Congenital neural abnormalities presenting with mirror movements in a patient with Klippel-Feil syndrome

Case report

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A young girl with Klippel-Feil syndrome presented with the onset of mirror movements in early childhood. Computerized tomography studies of her cervical spine and brain revealed fibrous diastematomyelia with duplication of the cervical spinal cord and an extra-axial midline posterior fossa cyst, together with the multiple cervical vertebral anomalies. Exploration of the posterior fossa lesion revealed it to be a dermoid cyst. The congenital spinal and cord abnormalities found in this case support the hypothesis that the Klippel-Feil syndrome may be associated with variable duplication of the spinal cord and that mirror movements may be related to impairment of pyramidal tract decussation.

KEY WORDS • diastematomyelia • Klippel-Feil syndrome • dermoid cyst • mirror movements • synkinesia

Mirror movements may occur when volitional movements of muscle groups on one side of the body are copied involuntarily by the identical contralateral muscle groups. This synkinesia represents an unusual disorder of motor control that may be hereditary or sporadic, or may occur with congenital or acquired neurological disorders. Cases of mirror movements occurring in patients with Klippel-Feil syndrome have been recorded previously, and in two of these postmortem examination has revealed malformation and dysplasia of the cervical spinal cord.

We report the occurrence of a similar congenital spinal cord abnormality, together with a posterior fossa dermoid cyst, in a young girl known to have Klippel-Feil syndrome who presented with mirror movements. The investigation of patients presenting with mirror movement is discussed with reference to the reported neural abnormalities associated with these disorders.

Case Report

This 2-year-old girl was first brought to us on December 5, 1979, for evaluation of a sore neck following a fall from a swing onto her head. She was the second child of Caucasian parents. Both parents were aged 32 years, and there was no family history of medical disorders. Pregnancy and delivery had been uncomplicated, and the child had walked at the age of 13 months. She had been investigated by an orthopedic surgeon for head tilt 1 year prior to admission, and was found to have congenital anomalies of the cervical spine.

First Admission. The major findings on initial clinical examination were a short, webbed neck and paucity of neck movement. The child held her cervical spine rigid and resisted all attempts at passive movement. The remainder of the examination revealed a developmentally normal child who was at the 50th percentile for height, weight, and head circumference. Roentgenograms of the cervical spine were reported to show fusion of the C2-5 vertebral bodies with spina bifida occulta at the level of the C-6 and C-7 vertebrae. There was no evidence of acute subluxation or fracture. The patient was thought to have the Klippel-Feil syndrome; she was treated with a soft collar and discharged 2 days later.

Second Admission. Six months later she was readmitted. She exhibited mirror movements of her upper limbs which the parents believed had become apparent approximately 3 months following the neck trauma, and were progressing. Clinical examination showed her neck to be short and tilted to the right. There was no
midline sinus or cutaneous pigmentation on the neck. The ears were low-set, and there was mild hypertelorism. Lateral cervical spinal movement was markedly restricted, although there was better preservation of neck flexion and extension. She had readily elicitable mirror movements, confined mainly to the hand and forearms. The mirror movement was of equal amplitude to the volitional movement and confined to the same muscle groups. The remainder of the neurological examination was normal.

Metrizamide myelography performed on August 18, 1980, showed widening of the cervical subarachnoid space, and the bone abnormalities (Fig. 1). Computerized tomography (CT) of the cervical spine, enhanced with intrathecal metrizamide, was performed on the same day. This study showed a septum extending anteriorly from the region of the bifid cervical spines, with division of the cord over 1.5 cm into two lateral segments (Fig. 2). An axial CT scan of the head showed a rounded midline cavity in the posterior fossa, of the density of cerebrospinal fluid, that displaced the fourth ventricle and did not fill with metrizamide (Fig. 3 left). These lesions were believed to be an extra-axial arachnoid cyst and fibrous diastematomyelia with hemicord formation.

Third Admission. Apart from her mirror movements she remained asymptomatic until November, 1981, when she was admitted with mild deterioration in her gait. There were no other symptoms. Clinical examination was essentially unchanged except for some mild gait ataxia. Repeat axial CT of the head showed enlargement of the posterior fossa lesion with no change in the ventricular size (Fig. 3 center). In view of the radiological and mild clinical progression, it was considered that the posterior fossa lesion warranted surgical exploration.

Operation. Posterior fossa craniectomy was performed on November 4, 1981. After opening the dura mater a large lesion filled with cheesy material and hair was found to be compressing the vermis and right cerebellar hemisphere. The contents of the dermoid cyst were removed, and the epithelial lining was dissected from the surrounding cerebellum and blood vessels using microsurgical technique. There was no evidence to suggest communication of the dermoid cyst with the skin or spinal canal inferiorly. Histological examination of the resected tissues confirmed the diagnosis of dermoid cyst, and there was no evidence of malignancy.

Postoperative Course. The patient's progress was uneventful. Although her gait returned to normal, the mirror movements were unchanged. Repeat axial CT of the head in November, 1982, showed complete extirpation of the lesion with no evidence of recurrence (Fig. 3 right).

