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A young girl with short stature and Sprengel deformity: A case report

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Abstract

Klippel-Feil syndrome (KFS) is a rare congenital disorder characterized by the classical triad of a short neck, limited neck mobility, and a low posterior hairline, though all features may not be present in every patient. We report a 5-year-old girl born to consanguineous parents who presented with poor growth and restricted neck movement for three years. Her height was 87 cm (SDS -4.6) and weight was 11 kg (SDS -2.8), indicating severe growth retardation. Clinical examination revealed a short neck, limited cervical movement, low posterior hairline, and congenital elevation of the left scapula. Intelligence was normal for age. Growth hormone stimulation test using clonidine showed a low peak response, while thyroid function tests were normal. Karyotyping revealed a normal female pattern (46, XX). Imaging demonstrated partial fusion of cervical vertebrae (C2-C4), Sprengel deformity, and cervical ribs. Ultrasonography showed absence of the left kidney with an infantile uterus, while echocardiography was normal. The patient was managed conservatively with multidisciplinary evaluation,growth hormone therapy, growth monitoring, physiotherapy, and genetic counseling. The prognosis is variable, and early identification of associated anomalies is essential for optimizing long-term outcomes. *[J Assoc Clin Endocrinol Diabetol Bangladesh,* January 2026; 5(1): *73-76]*

Keywords: Klippel-Feil syndrome, Short stature, Sprengel deformity, Growth hormone deficiency, Congenital cervical fusion

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Introduction

Klippel-Feil syndrome is a congenital disorder resulting from defective segmentation of the cervical spine and is associated with a wide spectrum of anomalies, including short stature and cardiovascular, neurological, genitourinary, and auditory abnormalities.[1-3] The exact etiology remains incompletely understood.[4] Mutations in the GDF6, GDF3, and MEOX1 genes have been implicated in its pathogenesis.[5] We report a case of Klippel-Feil syndrome in a young child highlighting the associated skeletal and endocrine findings in a resource-limited setting.

Case report

A 5-year-old female child born to consanguineous parents presented with poor growth for three years and restricted neck movement. She was born at term by normal vaginal delivery with no history of birth asphyxia or neonatal intensive care admission. All major developmental milestones (gross motor, fine motor, speech-language) were mildly delayed. There was no history of recurrent infections or chronic systemic illness.

Anthropometric assessment revealed severe growth retardation with height 87 cm (SDS −4.6) and weight 11 kg (SDS −2.8) (Figure-1). Clinical examination showed a short neck, low posterior hairline, restricted cervical flexion, extension, and rotation, and congenital elevation of the left scapula (Figure-2). Neurological examination revealed normal motor strength, intact sensation, normal deep tendon reflexes, and no gait abnormality.

Growth hormone stimulation test using clonidine showed a peak value of 3.8 ng/mL, suggestive of a subnormal

Figure-1: Stunted height 87 cm (SDS-4.6), weight 11 kg (SDS-2.8).

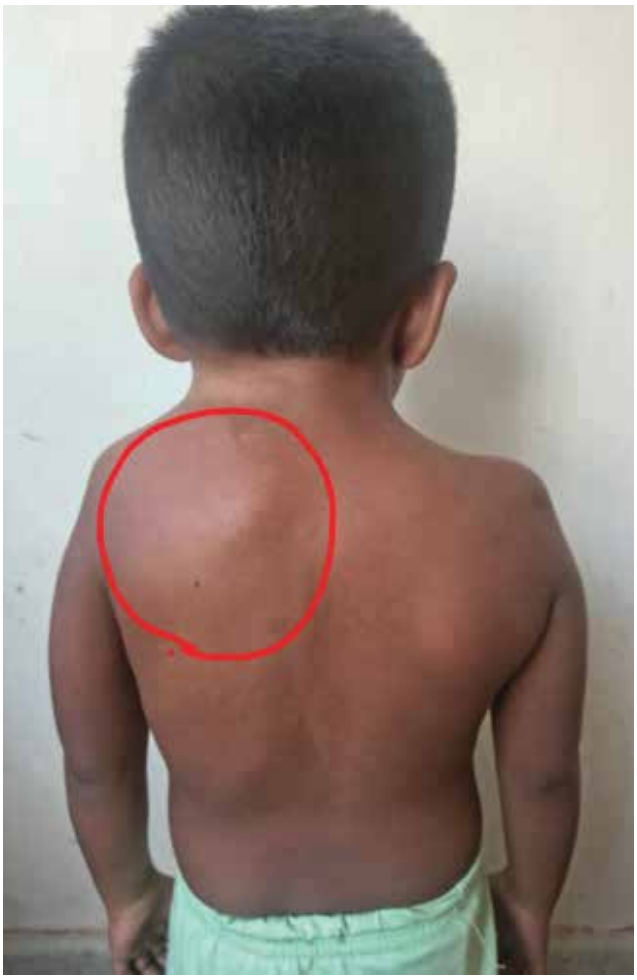

Figure-2: Short neck, Sprengel deformity of the left shoulder joint.

response. Thyroid function tests were normal (Table-I). Karyotyping showed a normal female pattern (46, XX). Ultrasonography of the abdomen revealed non-visualization of the left kidney with a small prepubertal (infantile) uterus. Echocardiography was normal. X-ray of the cervical spine demonstrated partial fusion of C2–C3 and C3–C4 vertebrae, Sprengel deformity of the left scapula, and a right-sided cervical rib (Figure-3). Bone age was delayed (3 years).

