

The Association of Chiari Type III Malformation and Klippel-Feil Syndrome with Mirror Movement: A Case Report

Mirror Movement ile Birlikte Olan Klippel-Feil Sendromunun Chiari Tip III Malformasyonu ile Birlikteliği

Fatih Serhat EROL, Necati UCLER, Huseyin YAKAR

Firat University, Faculty of Medicine, Department of Neurosurgery, Elazig, Turkey

Correspondence address: Fatih Serhat EROL / E-mail: fserol@yahoo.com

ABSTRACT

Basically Chiari type III malformation is a combination of encephalocele with of brain stem and cerebellar abnormality. Although Klippel-Feil syndrome may be associated with other congenital anomalies, this syndrome is mainly associated with varying degrees of cervical vertebral fusion anomalies. In this study, we reported the association of Chiari type III malformation and Klippel-Feil syndrome with the mirror movement by imaging studies. The main involvement in Chiari type III malformation and Klippel-Feil syndrome is in the craniocervical junction. In such a small area, the emergence of these complex pathologies in our case was remarkable. Our patient had reconstruction surgery of the posterior fossa and his encephalocele was excised successfully. Hydrocephaly and/or deterioration in the functions of other posterior fossa structures have not been seen in the patient's follow-up.

KEYWORDS: Chiari type III malformation, Klippel-Feil syndrome, Mirror movement, Syringomyelia, Craniocervical region

ÖZ

Temel olarak Chiari tip III malformasyonu beyin sapı ve serebellar anomaliler ile birlikte ensefaloselin kombinasyonudur. Her ne kadar Klippel-Feil sendromu diğer konjenital anomaliler ile birlikte olabilir de, bu sendrom çoğunlukla değişik derecelerde olan servikal vertebra füzyon anomalileri ile birliktedir. Bu çalışmada görüntüleme tetkikleri ile Chiari tip III malformasyonu, Klippel-Feil sendromu ile birlikte olan mirror hareket birlikteliğini gösterdik. Chiari tip III ve Klippel-Feil'de temel etkilenen bölge kranioservikal bileşkedir. Bu kadar dar bir bölgede, bu kompleks patolojilerin birlikte ortaya çıkması vakamızın dikkat çekici kısmı idi. Hastamıza başarılı şekilde posterior fossa rekonstrüksiyonu ve ensefalosel kesesinin eksizyonu yapıldı. Ameliyatı sonrası takiplerde hidrosefali ve/veya posterior fossa yapılarının fonksiyonlarında bozulma gibi herhangi bir komplikasyon hastada gelişmemiştir.

ANAHTAR SÖZCÜKLER: Chiari tip III malformasyonu, Klippel-Feil sendromu, Mirror hareket, Syringomyeli, Kranioservikal bölge

INTRODUCTION

The prognosis of Chiari type III malformation is much worse than other Chiari types. The association of Chiari type III malformation, Klippel-Feil syndrome (KFS), mirror movement, and syringomyelia has not been reported in previous studies. Mirror movement more frequently appears in KFS (10) and its pathophysiology is not known exactly. The suggested reasons for mirror movements are those abnormal development of pyramidal decussation and transcallosal inhibitory pathways (14) and/or effects of herniation of hind brain onto pyramidal decussation causing development of aberrant neuronal pathways and mirror movement (8). Syringomyelia may be seen in Chiari type III malformation and KFS, due to disruption of cerebrospinal fluid (CSF) circulation in these disorders. In addition to being very rarely seen, these pathologies in such a narrow region of the body, it was also remarkable that the

patient with this highly mortal Chiari malformation type reached the age of 4.

