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Klippel–Feil syndrome associated with a craniocervico-thoracic dermoid cyst

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Abstract

Background: Uncommonly, Klippel–Feil syndrome (KFS) has been associated with intracranial or spinal tumors, most frequently dermoid or epidermoid cysts. Although the associated dermoid cyst (DC) is usually located in the posterior fossa, isolated upper cervical DC has been reported. Extension from the posterior fossa to the upper cervical spine (C2) has been reported once. We report a rare case of KFS in association with a posterior fossa DC that extended down to the upper thoracic spine and review the current literature.

Case Description: A 47-year-old female with presented cervical myelopathy related to a cranio-cervico-thoracic DC in association with KPS-related cervicothoracic fusion (C2-T6) and thoracic kyphosis. The patient underwent complete tumor resection following sub-occipital craniectomy and C1-C4 cervical laminectomy. The patient exhibited complete resolution of symptoms with no tumor recurrence and no deformity at 6-year follow-up.

Conclusion: DC should be added to the list of congenital central nervous system abnormalities, which should be sought in patients with KFS. Therefore, the presence of a cystic lesion in the posterior fossa, the craniocervical junction or the anterior cervical spine should suggest the possibility of a DC in patients with KFS. In cases of cranio-cervical DC, the tumor may extend quite far down the spinal column (reaching the thoracic spine), as demonstrated in the present case.

Key Words: Congenital spinal malformation, craniocervical junction, dermal sinus, dermoid cyst, Klippel–Feil syndrome

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INTRODUCTION

Klippel–Feil syndrome (KFS) is a congenital spinal malformation characterized by the failure in segmentation of two or more cervical vertebrae due to an abnormal division of the mesodermal somites.[2,4,6,10] KFS may be associated with other developmental defects involving various tissues.[3,7,10] Uncommonly, KFS has been associated with intracranial or spinal tumors, most frequently dermoid or epidermoid cysts. Dermoid cyst (DC) development is thought to result from entrapment of dermal elements and their appendages within the closing neural tube, which occurs during the same period as somite development.[7,10] In these cases, the

DC is usually located in the posterior fossa, especially in children. Although its isolated occurrence in the cervical spine has been reported twice in patients with KFS, DC extension from the posterior fossa to the upper cervical spine (C2) has been reported only once.[5] We report a unique case of KFS in association with a posterior fossa DC that extended to the upper thoracic spine (T1) and review the current literature.

CASE REPORT

Clinical complaints and neurological findings

A 47-year-old female in good health was referred for progressive gait disturbance. At birth she had been operated for a palatal fissure and had been diagnosed with KFS at 1 year of age. Two months prior to presentation, the patient developed bilateral lower extremity paresthesias associated with progressive paraparesis.

On physical evaluation, the patient had a short webbed neck, a low occipital hairline, and restricted neck mobility. She did not report any symptoms and did not present any neurological deficits in her upper extremities. On examination she exhibited bilateral proximal paresis (4/5) in the iliopsoas and quadriceps muscles. Her motor deficit was accompanied by marked patellar hyperreflexia (4+), but without hyperactive Achilles responses (no clonus) or Babinski responses. Her sensory deficit included a loss of position and vibration appreciation in the distal lower extremities involving the ankles but not the knees, decreased superficial touch sensation in the L1-L3 distributions, and decreased pin-prick sensation on the left hemibody from the clavicle down and on the right hemibody from the lower thorax down. She also exhibited difficulty with tandem gait testing, falling to either side, while finger-nose-finger remained intact.

RADIOGRAPHIC STUDIES

Plain X-rays

Plain cervical radiographs (anterolateral, Lateral) demonstrated spontaneous/congenital fusion of all vertebral bodies from C2 down to T6 associated with significant 40° thoracic kyphosis centered at T4-T5. No instability was documented on flexion-extension studies throughout her spine, notably not at occiput-C1 or C1-C2 [Figure 1].

