


CARDIOVASCULAR FLASHLIGHT

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A rare case of ascending aortic stenosis

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A 48-year-old woman was referred to an in-hospital cardiology evaluation after performing a transthoracic echocardiography (TTE) that indicated a severe aortic valvular stenosis. She had a cognitive delay, short stature and long-term symptoms of fatigue for moderate exertion. No history of syncope or angina.

The TTE was repeated and revealed a tricuspid aortic valve with mild calcification, maintaining good opening; a left ventricular obstacle, apparently with a supra-valvular location, with a maximum velocity of 5.6 m/s and a mean gradient of 85 mmHg; left ventricular hypertrophy with good systolic function (Panels A and B). A transtoesophageal echocardiogram confirmed the good opening of the aortic valve (Panel C), but the proximal ascending aorta was poorly visualized. The cardiac magnetic resonance imaging confirmed the severe supra-valvular aortic stenosis, with nearly interruption of the aortic root (Panel D).

The case was discussed by a multidisciplinary team, having reached the probable diagnosis of Williams syndrome. This disease affects the elastin gene, leading to a generalized arteriopathy with diffuse or localized stenosis of large- or medium-sized arteries. The ascending aorta above the valve and the pulmonary arteries are one of the most affected locations. The cognitive delay and the short stature are characteristics of this syndrome. The patient underwent surgical correction with Brom’s technique — symmetric aortoplasty with three patches that expand Valsalva sinus (Panel E).

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