Craniocervical Anomalies: Chiari Malformation

17

Katrin Rabie, Francesco Cacciola, and Nicola Di Lorenzo

Introduction

The autopsy of a 17-year-old girl who died of typhoid fever was the first case of Chiari malformation described by Hans Chiari in 1891 [1]. Hans Chiari wanted to describe changes in the cerebellar region caused by hydrocephalus, she had however had no cerebellar or medullary symptoms prior to her death of typhoid fever; an incidental finding. Later on he went on to describe a case series consisting of 14 patients with Chiari malformation and speculated in other mechanisms such as insufficient bone growth or skull enlargement resulting in higher intracranial pressure [2]. He described the condition among other malformations of the craniocervical junction. Just 25 year later, in 1932, the first surgical attempt to correct the malformation was performed by Van Houweninge Graftidijk [3]. Much like the current surgical techniques he tried by resecting redundant cerebellar tonsils or resect the bone over the malformation and incise the dura. Given the early days of neurosurgery, his patients however didn't survive the procedure. By the end of 1930 and beginning of 1940 the condition had received further attention by several other publications in which adult cases of Chiari malformation was described either with or without hydrocephalus.

Today almost 1.5 century later we can still to some extent recognise the challenges and difficulties pioneers of neurosurgery faced and their struggle to find and refine surgical techniques to treat this condition. Our challenges have however changed in a fundamental way with overt use of imaging techniques leading to a surge in incidental findings with Chiari malformation being one of these findings. Just like Hans Chiari describing a condition that had to effect on the deceased girl and her symptoms, we can now face patients initially investigated for an unrelated condition or trauma and with no symptoms with a condition ultimately being an incidental finding.

It is fundamental to recognise that despite having varying degree of involvement of rhombencephalic derivate and hindbrain structures Chiari malformations are also a heterogeneous group of malformations with different underlying mechanisms and overlapping symptoms. Best treatment demands understanding of the various pathophysiological mechanisms involved in the specific case.

K. Rabie
Institution of Neuroscience and Physiology, Gothenburg University, Gothenburg, Sweden

F. Cacciola (✉)
Department of Neurosurgery, University Hospital of Siena, Siena, Italy

N. Di Lorenzo
Department of Neurosurgery, University Hospital of Florence, Florence, Italy

P. P. M. Menchetti (ed.), *Cervical Spine*, https://doi.org/10.1007/978-3-030-94829-0_17

Definition

Classic Chiari I malformation is a congenital condition with descent of cerebellar tonsils into foramen of magnum equivalent to or beyond 5 mm. Chiari malformation is found in 0.8–1% in hospital series [4, 5]. The prevalence seems to also be similar or lower among general population and is described as 0.2–1.7% in adults [6–8]. Asymptomatic cases are therefore almost always found among patients undergoing radiological imaging for various reasons specially among infants and young children. In children and infants, the extent of the caudal migration might decrease over time and asymptomatic children are therefore followed up until adulthood. There are also patients who do not have the classic descent of the cerebellar tonsils but who do have crowding in their posterior fossa with extensive syringomyelia and who do improve after decompression surgery of the posterior fossa.

In recent years the need to further describe the variety seen in Chiari I patients has led to description of two new entities within the Chiari I category namely Chiari 0 and Chiari 1.5. Chiari 0 describes the condition where a syringomyelia is present in absence of cerebellar herniation which resolves after posterior fossa decompression. Chiari 1.5 is a more severe form of Chiari I malformation in which the both the medulla and cerebellar tonsils are herniated below foramen magnum [9, 10].

Unlike Chiari I, patients with Chiari II suffer from neural tube defects such as myelomeningocele and encephalocele. Syringomyelia is common in this group as is hydrocephalus and a variety of other conditions involving the skull, bony spine, meninges, ventricles, spinal cord and various cerebral structures [11].

Pathophysiology

Early on it was hypothesized that hydrocephalus was related to and could cause Chiari malformation as evident in the early descriptions of Chiari. However later series found this association in less than 10% of the Chiari cases and hence a causal relationship between Chiari malformation and CSF disturbance has not been established [12].