Discussion

In a recent review of congenital mirror movements, Schott and Wyke emphasized the variability both in the expression of the abnormal synkinesia and the frequency of associated neural abnormalities. The disorder may be inherited as an autosomal dominant condition, although autosomal recessive patterns have been described, or may occur sporadically. However, of particular interest is the association of congenital mirror movements with skeletal craniocervical abnormalities, the Klippel-Feil syndrome, spinal cord malformation and dysplasia, and agenesis of the corpus callosum. In some of these cases, multiple neural abnormalities coexist. Consequently, the neurophysiological mechanism of the movement disorder has been variably suggested to result from failure of maturation of a motor inhibitory center that allows non-synkinetic movement to occur, loss or failure of suppression of bilateral motor cortical activity, and failure or compression of the medullary pyramidal decussation. This heterogeneity of possible functional origins of the disorder is reflected in the various neurological conditions with which acquired mirror movements may also be associated, such as cerebrovascular accidents, subarachnoid hemorrhage, cerebral tumors, and extrapyramidal disorders. The condition with which congenital mirror move-
Mirror movements in Klippel-Feil syndrome

FIG. 2. Computerized tomography scans of the cervical spine, enhanced with intrathecal metrizamide, demonstrating several abnormalities at the C-6 and C-7 vertebral levels. Both levels reveal spina bifida, posterior dislocation of the spinal cord, and widening of the subarachnoid space. There is a small meningocele (large arrow, left) and widening of the posterior median sulcus of the spinal cord (center left). Hemicord formation, with diastematomyelia is most evident in the scans at center right and right (arrows) although a fine septum can be seen (small arrow, left).

FIG. 3. Axial computerized tomography scans of the head demonstrating a midline ovoid low-density lesion in the posterior fossa that does not fill with metrizamide (left) and extends toward the inferior surface of the tentorium (center). The postoperative scan shows complete extirpation of the lesion with reexpansion of the cerebellum (right).

ments have most commonly been associated is the Klippel-Feil syndrome. To date, however, only two necropsy studies have been reported of patients with the combination of these disorders. Both patients had dysplastic and flattened spinal cords, with dorsal malformation and two prominent lateral neural columns separated by a deep anterior median fissure. There was also failure of pyramidal decussation, and no anterior commissure caudally. The similarities to our case are interesting. The high-resolution spinal CT in our patient enabled visualization of the cervical spinal cord malformation, although this was not detected by metrizamide myelography. The lamination of the cord into two hemicords at the site of the diastematomyelia in this case may well also be associated with impairment of rostral pyramidal decussation, and further cord malformation.

The cord duplication and cord malformations in these cases are probably related intimately to the surrounding mesodermal malformation that is the major manifestation of the Klippel-Feil syndrome. Gardner believed that the skeletal and neural abnormalities associated with the Klippel-Feil syndrome resulted from distortion of the somites by an overdistended neural tube. Subsequent splitting of the thin neural roof plate could result in an associated dorsal malformation of the cord. Splitting of both the neural roof and floor plates would result in hemicord formation. Erskine had earlier recorded that the Klippel-Feil syndrome represented a primary focal duplication of the cerebral axis with incomplete fusion, although he did not postulate the embryonic mechanism. Gardner's hypothesis appears to correlate well with the reported neural duplications, the cervical spina bifida, wide cervical canal, and meningocele for-
The validity of the distension of the neural tube hypothesis will no doubt be clarified by future investigation of patients with the Klippel-Feil syndrome by means of high-resolution spinal CT scanning.

Mirror movements are not regarded as an essential feature of this syndrome. Although they were clinically documented in 18% of cases in one large series of patients with the Klippel-Feil syndrome, electromyographic evidence of abnormal synkinesia could be elicited in 73% of one series of patients in whom the clinical expression of mirror movements was variable. This finding would suggest that a high incidence of underlying neural defects would be found in these patients.

The only report of necropsy findings in the brain of a patient with mirror movements and Klippel-Feil syndrome revealed a glioependymal third ventricular cyst and an abnormally thin corpus callosum. Agenesia of the corpus callosum has been associated with mirror movements, and in these patients impairment of interhemispheric transmission and other anatomical abnormalities of the pyramidal pathways are believed to underlie the abnormal synkinesia. Axial CT of the head in our patient discounted an associated callosal agenesis, but revealed an asymptomatic midline posterior fossa cyst that was initially considered to be an extra-axial arachnoid cyst. That this lesion was a dermoid cyst would relate it to a similar period in embryogenesis as the failure of cervical segmentation and cervical spine malformation. This would occur in the embryo at approximately 22 to 26 days. It is interesting to note that an intraspinal cervical dermoid cyst and an extradural cervical hemangiolipoma have also been described in patients with the Klippel-Feil syndrome.

With the ready availability of high-resolution CT scanning, it is desirable to investigate all children with congenital mirror movements with this modality, even if clinical examination is otherwise unremarkable. If there is an associated Klippel-Feil syndrome, investigation would appear mandatory to document the cerebral and spinal status; in this syndrome alone, hindbrain hernia, hydrocephalus, hydrayomyelia, syringomyelia, and meningocele have been reported. This case, the two necropsy cases alluded to, and others reported in the literature confirm that patients with either of these conditions may have many associated congenital neural abnormalities, some of which may be asymptomatic in early childhood. The further investigation of these patients may also add to our understanding of the neurophysiological pathways subserving this unusual neurological manifestation.

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References