

Complex surgical management of supraaortic stenosis in familial homozygous hypercholesterolemia: Aortic root replacement without coronary reimplantation: A Case Report

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Case Report

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Abstract

Background:

Homozygous familial hypercholesterolemia (HoFH) is characterized by accelerated valvular and vascular calcification. Supravalvular aortic stenosis (SVAS) is a severe complication in HoFH, often necessitating complex surgical intervention.

Case presentation:

We report a 33-year-old female with HoFH and severe aortic and SVAS who underwent aortic root and ascending aorta replacement without coronary reimplantation. The surgery was successful, and recovery was uneventful.

Conclusion:

This case illustrates an uncommon surgical approach to the management of advanced aortic root and supravalvular disease in HoFH, with favorable postoperative outcomes. It highlights the technical challenges posed by extensive calcification and the need for individualized surgical strategies when conventional reconstructive options are not feasible.

BACKGROUND

HoFH is an uncommon potentially fatal genetic disorder predisposing to premature ASCVD [1]. Progressive calcification in patients with HoFH can result in significant valvular stenosis and SVAS [2]. Treatment modalities include surgical or percutaneous aortic valve replacement (AVR). Extensive calcification or supravalvular involvement often necessitates complex root replacement and coronary reimplantation, associated with higher morbidity and mortality [3].

We report the case of a 33-year-old woman with HoFH who underwent complex aortic root and ascending aorta replacement, AVR, along with MVR, and CABG. Coronary reimplantation was not performed owing to severe calcific involvement.

CASE PRESENTATION

A 33-year-old female diagnosed with HoFH at 10 years of age was referred for evaluation of dyspnea, palpitations, and fatigue. She had been lost to follow-up and reported poor compliance with prescribed treatment. She was receiving statin therapy only. Preoperative lipid profile showed an LDL-C level of 560 mg/dL, HDL-C of 42 mg/dL, total cholesterol of 611 mg/dL, and triglycerides of 43 mg/dL. Genetic confirmation of HoFH was not available. Upon presentation she was hemodynamically stable.

Echocardiography showed EF 45%, impaired LV function, aortic valve stenosis (mean/peak gradient 44 mmHg/70 mmHg, AVA 0.7 cm², DVI 0.2), severe ischemic mitral regurgitation (SOR = 0.3 cm²), and mild

tricuspid regurgitation.

CT of the chest showed massive circumferential aortic root and STJ calcification, with a diameter of 16 mm × 16.4 mm consistent with SVAS. (Figs. 1 and 2)

Coronary angiography demonstrated severe triple-vessel disease, including left main and right coronary ostial stenoses.

Carotid angiography showed bilateral atherosclerosis without critical stenosis.

Surgical Procedure:

A median sternotomy was performed, and bilateral skeletonized internal mammary arteries, left radial artery and saphenous vein were harvested.

Cardiopulmonary bypass was initiated with hypothermia at 32° C. Myocardial protection was achieved via cold blood cardioplegia into the aortic root, repeated every 15 to 20 minutes via the coronary sinus, the saphenous vein and radial artery grafts, after completion of the distal anastomoses. Four bypass grafts were performed: RIMA Y graft with LIMA to obtuse marginal, LIMA to LAD, saphenous vein to diagonal, and radial artery to right coronary artery.

On aortotomy, severe circumferential SVAS and root calcification were evident. A Bentall procedure was required. Before root replacement, the thickened mitral valve was excised and replaced with a 29 mm ATS mechanical prosthesis.

The Bentall procedure started with excision of the severely calcified ascending aorta and root to the annulus. Extensive debridement of the aortic valve annulus and excision of the calcified valve were performed to avoid a catastrophic perivalvular leak. Coronary reimplantation was impossible due to heavy calcification; both ostia were ligated, and myocardial perfusion secured by bypass grafts. The aortic valve, root, and ascending aorta were replaced with a 20 mm ATS mechanical valve and 24 mm Dacron graft.

Cross-clamp and bypass times were 231 and 277 minutes, respectively.

