Klippel-Feil syndrome with multiple skeletal anomalies, Dandy-Walker spectrum, and occipital cephalocele—a rare presentation

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Background:
Klippel-Feil syndrome (KFS) is described as a congenital malformation causing fusion of at least two cervical vertebrae and is often characterized clinically by presence of triad of short neck, limited neck movements, and low posterior hair line. It occurs due to the failure of the segmentation of mesodermal somites during the 3rd to 8th week of intra-uterine life [1]. The exact etiology is unclear; however, certain genes such as GDF6, GDF3, MEOX1, and RIPP LY2 responsible for regulation of transcription and signaling pathways of somite development are thought to be causative in the pathogenesis [2]. The estimated incidence is 1 in 40,000–42,000 with a female predilection [2]. The disorder does not just show fused cervical vertebrae but can have multi-system involvement presenting as multiple skeletal and non-skeletal anomalies.

Case presentation:
A 6-year-old boy presented to our institute with complaints of restricted neck movements, deviation of the neck towards the right side, restricted range of motion at right shoulder joint, and deformity of thoracolumbar spine since birth. The patient was one of the three children of a couple from non-consanguineous marriage. In
antenatal history, the mother did not receive any ante-
natal care, and the child was born via normal vaginal de-
livery at home. There was no family history of spinal or
neural anomalies (Figs. 1, 2, 3, 4, 5, 6, and 7).

Physical examination demonstrated short neck, low
posterior hairline, torticollis with a head tilt towards the
right side and contralateral chin lift, elevated right scap-
ula, visible scoliotic deformity of the thoracolumbar
spine with convexity towards the left side. There was
also a soft tissue swelling in the occipital region of the
scalp.

The patient was referred to our department for radion.
graphs of chest and spine which revealed elevated right
scapula (Rigault’s classification grade II) and right-sided
hemivertebrae at D9 and L5 with resultant scoliotic
deformity with convexity towards the left side. Further
imaging evaluation with non-contrast computed tomog-
raphy (NCCT) was advised for better characterization of
vertebral anomalies and other associated skeletal de-
formities. Magnetic resonance imaging (MRI) of brain
and whole spine was also performed for characterization
of scalp swelling and to detect any associated central
nervous system (CNS) anomalies. Abdominal ultrason-
ography and echocardiography were done to rule out
renal and cardiac anomalies respectively which were
normal.

NCCT of the spine revealed scoliotic deformity of the
spine with convexity towards the left side in the dorso-
lumbar spine and convexity towards the right side in the
cervico dorsal spine. Multiple skeletal anomalies which
were not appreciable on plain radiographs were also de-
tected. These included CVJ anomalies (hypoplastic right
occipital condyle, type A defect of the posterior arch of
atlas and anterior arch cleft, hypoplastic odontoid
process with atlantoaxial instability, and spinal canal
stenosis), multiple segmentation and formation anomal-
ies of the spine (fused posterior elements of C4-C5 and
C5-C6; non-fusion of posterior elements of C4, C5, C6,
C7, and L5 (spina bifida occulta); right-sided hemiver-
tebrae at D1, D9, and L5 with hypoplastic left-sided pos-
terior elements of D9 and L5). Right scapula and right
clavicle were elevated lying between C5 and D4 levels
with a thick bony bar seen arising from the fused right-
sided neural arch of C5-C6 and extending posterolater-
ally towards superomedial part of elevated scapula (omo-
vertebral bone). Multiple rib anomalies were also
observed which included right-sided cervical rib, bifid
left-sided first rib, fused left-sided second and third ribs
near vertebral attachment, absent left 9th rib, and bilat-
eral 12th ribs.

