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THE KLIPPEL-FEIL SYNDROME*

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The Klippel-Feil syndrome originated from a case report by M. Klippel and A. Feil in 1912. They formulated the following conditions: (1) Limitation of head motion. (2) Low margin of head hair. (3) Absence of neck. They believed the etiology was intra-uterine inflammation or trauma. Their subject was a forty-six-year-old male, a tailor by trade. At necropsy they found the cervical vertebræ fused and having a posterior spina bifida occulta. The mass did not contain either axis or atlas and was thought to be formed by dorsal vertebræ as there were four pair of ribs, and but eight normal dorsal vertebræ below it.

In 1919, Feil expressed a belief that the high spina bifida was the original lesion and that pressure and trauma later in fœtal life caused the fusion and malformation. He recognized three types: (1) Complete absence of cervical spine; (2) partial numerical reduction of cervical vertebræ; and (3) associated partial reduction extending throughout the spine.

An earlier report of this syndrome was by J. Jackson Clarke, in 1906, before the Clinical Society of London. He reported a male four years of age with head fixed, with chin close to sternum and no movements of cervical spine. He stated, "skiagrams showed extensive abnormalities in the form of bones in the upper dorsal and cervical regions, and a cervical rib was present on each side." He claimed operative treatment followed by massage gave natural movement. There was, however, no report of the operative procedure.

The two cases that we will report fall into the second group of Feil's classification: the first case by actual numerical reduction of vertebræ, and the second case by reduction of vertebræ due to fusion.

Case I.—J. S., a male, aged eight years, was the sixth of seven children. (Figs. 1 and 2.) There were no existing abnormalities in the other members of the family. He had a normal birth at eight months. His deformed neck was noticed in the second week. He was weaned at five months. He had no serious illnesses. He is under-nourished and under-developed. His head rests low between his shoulders; the hair line is low on the neck; there is marked nuchal depression. He has a rounded dorsal kyphosis. His trapezii flare out from the base of his skull to his shoulders. His scapulæ are elevated. His chin rests close to his sternum. His nipples are relatively low. Flexion and extension of his head are practically normal. Rotation is possible to 25° in either direction. Lateral flexion is somewhat limited. All motions of the neck are without pain. There is bimanual synkinesia or associated movements of the hands. Scratching, patting and writhing motions are carried out by the opposite hand with mirror-like precision. Reflexes are normal. His teeth have serrated edges. The two lateral

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upper incisors are unerupted and the two lower lateral incisors are behind the middle incisors. There are no other gross abnormalities. The Mantoux Tuberculin Test and blood Wassermann are negative.

The Röntgen film shows but six cervical vertebræ with fusion between the bodies of the first and second and partial fusion with the third. The spinous processes of the second and third vertebræ are united as are those of the fourth and fifth. (Fig. 3.) There is a posterior spina bifida occulta of the third and fourth vertebræ. (Fig. 4.) The other spinal vertebræ are normal except for the first sacral which shows a posterior spina bifida occulta.

CASE II.—S. W., a male, aged ten years, was the second in four children. His mother was an epileptic, but there were no other abnormalities in other members of the family. His past history was essentially negative. His deformity was not observed until his second year.

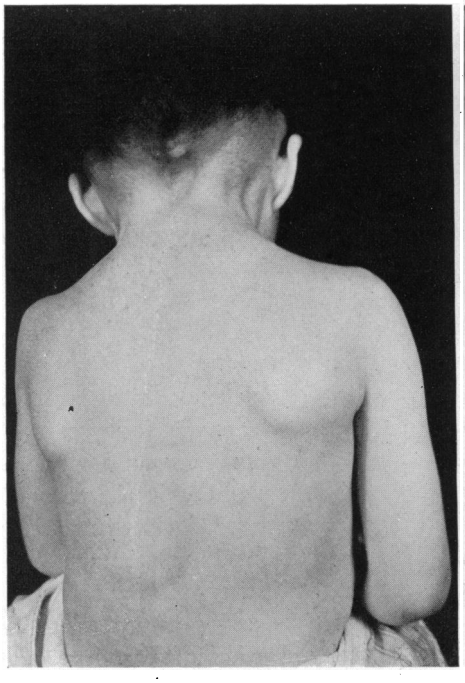

FIG. 1.—(Case I.) J. S. Head in flexion. Note low hair line, short neck, nuchal depression, winged trapezil, high scapulæ.

