Clinical Neurology and Neurosurgery 130 (2015) 155–158

Contents lists available at ScienceDirect

Clinical Neurology and Neurosurgery

journal homepage: www.elsevier.com/locate/clineuro

Case report

Chiari malformation type 1.5 in male monozygotic twins: Case report and literature review

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ARTICLE INFO

Article history:
Received 7 October 2014
Received in revised form
25 November 2014
Accepted 1 January 2015
Available online 8 January 2015

Keywords:
Chiari malformation
Syringomyelia
Twin
Posterior fossa decompression

Monozygotic twins both with Chiari malformation are extremely rare.

Chiari malformation type 1.5 has not been reported in monozygotic twins.

The twins developed different symptoms at different ages, with nine years apart.

1. Introduction

Chiari malformation (CM) is conventionally divided into four types (type I–IV) according to morphological characters. However, there are some patients in whom the morphological criteria of the four types do not strictly apply. One such group was described as Chiari malformation type 1.5 (CM 1.5), which referred to tonsillar herniation within a CM I but with addition of elongated brainstem [1].

Chiari malformation occurring in twins was rare and only a few cases have been reported. All these cases were diagnosed with CM I except one with Chiari malformation type 0 (CM 0) [4]. Here, we report on symptomatic twin brothers both with CM 1.5 and syringomyelia. To our knowledge, no such cases have been reported in the literature.

2. Case report

Patient 1 is the elder of identical twin 30-year-old brothers sharing one placenta. The patient presented to clinical attention with approximately 45° of dextroscoliosis 10 years ago, and subsequently an orthopedic surgery of spine was performed. There was no neurological dysfunction at that time. The patient was admitted to our department with a two-year history of walking difficulty and MR images demonstrated herniation of both the cerebellar tonsils and the brain stem, and syringomyelia extending from C2 level to thoracic region (Fig. 1). The cerebellar tonsils were 6 mm below the foramen magnum (basion–opisthion line, or MacRae line) and the obex was 7 mm inferior to the foramen magnum. To evaluate the angulation of the odontoid process, we measured the angle by drawing one horizontal line through a midpoint of the synchondrosis between the base and apex of the odontoid process and a second line through the midpoint of the synchondrosis and apex of the odontoid process on a midsagittal MR imaging, and the angulation of the odontoid process was 73°. To estimate the steepness of cerebellar tentorium, the tentorial angle was measured to be 87°. Brain computed tomography (CT) scan demonstrated no hydrocephalus.

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http://dx.doi.org/10.1016/j.clineuro.2015.01.003

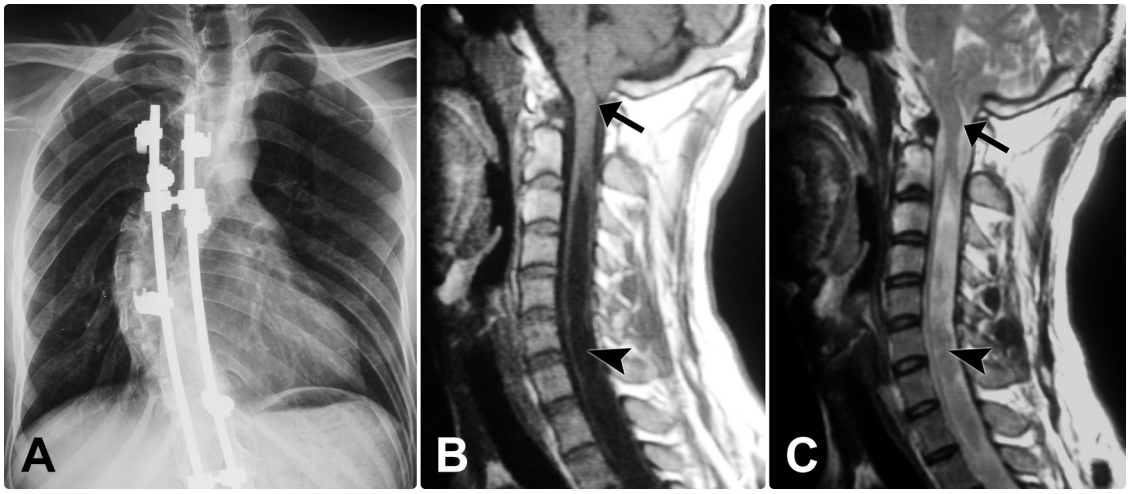

Fig. 1. X-ray of the patient 1 shows scoliosis (A). Preoperative sagittal T1-weighted (B) and T2-weighted (C) MR images demonstrate CM 1.5 with syrinx (arrowheads). The obex was below the foramen magnum (arrows).