Computed tomography findings

Computed tomography (CT) of the head, cervical, and thoracic spine confirmed congenital Klippel–Feil fusion of the cervical and thoracic vertebral bodies from C2 to T6, involving the anterior spinal column and posterior elements which were fused 'en bloc' [Figures 2 and 3]. Most notably, the CT documented a circumscribed lesion with heterogeneous texture enhancing after contrast infusion lying dorsal to the fourth ventricle and extending down to the T1 level. In the posterior fossa, the lesion obstructed the foramen of Magendie, thus resulting in secondary obstructive hydrocephalus. The head CT also documented a low lying venous confluence. A CT angiogram showed that both vertebral arteries were located anterior to the lesion [Figure 4].

Magnetic resonance imaging examinations

Cerebral and cervical magnetic resonance imaging (MRI) documented a craniocervical mass that measured 3.2 cm anteroposteriorly, by 8.4 cm rostrocaudally [Figure 5]. The tumor was heterogeneous with iso-, hypo- and hyper-intense areas on T1 and T2 weighted images. The lesion deformed the medulla, and displaced it caudally. The compressed cervical spinal cord measured 2.5 mm in largest anteroposterior dimension and was displaced anterolaterally. Furthermore, a sinus tract was suspected on the T2 weighted images

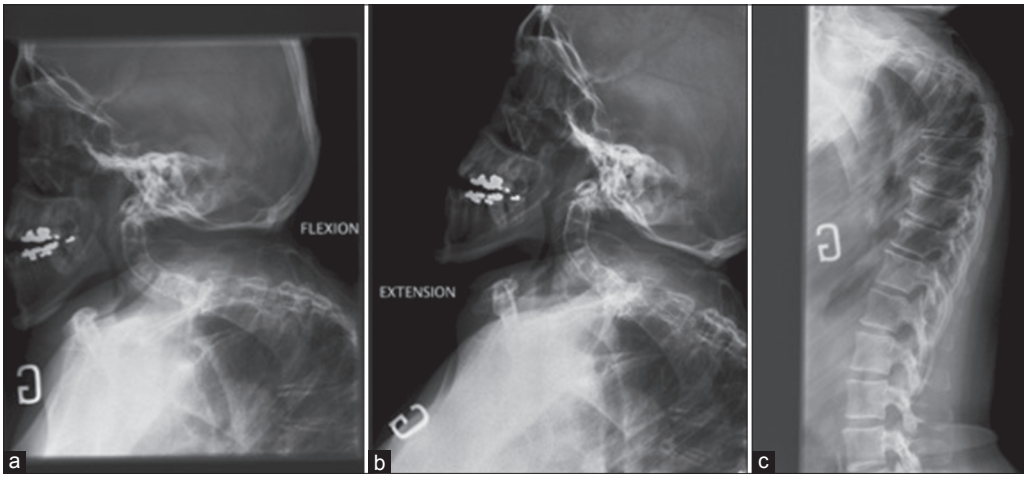

Figure 1: (a, b) Radiographs of the cervical and thoracic spine demonstrating fused vertebral bodies from C2 down to T6. No instability was documented on flexion-extension studies, (c) Significant kyphosis at the higher thoracic spine

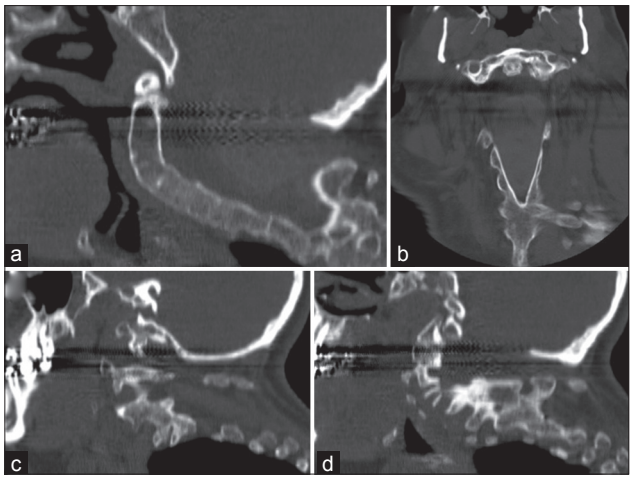

Figure 2: (a-d) Pre-operative cranio-cervico-thoracic CT scan showing a mass in the posterior fossa behind the fourth ventricle and below the cerebellum extending down behind the spinal cord to the posterior thoracic vertebra to T1. In the posterior fossa, it obstructed the foramen of Magendie, thus resulting in secondary obstructive hydrocephalus. In addition, fusion of the cervical and thoracic vertebral bodies from C2 to T6, involving the anterior spinal column and posterior elements is seen

involving the suboccipital region extending from the dermis to the suboccipital bone [Figure 5c].