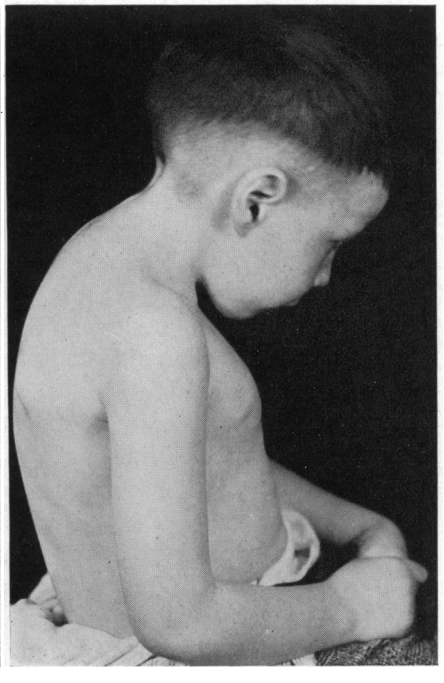

FIG. 2.—(Case I.) J. S. Showing prominent occiput, dorsal kyphosis.

He presents similar gross characteristics of the other case, but has a slight tendency toward a right torticollis. Rotation of the head is limited, other motions about normal. He does not have bimanual synkinesia. His von Pirquet test is positive, but blood Wassermann negative.

The Röntgen film shows seven cervical vertebræ with fusion of the first and second, and third and fourth vertebral bodies, with fusion of the spines of the second and third, and sixth, seventh cervical and first dorsal vertebræ. There is no spina bifida occulta. (Fig. 5.)

Etiology.—Of the sixty cases in the literature all have occurred spontaneously without history of familial malformations. The syndrome has occurred with about equal frequency in both sexes. De Beaujeur and Block

and also Ingelians and Piquet have reported hereditary syphilis as a factor. Both of these cases were deaf-mutes with mental deficiency. In the majority of reports syphilis was not questioned. Trauma has been mentioned as an explanation. The developmental deviations which take place before the third month of embryonic life are undoubtedly of influence.

Embryology.—Jamieson claims that ossification begins in the seventh week of fœtal life in the spinal arches and the tenth week in the bodies. Before the third month of development, therefore, distinct cervical characteristics essential to the shape of the adult osseous cervical vertebræ have occurred.

According to Bardeen, there are two bilaterally placed centres of chondri-

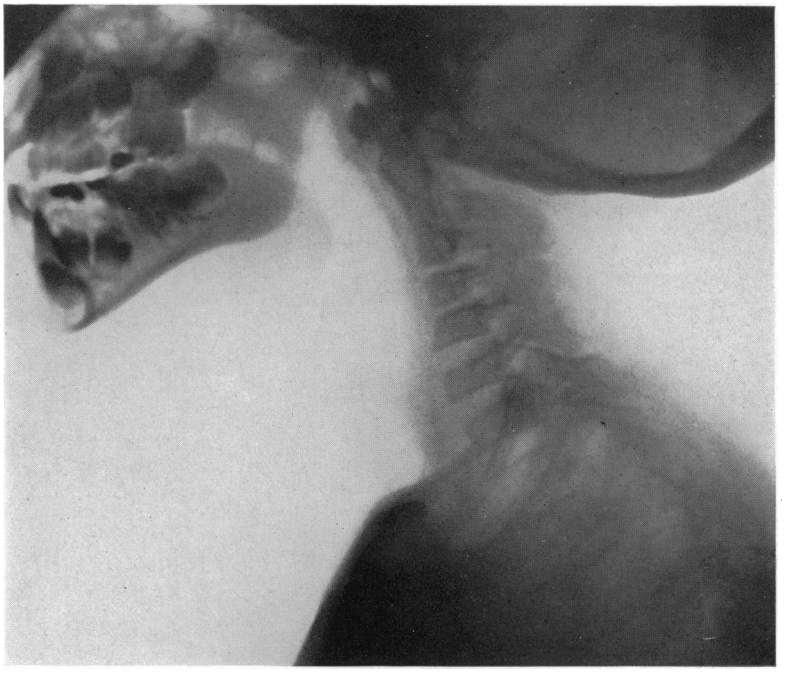

FIG. 3.—(Case I.) J. S. Six cervical vertebræ, fusion of first and second cervical bodies and fusion of spinous processes of second third and fourth and fifth.

fication for each of the vertebral bodies. Ventral fusion takes place before dorsal fusion. There are separate centres of chondrification for the neural processes from which develop the laminæ, articular and transverse processes.

The adontoid process represents the body of the first cervical vertebra. During the second month there is chondrification of the arches of the more cranial cervical vertebræ, at which time the atlas is fused to the axis and for a brief period; the bases of the neural arches of the axis and atlas, together with the tissue forming the occiput bone, become fused into a nearly continuous mass of pericartilage.

