

# Aortic stenosis of a bicuspid aortic valve in a patient with Klippel–Feil syndrome: a case report

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## Background

Klippel–Feil syndrome (KFS) is a rare congenital anomaly of the cervical spine, which is associated with a number of cardiovascular malformations, including coarctation of the aorta, bicuspid aortic valve (BAoV), and aortic aneurysm. Operative management of aortic stenosis of a BAoV in a patient with KFS has not been previously reported.

## Case summary

A 54-year-old Caucasian woman with known KFS presented to her local hospital for elective cholecystectomy. An ejection systolic murmur was found incidentally on preoperative workup, which was confirmed to be due to a severely stenosed BAoV. The cholecystectomy was cancelled, and the patient was referred to our centre and accepted for surgical aortic valve replacement (AVR) based on symptomatic and prognostic grounds. Anaesthetic review of cervical spine imaging showed fusion of the C2–C6 vertebral bodies and a desiccated bulging disc at C4–C5 but no significant foraminal narrowing in the lower cervical spine. Valve replacement with a mechanical aortic prosthesis resulted in an uneventful recovery and the patient was discharged home to follow-up.

## Discussion

We report the first case of severe aortic valve stenosis requiring AVR in a Klippel–Feil patient, in whom the aortic valve was confirmed to be bicuspid. This report provides further evidence of an association of KFS with BAoV and strengthens the case for screening and follow-up of KFS patients for BAoV and other cardiovascular pathologies, the consequences of which may be serious.

## Keywords

Klippel–Feil syndrome • Bicuspid aortic valve • Aortic stenosis • Aortic valve replacement • Case report

## Learning points

- Patients with Klippel–Feil syndrome have reported association with cardiovascular abnormalities, including ventricular septal defects, coarctation of the aorta, hypoplastic aortic arch, aortic root aneurysm, and insertion of pulmonary vessels.
- This case strengthens the association of Klippel–Feil with bicuspid aortic valves (BAoVs) and highlights the need for routine screening for BAoVs and other cardiovascular abnormalities, with careful follow-up and timely intervention for associated pathologies.

## Introduction

Klippel–Feil syndrome (KFS) was first clinically reported in 1912 by Klippel and Feil. Their findings of a triad of short or absent neck, severe limitation of head movement, and low posterior hairline characterize this syndrome which otherwise occurs in patients with a heterogeneous profile and is present in 1 in up to 50 000 live births.<sup>1</sup> Many anomalies are associated with KFS including, most commonly, spina bifida, deafness, and scoliosis. In addition, musculoskeletal, gastrointestinal, and cardiovascular systems may be involved. Cardiac anomalies are rare.

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Herein, we report the first of case a KFS patient presenting with severe aortic stenosis in a bicuspid aortic valve (BAoV) requiring valve replacement. This case strengthens the possibility of an association of KFS with BAoV.

## Timeline

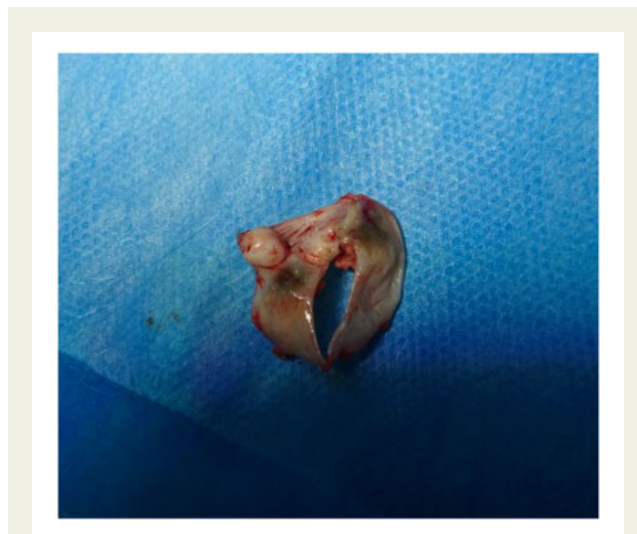
March 2017	Elective cholecystectomy cancellation due to incidental ejection systolic murmur finding on preoperative workup.
April 2017	Cardiac echo confirmed severely stenosed aortic valve with a peak gradient of 96 mmHg, a mean of 60 mmHg, and a valve area of 0.80 cm <sup>2</sup> . Moderate left ventricular function impairment.
June 2017	Preoperative consultant anaesthetist review due to potential problems with intubation from Klippel–Feil deformity.
August 2017	Elective surgical aortic valve replacement (mechanical prosthesis).
August 2017	Patient discharged and well at follow-up.