The patient was weaned from bypass with temporary inotropic support, extubated on postoperative day 3, and experienced transient atrial fibrillation managed with amiodarone. Echocardiography before discharge showed an ejection fraction of 40%, well-functioning aortic and mitral prostheses (mean gradients 7 mmHg and 8 mmHg), and no wall motion abnormalities. She was discharged in stable condition on day 13 and referred to an endocrinologist for follow up of the HoFH.

At 1.5 years of follow-up, the patient remained asymptomatic based on telephone assessment. Follow-up echocardiography demonstrated a left ventricular EF of 56% without regional wall motion abnormalities and normal function of both the aortic and mitral mechanical prostheses.

DISCUSSION

HoFH is characterized by aggressive ASCVD from early life [1]. Calcific deposits in the aortic root and cusps often cause SVAS, which markedly increases mortality risk [4].

However, due to the limited data available, the optimal treatment approach is largely expert-dependent. As far as we know, there have only been a few documented successful surgical procedures for SVAS in these patients. Among these, Kakuta et al. reported a patient with severe calcific involvement of the aortic root in whom standard coronary reconstruction was not fully feasible; the right coronary artery was ligated, while an 8-mm polytetrafluoroethylene graft was anastomosed to the left coronary button [3].

Another surgical option involves patch enlargement of the ascending aorta and the Valsalva sinus, which avoids the technically challenging dissection of the aortic root and reconstruction of the coronary buttons required in full root replacement procedures. However, enlargement of the aortic root necessitates a large longitudinal or spiral incision and suturing of the fragile, diseased aortic wall, making it more technically challenging than full root replacement using a valved conduit.

When the aortic annulus is of sufficient size to accommodate a prosthetic valve, resecting calcification at the sinotubular junction (STJ) and performing an aortic valve replacement is often the preferred approach. However, in cases where the annulus is narrow, alternative options include valve replacement with annular enlargement or complete root replacement [5–6].

In this case, upon cutting the aorta at the level of the ST junction, severe aortic stenosis and extensive calcification of the aortic root and annulus were noted. These findings made it unfeasible to replace the aortic valve without addressing the root and ascending aorta. Patch enlargement of the aortic root and annulus was also deemed impractical due to severe calcification, which rendered suturing a patch onto the diseased aortic tissue impossible. Consequently, an aortic root and aortic valve replacement procedure was performed, but without reimplantation of the coronary buttons, as they were heavily calcified. Despite the significant risk associated with not reimplanting the coronaries, this approach was chosen because four coronary bypasses were performed to ensure adequate myocardial perfusion. Nevertheless, complete dependence on bypass grafts for myocardial perfusion raises concerns regarding long-term graft patency and durability, and therefore careful long-term follow-up is required to assess the sustained reliability of this strategy.

Given the rarity of these cases, multicenter studies are needed to better define optimal management strategies and long-term outcomes.

Conclusion

This case highlights the complexity and challenges in the surgical management of extensive SVAS in HoFH, as well as the high-risk decision-making associated with not reimplanting the coronary buttons. It

is intended to demonstrate the clinical rational in this specific scenario and does not imply that this approach represents a new standard or technique to be routinely adopted.

Abbreviations

HoFH – Homozygous Familial Hypercholesterolemia

SVAS – Supravalvular Aortic Stenosis

ASCVD – Atherosclerotic Cardiovascular Disease

AVR – Aortic Valve Replacement

MVR – Mitral Valve Replacement

CABG – Coronary Artery Bypass Grafting

EF – Ejection Fraction

LV – Left Ventricle

CT – Computed Tomography

RIMA – Right Internal Mammary Artery

LIMA – Left Internal Mammary Artery

LAD – Left Anterior Descending (artery)

STJ – Sinotubular Junction

SOR – Surface of regurgitation

DVI – Dimensionless velocity index

Declarations

Participant Consent to Publish: Written informed consent was obtained from the patient for participation in the clinical case and for publication of the case report and any accompanying clinical information and images.

