THE TEXAS HEART INSTITUTE® JOURNAL

Case Reports

Mitral Valve Regurgitation in Klippel-Feil Syndrome With Related Thoracic Deformity

Yuki Endo, MD, PhD[1,2]; Yoshiei Shimamura, MD, PhD[1]; Hiroshi Niinami, MD, PhD[2]

[1]Department of Cardiovascular Surgery, Saitama City Hospital, Saitama City, Saitama, Japan
[2]Department of Cardiovascular Surgery, Tokyo Women's Medical University Hospital, Tokyo, Japan

Abstract

Klippel-Feil syndrome, characterized by congenital fusion of any 2 or more cervical vertebrae, is a rare disorder in which skeletal and other organ system–related abnormalities have been reported. This article reports a case of mitral valve regurgitation in a patient with Klippel-Feil syndrome and related thoracic deformity who underwent mitral valvuloplasty. Postoperatively, the mitral valve regurgitation disappeared, and there has been no recurrence for 3 years. This case highlights mitral valvuloplasty via median sternotomy as an excellent treatment for mitral valve regurgitation in a patient with thoracic deformity related to Klippel-Feil syndrome.

Keywords: Mitral valve insufficiency; Klippel-Feil syndrome; thoracic surgery; funnel chest

Case Report

Presentation and Physical Examination

The patient is a 30-year-old man with a heart murmur that was identified during a physical examination. He was referred to the reporting hospital for detailed examination and treatment. A transthoracic echocardiogram showed a prolapse of the median P2 segment of the posterior mitral valve leaflet and moderate to severe mitral valve regurgitation. At hospital admission, his height was 152.0 cm, his body weight was 40.3 kg, his body temperature was 36.3 °C, his pulse was 63/min, his blood pressure was 108/56 mm Hg, and his oxygen saturation was 100% on room air. Pulmonary sounds were clear, with no crackles; heart sounds were regular, with a systolic murmur. The patient's physical characteristics included a straight neck, a lowered occipital hairline, restricted neck motion, and pectus excavatum (Fig. 1A and Fig. 1B), which indicated Klippel-Feil syndnrome (KFS). There was no edema in the legs. Plain chest radiography revealed a cardiothoracic ratio of 51%, with slight cardiac dilation. An electrocardiogram revealed sinus rhythm, with a heart rate of 52/min and nonspecific ST-T segment changes. Blood chemistry tests showed a white blood cell count of 9.99 × 103/μL, a hemoglobin value of 14.5 g/dL, a platelet count of 26.6 × 104/μL, a creatine kinase value of 81 U/L, a lactate dehydrogenase value of 122 U/L, a C-reactive protein value of 0.10 mg/dL, and an N-terminal pro–brain natriuretic peptide value of 31 pg/mL. Pulmonary testing showed a vital capacity of 3.11 L (83.8%) and a forced expiratory volume of 2.22 L (80.4%). A computed tomographic scan showed left axis deviation of the heart; dilatation of the left atrium, left ventricle, and right ventricle; and pleural effusion. Scoliosis was also observed; the vertebral body cavity was narrowed, and the posterior vertebral bodies at C1-C2, C4-C5, T3-T4, T9-T10, and L2-L3 as well as the spinous processes were fused (Fig. 2A and Fig. 2B). Because of the patient's young age, coronary artery evaluation was performed by coronary artery computed tomographic examination, but no obvious, clinically significant stenosis or coronary artery anomaly

Citation: Endo Y, Shimamura Y, Niinami H. Mitral valve regurgitation in Klippel-Feil syndrome with related thoracic deformity. *Tex Heart Inst J*. 2024;51(1):e238282. doi:10.14503/THIJ-23-8282
Corresponding author: Yuki Endo, Department of Cardiovascular Surgery, Saitama City Hospital, 2460 Mimuro, Midori-ku, Saitama City, Saitama 336-8522, Japan (endo.yuki@twmu.ac.jp)

was observed. Transthoracic and transesophageal echocardiograms showed a left ventricular ejection fraction of 57% (modified Simpson method), a left ventricular end-diastolic/end-systolic dimension of 54/34 mm, a deviation of the median P2 segment of the posterior mitral valve leaflet, and torn chordae tendineae. Clear reversal of blood flow was also observed, with a vena contracta of 6 mm, and moderate to severe mitral valve regurgitation was shown.

