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Syringomyelia with Chiari malformation; 3 unusual cases with implications for pathogenesis

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Summary

Syringomyelia is an important cause of neurological deficit. Most cases of non-traumatic syringomyelia occur in association with a Chiari malformation. We present three unusual examples of syringomyelia with such an association. The first case is that of syringomyelia in a young woman with Marfan's syndrome, a spontaneous CSF leak and intractable intracranial hypotension. The second is a woman with long-standing lumbo-peritoneal shunt for pseudotumour cerebri who developed an acquired Chiari malformation. A young woman with a Dandy-Walker cyst that herniated into the upper cervical canal is the third case. These cases provide a basis for discussion of the pathogenesis and management of syringomyelia and the Chiari malformation in such cases.

Keywords: Syringomyelia; idiopathic intracranial hypotension; pseudotumour cerebri; Dandy-Walker malformation.

Introduction

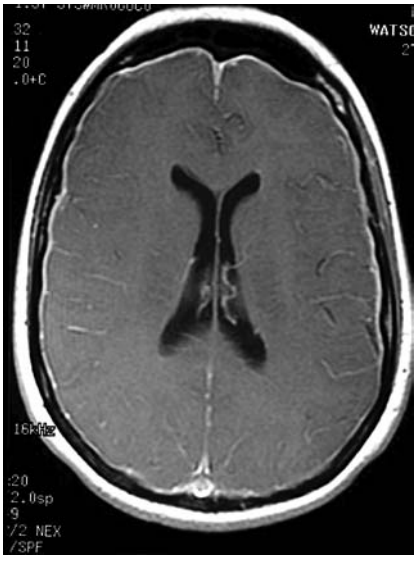
Syringomyelia is a condition in which cystic cavities develop within the parenchyma of the spinal cord. Neurological manifestations are varied from being asymptomatic to patterns of dissociated sensory loss to quadriplegia. Theories to explain the pathogenesis of the syringomyelia are many and varied. We present three unusual cases of syringomyelia secondary to different pathologies. All three cases occurred in combination with a Chiari I malformation. The first case occurred in association with a spontaneous CSF leak in a patient with Marfan's syndrome. This combination of pathologies has not been previously described. The second case developed in a patient with pseudotumour cerebri after prolonged lumbo-peritoneal (LP) shunting. The final case occurred secondary to herniation of a Dandy-Walker cyst into the upper cervical canal. These cases provide a basis for a discussion of the theories of pathogenesis and management of syringomyelia.

Case reports

Case 1

A 27 year-old woman was referred to this institution for assessment and management of intractable postural headache, neck pain, dizziness and nausea. These symptoms started 5 weeks previously and occurred every time she sat up and resolved immediately when she lay down. CT scan was reported as unremarkable. She had a background of Marfan's syndrome with the typical body habitus: she was 6′4″ tall with arachnodactyly, high arched palate, striae and positive wrist and thumb signs. Due to progressive aortic root dilatation she had undergone a Bentall's procedure 2 years previously and was currently anticoagulated with Warfarin.

The neurological examination was remarkably normal with no nystagmus, no cranial nerve abnormalities, no reflex or sensory changes. MRI of the brain demonstrated diffuse dural enhancement consistent with intracranial hypotension (Fig. 1a). There was tonsillar herniation (Chiari I "malformation") and the pons appeared flattened against the clivus (Fig. 1b). There was no hydrocephalus. MRI of the spine revealed a spinal syrinx extending from the C2/3 to the T7 vertebral level and there were dilated epidural veins in the cervical region (Fig. 1c). No discrete CSF leak was identified. There was evidence of lumbar dural ectasia with a thecal sac to vertebral body ratio of 0.58 (normal less than 0.47) at the L3 level [19]. The decision was made not to proceed with radio-isotope cisternography or with a blood patch because of the risks of ceasing Warfarin. Consequently the patient was managed conservatively with bed rest and her symptoms gradually resolved. A follow-up MR scan 4 months later demonstrated no change in the degree of tonsillar herniation or dural enhancement. However the syrinx had collapsed and the patient remains well under clinical observation.