It appears that the malformation is determined before the third month of fœtal life. The posterior spina bifida is caused either by the later fusion

of the posterior chondrification centres for the vertebral bodies, or by the lack of fusion of the laminæ chrondrification centres. Due to faults in these laminæ chondrification centres, fusion of adjacent spinous processes occur. The apparent or actual reduction of cervical vertebra is brought about by faulty or complete fusion of the body chondrification centres in forming the continuous mass of pericartilage with the occiput. An extension of this abnormal fusion probably accounts for the changes which may appear in the upper dorsal region.

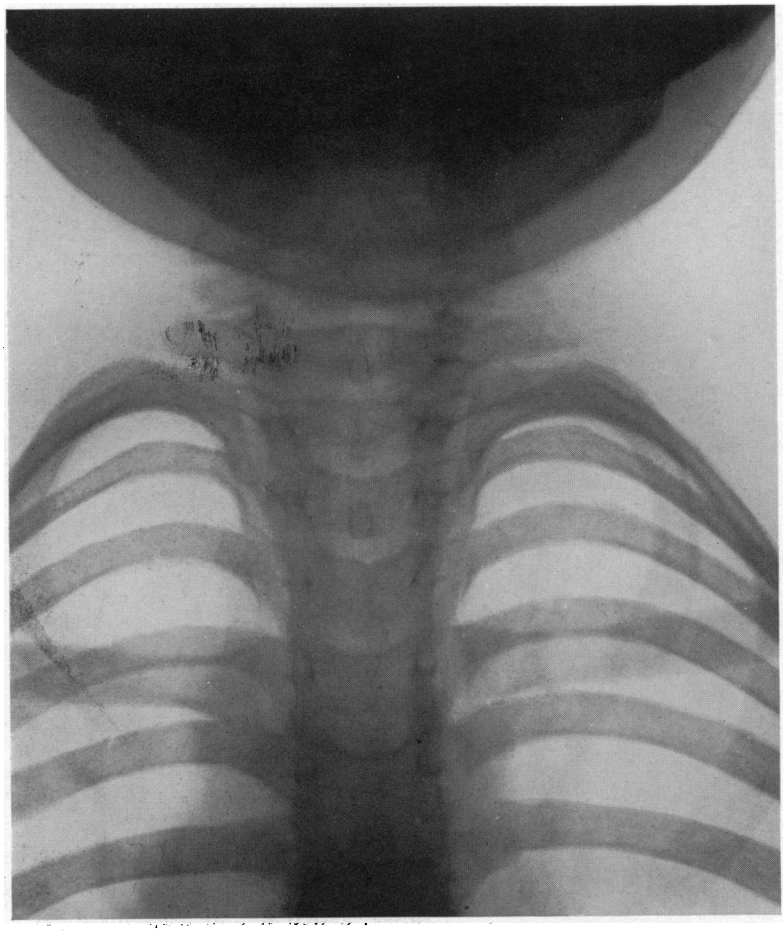

FIG. 4.—Posterior spina bifida occulta of the third and fourth cervical vertebræ.

Pathology.—The following additional variations occur in the reported cases of Klippel-Feil syndrome: (1) Usually fusion of atlas to occiput. (Heidecker.) (2) Fusion of first three vertebral bodies with fusion of spines of third, fourth and fifth cervical vertebræ. (Guillain and Mallaret.) (3) Fusion of the first and second cervical vertebræ with third intact and fourth, fifth and sixth fused. (Pierre Ingelians.) (4) Fusion of third, fourth, fifth and sixth cervical bodies and fusion of sixth and seventh cervical

and first and second dorsal spinous processes. (Elouson.) (5) Reduction to four cervical vertebræ. (Lavastine and Miget.) (6) All cervical vertebræ fused in one mass with four cervical ribs and reduction of dorsal vertebræ to eight. (Klippel-Feil.) (7) A posterior spina bifida occulta which may extend from occiput to thorax. (Nobel and Frawley.) (8) Fusion of six upper dorsal vertebræ. (Pytel and Saevic.) (9) A fusion of first and second right ribs and two ribs arising from the fourth left dorsal vertebra. (Ingelrans and Piquet.) (10) Fusion of the fifth lumbar and sacrum. (Ingelrans and Piquet.) (11) Dorsal spina bifida occulta and sacral rachichisis. (Ingelrans and Piquet.) (12) Oblique bodies of cervical dorsal vertebræ with a hemivertebra and unfused laminæ. (Ingelrans and Piquet.)