Medical History

The patient had had a heart murmur since high school. He had a history of hospitalization for short stature in childhood but no history of treatment for heart disease.

Technique

The mitral valve repair was performed via a median sternotomy because of the short sternal-intervertebral distance of 49 mm (Haller index, 4.92) at the left atrium, which made minimally invasive cardiac surgery difficult to perform via the right intercostal space. Anticoagulation was not performed preoperatively because there were no cardiac arrhythmias.

After induction of general anesthesia and endotracheal intubation, a median sternotomy was performed. Cardiopulmonary bypass was established by cannulating the ascending aorta and devascularizing the superior and inferior vena cava. Cardiac arrest was obtained with antegrade cardioplegia. The mitral valve was observed via a right-left atrial approach, and the P2 segment of the posterior mitral valve leaflet was prolapsed as a result of a chordae tendinea tear (Fig. 3A and Fig. 3B). Partial quadrangular resection of the median posterior leaflet was performed, the annulus at the base of the resected cusp was sutured using 3-0 Prolene (Ethicon), and the leaflet at the commissure was sutured using 5-0 Prolene. Annuloplasty was performed with a 28-mm CG Future annuloplasty ring (Medtronic), and good valve coaptation was confirmed by a regurgitation test. The left atrium was closed using a continuous suture.

Key Points

- Characteristic physical findings (straight neck, lowered occipital hairline, and restricted neck motion) may indicate KFS.
- Klippel-Feil syndrome is a rare congenital disease accompanied by skeletal and vascular malformations such as thoracic deformity and congenital heart disease.
- In the first such report in the literature, this case shows a cardiac surgical approach can be an important therapeutic strategy for valvular disease associated with thoracic abnormalities in congenital disease.

