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UNVEILING THE FUSION: AN ATYPICAL PRESENTATION OF KLIPPEL-FEIL SYNDROME

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ABSTRACT

Klippel- Feil syndrome is a congenital condition characterized by the abnormal fusion of two or more cervical vertebrae, resulting in a short neck. The condition presents with a classical triad of a short neck, low posterior hairline and limited mobility of the neck.The condition has been associated with several spinal and extra-spinal anomalies. The diagnosis usually involves a comprehensive evaluation, including physical examination, radiological imaging and genetic studies. Treatment is usually conservative involving physical therapy and pain management. Here, we report a case of a 5 year old boy who presented to us with complaints of short stature and limited neck mobility. On examination the child had kypho-scoliosis, right sprengel deformity, low posterior hairline, short neck and limited neck mobility. MRI of cervical spine with whole spine screening was suggestive of congenital block vertebrae at C2 - C4 levels and localized area of myelomalacia within the posterior aspect of the cervical cord at C2, C3 levels with mild kypho-scoliosis of cervico-dorsal area. The child had no neurological or extraspinal involvement. The child was managed conservatively.

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INTRODUCTION

Klippel-Feil (KF) syndrome was described by Maurice Klippel and Andre Feil in 1912 in a patient with congenital fusion of cervical vertebrae.[1]KF occurs in one out of 42,000 births and 60% occurs in girls.[2]KF is a complex, congenital condition that occurs due to abnormal fusion of two or more vertebrae due to failure of proper segmentation during early fetal development. The syndrome is classically a triad of short neck, low posterior hairline and limited range of neck movements. The triad is present in less than 50% of the cases.[3]Type I KF syndrome involves a single congenitally fused segment whereas type II involves multiple, non-contiguous congenitally fused segments and type III consists of multiple, contiguous, fused segments. Most common is type II deformity that occurs in about 50% and type I and III occur in 25% each.[3]KF syndrome has been associated with multiple spinal and extra-spinal abnormalities likespinal stenosis, neurological deficits, cervical spine deformities, and instability. Almost all cases of KF syndrome occur sporadically but a close evaluation of first degree family is recommended.

Since KF syndrome is rare with only limited case reports from India, we are reporting the present case to add to the literature on this condition.

CASE REPORT

A 5 year old boy presented to our outpatient department (OPD) with complaints of short stature and limited neck mobility, as noticed by the parents at the age of 2 years. The child had no neck pain or trauma. There was no antenatal history of fever, hypertension or anydrug intake. The child was born out of a non- consanguineous marriage by normal vaginal delivery at term and there was no significant peri- natal or post- natal history. There was no history of any language, social or motor developmental delay. Diet history revealed low calorie and protein intake (950kcal/day, 25 g/day respectively) for age. On examination the blood pressure was 98/60 mm Hg (both systolic and diastolic BP between 50-90th percentile) with a pulse rate of 92/min. On general examination there was a low posterior hair line, short neck with neck deformity (right> left), right trapezius upper border was shorter as compared to left, sprengel

deformity was presenton right side and there was kypho-scoliosis of the cervico-dorsal vertebrae (Fig 1A,1B). The range of movement of cervical spine was limited; the child could perform full flexion but extension, rotation and lateral bending could be performed only partly. The intelligence quotient was normal in all domains.

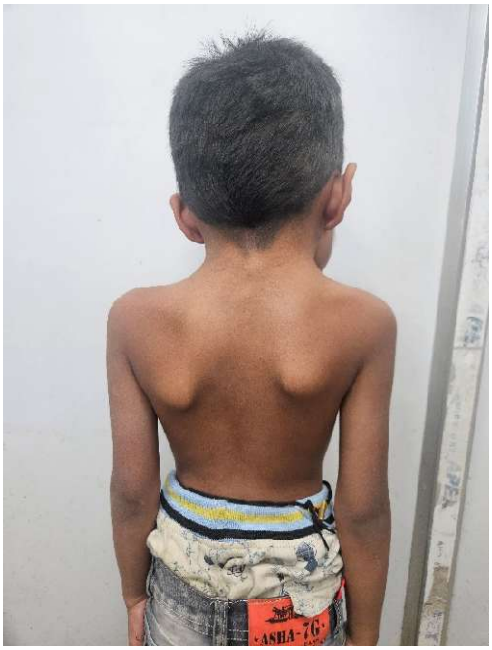

Fig. 1. A- Image of the patient showing the right Sprengel deformity and kyphoscoliosis

