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Klippel Feil Syndrome with Sprengel Deformity: A Case Report and Literature Review

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Abstract: Klippel-Feil syndrome (KFS) is a rare disorder characterized by the presence of congenital fusion of at least 2 cervical vertebrae.Patients with KFS often have a variety of congenital malformations, with Sprengel deformity being the most common.The etiology of KFS and Sprengel deformity is still unclear. A thorough evaluation and workup of the patient can help in the diagnosis of the disease. Because of different degrees of pathological changes and deformities, the treatments are quite different among all patients. In this review, we report the treatment course and prognosis of a patient with KFS and Sprengel deformity, aiming to provide a reference for the clinical management of this disease based on our treatment experience and literature review.

Keywords: Klippel-Feil Syndrome, Sprengel Deformity

Introduction

KFS is a rare disorder characterized by congenital fusion of part or all of the cervical spine [1, 2].The exact prevalence of KFS is unclear, ranging from approximately 1 : 40,000 to 1: 42,000 [3]; the ratio of female-to-male prevalence is approximately 3:4 [2].KFS was first clinically described in 1912 by Klippel and Feil. Short neck, low posterior hairline, and limited neck movement are the most typical clinical features of KFS, but less than 50% of patients demonstrate this clinical triad.The clinical diagnosis of KFS may be unreliable, so most sporadic cases were found incidentally on imaging [4].KFS is often associated with other congenital developmental malformations, including scoliosis, Sprengel deformity, spina bifida and visceral malformations . Sprengel deformity is one of the malformations most frequently associated with KFS, whose presence is up to 42% in KFS cases [5, 6]. In this paper, we focus on a case report of KFS with Sprengel deformity and briefly describe its diagnosis, management and prognosis.

Case Presentation

An 18-year-old female patient was hospitalized with "limitation of neck and shoulder movements and unequal height of both shoulders for more than 10 years". The patient, together with her family, complained of unequal height of both shoulders after birth, with the right shoulder being higher. The patient suffered no localized redness or swelling since the onset of the disease, no headache, dizziness and radiating pain in the upper extremities. Physical examination demonstrated scoliosis deformity of cervicothoracic segment, short and thick neck, low posterior hairline.The patient's right shoulder measured about 5cm higher than the left with obvious unequal shoulders. The right shoulder joint was limited in abduction and elevation activities, but other activities were not significantly restricted. Sensation and muscle strength of both upper limbs were not apparently abnormal. There was no significant abnormality in flexion and posterior extension.No abnormality was found in Eaton test and Spurling test. Additionally, normal reflexes were present; pathological reflexes were not elicited.

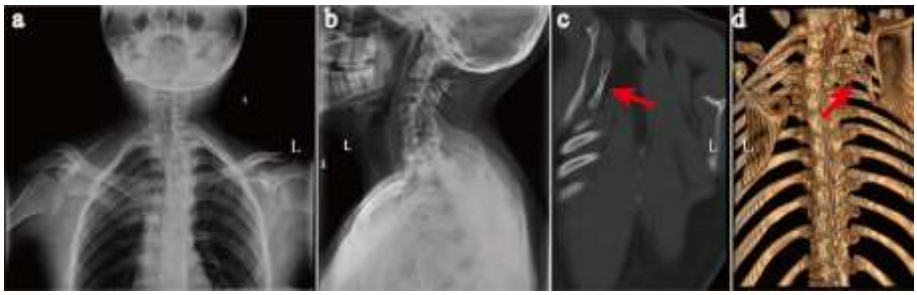

Fig. 1: (a) Scoliosis deformity of the cervicothoracic segment is present. Right scapula is positioned higher up to the level of the cervical spine. (b) Physiological curvature of the cervical spine is present. C4-6 vertebrae and attachments are fused, (c-d) CT+3D reconstructed images of the thoracic spine show the right T1-4 level with visible omovertebral bone.

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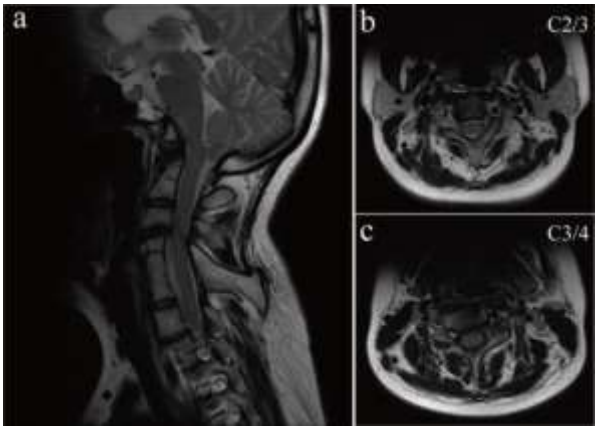

Fig. 2: MRI of cervical spine demonstrates: (a) disc degeneration at C2/3 and C3/4, (b-c) no abnormal signal in the spinal cord at C2/3 and C3/4 levels, and narrowing of the spinal canal at C3/4 level .