Abbreviations and Acronyms

KFS Klippel-Feil syndrome

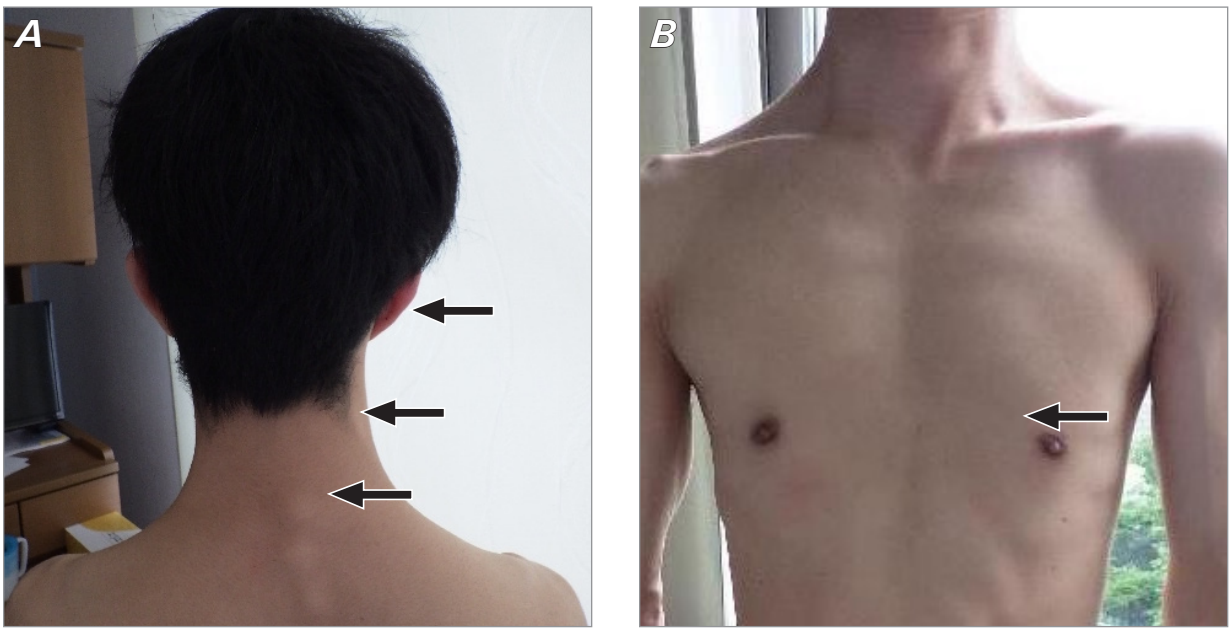

Fig. 1 Preoperative photographs show **(A)** physical findings of the patient on admission, indicating a low auricle, straight neck, and lowered occipital hairline (arrows), and **(B)** thoracic anomalies of scoliosis and pectus excavatum (arrow).

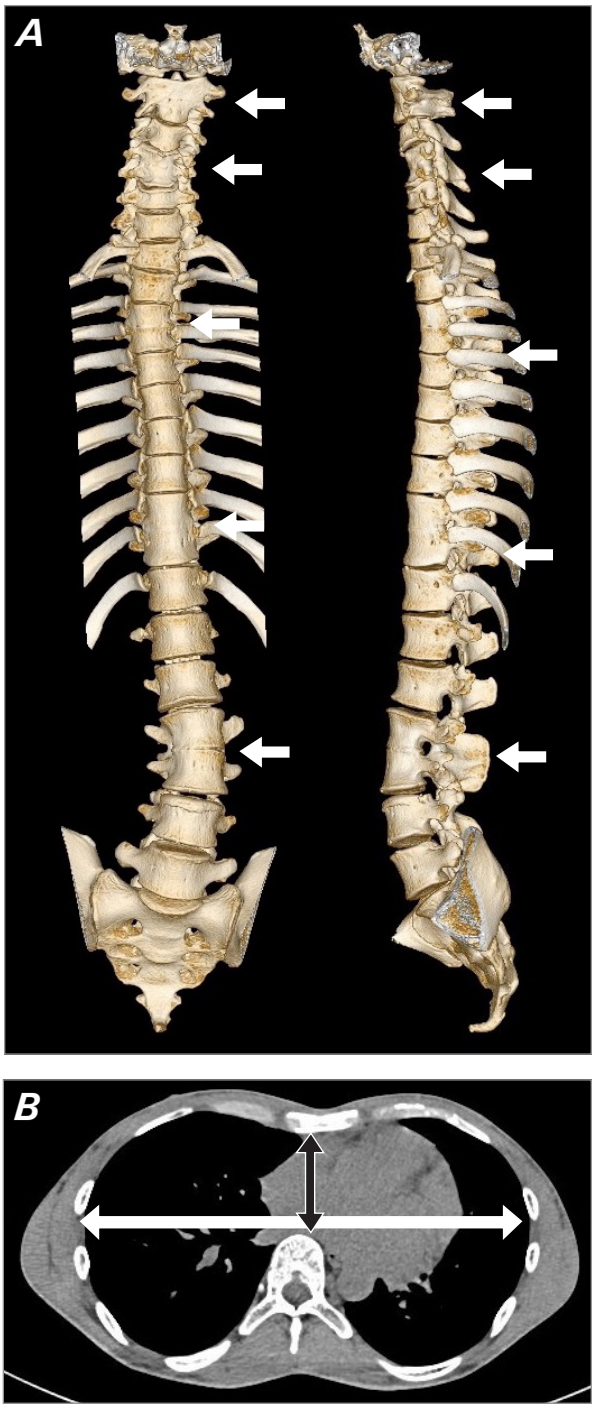

Fig. 2 Preoperative computed tomographic scans include **(A)** a 3-dimensional computed tomographic scan of the frontal (left) and sagittal (right) views of the spine showing scoliosis and fusion of the spinous processes of the posterior vertebral bodies at C1-C2, C4-C5, T3-T4, T9-T10, and L2-L3 (arrows) and **(B)** a transverse chest tomographic scan that shows the short distance between the sternum and vertebrae. The Haller computed tomography index was calculated by dividing the thoracic transverse diameter (white double-headed arrow) by the distance between the sternum and the vertebrae (black double-headed arrow).