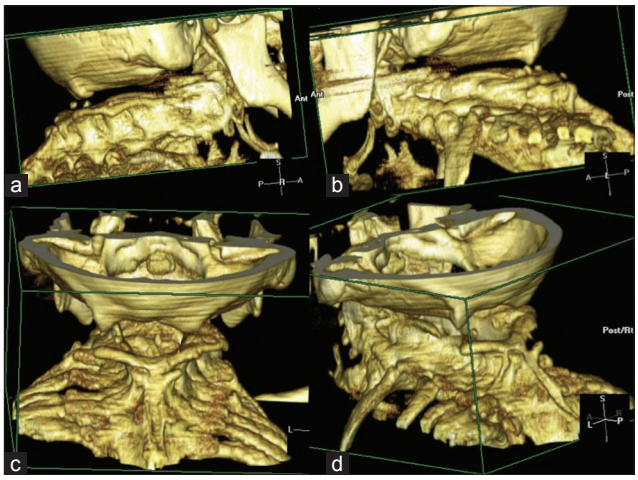

Figure 3: (a-d) Preoperative CT scan with 3D-reconstruction showing the osseous anomalies, including C2 to T6 'en bloc' fusion

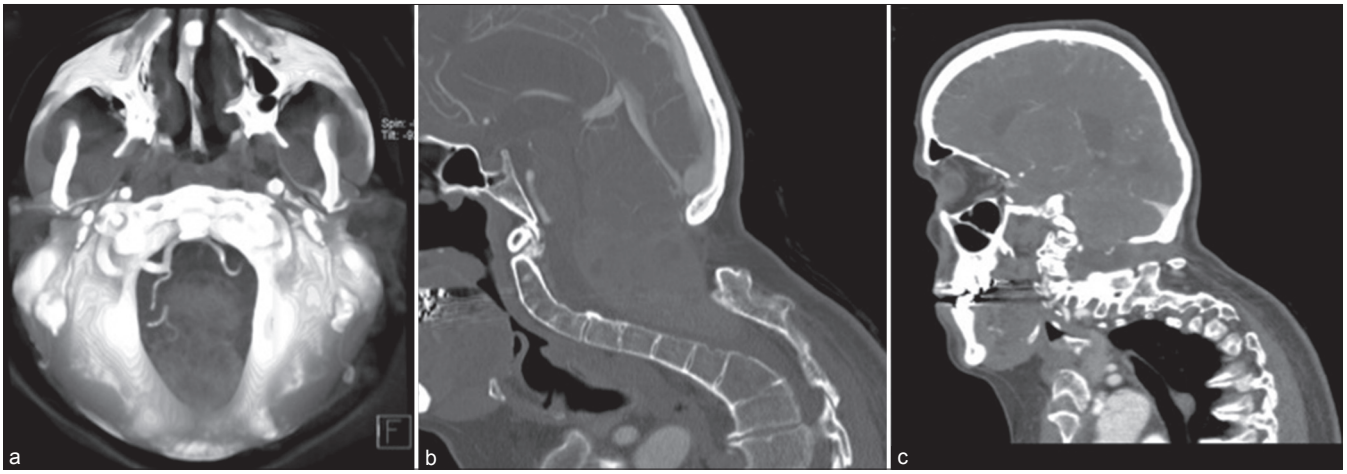

Figure 4: (a-c) CT-angiography showing the vertebral arteries located anterior to the lesion in addition to a low-lying venous confluence

