

The following research summary was prepared by ASF's Volunteer Research Program Chair and Board of Directors member, B. André Weinstock, PhD, MSAS.

Kamiar A, Alitter Q, Capcha JMC, et al. <u>Ascending aortic aneurysm and histopathology in Alport</u> <u>syndrome: a case report</u>. *BMC Nephrology*. Online 2023, October 12. **DOI**: https://doi.org/10.1186%2Fs12882-023-03345-5 (open access).

There is currently insufficient clinical evidence to confirm whether Alport syndrome (AS) is an independent risk factor for an aortic aneurysm. However, there is a growing body of tangential and anecdotal evidence that AS, as a collagen disease, may have some relationship with aortic disease that could be similar to other collagen disorders such as Ehlers-Danlos and Marfan syndrome.

The recently published Kamier 2023 case report presents a thorough survey of all cases of aneurysms in patients with AS that have been published to date. The survey attempts to provide context to the remarkable and unique histopathologic analysis of an aortic tissue sample that was obtained in 2021 from an AS patient who underwent elective repair of an ascending aortic aneurysm.

Alport Syndrome Foundation (ASF) and our educated and engaged patient community was essential in facilitating the effort that resulted in this case report and subsequent survey. ASF considers this paper to be the first small step in a larger systematic effort to understand the clinical risks of aortic disease in patients with AS. Therefore, ASF is committed to immediately prioritizing research funding and efforts towards evaluating whether and/or how much of a risk patients with AS may have of developing an aortic aneurysm.