Neurologic examination was normal aside from motor weakness of the lower extremities, and the muscle power was 4/5 according to the UK Medical Research Council (MRC) scale. Intending for an adequate decompression, a posterior fossa decompression procedure including a small-bone-window sub-occipital craniectomy (diameter, 2.5–3 cm) and a C-1 laminectomy (1.5–2.0 cm) was performed. The dura mater was opened with the arachnoid intact, and then it was grafted with the autologous graft to enlarge the cistern magna. During the four-year period of follow-up, no obvious improvement in neurological symptoms was observed. No MR imaging was done postoperatively because of the metal instrument within the body, in which case no radiological evidence of reduction in syrinx size was obtained.

The younger brother presented to us three years ago with an 11-month history of left upper limb weakness. Neurologic examination revealed sensory deficits and weakness of the left upper limb, and the muscle power was MRC grade 3/5. No scoliosis was noted. Cervical MR images revealed an 8-mm descent of the cerebellar tonsils below the foramen magnum, and herniation of the brain stem with the obex 10 mm inferior to the foramen magnum; syringomyelia from C2 level to thoracic region was also observed (Fig. 2A). The angulation of the odontoid process was 70° and the tentorial angle was 102°. Brain CT scan showed mild ventricular dilation with Evan's ratio of 0.33. Evan's ratio was defined as the maximum width of the frontal horns divided by the maximum width of the inner skull at the same plane. Fundus examination demonstrated no papilledema and there was no symptom of raised intracranial pressure (ICP). In consideration of the hydrocephalus and potential risk of durotomy-related hydrocephalus aggravation after losing support, a simple posterior fossa decompression including a small-bone-window sub-occipital craniectomy (diameter, 2.5–3 cm) and a C-1 laminectomy (1.5–2.0 cm) without duroplasty was performed. The patient stated improvement in motor weakness of the left upper extremity after surgery. During the three-year follow-up period, cervical MR images showed no reduction of syringomyelia (Fig. 2B), and brain CT scan demonstrated no change in ventricular volume with the same Evan's ratio (0.33).

The clinical and radiological manifestations were summarized in Table 1. In addition, the birth of the twins was the fourth delivery of their mother, and it was uneventful during labour and delivery. The parents and the siblings of the twins suffered no neurological dysfunctions.

3. Discussion

CM 1.5 is thought to be less common than CM I although the exact incidence is still unknown. It is reported that there was no single sign or symptom peculiar to CM 1.5, but the incidence of unresolved syringomyelia after posterior fossa decompression was nearly two-fold higher in CM 1.5 (13.6%) than CM I (6.9%) [1]. To our knowledge, this is the first report that describes CM 1.5 in monozygotic twins.

In literatures, we are aware of eleven reports describing Chiari malformation in monozygotic twins (Table 2). These reported cases can be divided into two groups according to the clinical and imaging manifestations: concordance group with similar symptoms and radiological appearances, and discordance group with disparate symptoms or MR manifestations. In the eleven reported pairs, there were five pairs of twin brothers and six pairs of twin sisters, with an average age of 19 years. All patients were diagnosed with CM I except one with CM 0. Out of the eleven pairs, five presented with similar clinical symptoms and MR manifestations, while the other six pairs were discordant. Three pairs had syringomyelia in both twins, three pairs had syringomyelia in only one out of each pair, and five pairs were without syringomyelia. Additionally, one pair was associated with Spondylo-epiphyseal dysplasia (SED) [5], another pair with Rubinstein–Taybi syndrome (RTS) [2].

Twin studies, especially the studies of concordant twins, suggest genetic factors may be involved in the etiology of Chiari malformation. Meanwhile, the cases of discordant twins support the idea that non-genetic factors are also significant. In one case report of twin sisters with CM I, one had birth trauma and developed syringomyelia requiring intervention [3]. While the other twin sister, whose delivery was uneventful, was asymptomatic without

Table 1
The radiological manifestations of the twins.

Patient	Type of Chiari malformation	Syrinx	Scoliosis	Length of tonsillar herniation (mm)	Length of obex herniation (mm)	Angulation of odontoid process (°)	Tentorial angle (°)	Evan's ratio
Patient 1 (elder)	CM 1.5	+	+	6	7	73	87	0.27
Patient 2 (younger)	CM 1.5	+	–	8	10	70	102	0.33