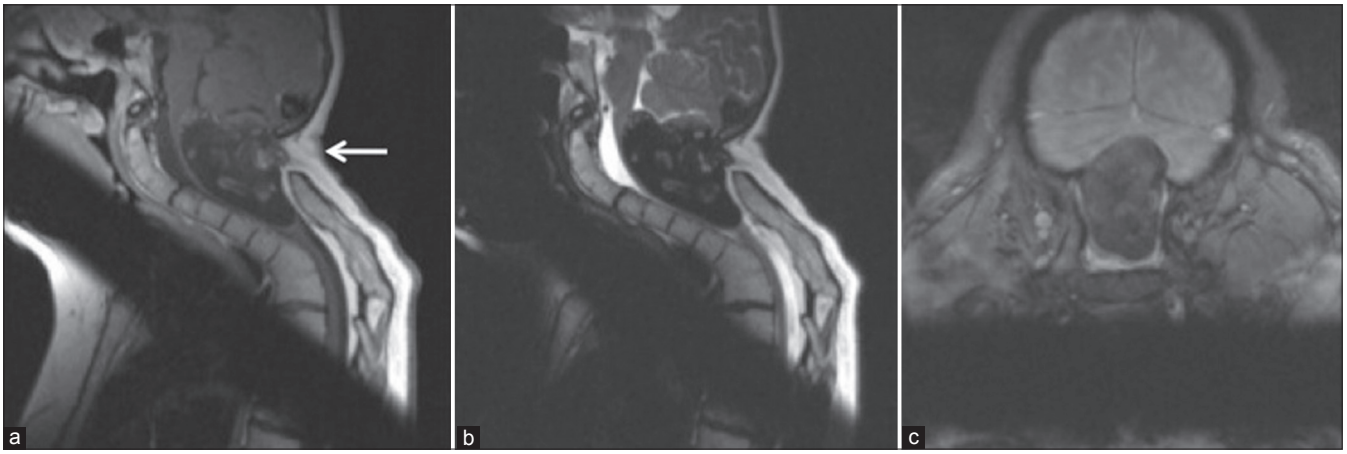

Figure 5: Pre-operative magnetic resonance imaging, T1WI sagittal (a), T2WI sagittal (b), and Stir axial (c) sections documenting an expansive midline lesion extending from the cranio-cervical junction down to T1. The dermal sinus tract, extending from the dermis to the suboccipital bone below the occipital protuberance is identified on T1WI and T2WI (arrow)

Cervical surgery

The patient underwent awake fiberoptic endotracheal intubation and prone positioning. There was no electrophysiological monitoring. During preparation of the sub-occipital region, a small dimple located below the occipital protuberance and connecting to a sinus tract during preparation of the sub-occipital region, a small dimple located below the occipital protuberance and connecting to a sinus tract was identified [Figure 5c] was clearly identified. A sub-occipital craniectomy and cervical laminectomy was performed to expose the lesion. However, the normal anatomy was distorted as the posterior elements of the cervical spine extending into the thoracic spine, including the C2-T6 spinous processes, laminae and facet joints were replaced by a unified mass of bone. After performing a laminectomy from C1 to C4 and opening the dura, a large midline cystic mass filled with thick yellowish fluid, hair and keratin debris was found [Figure 6]. The tumor was completely removed using the operative microscope and microinstrumentation. No fusion or subsequent instrumentation was required. The histopathological examination confirmed the diagnosis of a dermoid tumor.