CASE REPORT

A 4-year-old boy was referred to our clinic for occipital swelling that was present from birth and had not any discharge. Her parents stated that patient had similar movements in upper extremities and frequent falls. The patient's growth and development stages were normal. On physical examination, motor forces of the patient's upper extremities were 4/5, and head circumference was in normal ranges. Mirror movement was found in upper extremities' movements, and the patient's parents notified us that these movements were present from birth. By inspection and palpation, the swelling was soft in consistency in 4x3x3 cm and its surface was covered with normal skin in lower occipital region. Short neck and low hairline were observed in the patient (Figure 1). The range of

motion in his neck was determined as limited. On examination, other systems were normal. Brain computed tomography (CT) and 3-dimensional CT (3-D CT) studies showed herniation of cerebellum and meninx through in midline occipital bone defect with 3.5 cm diameter, cervical angulation at C5-6 level, fusions of C4-5 and C5-6 vertebrae corpuses, a defect at craniocervical junction in posterior midline and fusions and rotoscoliosis of cervical spinous processes in cervical region (Figure 2, 3). Craniocervical magnetic resonance imaging (MRI) studies showed encephalocele in occipital region, fusion of C4-5 and C5-6 vertebrae, and syringomyelia in the craniocervical junction (Figure 4).

In terms of anesthetic technique, patient's intubation was done carefully under general anesthesia. The encephalocele localized to the lower occipital region was repaired. During surgery, while gliotic cerebellar tissue and arachnoid membrane containing CSF were excised carefully, operational blood was not allowed to enter into CSF circulation. Decompression of posterior fossa was made with duraplasty. The result of excised tissue during surgery sent to the pathology laboratory was reported as a gliotic tissue. The patient's early and 2-years follow-up with cranial and cervical MRI were uneventful and no hydrocephaly and/or other posterior fossa complications occurred, but the patient's frequent falls were only decreased relatively, and the mirror movement disorder was not changed (Figure 5).

DISCUSSION

Chiari type III malformation is the rarest form of Chiari malformations (3). The natural prognosis of the patients with

this malformation is not known exactly (5), but the prognosis is often regarded as poor. On the other hand, Raimondi indicated that the presence of the cervical or occipital encephalocele was not necessarily associated with a poor prognosis, but the functional prognosis remains severe, and concluded that surgical care should be undertaken if possible (12). Headache, syncope, impairment of eye movements, sensory losses, motor deficits, and cerebellar ataxia can occur, due to the tissues compressed in foramen magnum. The characteristic lesion is an upper cervical meningoencephalocele containing cerebellar tissue (2). However this may be an isolated lesion, or can be associated with Chiari malformations, Dandy-Walker complex, cerebellar dysplasia, diastometamyelia or



Figure 1: The photography of the patient's posterior craniocervical region showing swelling in 4x3x3 cm and its surface covered with normal skin.



Figure 2: Axial brain CT image showing herniation of cerebellum and meninx through in a midline occipital bone defect.



Figure 3: Posterior craniocervical 3-D CT image showing cervical angulation at C5-6 level, fusions of C4-5 and C5-6 vertebrae corpuses, a defect at craniocervical junction in posterior midline and fusions and rotoscoliosis of cervical spinous processes in cervical region.



Figure 4: Mid-sagittal T2-weighted MR image showing encephalocele in the occipital region, fusion of C4-5 and C5-6 vertebrae, syringomyelia in craniocervical junction.

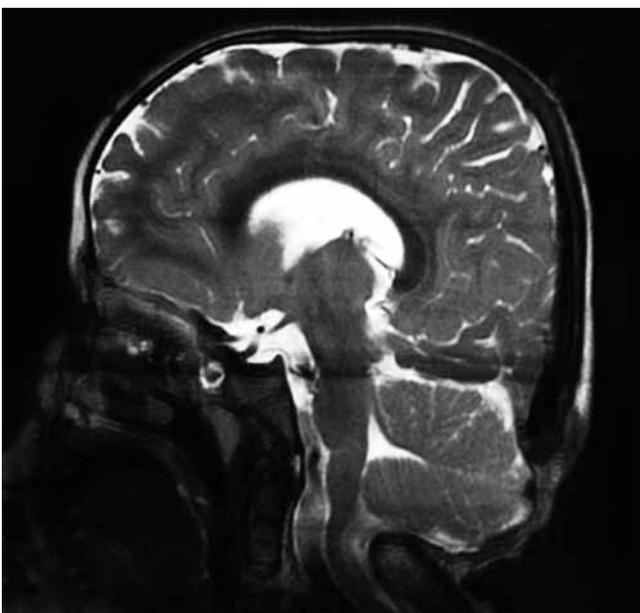


Figure 5: Postoperative T2-weighted sagittal MRI showed no progressive abnormality and hydrocephalus.