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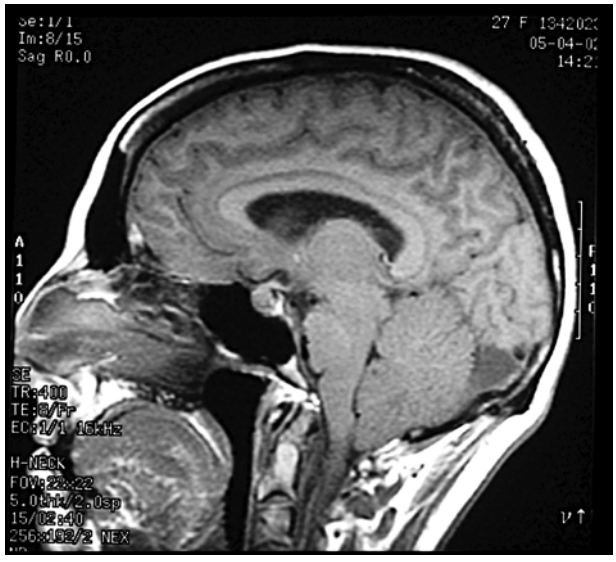

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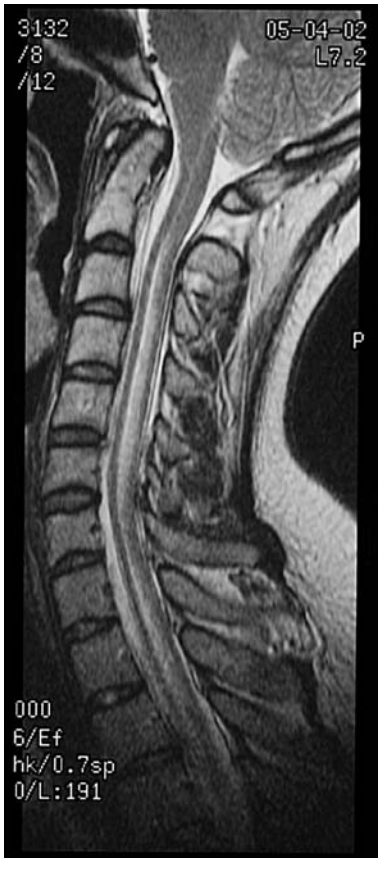

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Fig. 1. (a) T1-weighted axial MR image with gadolinium demonstrating meningeal enhancement consistent with intracranial hypotension. (b) T1-weighted sagittal MR image demonstrating the secondary Chiari I malformation in a patient with Marfan's syndrome and spontaneous CSF leakage. (c) T2-weighted sagittal MR image showing the extensive cervico-thoracic syrinx

Case 2

A 42 year-old woman was diagnosed with pseudotumour cerebri on the basis of headache papilloedema and high lumbar CSF pressure and treated with a lumbo-peritoneal shunt 13 years previously. She remained until 4 months before presenting with deteriorating balance and diplopia. On examination she had a mixed downbeating and torsional nystagmus, a skew deviation, absent tendon reflexes, impairment of pain and temperature sensation on the right arm and chest from C3 to about T8 and slight weakness without wasting in the right hand; the plantar reflexes were flexor. MR demonstrated descent of the cerebellar tonsils to the level of C1, about 10 mm (Fig. 2a). There was an extensive cervico-thoracic syrinx from C1-T11 (Fig. 2b).

