypertension, she proceeded to magnetic resonance imaging of her aorta. This indeed demonstrated a post-ductal interrupted aorta (Fig. 1) with extensive collateralization and enlargement of the internal mammary arteries (Fig. 2). Currently, she is awaiting surgical repair of the interrupted aorta with fertility treatment postponed until after surgery.

Cardiovascular disease is the commonest cause of death in Turner syndrome, with life expectancy reduced by approximately ten years [1, 2]. Aortic valve disease (particularly bicuspid valve morphology) and aortic coarctation are the commonest cardiovascular anomalies. Aortic root dilatation, aortic dissection, hypertension, mitral valve prolapse,

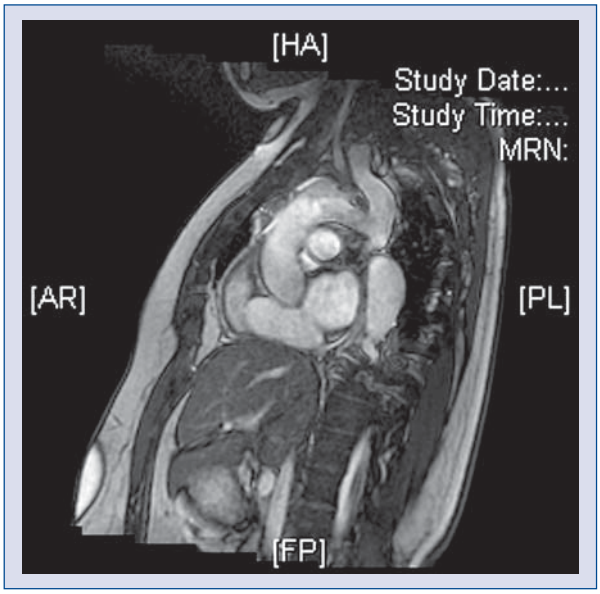

Figure 1. Magnetic resonance imaging showing a complete interruption of the descending aorta.

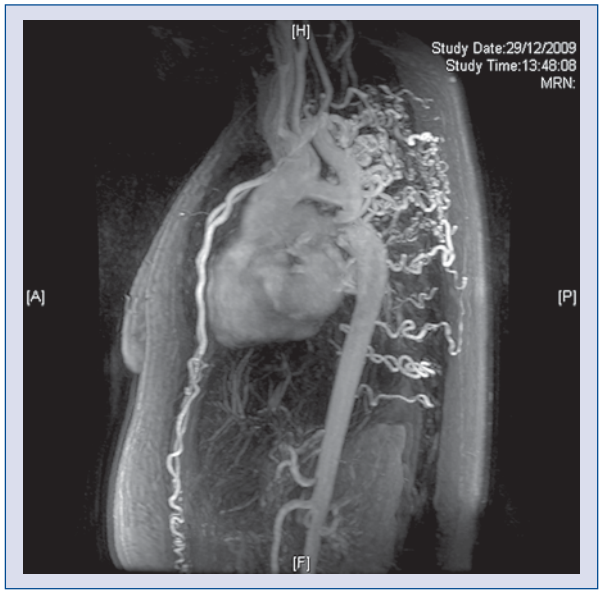

Figure 2. Magnetic resonance angiography showing extensive intercostal collateralization and enlargement of the internal mammary arteries.

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Received: 12.11.2010 Accepted: 10.01.2011

partial anomalous venous drainage and ischemic heart disease are also encountered more commonly than in the general population. However, the prevalance of cardiac anomalies differs considerably between females with X chromosome monosomy (45X) and those with mosaicism (45X//46XX) [3].

The prevalence of cardiovascular malformations is about 38% in monosomy and 11% in mosaicism. Aortic valve disease and coarctation occur at frequencies of 28% and 17% in monosomy, compared to 7% and 2% in mosaicism, respectively. Complete aortic interruption, rather than coarctation, has rarely been described in Turner syndrome [4] and we believe this is the first reported case in a mosaic patient.

Acknowledgements

The authors do not report any conflict of interest regarding this work.

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