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Genotype-phenotype linkage in Marfan syndrome: are FBN1 variants related to prognosis?

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Introduction: Marfan Syndrome (MS) is an autosomal inheritance disorder in which aortic dilatation, that may lead to aortic aneurysm or disection, is the main cause of morbidity and mortality. Variants in Fibrilline-1 (FBN1) are found in more tan 90% of MS cases.

Purpose: Our aim was to establish the possible relationship between genotype and phenotype in a big cohort of patients with suspected MS.

Methods: We conducted a prospective cohorts study in which all FBN1 variant carriers who were attended in our Familial Aortopathies Unit between 2010 and 2016 were included.

Results: Variants in FBN1 were found in 90 patients among 56 non-related families. Of a total of 56 FBN1 variants, 32 (57.15%) have been for the first time reported in our study. 30% of patients had been surgically intervened due to aortic aneurysm or type A disection. A majority of the cohort with aortic events presented truncating variants (nonsense or frameshift) (69%, vs 24.1% missense and 6.9% intronic), and they happened at a younger age than patients with missense variants (median age 33.5 vs 39.5 years-old, respectively).

Conclusions: Patients with truncating mutations presented a higher incidence of aortic events, and they ocurred at a younger age. Thus, genetic findings may be important for risk stratification and clinical manage in patients with suspected MS.