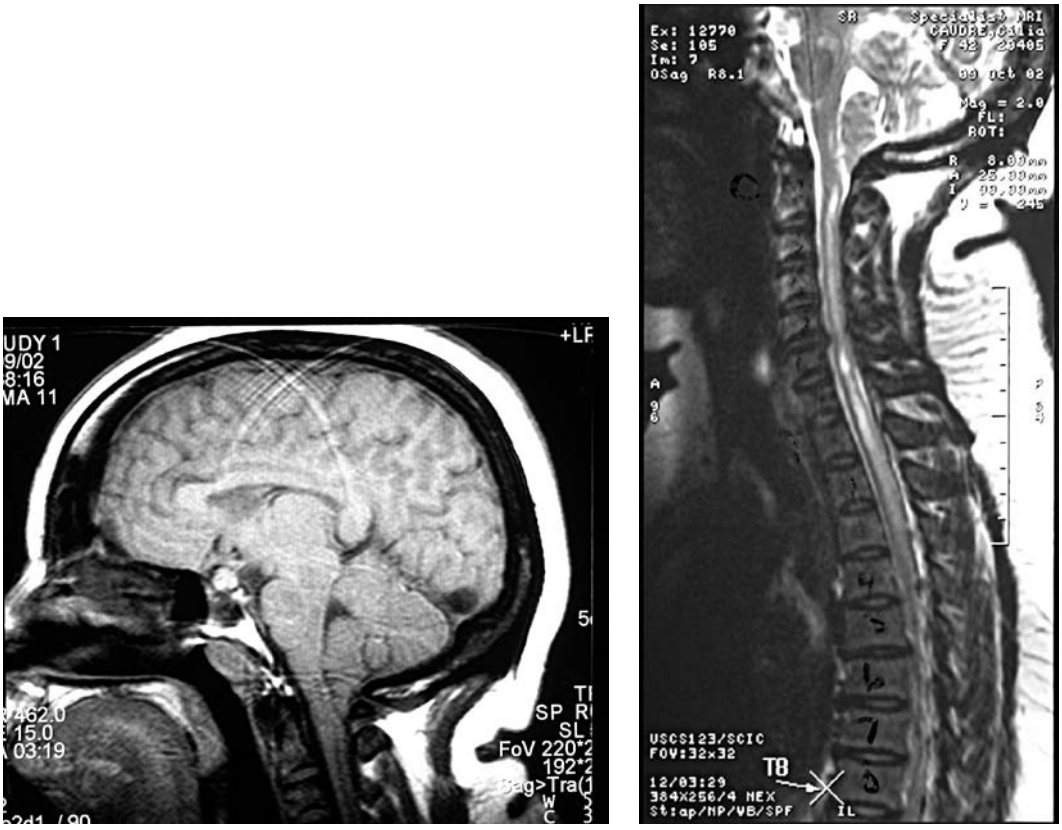

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Fig. 2. (a) T1-weighted sagittal MR image demonstrating the acquired Chiari I malformation in a patient with a lumbo-peritoneal shunt for pseudotumor cerebri. (b) T2-weighted sagittal MR image showing an extensive spinal syrinx

It was decided to direct treatment at the Chiari I malformation given the nature of her symptoms. A modified bilateral suboccipital craniectomy was performed with removal of the foramen magnum rim. The posterior arch of C1 and the superior half of the C2 lamina were also removed. On durotomy the cerebellar tonsils were noted to be at the level C1. The arachnoid was dissected to ensure free flow of CSF from the fourth ventricle. A previously prepared pericranial patch was sewn in as a duroplasty. The bone flap was replaced and the wound closed in the standard manner.

Symptoms improved and 4 months later the lumbo-peritoneal shunt was clipped to assess whether it was still necessary. Two months after clipping of the shunt she returned with headache and papilloedema so the shunt was reconnected. Since that time she has continued to improve.

A follow-up MR scan revealed some cerebellar descent through the enlarged foramen magnum with continued herniation of the cerebellar tonsils. The syrinx had collapsed although a tiny amount of CSF could still be seen within the collapsed syrinx cavity.