Comparisons of posterior cranial fossa of Chiari I patients and normal population has shown that the posterior cranial fossa is smaller in Chiari I patients compared to normal controls [13, 14]. In a recent study these differences has been shown to be more prominent among men than women despite the higher prevalence of Chiari I malformation in women than men [15]. A smaller posterior cranial vault is also observed in Chiari malformation cases associated with other conditions such as multisuture craniosynostosis, platybasia, neurofibromatosis type I, familial vitamin D-resistant rickets and acromegaly [16–19]. It has been hypothesized that mesodermal defects can cause a smaller posterior cranial vault which in turn can cause cerebellar tonsillar herniation [20, 14].

Signs and Symptoms

Chiari I patients can be asymptomatic as Chiari I can be found as an incidental finding. Symptoms usually have a gradual onset and acute onset is unusual.

The most common type of symptom in both adults and children is occipital and or cervical headache or pain which is either exacerbated or elicited by Valsalva manoeuver or Valsalva-like strain such as laughing and coughing. The headache should typically be of short duration (seconds to minutes). The proposed mechanism for the headache is disturbed cerebrospinal fluid dynamics and raised intrathecal pressure and worsening of the crowding in posterior fossa [21]. In young children and infants this presentation can be in the form of irritability or inconsolable crying [22].

Headache is a very common symptom in general population and among children and adolescents and therefore a thorough investigation of the origin of the headaches are necessary if the history is inconsistent with strain-related headache. Aside from headaches, medullary symptoms can be found and be prominent in patients with syringomyelia. The symptoms are that of

classical medullary signs and symptoms with progressive limb weakness, hyperreflexia, balance- and gait disturbance. Paediatric patients may also present with failure to thrive, sleep apnoea, hoarseness, snoring or arching back [22]. Scoliosis can also be a manifestation of Chiari I in children and is usually accompanied by syringomyelia.

Other types of symptom include dizziness, swallowing difficulties, sinus bradycardia and autonomic dysfunction, difficulties with hand coordination, nystagmus and visual impairment [12].

Syringomyelia

Syringomyelia, a fluid filled cavity within the spinal cord, is one of the most common findings associated with Chiari I malformation. First description of syringomyelia has been accredited to Stephanus who in 1545 described a cavitation in "the interior substance of the marrow of the back" containing a red brown fluid. This description is more befitting of a post-haemorrhagic cavity than true syringomyelia. A cystic cavity in spinal cord found in connection with hydrocephalus was first described by Brunner in 1688. The term itself was first applied by Ollivier D'Angers in 1827 who thought of the central canal as a pathological finding [23].

Syringomyelia can have various different causes with different underlying pathophysiological mechanisms. The condition can be tumour related, congenital, inflammatory or traumatic in nature and treatment should always if possible treat the underlying condition such as in the case of Chiari malformation or tethered cord. As previously mentioned, patients with Chiari 0 are patients in whom a syringomyelia is detected without tonsillar herniation but with posterior fossa crowding. In these patients the treatment of the underlying cause which is believed to be a functional obstruction of the foramen magnum with posterior fossa decompression and duraplasty can typically resolve the underlying syringomyelias.

Surgical Treatment

The surgical treatment of Chiari I malformations is very much in line with what forefathers of neurosurgery once proposed. Suboccipital decompression and duraplasty is the standard surgical method for treatment of Chiari I malformations. The procedure is often combined with C1 laminectomy but laminectomy to achieve a good decompression of structures oftentimes pushed beneath the level of foramen Magnum. C1 laminectomy should however be limited to minimal manipulation of facet joints and their capsule to avoid future complications and swan neck deformity. In recent times the use of suboccipital decompression alone without duraplasty has been performed and advocated because of the procedure's low rate of complications and easy mobilisation and discharge of patients. CSF leakage remains the most common complication of suboccipital decompressions combined with duraplasty and is avoided with bony decompression alone. The so called "bony only" decompressions are however mostly used in children without syringomyelia [24]. The procedure is then combined with C1 laminectomy, resection of atlantooccipital membrane and scoring of the outer layer of the dura. A recent study found that this procedure also had a favourable long-term outcome in children [25]. For adults the matter remains controversial as no high quality study has been performed on adult patients comparing the two procedures [26]. The preferred surgical method for adults remain suboccipital craniotomy with duraplasty.