Weaning the patient from cardiopulmonary bypass went smoothly, and transesophageal echocardiograms confirmed the disappearance of mitral valve regurgitation. The pericardium was closed as much as possible, the access incision was closed by the standard method, and surgery was completed. Operative time was 4 hours, 12 minutes; cardiopulmonary bypass lasted 127 minutes; and aorta cross-clamp time was 82 minutes. Pathologic examination showed organized fibrosis and irregular thickening of the mitral valve, with no inflammatory cells observed. Extubation was performed 4 hours after surgery, and the patient was transferred to a general ward the day after surgery. Transthoracic echocardiography performed again on postoperative day 7 confirmed the disappearance of mitral valve regurgitation. The patient was discharged on postoperative day 10.

Outcome

In this case of mitral valve regurgitation in a patient with KFS with related thoracic deformity, mitral valvuloplasty was performed, and the patient had a good postoperative course. Although approaches in cardiac surgery remain controversial, valvuloplasty via median sternotomy can be an effective treatment for thoracic abnormalities in patients with a short sternal-vertebral distance as a result of rare congenital disease.

Latest Follow-Up

At 2 years after surgery, the patient has shown no remarkable change. There has been no recurrence of mitral valve regurgitation on transthoracic echocardiograms.

Discussion

Three main symptoms characterize KFS: a short neck, a lowered occipital hairline, and a limited range of motion in the neck with congenital fusion of the cervical spine. It has remained a rare congenital disease ever since Klippel and Feil[1,2] first reported a case in 1912. Although less than 50% of patients with KFS have the 3 main features, skeletal abnormalities in which 2 or more cervical vertebrae are congenitally fused are called the *KFS group* and are classified into 3 types, according to the degree of fusion, including deformity of not only the cervical vertebrae but also of the thoracic and lumbar vertebrae.[1] Skeletal and vascular malformations have also been reported in KFS, including in cardiac surgery case reports