Case 3

A 29 year-old woman was referred for assessment of intermittent occipital headaches, bilateral upper limb paraesthesia and radiological abnormalities of the craniocervical junction. The paraesthesia and numbness in the hands had initially occurred post-partum and would be exacerbated by exertion. At age 13 months she had undergone an occipital reconstruction for an occipital meningocele using an autologous rib graft. She had made a good recovery without developmental abnormalities.

Neurological examination including fundoscopy was normal. MRI demonstrated a Dandy-Walker malformation with agenesis of the cerebellar vermis (Fig. 3a). The Dandy-Walker cyst protruded into the upper cervical spinal canal producing a block to CSF flow in the subarachnoid space. A syrinx was present in the cervical spinal cord (Fig. 3b). There was mild hydrocephalus which was probably long-standing. It was felt that her symptoms were spinal cord in nature, and in order to treat the syrinx and prevent progression of the disease, we chose to treat the Dandy-Walker cyst, decompress the foramen magnum and perform a duroplasty.

At operation, the foramen magnum rim and a section of occipital bone was removed, along with the posterior arch of C1 and the superior half of the C2 lamina. On durotomy the Dandy-Walker cyst immediately became apparent. Multiple arachnoid adhesions were encountered in the region of the foramen magnum. The spinal subarachnoid space was opened with free flow of CSF. The Dandy-Walker cyst was then widely fenestrated into the spinal subarachnoid space. The obex was noted to be invaginated by the fourth ventricular cyst. A duroplasty was performed and the wound closed in the standard manner.

Post-operative MR scans confirmed complete collapse of the syrinx with free flow of CSF around the dorsal spinal cord. Her clinical course was complicated by the development of delayed onset dorsal column dysfunction which settled spontaneously by the post-operative visit at six weeks. The post-operative deficits may reflect a change in CSF circulation dynamics and/or local parenchymal shift with its associated stress and strain after collapse of the long-standing Dandy-Walker cyst. Her