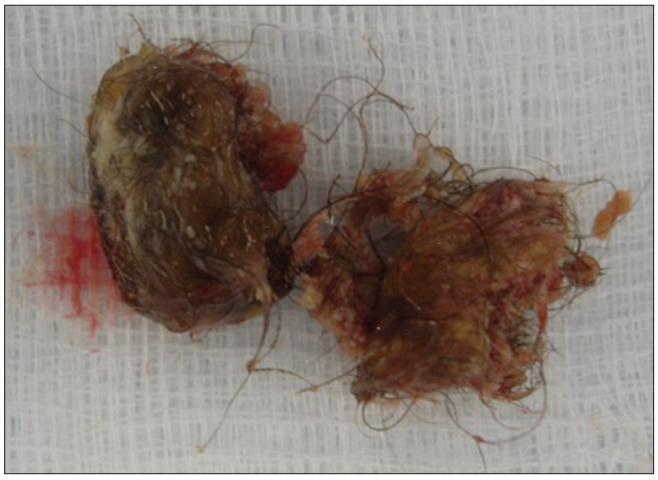

Figure 6: Intraoperative image of the dermoid cyst

Postoperative course

Post-operatively, the patient's motor exam was normal and her sensory exam, notably superficial touch and proprioception modalities, significantly improved. The MRI performed 4 days after surgery documented complete resection of the DC [Figure 7]. Additionally, dynamic X-rays performed within the first postoperative week documented no instability. Six years postoperatively, the patient is doing very well, and remains symptom-free, without evidence of deformity on plain radiographs [Figure 8] or tumor recurrence on follow-up MRIs studies [Figure 9].

DISCUSSION

Klippel–Feil syndrome triad and other congenital anomalies

KFS was initially described as a triad of short neck, limited neck mobility and low posterior hairline due to the congenital fusion of two or more cervical vertebrae. The classic triad is seen in approximately 52% of patients with KFS.[3] Patients with KFS also present abnormalities affecting other organ systems.[3,7,10] Cardiovascular anomalies have been detected in 4.2-14%, while genitourinary anomalies are identified in up to 64% of patients with KFS.[3,7]

Neurological anomalies seen with KFS

Mulitple neuronal tissue anomalies have also been described in patients with KPS. These include: diastematomyelia, syringomyelia, agenesis of the corpus callosum, meningocele, cervical occult spina bifida, intramedullary lipoma, extradural hemangiolipoma.[1] Intracranial or spinal tumors associated with KFS are believed to be rare.

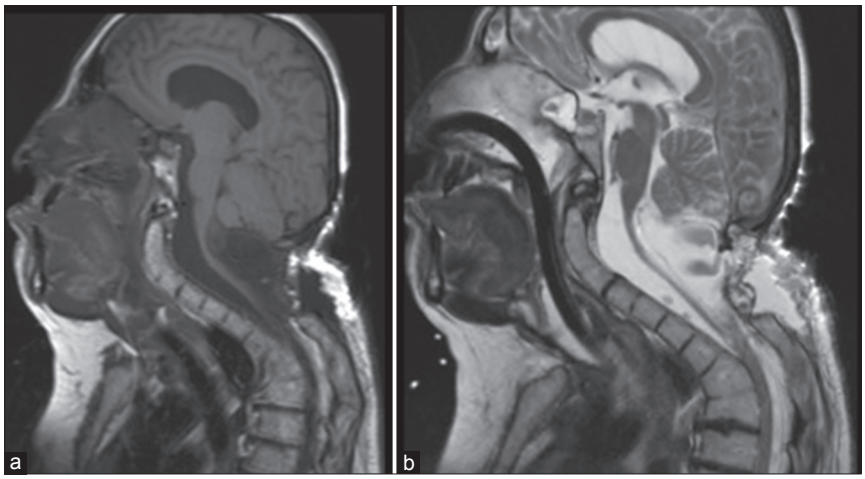

Figure 7: Postoperative magnetic resonance imaging, T1WI sagittal (a), T2WI sagittal (b) sections and T1WI axial

However, a review of the literature revealed 26 cases (including the present case) of KFS associated with DC. Together, these series include 16 females and 10 males, averaging 21.6 years of age (range 1-61 years) [Table 1].[1,2,4-6,8,9] DCs were localized in the posterior fossa in 22 cases, 1 case extended to the cranio-cervical junction. We add this unique case of a posterior fossa DC extending intradurally to the T1 level in a patient with KFS.

Of interest, two other cases showed DC located purely within the anterior spinal cervical canal.

Location and extend of excision of dermoid cyst in adults vs. children

The DC was located purely within the posterior fossa in 100% (n = 14) of the pediatric KPS cases compared with 66% of the DCs in the adult patients (8/12).

Complete Dc removal was achieved in 83% (20/24) of cases that detailed the extent of resection. Most patients (except two cases) did not require fusions as they did not demonstrate instability.

