Severe aortic regurgitation in Ehlers-Danlos syndrome type IV

A 47 year old white man was hospitalised for cardiac examination. A blowing diastolic heart murmur prompted us to perform echocardiography during which a severe aortic regurgitation caused by an enlarged aortic root accompanied by a dilated ascending aorta (maximal diameter 52 mm) was visualised (panels A–C). No findings indicated mitral valve prolapse.

The patient’s clinical features (panels D and E) together with his family history (his father died from a distended abdomen after cholecystectomy, a spontaneous intestinal perforation caused his brother’s death, and his sister died suddenly of a ruptured iliac artery) suggested a hereditary connective tissue disorder such as Marfan syndrome or Ehlers-Danlos syndrome (EDS). Indeed the subsequent genetic examination (panel F) confirmed a causative mutation in the COL3A1 gene coding for type III procollagen found in EDS type IV. This rare autosomal dominant disorder leads to an impaired synthesis, secretion, or structure of collagen type III. Since type III collagen is crucial for the development of such organs as the cardiovascular system, intestine, and skin this disorder is clinically characterised by a vulnerable, translucent skin and by premature death from vascular and bowel rupture. Interestingly aortic regurgitation is assumed to be extremely rare in association with EDS type IV.

After ruling out a coronary artery disease the patient received a valve bearing conduit of the ascending aorta. After this pre-emptive operation he recovered uneventfully.

The conspicuous habitus (facial features combined with a fragile, bruisable and translucent skin, panels D and E) guided the subsequent diagnostic steps. Hyperelasticity of skin and joints was absent. Panel F depicts the pedigree of the index patient.
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*Heart* 2005 91: 126
doi: 10.1136/hrt.2004.035097

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