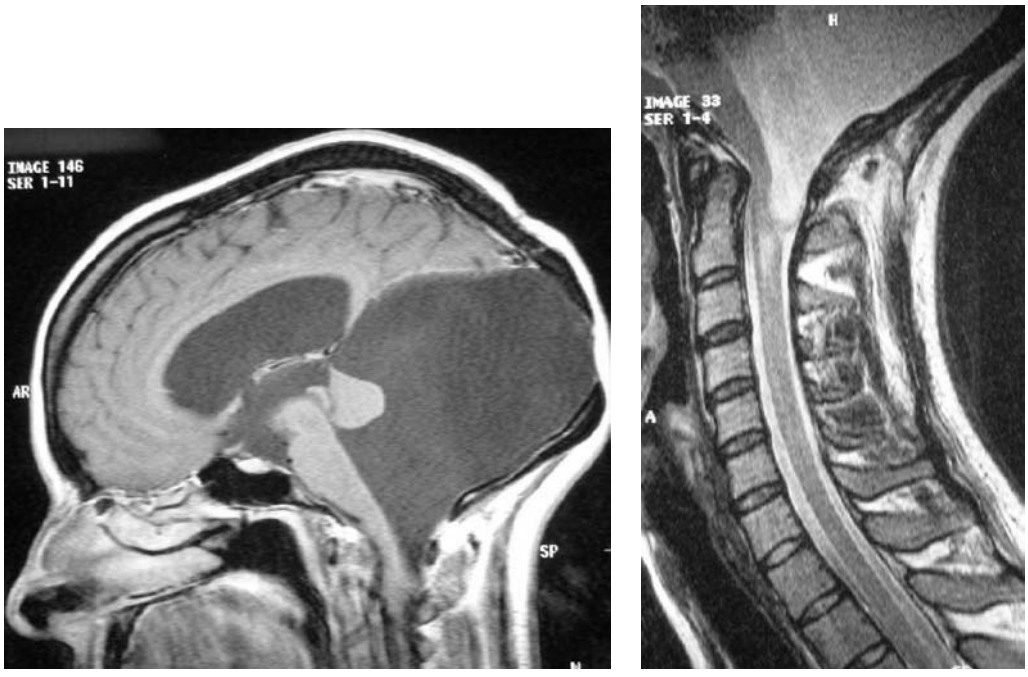

Fig. 3. (a) T1-weighted sagittal MR image showing a large posterior fossa cyst consistent with a Dandy-Walker malformation. The inferior aspect of the cyst herniates through the foramen magnum causing a type of Chiari malformation. (b) T2-weighted sagittal MR image showing the herniated Dandy-Walker cyst at the foramen magnum. There is a small upper cervical syrinx

initial symptoms completely resolved and MRI at six months showed complete resolution of the syrinx. She continues to undergo clinical and radiological surveillance.

Discussion

These three cases of syringomyelia with unusual associations provide a basis for evaluating theories of the pathogenesis and management of syringomyelia.

Marfan's syndrome is a known cause of spontaneous intracranial hypotension (SIH) [5, 17, 22]. An underlying weakness in the spinal thecal sac CSF is the potential site for a CSF leak since dural ectasias, meningoceles and arachnoid diverticula are common in Marfan's syndrome [20]. While most patients with Marfan's syndrome display a mutation of the *FBN-1* gene found on the long arm of chromosome 15. However, Schrijver *et al.* [22] have recently described a group of patients with Marfan's syndrome and spontaneous CSF leakage who do not display *FBN1* gene mutation and proposed a new classification for this subgroup – microfibrillopathy. None of these patients in their series had syringomyelia. To date, only one case of syringomyelia with a secondary Chiari malformation due to spontaneous CSF leak has been described [23]. That patient did not have Marfan's syndrome. Therefore this is the first case report describing the association between Marfan's syndrome, spontaneous CSF leakage and syringomyelia.

In our case the need for continuing anticoagulation contraindicated the use of a diagnostic CT myelogram. We could not identify the site of CSF leakage on MR imaging. However, there are recent reports using MR techniques to localize the point of CSF leakage [14]. Atkinson *et al.* [1] and Sharma *et al.* [23] have emphasized the importance of both pachymeningeal enhancement and sagging of various brain structures on MR in combination with Chiari I malformation. Both features were prominent in our case. Our therapeutic options were also limited because of both the inability to localize the point of leakage and the need for continued anticoagulation. The conservative approach used in this patient was successful. However, when conservative therapy fails percutaneous [1, 5] and various open surgical procedures may be useful [11, 21]. We postulated that correction of the underlying CSF leak and spinal subarachnoid hypotension, would result in spontaneous resolution of the Chiari I malformation and therefore the syringomyelia. Atkinson *et al.* [1] have reported the success of such an approach for acquired Chiari I malformation in spontaneous CSF leakage without syringomyelia.

In regard to out second case, development of an acquired Chiari I malformation is a known risk of lumboperitoneal shunting. Welch *et al.* [26] reported five patients who developed Chiari I malformations after LP shunting – 2 of these patients also demonstrated syringomyelia. However, the association between the development of a Chiari malformation and in some cases a syrinx does not appear to depend on the underlying aetiology. Johnston *et al.* [12] reported acquired Chiari malformation and syringomyelia associated with LP shunting in cases of craniofacial dysostosis and communicating hydrocephalus, an occipital pseudomeningocele and pseudotumour cerebri.

The association between pseudotumour and syringomyelia may not be so straight-forward. Kushner *et al.* [13] described syringomyelia with no evidence of a Chiari malformation at autopsy in a patient who had had an LP shunt for a long period for communicating hydrocephalus. Fischer *et al.* [6] also reported two cases of syringomyelia without ACM after LP shunting. Johnston *et al.* [12] reported 2 of 19 cases in whom a syrinx was detected on MR imaging prior to any surgical treatment. In two cases of pseudotumour cerebri treated with LP shunting, a symptomatic syrinx, without a Chiari malformation, developed. One was treated with a syringopleural shunt and another with syringostomy and maintenance of the LP shunt. Both subsequently developed Chiari malformations.

