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## **Aortic dissection and moyamoya disease in Turner syndrome.**

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### **Abstract**

Aortic dilation and dissection are well-recognized cardiac abnormalities in women with Turner syndrome (TS), although the underlying pathophysiology is not fully understood. We report on a 46-year-old Hispanic woman who was previously diagnosed with moyamoya disease on magnetic resonance imaging after a presentation with stroke-like symptoms. Her features were consistent with TS and chromosome analysis revealed mosaicism in which 17% of the cells showed a pseudoisodicentric Y chromosome: 45,X (25)/46,X psu idic (Y)(11.2) (5). A preceding screening transthoracic echocardiogram had shown a bicuspid aortic valve (BAV) with an aortic diameter of 3.2 cm; at the time of moyamoya diagnosis, the aorta was 3.5 cm with mild aortic stenosis and mild aortic regurgitation. Four years later, the patient had had an acute aortic dissection, Stanford type A, which was repaired successfully. This case report is the third individual with TS associated with moyamoya disease and the first associated with dissection. The small number of cases does not allow detailed analysis other than noting patient age (two older than 40 years), karyotype (two others associated with isochromosome Xq), and associated cardiac risk factors (one with BAV). Although this may be a chance occurrence, we hypothesize that moyamoya disease could be a manifestation of the vasculopathy in TS. (c) 2010 Wiley-Liss, Inc.

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