Author Contribution

Dr. Ibrahim Salah El Din collected the clinical information, performed the literature review, and drafted the manuscript. Dr. Ziad Khabbaz critically revised the manuscript, supervised the work, and approved the

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Figures

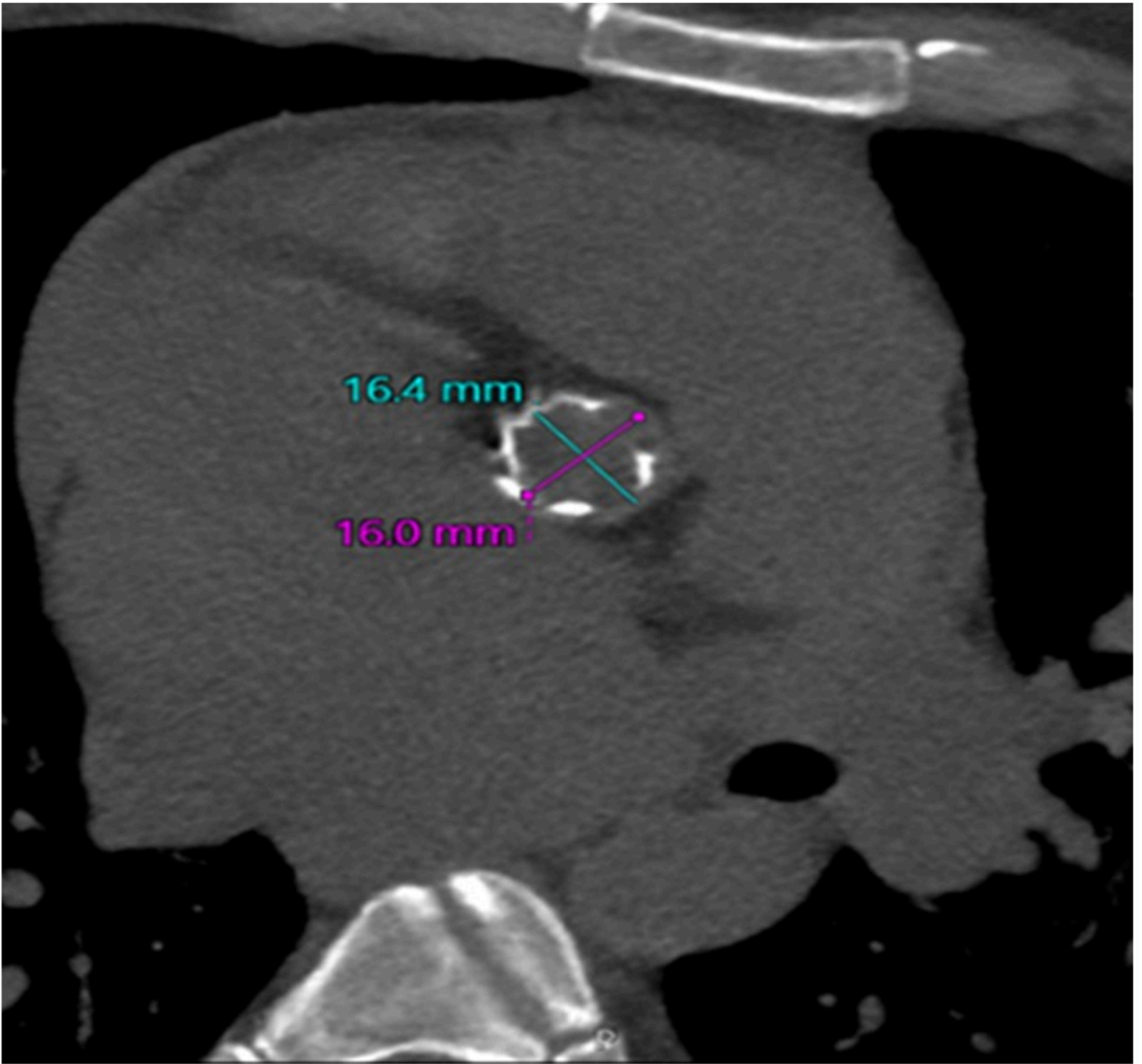


Figure 1

CT scan showing severe calcification of the STJ, with a diameter of 16 mm × 16.4 mm, consistent with SVAS.