In respect to treatment, correction of the supposed over drainage of CSF from the spinal subarachnoid space should correct the condition. In the report by Johnston *et al.* [12], resiting the LP shunt to the ventricular system was useful, although this was more likely to be successful in cases of communicating hydrocephalus than pseudotumour. Sullivan *et al.* [25] reported resolution of a syrinx in a patient with pseudotumour cerebri and an LP shunt. The difficulty in resiting the shunt in pseudotumour cerebri is well known. The ventricles are often small and these patients are prone to develop slit-ventricles and recurrent obstruction. Shunting from the cisterns has been used to overcome this problem but is more difficult to perform and revise. We chose to perform a craniocervical decompression as the first procedure and left the LP shunt functioning in order to minimize the risks of CSF leakage. As it had been many years since her initial presentation, we decided to clip the LP shunt in a delayed fashion. This however was unsuccessful and it was unclipped.

Syringomyelia in association with a Dandy-Walker malformation is rare. Hammond *et al.* [9] recently reported a case and reviewed the literature. Only 17 cases had been reported. Milhorat [15, 16] described five patients with the association between the Dandy-Walker cysts and the communicating type of syringomyelia where there is communication between the fourth ventricle and the syrinx cavity via the central canal.

Shunting of the fourth ventricle or lateral ventricles, if the aqueduct is patent, may result in successful treatment of both the cyst and the syrinx. The mechanism of action may be to halt CSF flow through the obex into the central canal and/or reduce the herniation of the Dandy-Walker cyst restore normal CSF dynamics to the region of the foramen magnum. It is interesting that in our case that, at operation, arachnoid adhesions around the herniated Dandy-Walker cyst required dissection. This has not been previously described. While shunting the fourth ventricle may have been successful in this patient, the presence of such adhesions is likely to have resulted in failure of this treatment.

By contrast there have been several reports of posterior fossa cysts with herniation into the upper cervical canal [3, 4]. Interestingly all three cases reported by Banna *et al.* [3] were found to have dense arachnoid adhesions around its lower part. In addition there was no communication between these inferocerebellar arachnoid cysts and the fourth ventricle. A similar case was described by Canno *et al.* [4]. All patients were treated successfully, two with cyst-peritoneal shunts and two with suboccipital craniectomies and foramen magnum decompressions.

Numerous theories surrounding the pathogenesis of syringomyelia have been suggested. Both theories in which alterations in CSF dynamics and those in which parenchymal pathology, for example ischaemia or dysraphism, are the fundamental cause of syringomyelia have been described. Gardner [7, 8] suggested that CSF was forced along the central canal of the spinal cord resulting in its cavitation. This was primarily a result of a blockage of CSF outflow from the foramina of the fourth ventricle. By contrast, Williams [27] and later Ball and Dylan [2] suggested that, a block to the free flow of CSF at the foramen magnum causes a dissociation of the cranial and spinal CSF pressures. During Valsalva, coughing or straining, CSF pressure in the spinal subarachnoid space is raised as the epidural veins are engorged. A blockage at the level of the foramen magnum results in an increased pressure in the spinal subarachnoid space which forces CSF through the perivascular (or Virchow-Robin) spaces and into the central canal. Williams [28] was able to measure

a pressure differential between the compartments during Valsalva. However, this was unable to be replicated by Heiss *et al.* [10]. Oldfield *et al.* [18] and later Heiss *et al.* [10] focusing on the combination of Chiari I malformation and syringomyelia used cine MR imaging and intraoperative ultrasound they were able to document the piston-like motion of the cerebellar tonsils and motion of the spinal cord throughout the cardiac cycle. They demonstrated that during cardiac systole the spinal cord containing the area of the syrinx collapsed and then expanded during diastole. They concluded that the CSF pulse-pressure wave produced during cardiac systole, that was normally absorbed by passage into the spinal subarachnoid space, was transmitted to the cerebellar tonsils which also moved down during systole and produced a large pressure wave in the upper cervical region, compressing the cervical cord in this region and which resulted in the passage of CSF into the central canal of the spinal cord.

