

Primary Non-Aortic Lesions Are Not Rare in Marfan Syndrome and Are Associated with Aortic Dissection Independently of Age

PURPOSE: The study sought to estimate the prevalence of primary non-aortic lesions (PNAL) unrelated to extension of aortic dissection (AD) in a cohort of patients with Marfan syndrome (MFS).

METHODS: Adult patients presenting with pathogenic FBN1 mutations and an available pan-aortic contrast-enhanced CTA in eight French MFS clinics from April to October 2018 were included. Clinical and radiological data, particularly the presence of aortic lesions and PNAL (including aneurysm and ectasia), were retrospectively analyzed.

RESULTS: Out of 138 patients, 28 (20.3%) had PNAL. In total, 27 aneurysms in 13 patients and 41 ectasias in 19 patients were reported mainly in the subclavian, iliac, and vertebral segments. Four patients (31%) with aneurysms and none with ectasia required prophylactic intervention during follow-up (median: 46 months). In multivariate analysis, factors associated with PNAL were history of AD (OR = 3.9, 95%CI: 1.3-12.1, $p = 0.018$), history of previous descending aortic surgery (OR = 10.3, 95%CI: 2.2-48.3, $p = 0.003$) and age (per 10 years OR = 1.6, 95%CI: 1.1-2.4, $p = 0.008$).

CONCLUSION: PNAL is not rare in MFS patients with evolutive aortic disease. Natural history may differ between aneurysms and ectasia, emphasizing the need for standardized definitions and systematic screening for PNAL.