As an alternative to suboccipital craniotomy and duraplasty, an extra arachnoidal craniocervical decompression has been successfully practiced at the institution of the senior author [27]. Using this technique the arachnoid membrane is left intact and duraplasty is not performed. The dura is left open with the dural slits stiched laterally to the muscles. The technique is briefly described here, Figs. 17.1 and 17.2. Patients are positioned in sitting position for this procedure. A suboccipital craniotomy and C1 laminectomy of at least

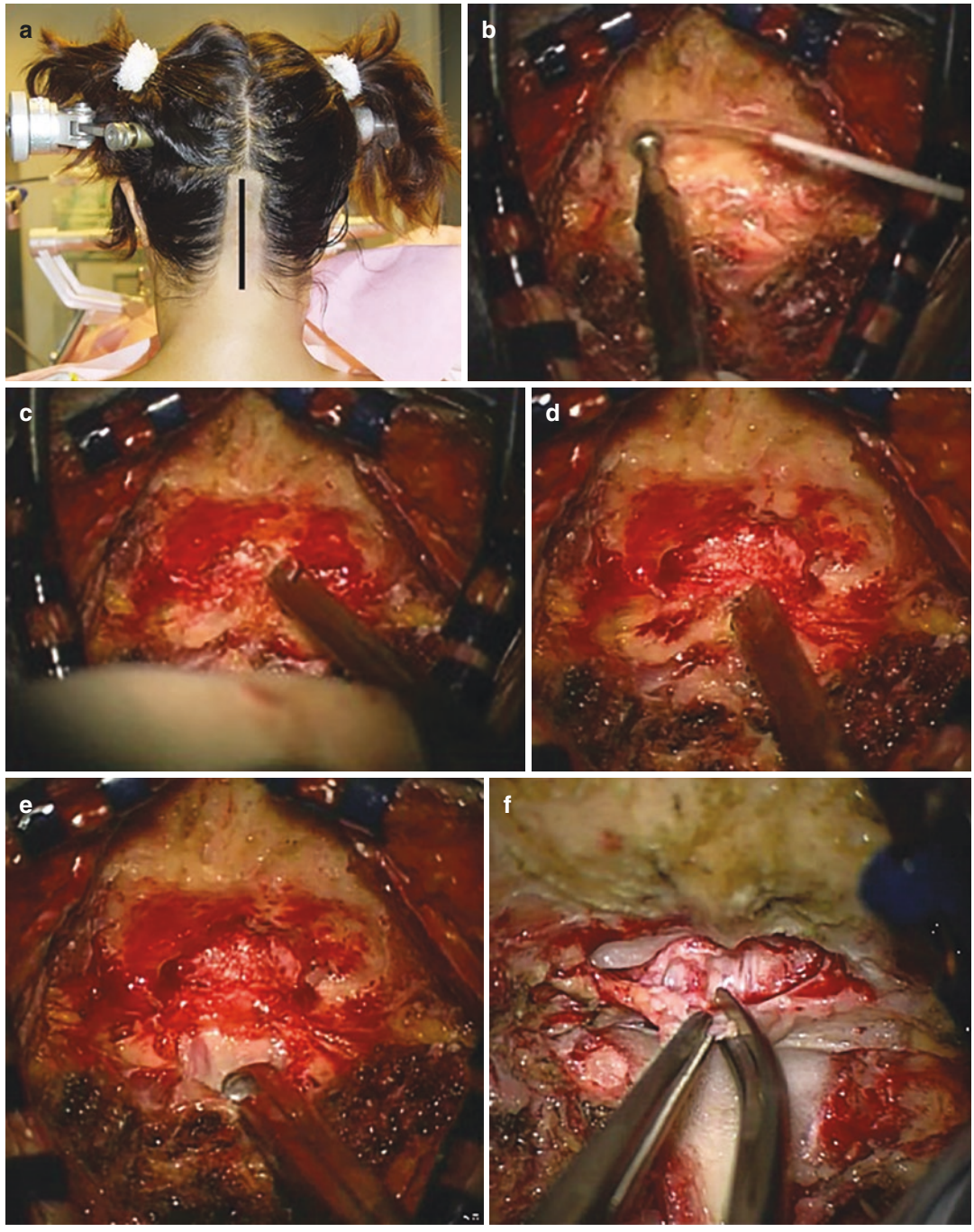

Fig. 17.1 (**a**) Patient is positioned in sitting position with a midline trichotomy. (**b**–**f**) Stepwise suboccipital craniotomy and opening of the posterior atlantooccipital membrane. (**g**) Opening of the dura with angled dural dissector. (**h**, **i**) Stiching of the dural to laterally to the muscles