Considering our three cases, all had a type of Chiari I malformation. The first two cases however had acquired Chiari I malformations secondary to either a CSF leak or an over-draining LP shunt. According to the mechanism proposed by Oldfield [18] and Heiss *et al.* [10], the Chiari malformation transmits an increased pulse pressure to the subarachnoid space of the upper cervical cord, compressing it and driving CSF via perivascular spaces into the central canal. However, CSF pressure in the spinal subarachnoid space in patients with spontaneous CSF leakage is typically low and the same might be said for an over-draining LP shunt. A low pressure might reduce the pressure transmitted via the piston-like action of the cerebellar tonsils given the increased compliance in this low pressure space and the provision of an alternative route for CSF to escape. Conversely however, the amplitude of the pressure wave may be accentuated by the pressure differential as a low pressure in the spinal subarachnoid space might allow greater excursion in the downward movement of the tonsils. Interestingly the finding that perivascular flow continues in the presence of a reduced spinal subarachnoid CSF pressure in the spines of normal sheep [24] may have implication for these two cases. Also, as noted previously, patients with pseudotumour may develop syringomyelia in the absence of a Chiari I malformation suggesting other factors may be operating in some cases. The pathogenesis of the syrinx in the third case appeared to be associated with a foramen magnum obstruction due to combination of tonsillar herniation and herniation of the enlarged fourth ventricular cyst in to the foramen. The cystic fourth ventricle did not appear to communicate with the central canal in this case. Rather the tissue forming the dorsal wall of the top of the spinal cord was pushed interiorly closing the obex. Thus despite an apparent association between communicating syringomyelia and Dandy-Walker malformation [15], this case appears to be a noncommunicating form of syringomyelia. Interestingly symptoms developed immediately after childbirth – this was a normal vaginal delivery. There was no epidural anaesthetic to inadevertently penetrate the thecal sac and lower the spinal CSF pressure. But association between this period of straining and onset of symptoms suggests a role for venous distention in the pathogenesis also.

Conclusions

Continued investigation into the pathogenesis of syringomyelia is important. Impressive results with the application of newer imaging techniques have provided an increase in our knowledge base and plausible explanations of pathogenesis. However, especially in atypical circumstances thought should be given to a range of potential pathogenic mechanisms as selection of patients for surgery and the type of surgery and its ultimate success depends upon the pathological mechanism of the syrinx and its associated CSF dynamics.

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Comment

Owler *et al.* present 3 interesting patients with unusual Chiari I Malformations and syringomyelia. As to the treatment strategy in each of these patients, I fully agree with their management. Patients with Chiari Malformations are a very diverse population and treatment has to be adopted individually depending on the pathomechanism how the Chiari Malformation originated. This report is an excellent example how treatment can be successful even if complex problems such as a pseudotumor cerebri or a Dandy-Walker cyst have to be taken into consideration.

The implications for the pathogenesis of a syrinx in these patients are well presented and discussed. I fully agree that Oldfield's and Heiss' explanation for syringomyelia associated with Chiari Malformations is not completely satisfying for these patients. With obstruction of CSF pathways we could demonstrate increased pressure gradients between spinal cord parenchyma and subarachnoid space as well as within the spinal subarachnoid space in an animal model [2] supporting the authors assumptions. The interaction between CSF flow and extracellular fluid dynamics appear to hold the key for understanding the pathophysiology of syringomyelia [2].

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