Based on clinical, radiological, and laboratory findings, a diagnosis of Klippel–Feil syndrome was made. The patient was managed conservatively with growth hormone therapy, physiotherapy for neck mobility, nutritional optimization, growth monitoring, and counseling of the family regarding prognosis and genetic implications. She is currently under follow-up.

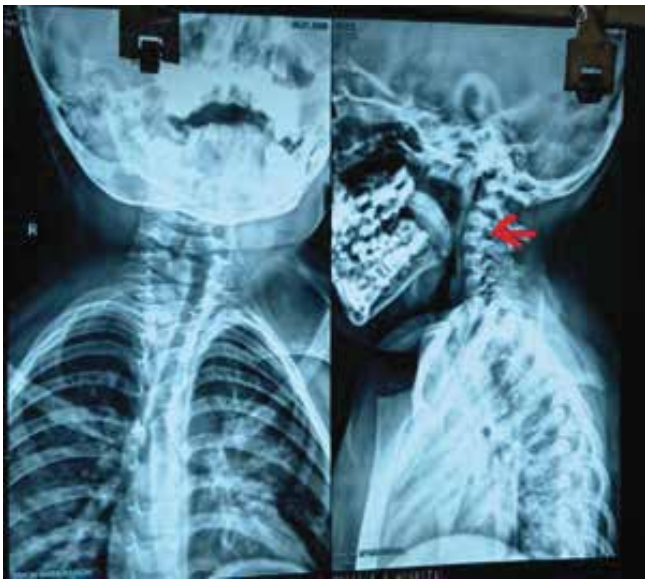

Figure-2: X-ray revealed partial fusion of cervical vertebrae C2-C3 and C3-C4, Sprengel deformity and cervical ribs.

Table-I: Laboratory investigations of the patient

Name of investigations	Result	Reference value
Plasma glucose level (mmol/L)	5.5	<7.8
Serum Sodium (mmol/L)	142	135-148
Serum Potassium (mmol/L)	4.78	3.5-5.5
Serum Chloride (mmol/L)	109.6	98-108
Serum creatinine (mg/dl)	0.7	0.7-1.3
Serum TSH (μIU/ml)	1.86	0.3-5
Serum FT4 (pmol/ml)	13.48	12-22.0
Serum clonidine stimulated Growth hormone (ng/ml)	3.80	>10
Karyotyping	46, XX	-
Ultrasonogram of the whole abdomen	Non-visualized left kidney	-
Color Doppler Echocardiography	Normal study	-

TSH: thyroid stimulating hormone, FT4: free T4

Discussion

Klippel–Feil syndrome is a rare congenital disorder with an estimated incidence of 1 in 40,000-42,000 births and a slight female predominance.[6] The classical clinical triad is present in fewer than half of affected individuals.[7] According to the Samartzis classification, our patient fits Type II, characterized by multiple non-contiguous fused cervical segments.

Several features commonly associated with KFS-such as hearing impairment, congenital heart disease, scoliosis, and neurological deficits-were not present in this patient. The patient's short stature may be attributed to a combination of skeletal abnormalities and possible growth hormone deficiency, although GH deficiency is not a classical feature of KFS and may represent an associated or coincidental finding.

Differential diagnoses considered included Turner syndrome, skeletal dysplasia, and other syndromic causes of short stature; these were excluded based on karyotype and clinical features. The infantile uterus was appropriate for prepubertal age, though long-term follow-up is required to assess pubertal development.

Growth hormone therapy was considered; however, due to limited resources, lack of advanced imaging such as pituitary MRI, and uncertainty regarding long-term benefit in KFS, a cautious approach was adopted. Limitations of this case include the inability to perform advanced imaging such as CT with volume-rendering or MRI of the cervical spine.

This case is unique due to the coexistence of Klippel-Feil syndrome, severe short stature, growth hormone deficiency, unilateral renal agenesis, and infantile uterus in a young child from a resource-limited setting.

Conclusion

Klippel–Feil syndrome requires a high index of suspicion and comprehensive evaluation for associated anomalies. Early diagnosis, multidisciplinary management, and careful endocrine assessment are essential. Children presenting with short stature and neck abnormalities should be evaluated for Klippel–Feil syndrome and associated systemic anomalies, even in resource-limited settings.

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Disclosure

The authors have no conflicts of interest to disclose.

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Data Availability

Any inquiries regarding supporting data availability of this study should be directed to the corresponding author and are available from the corresponding author upon reasonable request.

Ethical Approval and Consent to Participate

Written informed consent was obtained from the patient's attendant. All methods were performed in accordance with the relevant guidelines and regulations.

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