## Case presentation

A 54-year-old Caucasian female patient with a known history of KFS was admitted for elective cholecystectomy at her local hospital. She admitted to mild breathlessness and chest pains on exertion. Past medical history revealed a long series of investigations and interventions for her spinal disorders. Previous spinal magnetic resonance imaging showed deformity of the cervical spine, with fusion of the C2–C6 vertebral bodies and a desiccated bulging disc at C4–C5 but no significant foraminal narrowing in the lower cervical spine.

On examination, she had a loud systolic murmur with no evidence of heart failure clinically. Cardiac echo confirmed severely stenosed aortic valve with a peak gradient of 96 mmHg, a mean of 60 mmHg, and a valve area of 0.80 cm<sup>2</sup>. The aortic valve was reported to be bicuspid. There were no intracardiac shunts or other cardiac pathology. The aortic root and ascending aorta were of normal dimensions. The left ventricular ejection fraction was moderately impaired at 40%. Elective cholecystectomy was deferred, and the patient was referred for aortic valve replacement (AVR). She was accepted for surgery on symptomatic and prognostic grounds. Preoperative coronary angiography showed unobstructed coronary arteries. Anaesthetic review with spinal X-rays were requested to ensure safe tracheal intubation for general anaesthesia due to the nature of the patient's spinal deformity (Figure 1).

Preoperative TOE (transoesophageal echocardiogram) confirmed a heavily calcified and stenosed BAoV with a normal aortic root and ascending aorta diameters (Figure 2). The left ventricle was hypertrophied. At operation, the severely stenosed aortic valve was confirmed to be a Type 0 BAoV. The patient underwent AVR with a 21 mm mechanical prosthesis (Sorin Biomedica, Italy). The operation was uneventful, and the patient was



**Figure 1** Preoperative lateral cervical X-ray image with multiple cervical vertebral body deformities and fusions consistent with the known Klippel–Feil syndrome.

discharged home on the 6th post-operative day. A transthoracic echocardiogram prior to discharge confirmed no aortic regurgitation or para-prosthetic leak.

Histopathology of the excised valve leaflets revealed fibrosis, myxoid degeneration, areas of calcification, and neo-vascularization (Figure 3). Unfortunately, no genetic testing was performed to establish the possible link between BAoV and KFS.

At 24 months post-operatively, the patient is well and without complications.

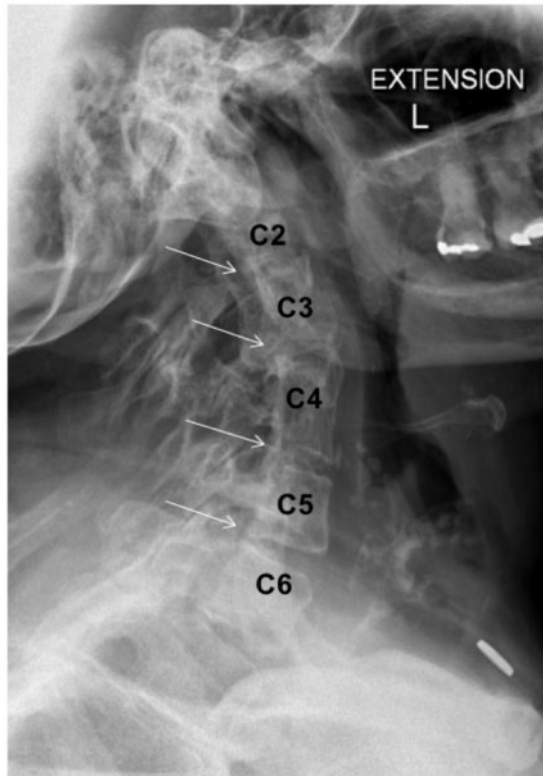
## Discussion and conclusion

Klippel–Feil syndrome is an inherently rare congenital anomaly. The syndrome is defined as the congenital fusion of two or more cervical vertebrae and is postulated to result from faulty segmentation along the embryo's axis during the 2nd to 8th week of gestation.

The importance of recognizing KFS lies in the fact that there is a strong association with many other abnormalities. Cardiovascular anomalies have been recognized in 4.4–14% of cases, most commonly ventricular septal defects, coarctation of the aorta, hypoplastic aortic arch, aortic root aneurysm, and abnormal insertion of pulmonary vessels.<sup>2,3</sup> Although no definite sex predominance has been proven for the congenital cervical anomaly, the cardiovascular association is more common in females.

Presentation of aortic stenosis in BAoV in the 5–6th decade is well-documented.<sup>4</sup> It is possible that the BAoV pathology and KFS occurred concurrently and that the KFS may not be associated with the BAoV. Thus, genetic testing in this patient would have been helpful to identify markers of BAoV and aortic valve calcification such as altered NOTCH-1 gene expression.<sup>5</sup> Unfortunately, the patient declined.

Sabol et al.<sup>6</sup> report the case of an asymptomatic 51-year-old woman with KFS found to have brachial blood pressure asymmetry on routine check-up. Subsequent TOE identified a BAoV with mild



**Figure 2** Mid-oesophageal right ventricular outflow transoesophageal echocardiogram image demonstrating a bicuspid aortic valve with calcification of the leaflets. (A) Diastole and (B) systole.