MRI brain revealed isolated inferior vermian hypopla-
sia with a cerebrospinal fluid (CSF)-filled cystic structure
communicating with the fourth ventricle in posterior
fossa. The findings were consistent with Dandy-Walker
spectrum. There was also the presence of an occipital
scalp swelling with areas of CSF signal intensity, which
is seen to communicate with enlarged posterior fossa
cyst through a calvarial defect. There was also associated
vertical orientation of straight sinus. These findings
were consistent with atretic cephalocele. MRI spine revealed
kinking of cervicomedullary junction, obliteration of
both anterior, and posterior subarachnoid spaces; how-
ever, normal signal intensity is noted within the spinal
cord. USG abdomen did not reveal any abnormalities.

![Fig. 1](image_url)  
**Fig. 1** a Photographs of a 6-year-old patient with Klippel-Feil syndrome and Sprengel deformity showing the clinical profile of the patient showing short neck and elevation of right scapula. b There is also torticollis with head tilt towards right side, visible scoliotic deformity of thoracolumbar spine and a scalp swelling. c AP radiograph of chest and abdomen shows Sprengel deformity with hemivertebra at D1, D9, and L5 and resultant scoliotic deformity.
Echocardiography was also normal. The patient was being considered for genetic testing for genes associated with KFS; however, due to financial constraints of the patient, it could not be done.

The patient was operated for Sprengel deformity following which he had significant improvement in range of motion at the shoulder joint. The parents were explained about the prognosis of associated anomalies and are put on follow-up for further management of scoliosis and planned excision of atretic cephalocele.

Discussion
In the year 1912, Maurice Klippel first described Klippel-Feil syndrome (KFS) as a congenital malformation causing fusion of cervical vertebrae which can involve either two vertebrae or entire cervical spine. In the same year, Feil classified KFS into 3 types based on the degree of involvement as type I (massive fusion of cervical and upper thoracic vertebrae), type II (fusion of 2 or more vertebrae with associated atlanto-occipital assimilation, hemivertebrae, or any other anomaly of the cervical spine), and type III (fused cervical vertebrae with lower thoracic or lumbar vertebrae). Diagnosis can be suggested by classical findings described as a triad of restricted neck movements, short neck, and low posterior hairline; however, complete triad is seen in only about 50% of the patients [3].

Various osseous and non-osseous anomalies are associated with KFS, the most common being scoliosis seen in 70% of cases [4]. Other commonly associated skeletal anomalies include Sprengel deformity (20–30%), spina bifida occulta (45%), and craniovertebral junction anomalies; all of these were seen in our patient [5]. Due to fusion anomalies (fused posterior elements of C4-C5 and C5-C6 vertebrae) and multiple hemivertebrae at D1, D9, and L5 seen in the patient, our patient fit the criteria for type II KFS [3].

Fig. 2  

![Sagittal reformatted non-contrast CT image of the cervical spine shows fusion of posterior elements of C4-C5 (white arrow).](image1)  

![Sagittal reformatted non-contrast CT image of the cervical spine shows fusion of posterior elements of C5-C6 (white arrow).](image2)  

![Coronal reformatted CT image shows elevated right scapula with its superomedial angle lying between C5 and D2 vertebral levels suggestive of grade II Sprengel deformity (red arrow).](image3)  

![Axial bone window CT image shows a thick bony bar arising from fused C5-C6 neural arch and reaching up to the superomedial angle of scapula suggestive of omovertebral bone (yellow arrow).](image4)
Sprengel deformity is classified radiographically into 3 grades based on the position of the superomedial angle of scapula, given by Rigualt [6]. This includes grade I (between D2 and D4 transverse processes), grade II (between C5 and D2 transverse processes), and grade III (above C5 transverse process). Our patient belonged to grade II Sprengel deformity. Sprengel deformity is often fixed with adjacent vertebrae by presence of omovertebral bone which limits the necessary scapular rotation needed for overhead arm abduction. Omovertebral bone was seen in our patient extending from neural arch of C5-C6 to superomedial part of elevated scapula. The patient underwent resection of omovertebral bone following which he gained a significant range of motion at shoulder which was previously limited.