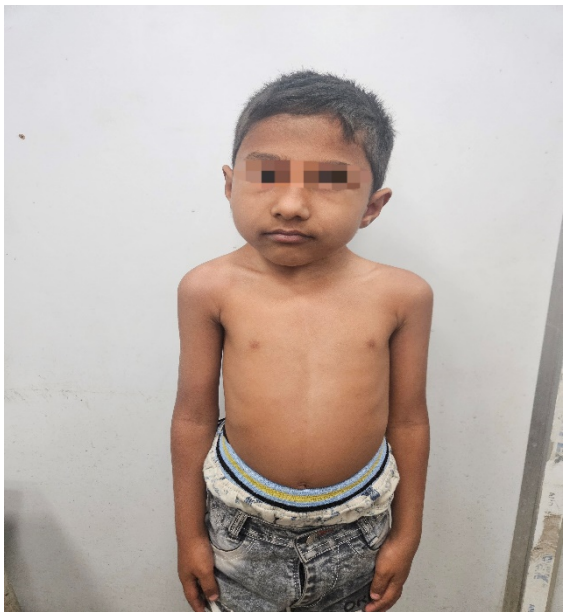

Fig. 1. B- Right neck deformity

Table 1. Anthropometric measurements of the patient

Anthropometric Parameter (Unit)	Measurement	Standard deviation as Per Age & Sex
Height (cm)	102.3	-2.6 SD
Weight (Kg)	10	-3.5 SD
BMI (kg/m2)	10.7	Less than 3rd centile
Upper segment : Lower segment	1.06	
Target height (cm)	162.4	-1.46 SD
Arm span (cm)	102	
Right upper limb length (cm)	44.5	
Left upper limb length (cm)	46	
Head circumference (cm)	51	

Central nervous system examination revealed no neurological deficit and other systemic examination was normal.

Table 2. Biochemical and hormonal evaluation of the patient

Investigations (Units)	Patient's Value	Reference Range
Hemoglobin (g/dl)	13.2	13-16
Total counts (cumm)	8000	4000-10,000
Platelet count (lakh/cumm)	2.6	2.0-4.0
Serum creatinine (mg/dl)	0.4	0.4-1.2
Serum sodium	138	135-145
Serum potassium	4.1	3.5-4.5
Total bilirubin (mg/dl)	0.4	0.3-1.2
Direct Bilirubin (mg/dl)	0.2	0-0.3
Alanine aminotransferase(ALT) (U/L)	15	15-45
Aspartate aminotransferase (AST) (U/L)	26	15-45
Alkaline phosphatase (ALP) (U/L)	253	45-160
Free T4 (ng/dl)	1.9	0.89-1.8
TSH (mIU/ml)	0.9	0.3-4.6
Serum calcium (mg/dl)	10.3	8.9-10.5
Serum phosphate (mg/dl)	6.3	3.0-4.5
Serum vitamin D (ng/ml)	12.6	>30
Serum IGF-1(ng/ml)	103 (50th -75th centile for age)	34.2-203.9

TSH- Thyroid stimulating hormone, T4- thyroxine, IGF-1- insulin like growth factor-1

Fig.2- Image showing short height on anthropometric measurement

X- ray spine was suggestive mild kypho- scoliosis of cervico- dorsal area of the spine (Fig.2.A, 2.B). MRI of cervical spine with whole spine screening showed congenital block vertebrae at C2- C4 levels and localized area of myelomalacia within the posterior aspect of the cervical cord at C2, C3 levels with mild kypho-scoliosis of cervico-dorsal area (Fig. 3).

