Surgical management of a hypoplastic distal aortic arch and coarctation of aorta in a patient with Klippel–Feil syndrome, ascending aortic aneurysm and bicuspid aortic valve

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Abstract
Klippel–Feil syndrome has been associated with cardiovascular malformations, but only 3 cases have been reported to be associated with aortic coarctation and surgical management is not defined. A 51-year old woman with Klippel–Feil syndrome associated with an aneurysm of the ascending aorta, hypoplastic aortic arch and aortic coarctation at the level of the left subclavian artery presented with shortness of breath 2 years after diagnosis. Imaging identified interim development of a 7.2-cm aneurysm at the level of the aortic coarctation. She underwent surgical repair with a Dacron interposition graft under hypothermic circulatory arrest. She continues to do well 18 months following repair.

Keywords: Klippel–Feil syndrome • Hypoplastic distal arch • Aortic coarctation

INTRODUCTION
Klippel–Feil syndrome was originally defined by Klippel and Feil in 1912 [1]. This syndrome occurs in a heterogeneous group of patients unified only by the presence of segmentation defects of the cervical spine, often associated with a triad of short neck, low posterior hairline with neck webbing and decreased cervical motion. A hypothesis of vascular aetiology has been proposed that posits that this anomaly results from defective blood flow through the vertebral arteries during embryogenesis. Based on one study, 25% of patients with occipito-cervical anomalies were found to have significant defects in their vertebral artery anatomy [2], lending support to this hypothesis. Many other malformations have been reported in association with Klippel–Feil syndrome, including rib anomalies, syringomyelia and cardiac malformations. It remains unknown if these associated defects are part of the Klippel–Feil syndrome, or if they are manifestations of associated, but different disease processes. Cardiac anomalies are rare, seen only in 4–5% of patients afflicted. Aortic defects associated with Klippel–Feil syndrome are also rarely seen, with only 3 case reports available in the literature describing association of this syndrome with aortic coarctation [3, 4].

A standardized approach to surgical management of aortic defects in the setting of Klippel–Feil syndrome has not been defined, with only one study describing a Dacron patch angioplasty repair of aortic coarctation and associated hypoplastic aorta in a 13-year old boy with this syndrome [4]. Here, we describe a successful repair of a hypoplastic aortic arch, coarctation and aneurysm in a patient with Klippel–Feil syndrome.

CASE
A 51-year old woman with a diagnosis of Klippel–Feil syndrome initially presented to our hospital for further evaluation after a brachial blood pressure asymmetry was diagnosed by her primary care physician. Subsequent computed tomography (CT) angiography showed a 4.7-cm dilatation of the ascending aorta, hypoplastic distal arch and descending thoracic aorta and aortic coarctation at the level of origin of the left subclavian artery (Fig. 1A). Transoesophageal echocardiogram demonstrated a bicuspid aortic valve with mild aortic regurgitation. Significant aortic stenosis was excluded. Clinically, the patient was asymptomatic at the time and she opted against any surgical intervention.

Fourteen months later, the patient presented with worsening shortness of breath. Computer tomography angiography at this time demonstrated progression of disease, with new findings of dilatation of the descending thoracic aorta distal to the coarctation and an interim development of a 7.2 × 3.4 cm aneurysm (Fig. 1B). Owing to the anatomical progression of disease and development of new symptoms, a decision was made to proceed with surgical repair.

Open repair was undertaken via a left posterolateral thoracotomy in the fourth intercostal space. The resection of the diseased aortic segment and the proximal anastomosis (tubular graft 22 mm) just distal to the left carotid artery were performed under circulatory arrest. The distal anastomosis in the mid-thoracic aorta was then performed (Fig. 2A). One intercostal artery and the left subclavian artery were reimplanted to the graft. There were no postoperative complications and the patient was discharged...
home on Day 12. A CT scan prior to discharge demonstrated an intact repair of the thoracic aorta, as did a CT scan 6 months after her surgery (Fig. 2B). The ascending thoracic aortic aneurysm, which was not repaired during the surgery, remains stable at 4.7 cm and although future corrective surgery is indicated based on current European guidelines, the patient opted against it and at present remains asymptomatic.

DISCUSSION

Klippel–Feil syndrome is a rare congenital anomaly. While it has been reported to be associated with multiple other organ system anomalies, the putative factors linking the disease-defining cervical spine segmentation defects with the various other malformations frequently seen have not been identified.

Our patient initially presented with the Klippel–Fein syndrome–characteristic physical findings of cervical spine fusion (synostosis), neck deviations, hearing impairment, scoliosis, deformed rib cage and renal insufficiency. We also found several associated defects in the vasculature including aneurysm of the ascending aorta, hypoplastic aortic arch and aortic coarctation. In addition to Klippel–Feil syndrome, we consider a co-diagnosis of Turner syndrome; however, karyotype testing ruled out the latter. Fourteen months after the initial diagnosis, patient’s condition worsened and we found an interim development of a large aneurysm at the level of the aortic coarctation.

Open surgical repair of aortic coarctation has been well documented and includes interposition graft placement, patch aortoplasty, arch augmentation and an extra-anatomical repair using a jump graft bypass tunneled through the retromediastinum. An endovascular approach has also been explored and angioplasty with stenting has been shown to be a viable alternative to an open approach, especially in patients with high surgical risk, and although long-term data are currently lacking, the short-term results are encouraging.

In the setting of Klippel–Feil syndrome, the associated abnormalities, particularly the decreased mobility of the neck, intrinsic weakness of the aortic wall and association with tortuous and underdeveloped iliofemoral arterial segment make open surgical and endovascular repair challenging. Similarly, some of the difficulties we observed intraoperatively included stiffness of the neck, requiring bronchoscopy-guided intubation; narrow fourth intercostal space, necessitating partial fourth rib resection and small bilateral iliac arteries, which precluded our attempting endovascular repair.

The presence of hypoplastic aortic arch and aneurysm at the level of coarctation excluded the possibility of an extra-anatomical bypass as an option and we opted for a repair by interposition grafting. Intraoperatively, the aneurysm was found to be cephalad to the coarctation of the aorta. Owing to prominent scarring, we were unable to detect any local abnormalities, such as a weakened or dilated aortic wall, to explain either the pseudoaneurysm or the aneurysm formation. We have performed a macroscopic inspection only.
CONCLUSION

Surgical solution to aortic malformations in the setting of Klippel-Feil syndrome remains undefined and will likely need to be determined on a case-by-case basis. Our patient with this syndrome suffered from several cardiovascular anomalies and presented with worsening of symptoms. Our surgical approach was dictated by patient’s anatomical features.

Conflict of interest: none declared.

REFERENCES