CASE REPORT

Postsurgical pneumoencephaly in a patient with Klippel-Feil syndrome

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SUMMARY

A 49-year-old woman presented with occipital headaches, dizziness, photophobia and vomiting for 2 weeks' status post posterior scalp mole removal by a general surgeon. The physical examination revealed occipital lesions with foul smelling purulent discharge mixed with clear fluid drainage, webbed neck with decreased range of motion, facial asymmetry and a low posterior hairline. A CT of head showed occipital skull defect providing a communicating pathway for scalp infection, an atlas fusion defect and an

extensive pneumoencephaly. Further imaging showed low-lying cerebellar tonsils and fusion of the body of second and third cervical vertebrae. Klippel-Feil syndrome was diagnosed and the patient was successfully managed with duraplasty of occipital dura, placement of a lumbar drain and antibiotics. In patients with findings suggestive of Klippel-Feil syndrome and lesions proximal to the CNS, appropriate neuroimaging and possible neurosurgical consult should be considered prior to any surgical intervention.

Figure 1 (A) Webbed neck and facial asymmetry. (B) Low lying hairline and scalp wound status post duraplasty. (C) The brain and cervical spine MRI and fused bodies of the second and third cervical vertebrae and low lying cerebellar tonsils. (D) The CT scan of the head without contrast image and square-shaped occipital bone defect and pneumoencephaly. (E) The CT scan of the head without contrast image and anterior atlas fusion defect. (F) Pneumoencephaly and chronically dilated lateral ventricles.

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BACKGROUND

Klippel-Feil syndrome (KFS) is a congenital bone disorder characterised by fusion of two or more vertebrae of the cervical spine. Patients with this syndrome have distinct clinical features including a short webbed neck, a limited range of motion of the neck and a low hairline at the back of the head. These patients may also display cleft palate, short stature, deafness and cardiovascular abnormalities. Cases of KFS can be sporadic or inherited in autosomal dominant or autosomal recessive pattern.¹ The differential diagnosis of atlantoaxial instability should include Down syndrome, mucopolysaccharidosis type IV also known as Morquio syndrome and isolated cases of atlantoaxial instability.^{2–3} The differential of the short webbed neck should include Turner syndrome and Noonan syndrome.^{4–5} The interested reader is referred to references above for further discussion of these disorders.

In this article, we present a patient with KFS, who developed pneumoencephaly due to lack of knowledge about this genetic diagnosis.

CASE PRESENTATION

A 49-year-old woman presented with the new-onset occipital headache, dizziness, photophobia, nausea and vomiting for 2 weeks' status post three occipital mole removal by a general surgeon. The vital signs at presentation were significant for a fever of 102.6°F and a tachycardia with a heart rate of 106 bpm. The physical examination was significant for facial asymmetry, webbed neck with decreased range of movement (figure 1A), a low-lying hairline (figure 1B) and purulent foul smelling discharge from occipital lesions mixed with clear coloured fluid drainage. The initial laboratory tests were significant for a leucocyte count of 16.5 (4.0–11.0K/mm³) and sodium of 120 (135–145 mmol/L). Blood culture and wound cultures were sent. A CT scan of the head without contrast was carried out and showed square-shaped occipital skull defect with well-defined clear margins, a large open wound in the occipital area, external to bone defect, with soft tissue fluid collections, extensive pneumoencephaly (air in skull; figure 1C–E), anterior atlas vertebrae fusion defect (figure 1D) and dilated lateral and third ventricles with compressed fourth ventricle. Cerebrospinal fluid (CSF) studies revealed CSF glucose 35 mg/dL (50–75), CSF protein 450 mg/dL (15–40) and CSF white cell count 6000/mm³ with predominant neutrophilia. Meropenem and vancomycin were started for suspected meningitis and cerebrospinal fluid leak. Further imaging including MRI of the cervical spine and brain showed fusion of the body of second and third cervical vertebrae, low lying cerebellar tonsils (figure 1F) and leptomeningeal effacement consistent with diagnosis of meningitis. Diagnosis of KFS was made, a lumbar drain was placed and an occipital duraplasty was performed. During surgery, three 2–3 mm small holes in the dura were noted, which were repaired with interrupted 4–0 Nurodon sutures followed by extradural application of DuraGen and BioGlue. On day 5, wound culture and CSF culture grew *Pseudomonas aeruginosa* susceptible to quinolones, the lumbar drain was removed and the patient was discharged home on oral ciprofloxacin.

OUTCOME AND FOLLOW-UP

A follow-up CT scan at 2 months showed a complete resolution of pneumoencephaly.

DISCUSSION

KFS is a complex syndrome of osseous and visceral anomalies which was described for the first time in 1912.⁶ KFS includes a

classical triad of webbed neck, limited range of neck movement and a low posterior hairline.⁷ Three types of KFS have been described⁸: type I with massive fusion of many cervical and upper thoracic vertebrae into bony blocks; type II with fusion of only one or two interspaces, usually C2–C3 or C5–C6; type III with cervical fusion and lower thoracic or lumbar fusion, often associated with multiple organ anomalies and subsequent neurological compromise. Our patient had classical features of type II KFS and had facial asymmetry, which is also another characteristic feature of type II KFS.^{9–10} In addition to osseous and other visceral anomalies, KFS may also include neural tube defects resulting in multiple CNS anomalies,¹¹ as seen in our case. Our patient had congenital meningocele due to occipital skull defect, and a lack of knowledge for this genetic diagnosis at the time of surgical intervention by a general surgeon resulted in iatrogenic occipital dura damage resulting in CSF leak. It is believed that continued CSF leak combined with inability of the choroid plexuses to compensate for the CSF loss results in a negative intracranial pressure leading to air entry into the subarachnoid space¹² and thus pneumoencephaly. Clinically, pneumoencephaly may present from simple headache, to altered consciousness,¹³ to hemiparesis, seizures and cardiovascular collapse¹⁴ resulting in a neurosurgical emergency.¹⁵ However, most cases of pneumoencephaly resolve spontaneously, and conservative management should be provided. Non-operative management involves oxygen therapy, keeping the head of the bed elevated, prophylactic antimicrobial therapy, analgesia, frequent neurological checks and repeated CT scans.¹⁶

Learning points

- ▶ Klippel-Feil syndrome is a rare disease classically presenting as webbed neck, limited neck movement and low posterior hairline.
- ▶ Three major types of Klippel-Feil syndrome are recognised.
- ▶ Patients with cutaneous lesions proximal to the nervous system as well as physical manifestations consistent with genetic syndromes such as Klippel-Feil must have thorough preoperative work-up, including neuroimaging and possible neurosurgery consultation prior to instrumentation or further management so as to avoid any catastrophic outcome.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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