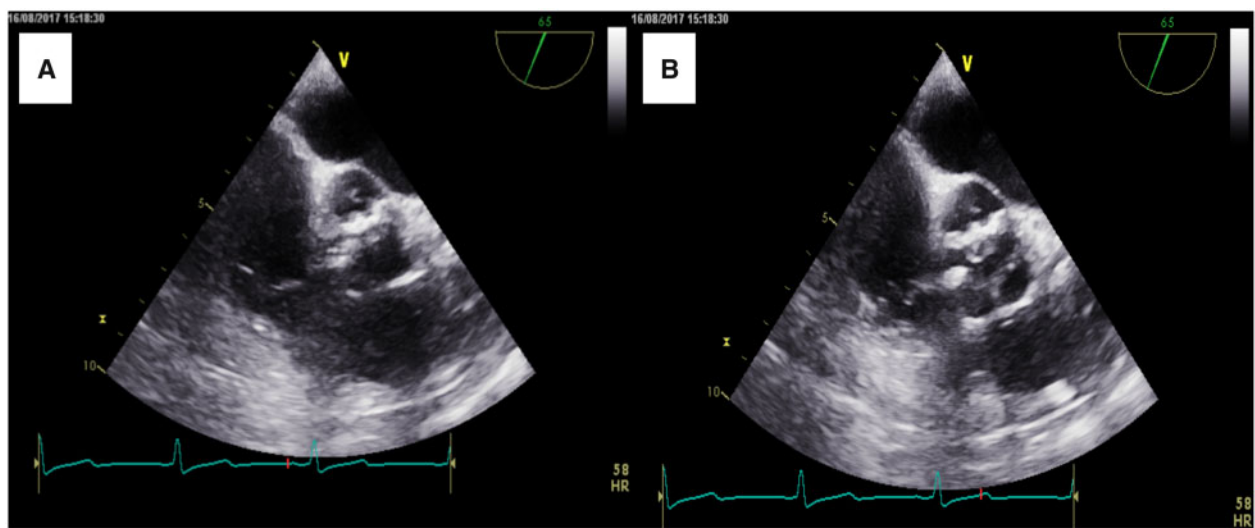
aortic regurgitation. Computed tomography identified an aneurysm of the ascending aorta, hypoplastic aortic arch, and aortic coarctation at the level of the left subclavian artery. She underwent a successful repair of the hypoplastic aortic arch, coarctation, and aneurysm. The valve was not replaced. The authors do not report long-term follow-up of the patient with regards to her BAoV and possible progression to aortic stenosis.

In only the second report of these pathologies, Bayam *et al.*<sup>7</sup> described the case of a 21-year-old man with known KFS under routine investigation for hypertension. On clinical examination, brachial blood pressure asymmetry was noted. Subsequent TOE revealed a BAoV, an aortic aneurysm of sinus of Valsalva (5.8 cm) and coarctation of the arch of aorta with post-dilation of the descending aorta. The patient underwent an aortic valve and ascending aorta replacement, but the reasons for the AVR are not described.

Further reports of additional cardiovascular abnormalities in patients with known KFS have been described in younger age groups in a case series by Bejiqi *et al.*<sup>3</sup> of four children aged from 2 days to 12 years.

This is the first report of a KFS patient with severe aortic stenosis of a BAoV, requiring AVR. Although there is a strong and well-documented association of ascending aortic aneurysm with BAoV,<sup>8,9</sup> the ascending aorta in our patient was of normal calibre with no associated coarctation of the aorta or other cardiovascular pathology. Nevertheless, early recognition of the mentioned cardiovascular abnormalities is of significant importance in all patients, as they may result in life-threatening complications.

Cardiovascular abnormalities in patients with KFS can present at a wide age range. Regular screening for BAoV and other cardiovascular abnormalities with thorough clinical examination and cardiac echo investigations should be considered in all patients with KFS.



**Figure 3** Post-operative excised stenosed bicuspid aortic valve.

## Lead author biography



Rory F.L. Hammond was born in 1995 in the UK. He completed a BSc in Experiment Pathology at Barts and the London Medical School and is currently in his final year of medicine. He has an interest in Cardiothoracic Surgery, in particular surgical cancellations and valvular heart disease. He has been involved in research at the Barts Heart Centre.

## Supplementary material

[Supplementary material](#) is available at *European Heart Journal - Case Reports* online.

**Slide sets:** A fully edited slide set detailing this case and suitable for local presentation is available online as [Supplementary data](#).

**Consent:** The authors confirm that written consent for submission and publication of this case report including images and asso-

ciated text have been obtained from the patient in line with COPE guidance.

**Conflict of interest:** none declared.

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