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Hereditary thoracic aortic disease associated with cysteine substitution c.937T > G p.(Cys313Gly) in *FBN1*

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A 56-year-old male was diagnosed with a type A aortic dissection, followed by a type B dissection 3 years later. There were no other signs indicating a familial connective tissue disorder. Pathogenic variant c.937T > G p.(Cys313Gly) in *FBN1* [(NM_000138.4), Online Mendelian Inheritance in Man (OMIM) entry *134797] was identified by DNA testing, consistent with Marfan syndrome (OMIM entry #154700). The variant was identified in 21 out of 53 tested relatives (Fig. 1). A thoracic aortic aneurysm was diagnosed in eight relatives carrying the variant, three of whom met the criteria for preventive surgery. One of the deceased obligate carriers probably had a thoracic aortic aneurysm. Most mutation carriers had a systemic score [1] of zero or one, although the highest score was four. As illustrated by this image, *FBN1* variant c.937T > G p.(Cys313Gly) can cause isolated aortic disease. Timely recognition of individuals with a pathogenic *FBN1* variant is highly important, as it enables the prevention of severe cardiovascular complications [2, 3].

Conflict of interest E. Overwater, K. Van Rossum, M.J.H. Baars, A. Maugeri and A.C. Houweling declare that they have no competing interests.

Ethical standards Written informed consent for publication was obtained from all the carriers of FBN1 variant c.937T > G p.(Cys313Gly).

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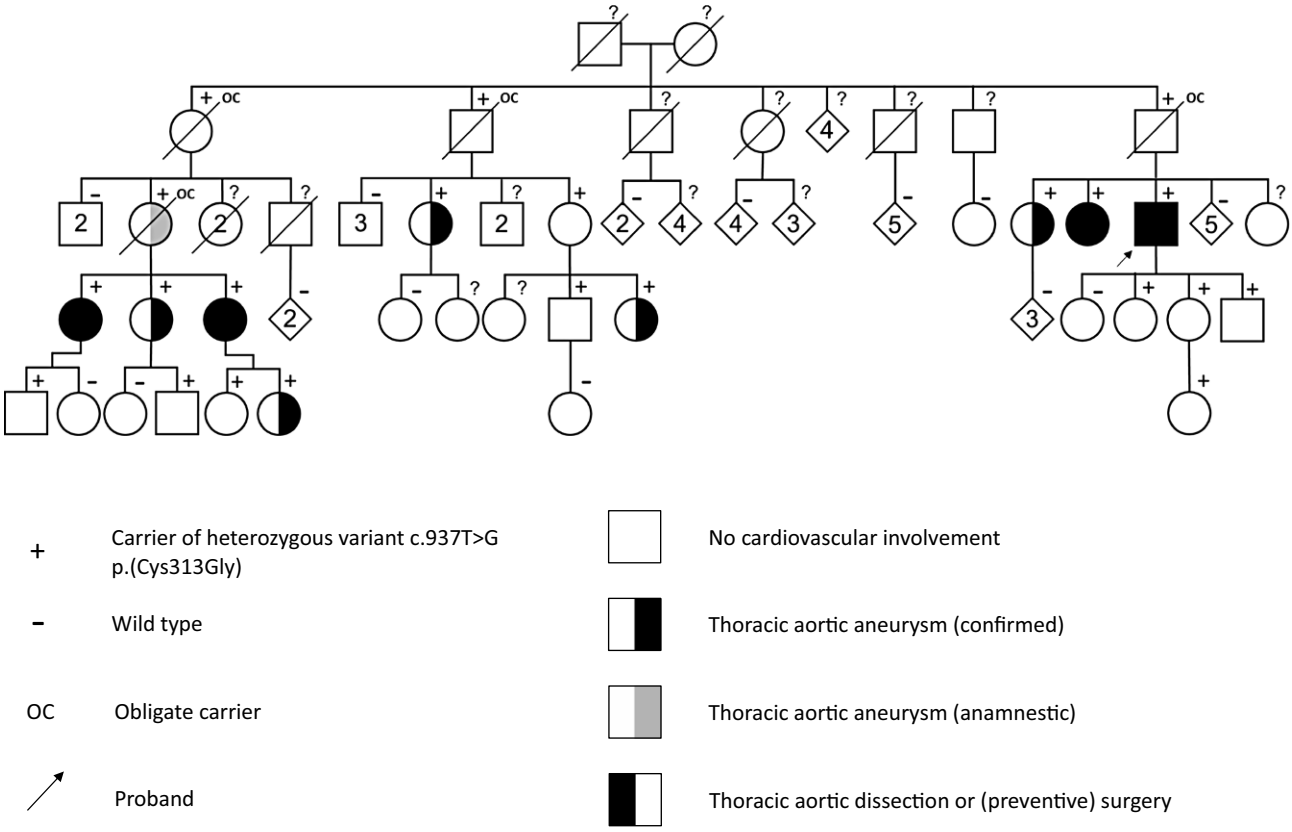

Fig. 1 Pedigree depicting thoracic aortic aneurysms and/or dissections. *Squares* represent males, *circles* indicate females