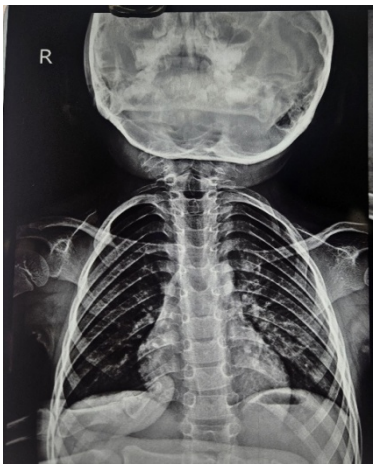

Fig. 3. A- X-ray chest postero-anterior (PA) view showing kypho-scoliosis

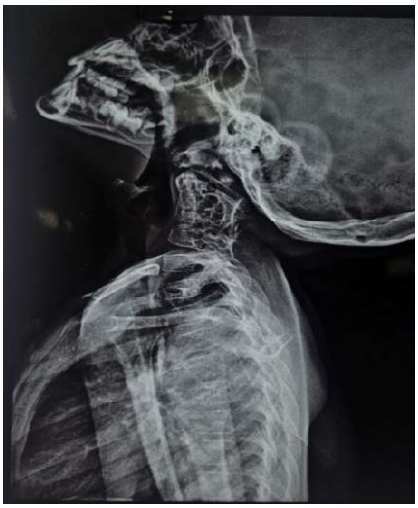

Fig. 3. B- X-ray cervical spine showing block vertebrae at C2-C4

Ultrasound abdomen and pelvis and 2D Echocardiography were found to be normal. Neuromedicine and Neurosurgery opinion was sought and since the child had no neurologic deficit on examination, conservative management by corrective cervical collar, neck and shoulder exercises for 5-6 times/day was advised. The prognosis was explained in detail to the parents of the child.

DISCUSSION

Our patient is a 5 year old boy who presented with short stature and restricted neck mobility without any neurological deficit. MRI of cervical spine with whole spine screening was suggestive of congenital block vertebrae at C2- C4 levels and localized area of myelomalacia within the posterior aspect of the cervical cord at C2, C3 levels with mild kypho-scoliosis of cervico- dorsal area.

He was diagnosed with KF syndrome and managed conservatively as there was no neurological deficit. KF syndrome is also known as synostosis of the cervical spine. It occurs due to failure in normal segmentation of cervical mesodermal somites during embryonic development.[4] This usually occurs at 2 to 8 weeks of gestation.[5]The most common presentation is short neck, torticollis and restricted neck movements. It has been proposed that the syndrome occurs due to a combination of genetic and environmental factors, though the cause remains unknown.Various genetic mutations like GDF6 and GDF3 with autosomal dominant inheritance, and MEOX1 with autosomal recessive inheritance have been reported in KF syndrome. [3]The syndrome has spinal and other associated abnormalities like anomaly of kidneys (renal aplasia), ribs, cleft palate, jaw abnormalities like mandibular hyperplasia, respiratory abnormalities and congenital heart anomalies (coarctation of aorta, atrial, ventricular septal defects, transposition of great arteries, situs inversus and vertebral artery anomalies).[6]A variety of other syndromes like fetal alcohol syndrome, Goldenhar syndrome, syringomyelia and anomalies of extremities might be associated.[7][8]The complex disease requires lifelong treatment by multidisciplinary teams.

CONCLUSION

Klippel- Feil syndrome usually has a female predominance and the classical triad occurs in less than 50% Our patient was a boy with the classical triad with characteristic imaging findings without any neurodeficit. This case report will add to the existing information on this rare syndrome.

REFERENCES

1. Jones, K.L., Jones, M.C., Del Campo, M. 2021. Smith's recognizable patterns of human malformation. *Elsevier Health Sciences*. Mar 2.
2. Stephen, R. Pledger. *Ortho Secrets.* 3rd Ed 2003. Cervical Spine Disease.
3. Agarwal, A.K., Goel, M., Bajpai, J., Shukla, S., Sachdeva, N. Klippel Feil Syndrome: A Rare Case Report. *J Orthop Case Rep*. 2014 Jul-Sep; 4(3):53-5.
4. Vaidyanathan, S., Hughes, P.L., Soni, B.M., Singh, G., Sett, P. Klippel-Feil syndrome - the risk of cervical spinal cord injury: A case report. *BMC FamPract.* 2002; 3:6.
5. Boraz, R.A., Irwin, D.H. and Van Blarcom, C. The dental rehabilitation of a patient of klippel-feil syndrome and sprengel's deformity. *Spec Care Dentist.* 1986; 6(1):22–4.
6. Schilgen, M., Loeser, H. Klippel-Feil anomaly combined with fetal alcohol syndrome. *Eur Spine J.* 1994; 3(5):289–90
7. Stadnicki, G., Rassumowski, D. 1972. The association of cleft palate with the Klippel-Feil syndrome. *Oral Surg Oral Med Oral Pathol.,* 33(3):335–40.
8. Tredwell, S.J., Smith, D.F., Macleod, P.J., et al. 1982. Cervical spine anomalies in fetal alcohol syndrome. *Spine.,* 7(4):331–4.

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