KFS (6). As in our case, encephalocele is most commonly seen in Chiari type III malformation (4). The cerebellum, occipital lobes or CSF also are common encephalocele contents, and the medulla and pons may sometimes be present in it depending on the herniation degree. We think that symptoms and prognosis may change according to brain tissue within encephalocele. Because of the presence of necrosis, gliosis, fibrosis and heterotopias, herniated tissue is usually abnormal and nonfunctional (11). In our case, the amount of herniated tissue was less, and also during surgery only gliotic tissue was excised with viable tissue carefully preserved. These situations may help explain how the patient reached the age of 4 years and the postoperative good prognosis. We think that because of infection, trauma, and spontaneous rupture of cyst and the other potential risks, cyst repair must be performed as early as possible. After primary repair of the lesion in the treatment and the patient's close follow-up for hydrocephalus, ventriculoperitoneal shunting may be necessary with high probability (3). However, hydrocephalus did not develop in our patient's follow-up. The CSF circulation is seriously impaired in Chiari malformations, and this impairment may show itself as hydrocephalus, syringomyelia. In our case, there was an upper extremity paresis associated with syringomyelia, but when compared with other serious complications such as respiratory insufficiency, this paresis was tolerable.

The classic clinical triad of short neck, low hairline, and restricted neck motion is present in less than 50% of patients with KFS (15). This syndrome may be associated with other organ system anomalies, skeleton system anomalies like scoliosis, torticollis, Sprengel anomaly, genito-urinary tract anomalies, facial asymmetry, cardiovascular anomalies, and brain stem anomalies (9). So far we did not find any Chiari III malformation association with KFS and mirror movement in the literature.

Mirror movements refer to involuntary movements, which occur in a muscle group or limb on one side of the body in response to an intentionally performed movement in the corresponding contralateral muscle group or limb (13). Mirror movements may be physiological, congenital, or acquired. The exact pathophysiology of this disorder is unknown, but it is most commonly seen in KFS, and can be associated with Kallmann's syndrome, agenesis of the corpus callosum, basilar invagination of the skull, spina bifida occulta, Friedrich's ataxia, Usher's syndrome and hemiplegic cerebral palsy (14). We previously reported that a band tissue that extends from posterior spinal to meningocele cord could cause mirror movement (7). Scoliosis, spina bifida, omovertebral bone, renal-otological disorders, torticollis, and psychiatric diseases that may accompany KFS but were not present in our case.

Syringomyelia can be seen in Chiari type III malformation and KFS. Both pathologies cause syringomyelia, due to disruption of CSF circulation. In the treatment of syringomyelia, the goal of the operation is to enlarge the posterior fossa and to re-create the cisterna magna, thereby permitting normal flow

of CSF from the posterior fossa to the cervical subarachnoid space (1). In present case, cervical syringomyelia and paresis in the patient's upper extremities were present but it was not clear that which basic factor caused the syringomyelia. We think that this syringomyelia was caused by KFS, due to no change in syringomyelia found by follow-up with post-operative MRI. If the Chiari type III malformation had caused the syringomyelia, the syringomyelia could have improved.

It was interesting that rarely seen serious Chiari type III malformation and KFS which involved the very narrow region of 4-year-old patient's body caused non-serious complications. Another important point to emphasize is that our operation technique in this case prevented the hydrocephalus that was expected as a high possibility. Currently we do not have adequate embryological and physiopathological data to explain so many structural disorders in such a narrow area of the craniocervical region and the concurrence of these pathologies may therefore only explained in the future.

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