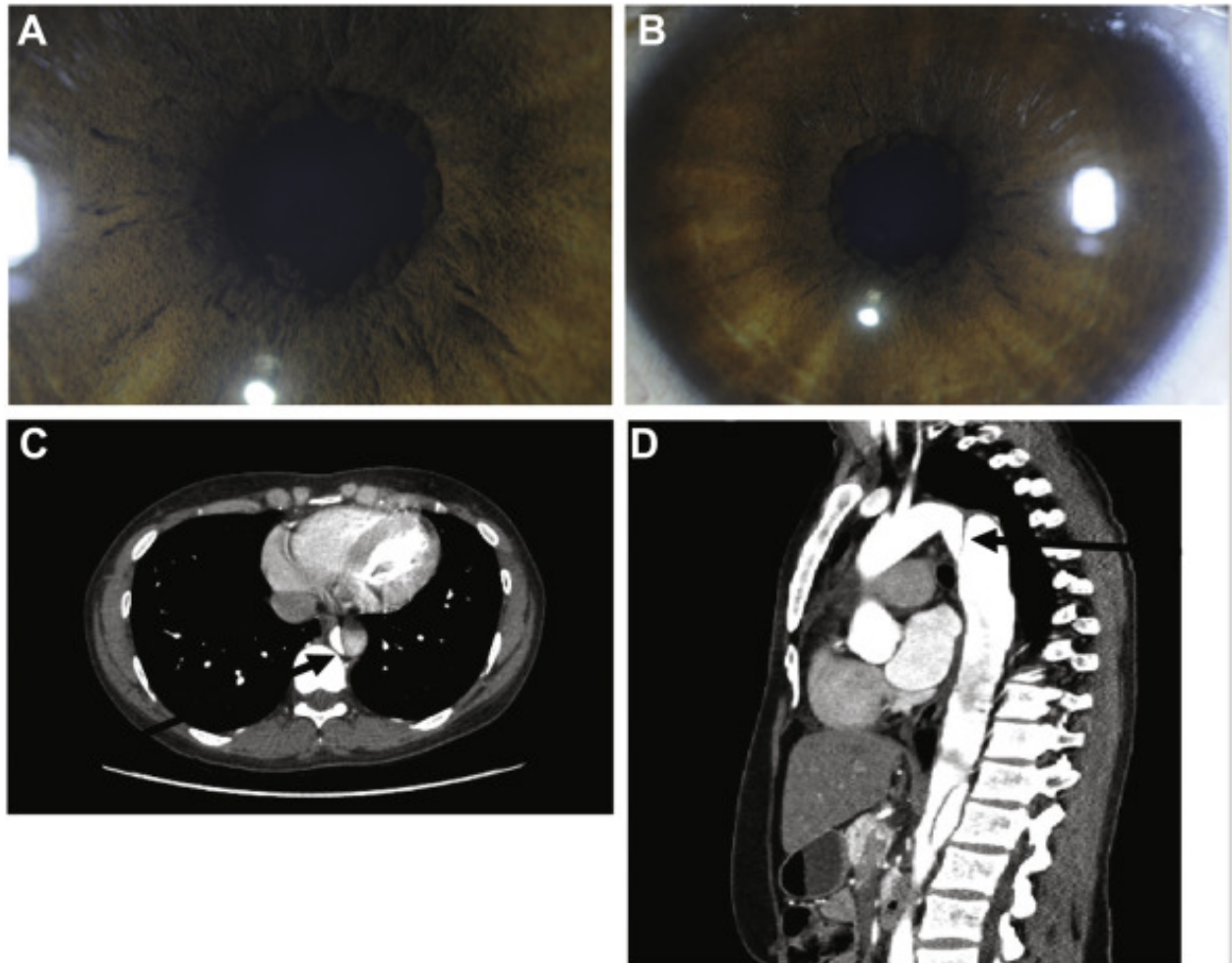


Iris Flocculi Investigated for Familial Thoracic Aortic Aneurysms and Dissections



Slit-lamp examination revealed bilateral iris flocculi (Fig A, right eye; Fig B, left eye), and pigment epithelial excrescences at the pupillary border in a 35-year-old patient. Computed tomography angiography with axial (Fig C) and sagittal (Fig D) views diagnosed a type B aortic aneurysm (right aberrant subclavian artery to infrarenal artery). The patient carried a heterozygous, pathogenic variant in the *ACTA2* gene (c.445C>T, p.Arg149Cys) that causes familial thoracic aortic aneurysms and dissections (TAAD). No other family members had TAAD. She did not have livedo reticularis, which is often

observed in this condition. Similar iris findings may occur in familial amyloidotic polyneuropathy and with chronic topical treatment using acetylcholinesterase inhibitors (Magnified version of Fig A-D is available online at www.aajournal.org).