Case Study

A Comprehensive Approach to the Diagnosis and Management of Klippel Feil Syndrome

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ABSTRACT

Klippel-Feil Syndrome (KFS) is a rare genetic disorder characterized by the abnormal development of the cervical spine, leading to the fusion of two or more cervical vertebrae. The syndrome presents diverse symptoms, including limited neck movement, chronic pain, and neurological manifestations such as limb numbness or weakness. The severity of KFS can vary significantly, and treatment primarily focuses on symptom management and preventing complications such as scoliosis or spinal cord compression. Surgical interventions are often necessary for patients with complex forms of the syndrome. Interestingly, Chiari 1 malformation, a cranial anomaly affecting the brainstem, can coincide anatomically with KFS. In this case report, we present the case of a 9-year-old patient who sought medical attention due to persistent, unchanging neck pain. The patient's medical history was notable for developmental delays and cervical restraint observed during physical examination. Magnetic resonance imaging (MRI) findings revealed hydrocephalus and brainstem descent, indicating the presence of Chiari 1 malformation. Comprehensive MRI and CT scans were performed, and a management plan was formulated, primarily involving cranial surgery and physiotherapy. Implementation of the treatment approach resulted in significant improvement in the patient's symptoms. This case highlights the significance of considering Chiari 1 malformation as a potential comorbidity in patients diagnosed with KFS who present with persistent neck pain. Early detection and appropriate management of both conditions are crucial for achieving favorable outcomes and enhancing the quality of life for affected individuals. Understanding the complex interplay between KFS and Chiari 1 malformation is essential for providing comprehensive care and tailored treatment strategies. Further research is warranted to elucidate the underlying mechanisms linking these two conditions and to explore optimal management approaches for patients with dual pathology. By reporting this case, we contribute to the existing literature and increase awareness among healthcare professionals regarding the potential coexistence of KFS and Chiari 1 malformation. Continued efforts in identifying associated anomalies and optimizing therapeutic interventions will aid in improving patient outcomes and ensuring optimal care for individuals affected by these conditions.

Keywords: Brainstem descent, Cervical spine abnormalities, Chiari 1 malformation, Klippel-Feil Syndrome

1. Introduction

Klippel Feil syndrome (KFS) is a rare congenital disorder characterized by a triad of low posterior hairline, short neck, and limited range of motion of the cervical spine (1, 2). With an estimated incidence of approximately 1 in 40,000 individuals, KFS predominantly affects females (1, 2). However, it is important to note that less than half of the patients exhibit complete triad findings (3). The KFS is often incidentally detected on cervical imaging following a traumatic event, and its clinical presentation can vary widely, ranging from asymptomatic cases to severe spinal fusion and accompanying visceral abnormalities, necessitating a multidisciplinary approach. Additionally, KFS has been associated with Chiari 1 malformation, which involves the herniation of a portion of the cerebellum through the foramen magnum (4). The syndrome frequently goes unnoticed due to its high asymptomatic rate, and it is frequently linked to comorbidities such as scoliosis, spina bifida occulta, renal abnormalities, deafness, rib deformity, synkinesis, and congenital heart disease (5). In Pakistan, the first reported case of KFS exhibited an unusual association with situs inversus, highlighting the diverse manifestations of the disease (6). Furthermore, a recent case report documented the occurrence of KFS in Pakistan, where it was found to be accompanied by cervical myelomeningocele and thoracic syringomyelia (7). In this report, we present the case of a 9-year-old patient with a history of encephalocele in infancy who subsequently presented with classic KFS and an associated Chiari 1 malformation.

2. Case Presentation

In January 2023, a 9-year-old female patient referred to National Institute of Child Health in Karachi, Pakistan, with a constant neck pain that persisted regardless of rest or neck movement. Despite seeking help from multiple healthcare professionals, the patient's symptoms remained unchanged and did not improve with vitamin D supplementation and physiotherapy. The patient's mother denied any complications during pregnancy or substance abuse but reported that the patient exhibited normal physical development, albeit with some delays in reaching milestones. The patient's appearance was slightly different from her peers at school.