Management

With limited neck and shoulder movement and unequal height of both shoulders, this in-patient underwent more ancillary investigations: blood and urine tests did not suggest significant abnormalities; urological ultrasound suggested: left renal agenesis; X-ray showed: scoliosis deformity of the cervicothoracic segment was present (Figure 1a, Figure 3c); the right scapula was positioned approximately 5.18 cm higher than the left (Figure 3c), and C4-6 vertebrae and attachments were fused (Figure 1b). CT of the thoracic spine with 3D reconstruction showed that omovertebral bone formation was visible at the right T1-4 level (Figure 1c, d). MR of the cervical spine suggested that no significant signal shadow was seen in the spinal cord at the levels of C2/3 and C3/4, and narrowing of the spinal canal at the level of C3/4 was found.

The patient was diagnosed with: 1. Klippel Feil syndrome and Sprengel deformity; 2. left renal agenesis, based on the patient's medical history, physical examination and relevant tests. The patient, aged18, was skeletally mature and did no present with neurological impairment. Considering the patient's urgent need to resolve the shoulder deformity and shoulder mobility, we recommended that "right clavicle osteotomy and fusion in situ fixation + right omovertebral bone resection+ right scapula internal superior margin release + right scapula descent" should be performed.

Surgical procedure: 1. After successful anesthesia, the patient was placed in the supine position, and the right shoulder operative field was disinfected; skin preparation and surgical draping were completed. 2. An oblique incision was made along the surface of the right mid-clavicle along its course, approximately 10 cm long, and the skin, subcutaneous, deep fascia and periosteum were incised. After stripping the periosteum, the mid-clavicle was identified, and the clavicle was truncated at the distal and proximal 1/3, respectively, and the incision was sutured.3. The patient was transferred to the prone position, the right shoulder and right upper extremity were disinfected in the operative field, and surgical draping was completed. A longitudinal "S"-shaped incision was made from the level of cervical 5 to the level of T8 to reveal the fused C4-7 spinous process, exposing and resecting the omovertebral bone. The trapezius muscle was incised, and the supraspinatus, levator scapulae and rhomboideus were dissected from top to bottom along the superior and medial edges of the scapula, respectively, at the point of termination on the scapula. Structures such as the serratus anterior and subscapular muscle were separated anterior to the scapula, and the medial superior scapular angle was excised. Press down and internally rotate the scapula until the right mesoscapula and the left mesoscapula are at the same level. The subscapular Angle and the medial edge of the scapula are drilled separately, and the previously stripped muscle stops are fixed with sutures to the scapula in their new positions, respectively.Layered suture incision is made.4. The patient is then transferred to a supine position, and surgical instruments accessed from the previous surgical incision. The mid clavicle was identified after stripping the periosteum, the original clavicle osteotomy was reset, and the clavicle was fixed with a 5-hole reconstruction plate and 4 screws. The incision was repeatedly rinsed with saline, the layers were sutured, and the incision was covered with a sterile dressing.5. The operation was uneventful, with minimal intraoperative bleeding.

Postoperatively, the shoulder joint was splinted for 2 weeks, followed by active assistance and passive activities for shoulder rehabilitation therapy; resistance training was initiated 4 weeks, follow-up after 2 months. It can be seen that the patient's shoulders were basically flush bilaterally (Fig. 3c, f), the right upper limb abduction mobility increased (Fig. 3a, d), and the combing movement could be completed without the lateral neck (Fig. 3b, e).

