**A CASE OF KLIPPEL–FEIL SYNDROME: CLINICAL PRESENTATION AND MANAGEMENT**

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 **ABSTRACT**

Klippel-Feil Syndrome (KFS) is a rare congenital ailment characterized by the fusion of cervical vertebrae, often associated with different musculoskeletal and systemic abnormalities. We present the case of a 12-year-vintage male who was admitted with a -month history of neck pain, frame aches, three days of fever, and two days of cough. The affected person had a substantial clinical history of recurrent respiration infections from early childhood. Diagnostic imaging revealed several feature features of KFS, together with cervical vertebral fusion (C1), hypoplastic ribs, and an ectopic, malrotated left kidney. Laboratory findings indicated mild anemia with low hemoglobin, microcytic hypochromic indices, and barely altered differential leukocyte count, however, no considerable renal or liver disorder turned into noted. Given the affected person's musculoskeletal and renal abnormalities, alongside recurrent respiration infections, KFS became suspected and confirmed via imaging research. Treatment included antibiotics (Amoxicillin Clavum) for respiratory contamination, ache control (Acetaminophen), bronchodilators (Levosalbutamol), and different symptomatic care. The case highlights the significance of early analysis and a multidisciplinary approach to manipulate the complexities related to KFS, together with musculoskeletal deformities, renal anomalies, and respiration complications. Long-term observation is necessary for monitoring and addressing capability complications such as renal dysfunction, persistent breathing issues, and anemia. This case underscores the need for complete care to enhance the satisfaction of lifestyles and prevent lengthy-time period sequelae in patients with Klippel-Feil Syndrome.

**Keywords:**

Klippel-Feil Syndrome, Respiratory Infections, Neck Pain, Hypoplastic Ribs, Ectopic Kidney,

Pediatric Case Report, Anemia.

**INTRODUCTION**

Klippel-Feil Syndrome (KFS) is an unprecedented congenital ailment that results from the peculiar fusion of cervical vertebrae at some point of embryonic improvement. It is characterized by various degrees of cervical spine anomalies, which frequently lead to several musculoskeletal and neurological headaches. The hallmark medical features of KFS include a brief neck, restricted neck movement, low hairline, and a palpable fusion of the cervical vertebrae. However, the syndrome can also present with associated abnormalities in other organ systems, inclusive of the kidneys, coronary heart, and ears, complicating the clinical photograph and necessitating a complete diagnostic approach.

The pathogenesis of KFS is thought to be due to a disturbance inside the segmentation of somites during the early levels of embryonic development. This disturbance affects the normal formation of the vertebrae and, in a few instances, different systems derived from the mesoderm. The quantity of vertebral fusion can vary from a single vertebra to more than one segment of the cervical backbone. While a few people with KFS may also stay asymptomatic or gifted with minimum signs and symptoms, others may additionally broaden substantial practical obstacles due to spinal deformities, restrained neck mobility, and associated organ malformations.

One of the more tough aspects of KFS is the huge spectrum of related situations. Among these, renal anomalies are commonplace and might include ectopic kidneys, renal hypoplasia, or malrotation. These findings are vital as they'll result in similar complications such as urinary tract infections, renal insufficiency, or different renal-related pathologies. The presence of these abnormalities can significantly affect the long-term management and prognosis of affected people.

Respiratory problems can also rise in sufferers with KFS, regularly because of thoracic abnormalities like rib deformities or hypoplastic ribs, which can result in reduced lung growth and continual respiration infections. Such conditions may additionally require close tracking and specialized interventions to prevent or deal with respiratory headaches.

Despite its rarity, KFS is a crucial clinical entity that requires early recognition, thorough assessment, and multidisciplinary management. The remedy method is tailored to the person patient's symptoms and related abnormalities. Given the capacity for existence-length headaches, early diagnosis is critical to provide the most appropriate care, save you from a practical impairment, and enhance your high quality of life.