During the physical examination, it was observed that the patient had normal flexion of the thoracolumbar spine and normal shoulder abduction, adduction, and external shoulder rotation. However, there was restricted neck extension and lateral flexion at the cervical joint. Magnetic resonance imaging (MRI) revealed hydrocephalus and crowding of the foramen magnum with descent of cerebral tonsils, indicative of Chiari 1 malformation (Figure 1 A and B). A computed tomography (CT) scan showed a complete fusion of C2, C3, C4, and C5 vertebrae, with a rudimentary disc visible at the C5-C6 levels. Additionally, a hemivertebra was noted on the right side at D2. The anteroposterior diameter of the affected vertebral bodies was smaller than the diameter at the superior and inferior limits, indicating the presence of the "wasp-waist" sign (Figure 1C and D). Postsurgical changes, characterized by focal bony defects and skin thickness, were observed in the nape of the neck and occipital region (Figure 2).

Various differential diagnoses were considered, but an accurate diagnosis was established through radiographic and MRI scans. After consultation with neurosurgeons and plastic surgeons, conservative management with symptomatic pain relief was initially recommended. However, due to the lack of improvement with conservative therapy, surgical intervention was eventually pursued. The patient underwent cranial surgery for posterior fossa decompression and spinal surgery for cervical decompression.

Postoperatively, the patient was advised to avoid rigorous, sudden, or substantial movements for at least a month, gradually increasing the workload on the neck and head. Contact sports were to be avoided until permitted by the medical team. Weekly physiotherapy

sessions were scheduled to teach the patient exercises to perform at home. Regular follow-up visits were arranged to monitor the patient for any underlying issues, and periodic central nervous system (CNS) examinations were conducted to ensure no neurological complications or abnormalities.

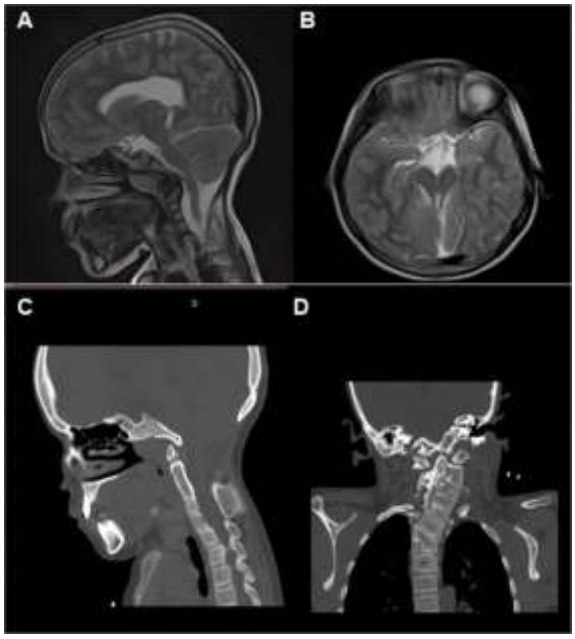

Figure 1: Magnetic Resonance Imaging (MRI) demonstrating Chiari 1 malformation.
A) Sagittal section of head MRI highlighting the cerebellar tonsillar herniation through the foramen magnum, characteristic of Chiari 1 malformation. The downward displacement of the cerebellar tonsils is visible.
B) Axial view of the same MRI showing the heart-shaped incisura, which is another typical feature of Chiari 1 malformation. The indentation in the cerebellum is visible, which is caused by the compression of the cerebellar tonsils against the brainstem.
C) Loss of cervical lordosis and fusion of cervical vertebrae in a patient with Klippel Feil syndrome (KFS). The fusion of cervical vertebrae is visible, resulting in limited neck motion and characteristic short neck appearance.
D) Hemivertebra, a congenital abnormality in which one half of a vertebra fails to form properly, is also noted on the right side at D2. This finding is often seen in patients with KFS and can contribute to spinal deformities such as scoliosis.

