

Klippel-Feil syndrome

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Abstract

We present a 38-year-old male patient who sustained a minor and superficial stab injury in the left flank as well as a laceration on the scalp overlying the right parietal bone.

On examination the classical triad of Klippel-Feil syndrome (short neck, low posterior hair line, limitation of neck movements) were observed. Further investigations revealed a plethora of congenital anomalies, including atlanto-occipital fusion, basilar impression, congenital fusion of C5 and C6 vertebrae, scoliosis with convexity to the right side, complete situs inversus, and bilateral pelvic kidneys.

Despite these multiple anomalies our patient still lives a relatively normal life and only sought medical help after minor injuries sustained during a brawl, for which he was treated in

the hospital and discharged the following day.

Case report

A 38-year-old man presented with a stab injury in the left flank and laceration of the head.

On examination he was short statured, with a short neck, low posterior hairline, and severe restriction of neck movement. There was no loss of consciousness. A stab injury in the left flank, and scalp laceration overlying the right parietal region of the skull were observed. Blood pressure was 120 mmHg, and pulse 72 beats/minute and regular.

Blood analysis showed sedimentation rate of 6 mm in the first hour, haemoglobin 11.0 g/dl. Urinalysis and hearing tests were normal. The presence of the classical clinical triad of Klippel-Feil syndrome, namely



Fig. 1. Lateral view of the skull and cervical spine. This shows atlanto-occipital fusion, and fusion of the C5 and C6 vertebrae, as well as basilar impression.

short neck, limitation of neck movement and low posterior hairline, stimulated a search for associated anomalies in this patient. Lateral skull and cervical spine X-ray showed atlanto-occipital fusion, congenital fusion of C5 and C6 and basilar impression (Fig. 1). The atlanto-occipital fusion was better demonstrated by the tomogram of the lateral skull and cervical spine (Fig. 2).



Fig. 2. Tomogram of the lateral view of the skull and cervical spine showing atlanto-occipital fusion.

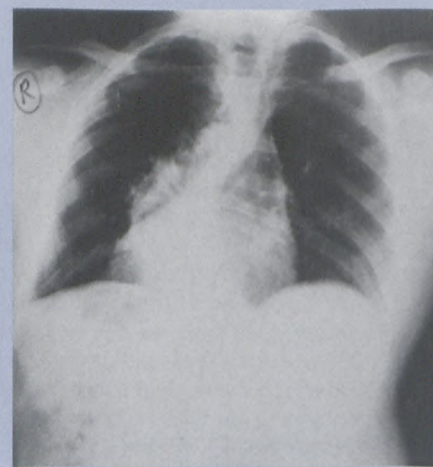


Fig. 3. Chest X-ray, PA-view. This shows marked

complete situs inversus scoliosis, and X-ray of the thoracic spine showed convexity to the right side (Fig. 3). Intravenous

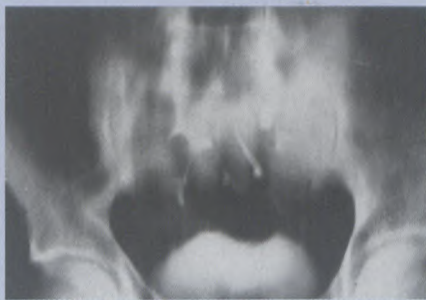


Fig. 4. Nephrotomogram, demonstrating the bilateral pelvic kidneys.

urography disclosed bilateral pelvic kidneys, illustrated by the tomogram of the intravenous urography (Fig. 4). The superficial stab injury in the left flank was sutured and the patient was admitted. He was however discharged from the ward the next day. Subsequent follow-up visits were uneventful.

Discussion

It is perhaps noteworthy that our patient presented with such a range of congenital mesodermal abnormalities and yet was still living a relatively normal life. To the best of our knowledge this case of Klippel-Feil syndrome with a plurality of other anomalies is the first to be documented. Klippel-Feil syndrome occurs in 1 of 42 000 births, with equal male to female ratio, and it is believed to have been recognised in an Egyptian mummy in 500 BC.¹ The classifications as described by Klippel-Feil are: (i) type 1 — a block fusion of all the cervical and upper thoracic vertebrae; (ii) type 2 — fusion of one or two pairs of cervical vertebrae, frequently the second to the third or the fifth to the sixth,

which is the most common type; and (iii) type 3 — fusion of the cervical and lower thoracic or lumbar spine.²

The syndrome appears to be genetically determined, and with type 2 anomalies there may be occipitalisation of the atlas,³ as was seen in this patient. Chamberlain's line is drawn from the back of the hard palate to the posterior lip of the foramen magnum, and basilar impression is implied when more than half the odontoid peg lies above it. Our patient had basilar impression because almost all the C2 vertebrae lay above Chamberlain's line (Fig. 1). Basilar impression implies an elevation of the floor of the posterior fossa, which is congenital in origin and is associated with Klippel-Feil syndrome or atlanto-occipital fusion.⁴ Other conditions include fragilitas ossium and cleidocranial dysostosis.

The main significance of basilar impression, which is usually symptomless, is in the neurological symptoms, which may sometimes result from pressure on and distortion of the brainstem, and obstruction of free cerebrospinal fluid circulation.⁴

The fusion of C5 and C6 vertebrae seen in our patient is common. The association of Klippel-Feil syndrome with complete situs inversus is a rarity. In 505 cases of Klippel-Feil syndrome reviewed, only two patients had complete situs inversus.³ In a review of 50 patients with Klippel-Feil syndrome, 60% had scoliosis, believed to be the most frequent anomaly.⁵ Unilateral renal anomaly is the most common renal anomaly, followed by malrota-

tion of the kidneys, renal pelvic and ureteral duplication, and simple renal ectopia.² Bilateral pelvic kidneys as found in this patient are very rare. All the five pelvic kidneys studied by Moore *et al.*² in Klippel-Feil syndrome patients were unilateral.

Other serious but less apparent anomalies include Sprengel's deformity, synkinesia,^{3,5} hearing impairment,^{3,5,6} and congenital heart disease.^{3,5,7} The cervical vertebrae and genitourinary tract differentiate at the same time and in the same vicinity (between the seventh and fourteenth mesodermal somites) in the embryo, therefore insult to the fetus between the fourth and eighth weeks of development could produce both genitourinary anomalies and the Klippel-Feil syndrome.²

References

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