Among other skeletal anomalies, our patient also had multiple rib anomalies which included cervical rib, adjacent fused ribs, bifid ribs, and absent ribs. Rib anomalies are seen rarely in association with KFS with few case reports in the literature.

Apart from skeletal anomalies, associated common visceral anomalies in KFS include renal anomalies which usually occur in the form of unilateral agenesis, CNS abnormalities such as meningocele, syringomyelia,
Fig. 4  

**a** Non-contrast sagittal reformatted CT image shows hypoplastic odontoid process with associated atlantoaxial instability and increased atlanto-dens interval.  
**b** Axial non-contrast CT image shows anterior arch cleft and midline cleft in the posterior arch of atlas suggestive of type A defect.  
**c** Coronal reformatted CT image shows hypoplasia of right occipital condyle (white arrow) with widened Schmidt Fischer angle.  
**d** Sagittal T2- weighted MRI image of the same patient showing kinking of cervicomedullary junction with obliteration of anterior and posterior subarachnoid spaces.

Fig. 5  

**a** Axial non-contrast CT image shows presence of hypoplastic right cervical rib (white arrow).  
**b** Axial non contrast CT image show presence of bifid left-sided 1st rib (white arrow).  
**c** Volume rendered three-dimensional CT images of the same patient demonstrating fused left 2nd and 3rd ribs (red arrow) and presence of aplastic left 9th rib and bilateral 12th ribs.
Fig. 6  
(a, b) Sagittal T1-weighted and T2-weighted MR images of the brain demonstrate Dandy-Walker spectrum with isolated inferior vermian hypoplasia with counter clock wise rotation of superior vermian remnant (white arrow).  
(c, d) Axial T2 weighted images of the same patient demonstrate a posterior fossa cyst communicating with the 4th ventricle.

Fig. 7  
(a) Non-contrast axial bone window CT image demonstrates a bony calvarial defect in the occipital bone.  
(b, c) Sagittal T1-weighted and axial T2-weighted MR images show an occipital scalp swelling with areas of CSF signal intensity within (yellow arrow). It is seen to communicate with posterior fossa cyst through the calvarial defect with associated vertical orientation of straight sinus (red arrow) suggestive of atretic cephalocele.
compressive myelopathy, diastematomyelia in the spinal cord and occipital cephalocele, Chiari malformation type I, and hydrocephalous in the brain have also been observed in patients with KFS. Auditory anomalies and congenital heart diseases are also commonly seen. The visceral findings seen in our patient were atretic occipital cephaloceles, compressive myelopathy, and Dandy-Walker spectrum. Dandy-Walker malformation is a rare association of KFS. In an extensive literature search, we found only two reports of association of Dandy-Walker malformation with Klippel-Feil syndrome, one of which is reported in antenatal USG and the other was a 6-day-old infant [7, 8]. Dandy-Walker spectrum refers to a group of disorders that do not fulfill criteria for Dandy-Walker malformation and are characterized by inferior vermic hypoplasia with poorly formed fourth ventricle communicating with posterior fossa cyst. The abnormality rarely causes any specific symptoms and is characterized only in imaging studies.

**Conclusion**
The radiologists while evaluating a case of Klippel-Feil syndrome should be aware of the full possible spectrum of associated skeletal and non-skeletal anomalies which guides the treatment planning and has prognostic significance. Therefore, our case report highlights the importance of knowledge of various common and uncommon associations of KFS to avoid missing significant anomalies while reporting.

**Abbreviations**
KFS: Klippel-Feil syndrome; CVJ: Craniovertebral junction; NCCT: Non-contrast computed tomography; MRI: Magnetic resonance imaging; AP: Anteroposterior; CNS: Central nervous system; CSF: Cerebrospinal fluid

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**Authors’ contributions**
MS obtained the patient data, analyzed the images, designed, and drafted the manuscript. MSS and GP revised the manuscript critically for important intellectual content. The authors read and approved the final manuscript.

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**Competing interests**
The authors declare that they have no competing interests.

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