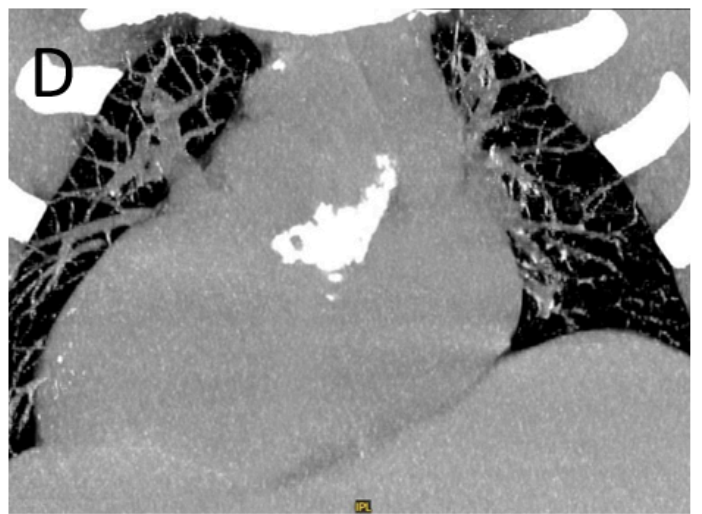
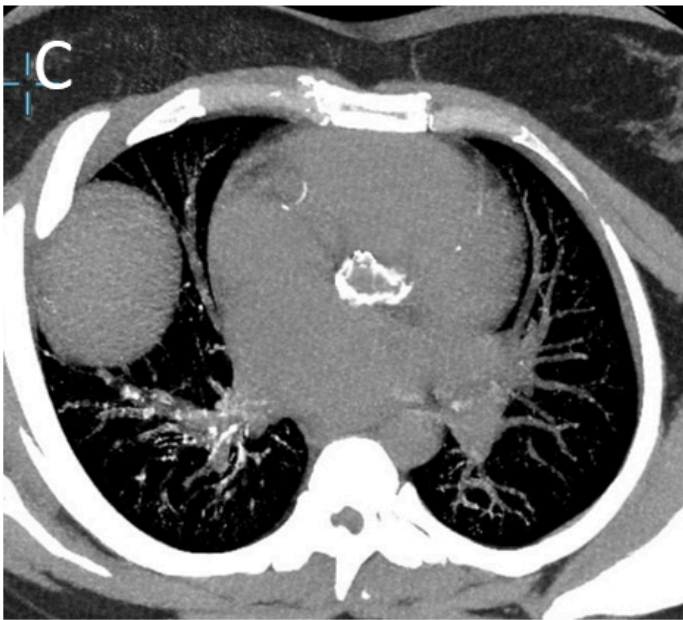
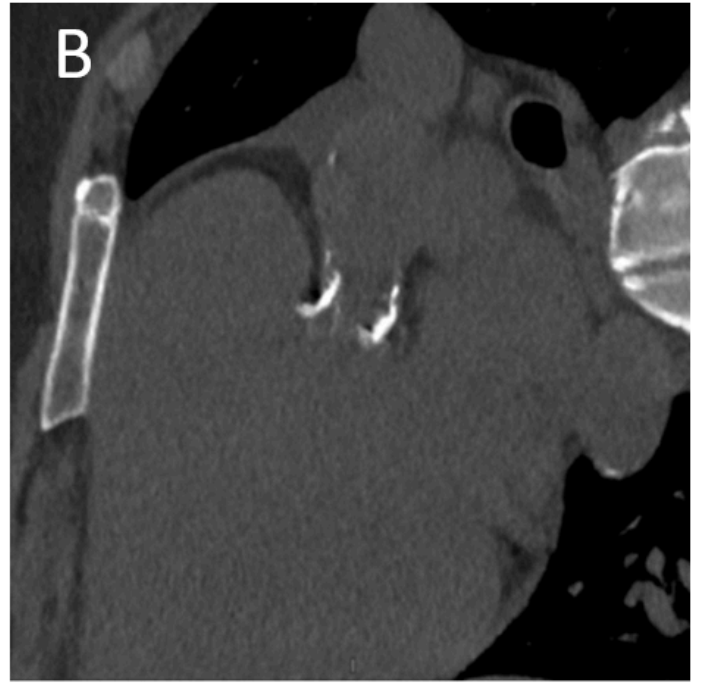


Figure 2

CT images revealing a severely calcified aortic root and STJ, highlighting the presence of significant SVAS due to circumferential calcific narrowing.