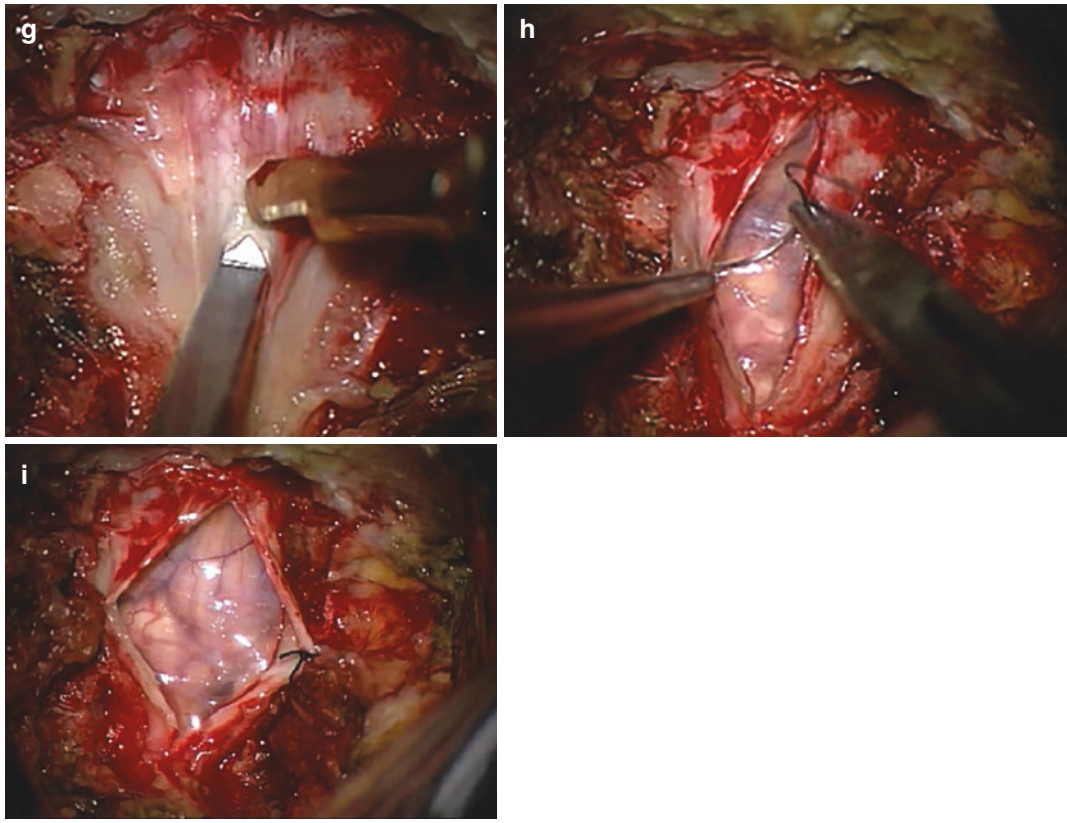

Fig. 17.1 (continued)

2 × 3.5 cm is performed after which the underlying posterior atlantooccipital membrane is opened. Underlying dura is opened meticulously using an angled dural dissector and paying attention to not violate the arachnoid membrane accidentally. The dura is then stitched to the muscles laterally and left open. In our series a resolution of the syringomyelia and good neurological outcome was found after a mean follow-up period of 44 months [27]. The senior author uses this procedure for younger adults due to its low risk of complications and good surgical outcome in that patient group.

