Klippel-Feil syndrome, or brevicollis, is a complex congenital disorder caused by the improper segmentation of cervical vertebrae.\textsuperscript{27} Neurological complications in patients with this disorder are mostly due to associated neural or bony malformations at the craniocervical junction.\textsuperscript{20,29} We present the very rare case of a patient with Klippel-Feil syndrome who presented with an intradural arachnoid cyst at the craniocervical junction. They also examine possible factors contributing to this association.

A 46-year-old woman presented with complaints of progressively worsening headaches and dizziness of 18 months’ duration. She also demonstrated mild bilateral upper-extremity weakness. Magnetic resonance imaging revealed fused cervical vertebrae and a dorsal intradural arachnoid cyst at the craniocervical junction, extending down to the fourth cervical level. Because of worsening myelopathy and the presence of brainstem compression, the patient underwent surgical excision of the arachnoid cyst, which was approached via a midline posterior suboccipital/upper cervical route. An endoscope was introduced through a gap between the occiput and fused upper cervical vertebrae, and the arachnoid cyst was widely fenestrated. Postoperatively, the patient has remained symptom free for more than 2 years with evidence of good radiological decompression.

The authors report a unique association between craniocervical arachnoid cyst and Klippel-Feil syndrome. To their knowledge, no other cases of this association have been reported in the literature. Arachnoid cysts should be part of the differential diagnosis in the presence of worsening myelopathic symptoms or pain in patients with Klippel-Feil syndrome.

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\textbf{Key Words} \hspace{2em} Klippel-Feil syndrome \hspace{2em} brevicollis \hspace{2em} arachnoid cyst \hspace{2em} craniocervical cyst

\section*{Case Report}

\textit{History and Examination.} A 46-year-old woman presented with complaints of progressively worsening headaches and dizziness of 18 months’ duration. A few weeks prior to her presentation, bilateral upper-extremity numbness and tingling also developed. She had a known medical history of Klippel-Feil syndrome, impaired hearing, and a surgically repaired cleft palate.

On examination, her neck was slightly extended and displayed a decreased range of active and passive motion. She also demonstrated mild upper-extremity weakness bilaterally (4/5) and diffuse upper- and lower-extremity hyperreflexia. Magnetic resonance imaging revealed autofusion between C1–3 and C5–6 vertebrae as well as a dorsal intradural arachnoid cyst at the craniocervical junction, extending down to the fourth cervical level (Fig. 1). The cyst caused significant craniocervical brainstem and upper cervical cord compression. There was no evidence of Chiari malformation, platybasia, or hydrocepha-
lus. The cyst did not communicate with the subarachnoid space. Because of the progressive myelopathy in the setting of brainstem and upper cervical cord compression, the patient was scheduled for surgery.

**Operation.** A midline posterior skin incision extending from the superior aspect of the inion to the level of C-4 was made, and subperiosteal dissection exposed the suboccipital region and upper cervical spine. A defect was found between the lower occipital bone and the fused upper cervical vertebrae. To minimize bony destabilization, the dura mater was opened through this defect without removing any bone from the upper spine or lower occipital bone. Upon opening the dura, a large cystic mass with clear fluid content was encountered. Using endoscopic assistance, the entire extent of the cyst was visualized, and the dorsal and lateral walls of the cyst were excised. The cyst was fenestrated and fluid was noted to rapidly egress from the cyst cavity. After evacuation of the cyst fluid, cyst compression on the surrounding structures was noted to have decreased. The endoscope was inserted into the cyst cavity, and direct visualization of the surrounding structures showed less compression and mass effect from the cyst, including the cerebellar tonsils, brainstem, and cervical cord. The anterior aspect of the cyst wall was intimately adherent to the dorsal surface of the brainstem and upper cervical spinal cord. Attempts to aggressively resect this ventral aspect of the cyst wall were not made, as we did not want to risk injury to this eloquent neural tissue.

**Postoperative Course.** Immediately postoperatively, the patient had marked resolution of her headaches and dizziness. She had an unremarkable postoperative course and was discharged home on postoperative Day 3. Postoperative imaging showed good surgical decompression and cyst fenestration. She has remained symptom free in the 2 years since surgery. There has been no radiological evidence of cyst recurrence thus far (Fig. 2).