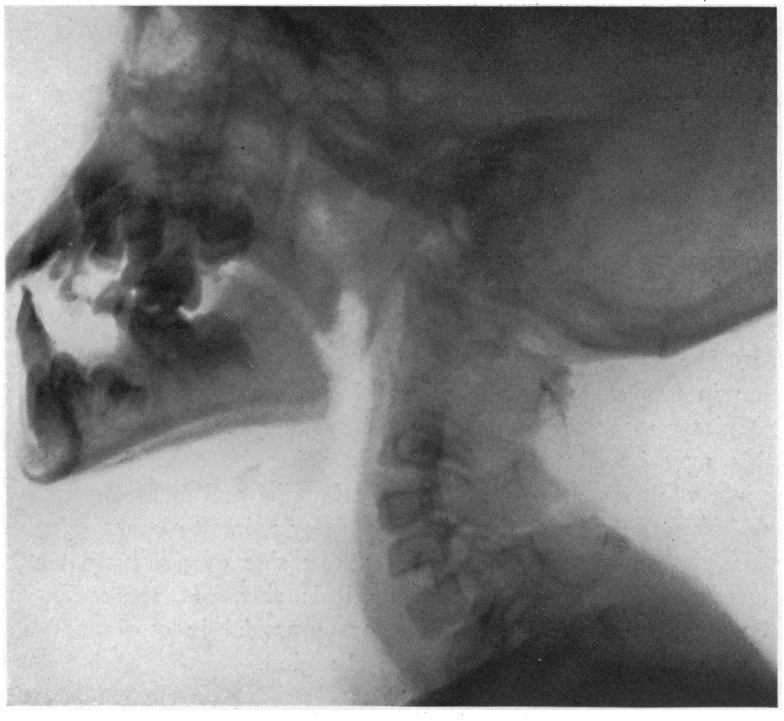

FIG. 5.—(Case II.) Fusion of first and second and third and fourth cervical bodies with fusion of the spines of the second and third and sixth and seventh cervical and first dorsal vertebræ.

Symptoms.—The physical characteristics are the apparent absence or shortness of neck, the low hair line on the back of the neck, the nuchal depression, the flaring trapezii, the high position of the shoulders, the prominence of occiput, the dorsal kyphos, the high scapulæ, the proximity of chin to sternum, the low nipple line, the limitation of head motion and freedom from pain.

The associated variations may be torticollis, asymmetry of face, scoliosis, Sprengle's deformity (Heidecker), absence of external auditory meatus (Ingelrans and Piquet), abnormalities of upper extremities—atrophy of left

forearm and hand (Pytel and Saevic), club hand (Ingelrans and Piquet), mental debility (de Beaujeur and Block), bimanual synkinesia or mirror movements (Bauman).

Diagnosis.—The cases may readily be mistaken for tuberculosis of the cervical spine. The differential diagnosis depends upon, first, the absence of rigidity; second, motion without pain; third, Röntgen film.

Treatment.—Heidecker states improvement in mobility after gymnastic exercise. Ryerson cites improvement in cosmetic effects in one case after division of the trapezius. Certainly, massage and stretching should be given a thorough trial early in the growth period and the associated deformities of torticollis scoliosis, club hand, *etc.,* corrected.

Prognosis.—Guillain and Mollaret describe a case, male, thirty-three years old, who developed a progressive spastic paralysis starting in the right leg and involving the left side twelve years later. Heidecker reports pain in old age due to plexus disturbances. All cases, however, which have come to necropsy have died of an acute infection. About fifteen adult cases have been reported. The oldest was seventy years.

Discussion.—There is no description of the cervical nerve abnormalities which would be expected with reduction and fusion of cervical vertebræ. There are few neurological symptoms reported. Bauman reported mental retardation, spasm of cervical muscles in two cases, and difficulty in swallowing or breathing in one case, and "mirror movements."

There are no previous accounts of these observations except mental retardation in two cases of mutism and hereditary syphilis. Our first case shows "mirror movements" or bimanual synkinesia. Purves-Stewart states that this may be physiological, especially in children, occasionally persisting into adult life. Rarely, it may be familial and is then regarded as a stigma of a neuropathical inheritance. Badgley points to imitative synkinesia in hemiplegics; voluntary movements of one side of the body tend to be reproduced symmetrically on the hemiplegic side. It, therefore, appears that the associated movements are extra-pyramidal in origin and are not caused by any abnormality in the cervical region.

CONCLUSIONS

(1) The Klippel-Feil syndrome is a developmental abnormality dating from the third month of fœtal life.

(2) Syphilis is incidental rather than etiological.

(3) Additional variations in other spinal vertebræ frequently occur.

(4) Other congenital abnormalities co-exist.

(5) Cases may be mistaken for cervical Pott's disease.

(6) The treatment is palliative.

(7) The deformity is not detrimental to longevity.

(8) "Mirror movements," bimanual synkinesia, are not characteristic of the condition.

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