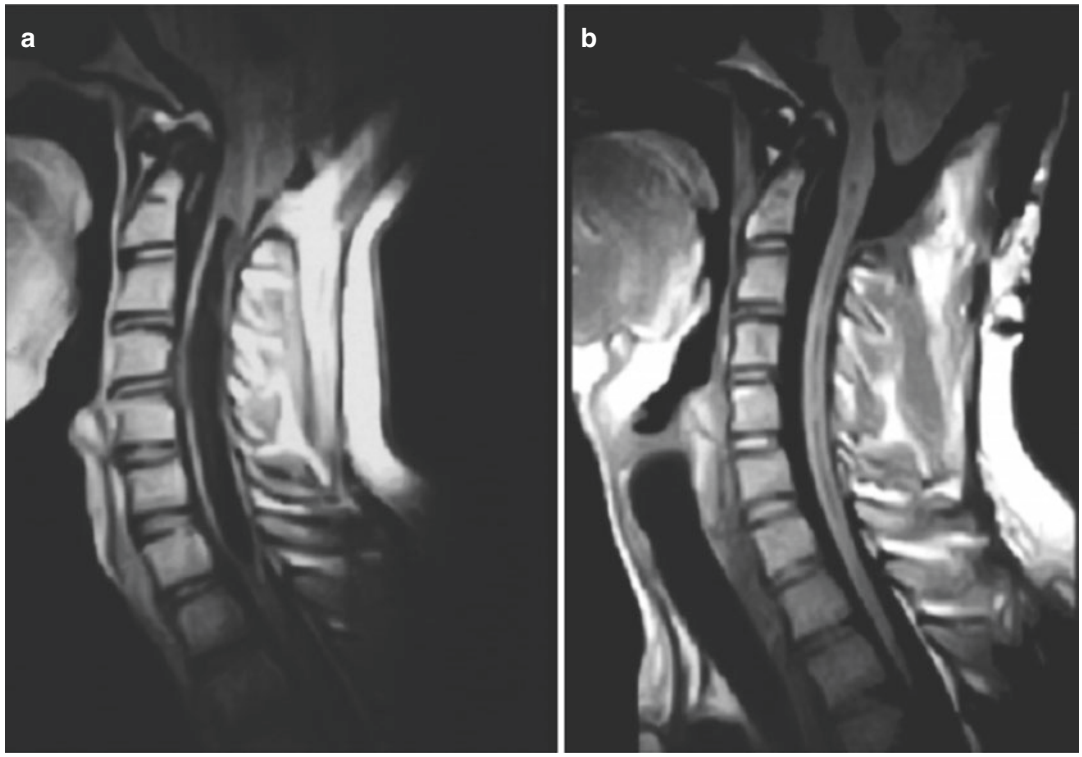

Fig. 17.2 (**a**) Preoperative images of the patient described in Fig. 17.1. (**b**) Postoperative imaging of the same patient

References

1. Chiari H. Ueber Veränderungen des Kleinhirns infolge von Hydrocephalie des Grosshirns1. Dtsch Med Wochenschr. 1891;17(42):1172–17.
2. Chiari H. UberdieVeränderungendesKleinhirns,derPonsund der Medulla oblongata in Folge von congenitaler Hydroceph- alie des Grosshirns. Denkschr Akad Wissensch Math Naturw Cl. 1895;63:71–116.
3. Van Houweninge Graftdijk CJ. Over hydrocephalus. Leiden: Eduard Ijdo; 1932.
4. Aitken LA, Lindan CE, Sidney S, Gupta N, Barkovich J, Sorel M, Wu YW. Chiari type I malformation in a pediatric population. Pediatr Neurol. 2009;40(6):449–54.
5. Meadows J, Kraut M, Guarnieri M, Haroun RI, Carson BS. Asymptomatic Chiari type I malformations identified on magnetic resonance imaging. J Neurosurg. 2000;92(6):920–6.
6. Vernooij MW, Ikram MA, Tanghe HL, Vincent AJ, Hofman A, Krestin GP, Niessen WJ, Breteler MM, van der Lugt A. Incidental findings on brain MRI in the general population. N Engl J Med. 2007;357(18):1821–8. https://doi.org/10.1056/NEJMoa070972.
7. Weber F, Knopf H. Incidental findings in magnetic resonance imaging of the brains of healthy young men. J Neurol Sci. 2006;240(1–2):81–4. https://doi.org/10.1016/j.jns.2005.09.008.
8. Haberg AK, Hammer TA, Kvistad KA, Rydland J, Muller TB, Eikenes L, Garseth M, Stovner LJ. Incidental intracranial findings and their clinical impact; the HUNT MRI study in a general population of 1006 participants between 50-66 years. PLoS One. 2016;11(3):e0151080. https://doi.org/10.1371/journal.pone.0151080.
9. Bordes S, Jenkins S, Tubbs RS. Defining, diagnosing, clarifying, and classifying the Chiari I malformations. Childs Nerv Syst. 2019;35(10):1785–92.
10. Azahraa Haddad F, Qaisi I, Joudeh N, Dajani H, Jumah F, Elmashala A, Adeeb N, Chern JJ, Tubbs RS. The newer classifications of the chiari malformations with clarifications: an anatomical review. Clin Anat. 2018;31(3):314–22.
11. Shrot S, Soares BP, Whitehead MT. Cerebral diffusivity changes in fetuses with Chiari II malformation. Fetal Diagn Ther. 2019;45(4):268–74.
12. Tubbs RS, Beckman J, Naftel RP, Chern JJ, Wellons JC 3rd, Rozzelle CJ, Blount JP, Oakes J. Institutional experience with 500 cases of surgically treated pediatric Chiari malformation type I. J Neurosurg Pediatr. 2011;7(3):248–56.
13. Houston JR, Eppelheimer MS, Pahlavian SH, Biswas D, Urbizu A, Martin BA, Bapuraj JR, Luciano M, Allen PA, Loth F. A morphometric assessment of