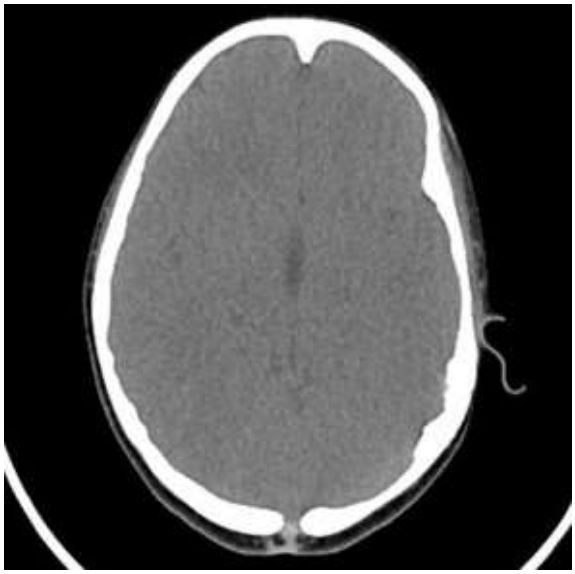

Figure 2: Radiograph demonstrating post-surgical changes in the patient's scalp secondary to previous encephalocele surgery in infancy. Overlying skin thickening and scarring is observed at the site of previous surgical intervention.

3. Discussion

Klippel-Feil syndrome (KFS) is a congenital abnormality of the skeletal system that results in the fusion of cervical vertebrae, which was first reported by Klippel and Feil in 1912 (8). The KFS is commonly inherited sporadically, autosomal dominant, or autosomal recessive (9). The KFS manifests with a variety of phenotypes and is associated with laryngeal and genitourinary tract involvement (10-12). The classification of KFS is based on the level of fusion and inheritance pattern (13). The most commonly used method for clinical classification is based on anatomic fusion patterns, as described by Samartzis et al. (14).

The KFS is often undiagnosed because of its high asymptomatic rate, but distinctive symptoms of facial anomalies, Sprengel deformity, and ocular pathology can aid in diagnosis (15). Diagnosis requires a thorough cervical spine examination, which assesses extension, rotation, and bending of the neck (16). X-ray imaging is a useful diagnostic tool to visualize fused vertebral segments. Atlantoaxial displacement can cause neck pain and torticollis, with cervical spine subluxation noted in 33% of KFS patients (17). Magnetic resonance imaging is used to diagnose associated neurologic abnormalities, such as cervical myelopathy and nerve root compression (18). The KFS is also associated with other conditions, such as scoliosis and Sprengel deformity (19).

Non-surgical management of KFS mainly focuses on pain management. Surgical management options depend on the severity of the disease, and fusion surgery is the most commonly performed procedure (20). However, surgery is often associated with significant morbidity, including nerve root injury, arterial injury, and spinal cord injury (21). Therefore, the management of KFS should be individualized based on the patient's condition, and appropriate counseling and multidisciplinary care should be provided.

In conclusion, KFS is a rare disorder characterized by cervical fusion and limited cervical spine motion. While often asymptomatic, KFS can be associated with various comorbidities, including Sprengel deformity, Chiari malformation, deafness, and renal agenesis. Our case report highlights a 9-year-old female with KFS and Chiari-I malformation who underwent surgical intervention to alleviate symptoms and improve her quality of life. Given the complexity of KFS and its comorbidities, a multidisciplinary approach is essential for effective management. Future research is necessary to better understand the underlying mechanisms of KFS and potential genetic factors contributing to its development. Our study contributes to the existing literature by providing insights into the clinical presentation, diagnostic evaluation, and comprehensive management approach, including surgical intervention, for this rare combination of KFS and Chiari-I malformation. Early diagnosis and appropriate treatment are crucial to prevent complications and improve outcomes in patients with KFS. Healthcare professionals must consider KFS as a possible diagnosis in patients with cervical fusion and associated symptoms.

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Ethics

The manuscript complies with the ethical recommendations of the Declaration of Helsinki of the World Medical Association (WMA). M.H, F.S, B.A, M.I contributed to the conception and design of the manuscript. M.H, M.I, B.A supervised the project. M.H, M.K.K, M.I, S.J.S provided the materials and contributed to data collection and processing. F.S, M.K.K, M.T.A, B.A, S.J.S contributed to the interpretation and analysis of the project. F.S, M.K.K, M.T.A, B.A, S.J.S contributed to the literature review and writing of the manuscript respectively. M.H, F.S, M.K.K, M.T.A, M.I, S.J.S critically revised the manuscript.

Conflict of Interest

The authors declare no conflict of interest.

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