

Simple Renal Cysts in Marfan Syndrome: Prevalence and Association With Aortic Events

BACKGROUND: A link between simple renal cysts (SRCs) and aortic aneurysms or dissection has been reported in the general population. Marfan syndrome (MFS) is associated with severe aortic disease, but very few data on SRCs exist in this population.

OBJECTIVES: The objectives were to evaluate: 1) the prevalence of SRCs in patients with MFS, compared to matched controls; and 2) the association between SRCs and aortic events in patients with MFS.

METHODS: Consecutive patients with MFS ascertained by a pathogenic variant in the fibrillin-1 gene who underwent complete computed tomography scans at our institution were included and matched 1:1 for age and sex with controls.

RESULTS: Between 2010 and 2016, 131 patients with MFS and 131 controls were included. The mean age was 40 ± 14 years, with 42% women. SRC prevalence was higher in patients with MFS: 41% vs 21% in controls ($P < 0.0001$). SRC prevalence increased with aortic disease severity: 59% in patients with dissection, 43% in patients with aortic aneurysm surgery, and 19% in patients with MFS but without aortic events, similar to 21% in controls ($P < 0.009$). In multivariable analysis, SRC presence in patients with MFS was independently associated with aortic dissection (adjusted OR: 2.30 [95% CI:1.00-5.32]; $P = 0.049$).

CONCLUSIONS: The prevalence of SRCs was significantly higher in patients with MFS compared to matched controls. SRCs were independently associated with aortic dissection in MFS. Prospective studies are needed to further evaluate whether SRCs could represent a marker of aortic disease severity in patients with MFS.