type I Chiari malformation above the McRae line: a retrospective case-control study in 302 adult female subjects. J Neuroradiol. 2018;45(1):23–31. https://doi.org/10.1016/j.neurad.2017.06.006.
14. Urbizu A, Poca M-A, Vidal X, Rovira A, Sahuquillo J, Macaya A. MRI-based morphometric analysis of posterior cranial fossa in the diagnosis of chiari malformation type I. J Neuroimaging. 2014;24(3):250–6.
15. Houston JR, Allen NJ, Eppelheimer MS, Bapuraj JR, Biswas D, Allen PA, Vorster SJ, Luciano MG, Loth F. Evidence for sex differences in morphological abnormalities in type I Chiari malformation. J Neuroradiol. 2019;32(6):458–66.
16. Rijken BF, Lequin MH, van der Lijn F, van Veelen-Vincent ML, de Rooi J, Hoogendam YY, Niessen WJ, Mathijssen IM. The role of the posterior fossa in developing Chiari I malformation in children with craniosynostosis syndromes. J Craniomaxillofac Surg. 2015;43(6):813–9.
17. Tubbs RS, Rutledge SL, Kosentka A, Bartolucci AA, Oakes WJ. Chiari I malformation and neurofibromatosis type 1. Pediatr Neurol. 2004;30(4):278–80.
18. Richards PS, Bargiota A, Corrall RJ. Paget's disease causing an Arnold-Chiari type 1 malformation: radiographic findings. Am J Roentgenol. 2001;176(3):816–7.
19. Ammerman JM, Goel R, Polin RS. Resolution of Chiari malformation after treatment of acromegaly. Case illustration. J Neurosurg. 2006;104(6):980.
20. Marin-Padilla M, Marin-Padilla TM. Morphogenesis of experimentally induced Arnold—Chiari malformation. J Neurol Sci. 1981;50(1):29–55.
21. Sansur CA, Heiss JD, DeVroom HL, Eskioglu E, Ennis R, Oldfield EH. Pathophysiology of headache associated with cough in patients with Chiari I malformation. J Neurosurg. 2003;98(3):453–8.
22. Grahovac G, Pundy T, Tomita T. Chiari type I malformation of infants and toddlers. Childs Nerv Syst. 2018;34(6):1169–76.
23. Batzdorf U. Historical aspects. In: Flint G, Rusbridge C, editors. Syringomyelia; a disorder of CSF circulation. Berlin: Springer; 2014. p. 1–9.
24. Massimi L, Frassanito P, Bianchi F, et al. Bony decompression vs duraplasty for Chiari I malformation: does the eternal dilemma matter? Childs Nerv Syst. 2019;35:1827–38.
25. Massimi L, Frassanito P, Chieffo D, Tamburrini G, Caldarelli M. Bony decompression for chiari malformation type I: long-term follow-up. In: Visocchi M, editor. New trends in craniovertebral junction surgery, Acta neurochirurgica supplement, vol. 125. Cham: Springer; 2019.
26. Förander P, Sjåvik K, Solheim O, Riphagen I, Gulati S, Salvesen Ø, Jakola AS. The case for duraplasty in adults undergoing posterior fossa decompression for Chiari I malformation: a systematic review and meta-analysis of observational studies. Clin Neurol Neurosurg. 2014;125:58–64.
27. Perrini P, Benedetto N, Tenenbaum R, et al. Extra-arachnoidal cranio-cervical decompression for syringomyelia associated with Chiari I malformation in adults: technique assessment. Acta Neurochir. 2007;149:1015–23.