Several hypotheses have been suggested to explain the occurrence of DC in patients with KFS. Shortening of the cervical spine due to Klippel–Feil abnormality may result in an abnormal flexure, altering tissue tension at the craniocervical junction favoring entrapment of ectodermal elements during neural tube closure resulting in the DC.[2,4]

CONCLUSION

Uncommonly, KFS has been associated with intracranial or spinal DCs. We report a unique case of KFS in association with a posterior fossa DC that extended to the upper thoracic spine (T1).

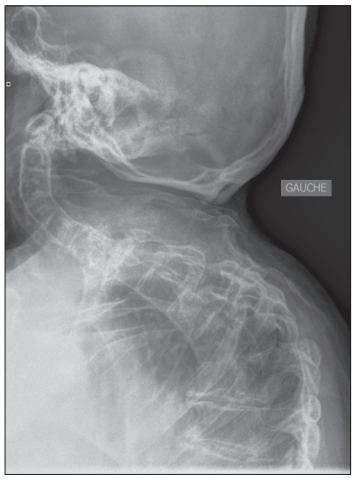

Figure 8: Postoperative plain lateral cervicothoracic radiograph demonstrating no new-onset deformity at 6-year follow-up

Table 1: Summary of reported cases of Klippel-Feil syndrome associated with dermoid cyst

Variables	No. (%)	No. cases where data is specified
Demographics		
Female sex	16 (61.5)	26
Avg age (range), yrs	21.6 (1-61)	26
Bony abnormalities		
Cervical fusion	26 (100)	26
Spina bifida	2 (7.5)	26
DC location		
PF only	22 (85)	26
PF and cervical spine	2 (7.5)	26
Cervical spine only	2 (7.5)	26
Outcome		
Follow-up (range), months	18 (3-60)	15
Initial GTR	20 (83)	24
Tumor recurrence	0 (0)	14

GTR: Gross total resection, F: Female, yrs : Years, No.: Number, DC: Dermoid cyst

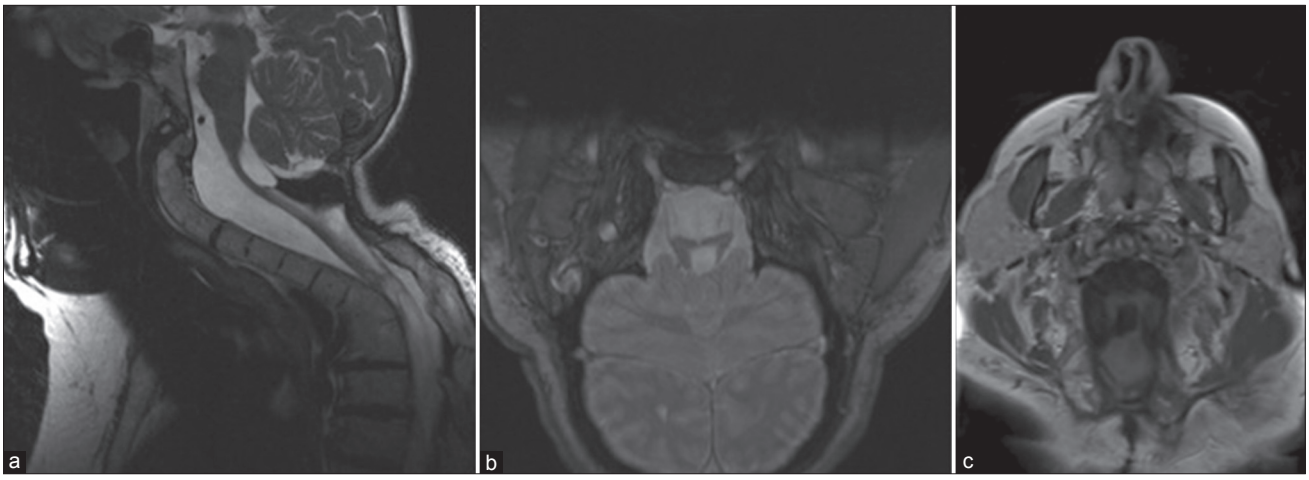

Figure 9: Postoperative magnetic resonance imaging, T2WI sagittal (a) T2WI axial (b) T1WI post-gadolinium axial sections demonstrating no recurrence at 6 years after surgery (c) Demonstrating no recurrence at 6-year follow-up

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