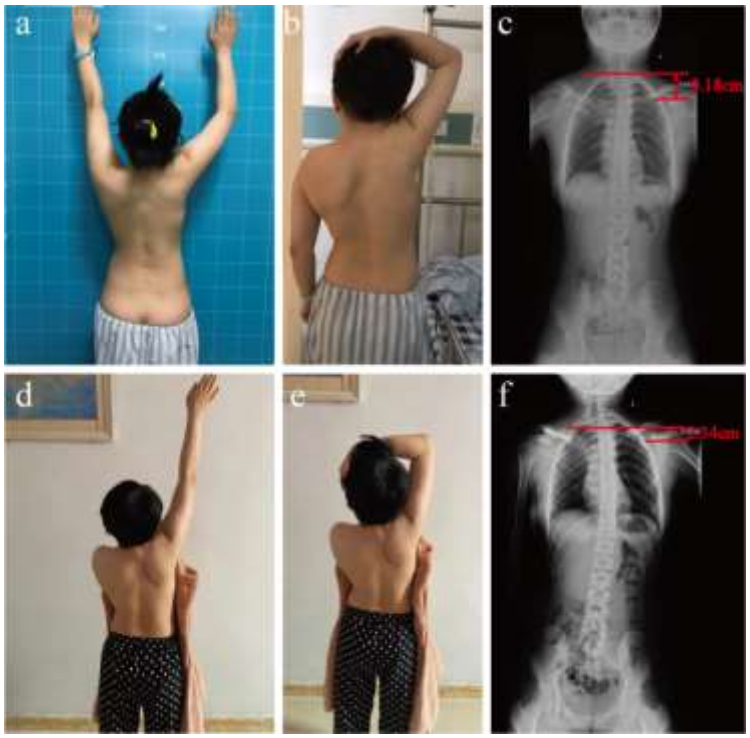

Fig. 3: Pre-surgery (a-c) vs. post-surgery (d-f)

Discussion

Studies have suggested that not all patients with KFS and congenital scoliosis require surgical treatment. One scholar conducted long-term follow-up of 32 cases of KFS. During 10 years of follow-up, only 7 patients developed neurological symptoms and 2 required surgical treatment [8]. Another scholar conducted long-term follow-up of 19 pediatric and adolescent patients with KFS. In a mean follow-up of 12.5 years, 5 patients were affected by neurological complications; 4 underwent surgery and 14 had no neuropathy [9]. It is generally accepted that patients with KFS without neurological symptoms require only conservative treatment, including regular review, restriction of activity, use of cervical orthoses, and traction as needed [10]. In contrast, indications for surgical treatment of KFS include persistent pain and neurological dysfunction where the pharmacological effects have failed, with an overall goal of increasing the cervical spine stability [11].

Sprengel deformity is the most common congenital shoulder deformity caused by abnormal descent of the scapula during embryonic development [12]. Sprengel deformity often involves deformities in the appearance of the shoulder and dysfunctional movement. In 1891, Sprengel drew attention to this deformity by describing it as congenital scapular elevation.Patients with Sprengel deformity are often associated with other congenital developmental deformities including: scoliosis, Klippel-Feil syndrome (KFS), rib anomalies, spina bifida, and limb deformities [13-15].Dino Samartzis et al. concluded that cervical fusion of multiple segments is more common in patients with KFS with Sprengel deformity. Therefore, patients with confirmed KFS should be thoroughly examined for the presence and severity of Sprengel deformity to assess factors that may put patients with KFS at an increasing risk of neurological injury [16].

Surgical treatment of Sprengel deformity is performed to improve the shape and function of the shoulder. The core of the surgical technique is resection of the protruding portion of the scapula and inferior translation of the scapula [12]. In patients aged >8 years, clavicle osteotomy prior to scapular repositioning can reduce the incidence of brachial plexus palsy [17]. Most patients with Sprengel deformity have the omovertebral bone that connects the scapula to the vertebrae. The presence of the omovertebral bone affects the range of motion of the shoulder joint and the neck, as well as the descent of the scapula [18, 19]. Therefore, removal of the omovertebral bone is an important part of the surgical treatment of Sprengel deformity. Surgical modalities that have been developed for the treatment of Sprengel deformity reportedly include the Woodward procedure [20], the Green procedure [17], the Bellemans and Lamoureux modified Green procedure [21], the Leibovic modified

Green procedure [22] and the Mears procedure [23]. Satisfactory results have been obtained with various surgical approaches. Among them, the Woodward procedure is probably the most widely used technique for the treatment of Sprengel deformity. This procedure focuses on the release and repositioning of the muscular initiation at the medial border of the scapula, as well as the removal of any omovertebral bone. The patient reported in this case was treated with the Woodward procedure, and during the clinical follow-up, satisfactory clinical outcomes were achieved postoperatively.

Conclusion

Early diagnosis and appropriate treatment can help patients with KFS avoid developing neurological symptoms or undergoing surgery. As the clinical diagnosis of KFS disease may be unreliable, a thorough evaluation and a complete physical examination for the patient can be helpful. Surgical treatment can improve shoulder shape and neck and shoulder mobility in patients with Sprengel deformity. Patients with a confirmed diagnosis of KFS should be thoroughly examined for the presence and extent of Sprengel deformity to assess factors that may put patients with KFS at increased risk of nerve injury and to manage them symptomatically.Patients with a confirmed diagnosis of KFS should be thoroughly examined for the presence and extent of Sprengel deformity, so that factors that may put patients with KFS at increased risk of nerve injury can be assessed, meanwhile, the abnormalities can be managed symptomatically.

