A 20-year-old male presented with exertional dyspnoea and history of premature coronary artery disease in father and paternal uncle. Clinical examination revealed bilateral corneal arcus (Panel A) and blood pressure of 144/84 mmHg in the right arm and 138/82 mmHg in the left arm. Electrocardiogram showed left ventricular hypertrophy by voltage criteria (Panel B). A grade 4/6 ejection systolic murmur was noted in aortic area radiating to carotid arteries. Two-dimensional echocardiography was suggestive of concentric left ventricular hypertrophy, grade 1 diastolic dysfunction and supra-valvular tubular narrowing of ascending aorta aortic with pressure gradients of 40/28 mmHg across this segment (Panel C). Computed tomographic (CT) aortogram revealed diffuse atheromatous involvement of aortic root and ascending aorta as the cause of stenosis (Panels D, E, and F). Non-obstructive atheromatous plaque was also noted in distal left main coronary artery, with rest of the coronary system being normal (Panel G). Serum cholesterol levels were 500.2 mg/dL (normal range 130–200 mg/dL), serum LDL 449 mg/dL (normal range 65–130 mg/dL) and serum triglyceride level 97.9 mg/dL (normal range 20–150 mg/dL). A diagnosis of familial hypercholesterolemia causing supra-valvular aortic stenosis was made and intensive statin therapy was initiated.

Premature malignant atherosclerosis is a known complication of familial hypercholesterolemia. Supra-valvular aortic stenosis, although rare, is a well-known manifestation of homozygous familial hypercholesterolemia. Detailed history taking and clinical examination is indispensable in diagnosing this rare metabolic disorder and preventing catastrophic complications.

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