In this case file, we present a 12-year-antique male patient who turned into admitted with neck pain, frame aches, fever, and a cough, observed through a record of recurrent respiratory infections. Diagnostic imaging found characteristic findings suggestive of Klippel-Feil syndrome, inclusive of cervical vertebral fusion, hypoplastic ribs, and renal anomalies. Through this case, we propose to spotlight the importance of thinking about KFS in pediatric patients with musculoskeletal, neurological, and systemic abnormalities and to talk about the management and capability challenges related to this rare syndrome.

**Case Report**

A 12-year-old male child was admitted to the pediatric department with the following clinical complaints:

- Chief Complaints: Neck pain, body aches for two months, fever for three days, and cough for two days.

- Past Medical History: The patient has a history of repeated hospital admissions for respiratory infections from the age of 3 months to 10 years.

**- Vital Signs:**

 - Blood pressure: 90/60 mmHg

 - Pulse rate: 72 bpm

 - Respiratory rate: 20 CPM

 The child appeared active.

- Physical Examination:

 - Cardiovascular system: No abnormality detected (NAD)

 - Respiratory system: Bronchial breathing with clear lung fields.

 **Diagnostic Imaging:**

- 2D Echo and Color Doppler:

**Impression:** Normal findings.

**- USG Abdomen:**

 Impression: Empty left renal fossa.

**- CT Chest**

**Impression:**

 - Hypoplastic right 1st rib with widened intercostal space.

 - Complete assimilation of the anterior and posterior arch of the C1 vertebra.

 - Aplastic right posterior elements and hypoplastic dens of C2 vertebra.

 - Small-sized ectopic and mal rotated left kidney.

These findings suggest a diagnosis of Klippel-Feil Syndrome (KFS), given the vertebral and renal anomalies.

**- Hematology:**

 - RBC: 4.4 million/cubic milliliter

 - WBC: 5.3 thousand/microliter

 - ESR: 5 mm/hr (within normal limits)

 - Differential leukocyte count showed a higher percentage of lymphocytes (62%) and lower neutrophils (24%).

**- Serum Biochemistry:**

 - Serum creatinine: 0.8 mg/dL (normal)

 - Urea: 20 mg/dL (normal)

- **RBC Indices:**

 - MCV: 73.1 fL (low, normal range 83-101)

 - MCH: 22.6 pg (low, normal range 27-31)

 - Hemoglobin: 10.0 g/dL (mild anemia)

 - Hematocrit: 32.4% (mild anemia)

- Urine Analysis: Pale yellow, clear, within normal limits.

 **Pharmacological Treatment:**

- Inj. Amoxicillin Clavum 600 mg IV, TID for respiratory infection.

- Tab Acetaminophen 300 mg, PO, QID for fever and pain relief.

- Syp. Levosalbutamol, Ambroxol, and Guaifenesin 5 ml PO, TID for cough and respiratory symptoms.

- Syp. Mecobalamin 5 ml PO, OD for neurological support.

- Tab. Albendazole 400 mg PO, HS for parasitic infection.

- Tab. Pantoprazole 20 mg PO, OD for gastrointestinal protection.

- Ear drops: Para dichloro benzene and Turpentine oil for ear-related issues.

**DISCUSSION**

Klippel-Feil Syndrome (KFS) is a congenital sickness commonly characterized by way of the odd fusion of two or more cervical vertebrae, resulting in numerous medical manifestations. This syndrome can present with a wide spectrum of symptoms, from moderate and non-unique to more extreme and debilitating situations. The actual path physiology of KFS stays doubtful, but it's miles commonly notion to result from odd segmentation of somites at some stage in early embryogenesis. This disease is usually diagnosed via medical features, radiological findings, and related anomalies. Early recognition and a multidisciplinary management approach are critical for premier affected person outcomes.

 **Clinical Features and Presentation**

The classic scientific features of KFS consist of a quick neck, low hairline, and limited neck movement because of the cervical vertebral fusions. In extreme instances, spinal deformities can result in neurological deficits, inclusive of radiculopathy, myelopathy, and even paralysis. Additionally, many patients with KFS have associated musculoskeletal abnormalities, together with scoliosis, rib abnormalities, or different vertebral anomalies. In this example, our 12-year-antique affected person was offered with nonspecific signs, consisting of neck pain, frame aches, fever, and cough. While those court cases are common in pediatric breathing infections, the presence of neck aches and frame aches raised suspicion for an extra systemic or congenital difficulty.