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References

1. M. Klippel, A. Feil, The classic: A Case of Absence of Cervical Vertebrae with the Thoracic Cage Rising to the Base of the Cranium (Cervical Thoracic Cage), Clin Orthop Relat Res (109) (1975) 3-8.
2. P.D. Pizzutillo, M. Woods, L. Nicholson, G.D. MacEwen, Risk factors in Klippel-Feil syndrome, Spine (Phila Pa 1976) 19(18) (1994) 2110-6.
3. M.N. Thomsen, U. Schneider, M. Weber, R. Johannisson, F.U. Niethard, Scoliosis and congenital anomalies associated with Klippel-Feil syndrome types I-III, Spine (Phila Pa 1976) 22(4) (1997) 396-401.
4. J. Gruber, A. Saleh, W. Bakhsh, P.T. Rubery, A. Mesfin, The Prevalence of Klippel-Feil Syndrome: A Computed Tomography-Based Analysis of 2,917 Patients, Spine Deform 6(4) (2018) 448-453.
5. A.R. Larson, K.D. Josephson, R.M. Pauli, J.M. Opitz, M.S. Williams, Klippel-Feil anomaly with Sprengel anomaly, omovertebral bone, thumb abnormalities, and flexion-crease changes: novel association or syndrome?, Am J Med Genet 101(2) (2001) 158-62.
6. M.F. Van Kerckhoven, G. Fabry, The Klippel-Feil syndrome: a constellation of deformities, Acta Orthop Belg 55(2) (1989) 107-18.
7. R.N. Hensinger, J.E. Lang, G.D. MacEwen, Klippel-Feil syndrome; a constellation of associated anomalies, J Bone Joint Surg Am 56(6) (1974) 1246-53.
8. S.M. Theiss, M.D. Smith, R.B. Winter, The long-term follow-up of patients with Klippel-Feil syndrome and congenital scoliosis, Spine (Phila Pa 1976) 22(11) (1997) 1219-22.
9. P. Rouvreau, C. Glorion, J. Langlais, H. Noury, J.C. Pouliquen, Assessment and neurologic involvement of patients with cervical spine congenital synostosis as in Klippel-Feil syndrome: study of 19 cases, J Pediatr Orthop B 7(3) (1998) 179-85.
10. M.J. Herman, P.D. Pizzutillo, Cervical spine disorders in children, Orthop Clin North Am 30(3) (1999) 457-66, ix.
11. P.L. Zhou, G.W. Poorman, C. Wang, K.E. Pierce, C.A. Bortz, H. Alas, A.E. Brown, J.C. Tishelman, M.B. Janjua, D. Vasquez-Montes, J. Moon, S.R. Horn, F. Segreto, Y.U. Ihejirika, B.G. Diebo, P.G. Passias, Klippel-Feil: A constellation of diagnoses, a contemporary presentation, and recent national trends, J Craniovertebr Junction Spine 10(3) (2019) 133-138.
12. E.J. Harvey, M. Bernstein, N.M. Desy, N. Saran, J.A. Ouellet, Sprengel deformity: pathogenesis and management, J Am Acad Orthop Surg 20(3) (2012) 177-86.
13. D.M. Ross, R.L. Cruess, The surgical correction of congenital elevation of the scapula. A review of seventy-seven cases, Clin Orthop Relat Res (125) (1977) 17-23.
14. M.E. Cavendish, Congenital elevation of the scapula, J Bone Joint Surg Br 54(3) (1972) 395-408.
15. C.L. Jeannopoulos, Congenital elevation of the scapula, J Bone Joint Surg Am 34 A(4) (1952) 883-92.
16. D. Samartzis, J. Herman, J.P. Lubicky, F.H. Shen, Sprengel's deformity in Klippel-Feil syndrome, Spine (Phila Pa 1976) 32(18) (2007) E512-6.
17. G. Andrault, F. Salmeron, J.M. Laville, Green's surgical procedure in Sprengel's deformity: Cosmetic and functional results, Orthop Traumatol- Sur 95(5) (2009) 330-335.
18. Y. Mikawa, R. Watanabe, Y. Yamano, Omoclavicular bar in congenital elevation of the scapula. A new finding, Spine (Phila Pa 1976) 16(3) (1991) 376-7.
19. J.F. Mooney, 3rd, D.R. White, S. Glazier, Previously unreported structure associated with Sprengel deformity, J Pediatr Orthop 29(1) (2009) 26-8.
20. W.G. Carson, W.W. Lovell, T.E. Whitesides, Jr. Congenital elevation of the scapula. Surgical correction by the Woodward procedure, J Bone Joint Surg Am 63(8) (1981) 1199-207.
21. M. Bellemans, J. Lamoureux, Results of surgical treatment of Sprengel deformity by a modified Green's procedure, J Pediatr Orthop B 8(3) (1999) 194- 6.
22. S.J. Leibovic, M.G. Ehrlich, D.J. Zaleske, Sprengel deformity, J Bone Joint Surg Am 72(2) (1990) 192-7.
23. D.C. Mears, Partial resection of the scapula and a release of the long head of triceps for the management of Sprengel's deformity, J Pediatr Orthop 21(2) (2001) 242-5.