**Discussion**

Failure of the mesoderm somites to fuse during the 3rd to 8th week of gestation leads to Klippel-Feil syndrome, whose incidence is approximately 1 case in 42,000 births. While most cases are sporadic, some instances of autosomal dominant and recessive inheritance patterns have also been described. The classic triad of a short neck, restricted neck movement, and a low posterior hairline, first described by Klippel and Feil in 1912, is only seen in approximately half of all patients with the syndrome.

The clinical presentation in patients with Klippel-Feil syndrome is quite variable, and multiple congenital conditions have been associated with this developmental anomaly. Hall et al. listed up to 38 conditions associated with the syndrome, such as syringomyelia, diastematomyelia, agenesis of the corpus callosum, synkinesis, cleft palate, and hearing disturbances. The pathogenesis of Klippel-Feil syndrome lies in the failed formation and the rearrangement of segmental cervical sclerotomes. Defective notochord and notochord signaling during the early stages of embryological development are seen as probable factors in the disorder's occurrence. Mouse models point toward members of the PAX gene family and Notch signaling pathway as possible etiological candidates.

Neural tube defects and spinal deformities have been associated with spinal arachnoid cysts. Altered CSF flow patterns and arachnoidal adhesions in these conditions are thought to lead to the formation of an arachnoid cyst. A slight disturbance in the development of the mesenchymal layer covering the notochord early on in development might be implicated as well. It is plausible that in the presence of disturbed anatomical development in Klippel-Feil syndrome, these factors play a part in the formation of an arachnoid cyst. Along the same lines, Akgün et al. reported an association between a cervical arachnoid cyst and the complex maldevelopment in Klippel-Feil syndrome.

Craniocervical junction arachnoid cysts are rare, and to our knowledge only 9 other cases have been reported to date. Although these cysts occupy an uncommon location, their pathogenesis does not seem to differ from their counterparts in the cervical spine or posterior cranial fossa. Some cases have documented a history of trauma or meningitis in patients. Among cases with primary arachnoid cysts, spinal defects were described in 2 patients: an enlarged foramen magnum and failed fusion of the posterior arch of C-1 in 1 patient and a cervical myelomeningocele in the other. The location of these cysts leads to symptoms associated with compression of the brainstem and spinal cord. Nonspecific signs of head or neck pain and dizziness are
Klippel-Feil syndrome associated with craniocervical arachnoid cyst

usually present. The occurrence of hydrocephalus is variable and, when present, is probably obstructive in origin.25 Treatment of arachnoid cysts in this location includes craniotomy with or without laminectomy and cyst drainage. The altered anatomy of the craniocervical junction in our patient allowed us access to the cyst without any bone removal. While most patients have been treated with fenestration and partial resection of the cyst, Price et al.25 performed complete excision and Shukla et al.28 performed cystoperitoneal shunting. None of the cases documented failure in the follow-up period. However, the best treatment modality in these patients is still debatable. Fenestration alone in posterior fossa arachnoid cysts has been shown to fail after scarring of the cyst wall in some patients.9 At the same time, complete excision might be challenging if the cyst is tightly adherent to the brainstem or spinal cord.52 Only long-term follow-up data in patients with cysts in this location will help to determine the best mode of treatment.

Conclusions

We report a unique association between a craniocervical arachnoid cyst and Klippel-Feil syndrome. Arachnoid cysts should be part of the differential diagnosis in the presence of worsening myelopathic symptoms or pain in patients with Klippel-Feil syndrome. Inherent abnormality in the flow of CSF at the craniocervical junction is the probable etiology for arachnoid cyst formation, and this abnormality may be more likely in patients with Klippel-Feil syndrome. When patients are asymptomatic, surgical intervention should be considered for brainstem decompression. If the cyst recurs, cyst-peritoneal shunt placement has been shown to be a successful salvage plan.

Disclosure

The authors report no conflicts of interest concerning the materials or methods used in this study or the findings specified in this paper.

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