The prognosis of KFS in our affected person turned into supported by characteristic imaging findings, inclusive of the fusion of the anterior and posterior arches of the C1 vertebra, hypoplastic C2 vertebra with aplastic posterior factors, and the presence of right hypoplastic ribs with widened intercostal spaces. The patient additionally had an ectopic and malrotated left kidney, a not-unusual renal abnormality related to KFS. These findings are in keeping with previous reports within the literature, which describe comparable musculoskeletal and organ system anomalies in KFS.

The patient's recurrent respiration infections in the course of early adolescence (from the age of three months to 10 years) had been specifically noteworthy. Such recurrent infections can occur because of numerous factors in KFS, consisting of thoracic deformities from hypo-plastic ribs or restrained chest enlargement due to spinal deformities. The hypoplastic first rib and the related widened intercostal areas ought to contribute to a reduced thoracic capacity, potentially impeding green breathing characteristics and predisposing the patient to persistent breathing infections.

 **Renal Abnormalities in KFS**

Renal anomalies are usually related to KFS, going on in up to 30-40% of affected people. These encompass conditions like ectopic kidneys, renal hypoplasia, and malrotation, as seen in this affected person. The ectopic and malrotated left kidney determined in the imaging is a key finding that boosts the suspicion of KFS in this case. These renal abnormalities can result in long-term complications, together with urinary tract infections, kidney stones, or maybe renal insufficiency in extreme instances. The empty left renal fossa seen on ultrasound further helps this renal anomaly and could explain the patient's past clinical records of recurrent urinary tract infections or different kidney-related problems that won't have been explicitly documented inside the case history.

Management of renal anomalies in KFS normally includes regular tracking for urinary tract infections (UTIs), renal function, and imaging studies to evaluate the placement and function of the kidneys. In this example, the absence of apparent urinary symptoms does not exclude the opportunity of underlying renal dysfunction, necessitating ongoing monitoring for potential long-term headaches.

 **Respiratory Manifestations and Rib Abnormalities**

The hypo-plastic right first rib and widened intercostal areas located on the chest CT test are full-size findings, as they'll be responsible for the reduced thoracic expansion and potentially chronic respiration issues. Respiratory complications in KFS can be a result of structural thoracic abnormalities, which may result in inefficient ventilation and predispose patients to recurrent infections or breathing difficulties. The patient's current presentation with fever and cough might be indicative of a breathing infection, which inside the context of KFS, might be exacerbated with the aid of those structural deformities. The management of such respiration headaches regularly involves supportive care, together with antibiotics for infections, bronchodilators, and chest physiotherapy. In extreme cases, surgical intervention to correct rib abnormalities can be taken into consideration, although it's miles hardly ever required in pediatric instances.

 **Hematological Findings and Anemia**

The laboratory effects for this patient confirmed a moderate microcytic hypochromic anemia, with a low hemoglobin stage (10 g/dL), low MCV (73.1 fL), and low MCH (22.6 pg). Anemia in KFS may be attributed to various factors, including chronic contamination, negative dietary intake, or renal disorder. Chronic illness anemia is commonplace in people with structural abnormalities and recurrent infections, each of which is found in this situation. Additionally, renal abnormalities, mainly in sufferers with ectopic or hypoplastic kidneys, can result in impaired erythropoiesis or persistent kidney disease, both of which could contribute to anemia.

A thorough evaluation of iron studies, in addition to renal characteristics, is warranted to determine the precise cause of this patient's anemia. Given the slight nature of the anemia, treatment may include iron supplementation and addressing any underlying causes, such as nutritional deficiencies or renal disorders.

**CONCLUSION**

This case highlights the importance of spotting the clinical features of Klippel-Feil syndrome, particularly, while there are related vertebral and renal abnormalities. Early prognosis and multidisciplinary control are essential for enhancing the pleasant of life and stopping complications. The patient's recurrent respiration infections can also be a result of underlying thoracic deformities. Ongoing control with supportive treatment and monitoring for potential complications is essential for this circumstance.

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