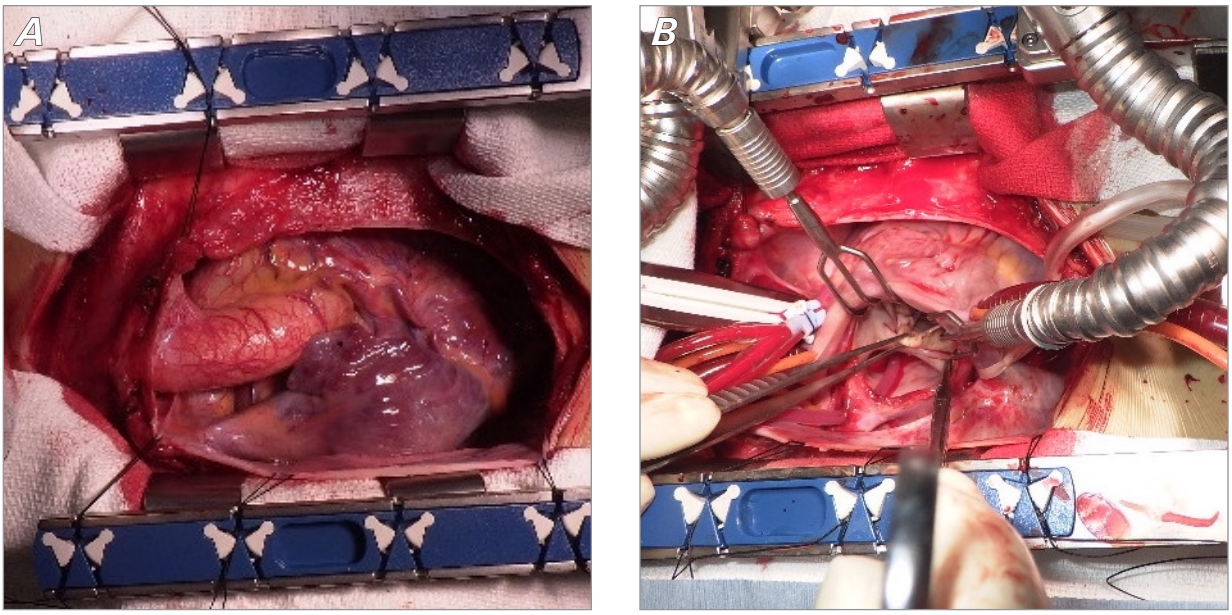

Fig. 3 Intraoperative photographs show **(A)** enlargement of the right atrium, which was observed after thoracotomy, and **(B)** a view of the mitral valve after the right-left atrial approach, which revealed posterior leaflet prolapse.

on congenital heart disease.[2,3] This case, in which mitral valve repair was performed for mitral valve regurgitation in a patient with a thoracic anomaly related to KFS, is reported along with a review of the literature.

The incidence of KFS is 1 in 40,000 to 42,000 people, and variations occurring in the *GDF6* and *GDF3* genes, which provide instructions for making proteins in the bone morphogenetic protein family and in regulating the growth and maturation of bone and cartilage, have been implicated in KFS.[1,2] Concomitant malformations reported include cranial base invagination in the central nervous system, kidney agenesis in the urinary system, and intestinal cysts in the digestive system; congenital heart disease occurs in approximately 4% of patients with this syndrome.[2-5] Complications of cardiac malformations include ventricular septal defect, pulmonary stenosis, endocardial cushion defect, patent foramen ovale, patent ductus arteriosus, and congenital bicuspid aortic valve.[2,4]

In this case of KFS with mitral valve regurgitation that required cardiac surgery, in addition to the characteristic physical findings in this patient (lowered occipital hairline, restricted neck motion, and pectus excavatum), a preoperative computed tomographic scan revealed bone fusions in the cervical, thoracic, and lumbar vertebrae and thoracic abnormalities (scoliosis and pectus excavatum), which led to the diagnosis. When the distance between the thoracic vertebrae is short as a result of an abnormality of the thoracic cage, as in the present patient, the risk of performing cardiac surgery via a right intercostal space is high because of the poor field of view and increased technical difficulty.[6,7] A mitral clip was not used because the patient was still young, and the transthoracic echocardiographic findings suggested that mitral valvuloplasty could be performed.

The thoracic deformity in KFS may also be a contributing factor to mitral valve regurgitation in adulthood. Klippel-Feil syndrome is associated with rib abnormalities and scoliosis at a high rate of 60% to 70%,[5,8] and mitral valve prolapse is also reported in patients with thoracic abnormalities such as pectus excavatum, straight back syndrome, and scoliosis.[9,10] These thoracic abnormalities cause mechanical compression of the heart, which can lead to deformation of the left atrium and mitral annulus resulting from morphologic abnormalities of the left ventricle. As a result, the positional relationship of the mitral valve system becomes abnormal, which is thought to cause mitral valve prolapse and mitral valve regurgitation. In addition to KFS, the present patient had thoracic abnormalities and mitral valve regurgitation because of cardiac displacement. Because cardiac surgery for mitral valve regurgitation in this patient was required not in childhood but in adulthood, the duration of the abnormality in his left heart morphology was considered to be involved in the development of his mitral valve regurgitation.

Article Information

Published: 26 April 2024

Author Contributions: Y.E. wrote and prepared the manuscript under the supervision of H.N. S.Y. performed the surgeries. The authors have read and approved the final version of the manuscript.

Conflict of Interest Disclosure: The authors have no conflicts of interest to declare.

Funding/Support: Not applicable.

Statement of Ethics: The procedure was approved by the Ethics Committee of Saitama City Hospital. All procedures performed on human participants were in accordance with the ethical standards of the institutional and national research committee and with the 1964 Declaration of Helsinki and its later amendments or comparable ethical standards. Written informed consent was obtained from the patient before all surgeries were performed.

Acknowledgments: We would like to thank Rise Japan LCC (rise-japan.rulez.jp) for English-language editing.

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