A teenage girl with numb fingers

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Case report and quiz

A 15 year old girl presented to the paediatric neurology clinic with a one year history of lower back pain which radiated to her right leg and a six month history of numbness and tingling in her right arm, particularly on the outer aspect. The patient described sharp, shooting pains down her right arm in to her fingers when coughing and sneezing, which lasted several seconds. There was no history of trauma to the skull or spine, no loss of sensations or pain in any other areas and there was no history of vomiting, bladder or bowel disturbance. Otherwise she was a fit and healthy girl whose development was entirely normal and whose immunisations were complete. The only significant family history was spinal stenosis in her father.

On examination she was apyrexial and haemodynamically stable. Cranial nerve examination was normal. Tone and power were normal in the upper and lower limbs. The biceps and supinator reflexes were absent in the right upper limb. Other reflexes were normal and plantar responses were flexor. Pain and temperature sensation was impaired over the C5, C6 and C7 dermatomes of the right arm. All sensory modalities were normal everywhere else. Her gait was normal.

Questions

What is the likely diagnosis and what investigations should be performed?

What abnormalities are seen on the MRI?

What is the treatment?

Answers

Differential Diagnosis

On a quick glance the sensory disturbance and areflexia could localise the problem to C5-7 nerve roots or brachial plexus. However, pain and temperature are selectively affected with preservation of dorsal column sensations and most importantly the symptoms are exacerbated by coughing and sneezing. This suggests that symptoms are worsened by brief increases in intracranial pressure (ICP) and must therefore be related in some way to CSF dynamics. In addition there is a history of lower back pain radiating down the left leg which is unexplained by a C5-7 root or brachial plexus problem. The diagnosis is therefore likely to be one related to changes in ICP.

The first investigation would be an MRI of head and spine (Figure 1). Only consider further investigations once the result is known.

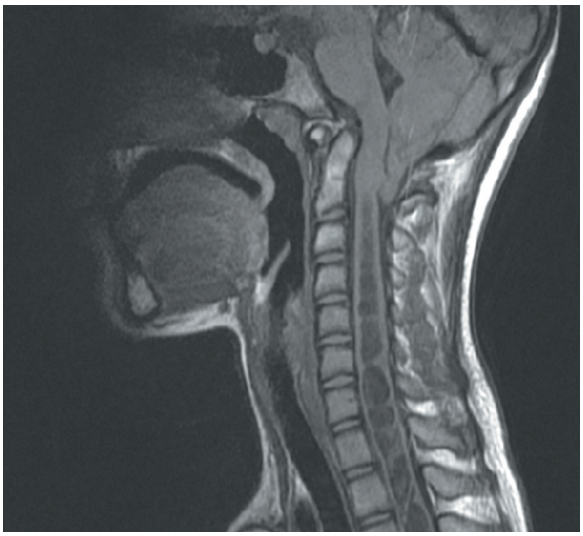

Figure 1. – *midline sagittal T1 weighted MRI showing Chiari 1 Malformation and a syrinx*

Abnormalities on the MRI:

The sagittal section shows two abnormalities. There is an Arnold-Chiari I malformation (cerebellar tonsils are protruding inferiorly through the foramen magnum) and extensive syringomyelia (the dark central cord cavity which in this image is visible from C2 down to T3).

The Treatment:

The treatment is a foramen magnum decompression undertaken by the neurosurgical team. The operation involved drilling four burr-holes through the occipal bone and then cutting between them. The posterior margin of the foramen magnum and posterior arch of C1 is then removed. The dura is expanded with a dural graft and then the bone flap is replaced more inferior to its original position and is fixed with titanium plates. The final aim of the operation is to create more space within the posterior fossa, which enables CSF to flow more freely and thus reduces CSF pressure. This decompression treats the syrinx and it should become smaller.

Discussion

Syringomyelia is a cavity within the spinal cord which may slowly increase in size. The most common sites are within the cervical and lumbar segments. The cavity may extend to involve the medulla and is called syringobulbia. The cavity is centrally placed in the cord and expands outwards, causing destruction of the anterior white commissure. The crossing fibres of the spinothalamic tracts are close to the expanding cavity anteriorly and are interrupted resulting in loss of pain and temperature sensation. In the early stages of the disease, involvement is usually asymmetric. The alpha motor neurones situated within the anterior horn cells more distant from the cavity are only affected when the lesion has become large; consequently muscle wasting is a late feature. The dorsal columns are unaffected, thus vibration and proprioception are spared.

Chiari One malformation (CM1) is used to describe cerebellar tonsil herniation of more than 5 mm though the foramen magnum[1], with associated obliteration of the subarachnoid spaces at the level of the foramen magnum. The CM1 has been an incidental finding on MRI in asymptomatic patients. However, when spinothalamic tract dysfunction is noted, syringomyelia is found in 50-70% of CM1 cases[2]. The majority of cases are sporadic, with most patients presenting from the second to fifth decades of life, although a few familial cases have been described in the last 100 years[2].

The Importance of Radiological Investigation

Neuro-imaging plays a fundamental role in the identification of the anatomical aspects of the CM1, with MRI representing the best modality. Prior to the advent of MRI, CT scanning was used but it never offered completely sensitive and specific images, even with the high-resolution, contrast-enhanced scans. However CT offers a sensitive method of demonstrating genetically associated bony abnormalities of the foramen magnum and the craniocervical junction, which can be associated with CM1[3].

As well as the position of the cerebellar tonsils, other radiological parameters should be used to assess the severity of the CM1. The most important is the extent of crowding of the neural structures within the posterior fossa and their impaction on the foramen magnum. It has been shown that those with CM1 tend to have smaller posterior fossa volumes, and that the smaller the volume the more significant the symptoms one would expect from the CM1. Symptoms are those of raised intracranial pressure and include headache which is worse on waking, particular postures and on coughing and vomiting. Conversely, it is also the case that those patients with smaller volumes achieve better outcomes during surgical decompression.

When investigating CM1 it imperative that the MRI should be extended to include the rest of the spinal cord. This is not only to exclude syringomyelia but also to detect an apparently ‘idiopathic’ scoliosis which can be associated with CM1 and a syrinx[4]. It is also important as a tool by which postoperative outcome can be measured. Chiari malformations are unlikely to ascend following surgery therefore this is not used as a primary outcome measure, but reduction in size of the syrinx is used.

The aetiology and pathophysiology

The genetic and acquired aetiologies thought to be associated with CM1 and syrinx.	
Acquired3,5,6	**Genetic2**
Cervical Spine Injury	Achrondroplasia
Following lumbar subarachnoid space shunting	Klippel-Feil Syndrome
Posterior fossa tumour or cyst Hydrocephalus	Primary basilar impression Goldenhar Syndrome
	Phakomatoses and spinal deformities
	e.g. neurofibromatosis 1, scoliosis and spina bifida

Klippel-Feil Syndrome – a condition where there is a reduced number and fusion of cervical vertebrae.

Goldenhar Syndrome – a rare syndrome which affects development of the face and also results in abnormal vertebral development.

Primary Basilar Impression – a radiological definition of an autosomal dominant form of craniovertibral invagination.

Phakomatoses – the word given to the group of neurocutaneous diseases, NF1 being an example.

The pathophysiology of syringomyelia is debated and there have been numerous hypotheses over the years which have been hotly contested. However, there is one explanation which has become generally accepted[7]. Firstly the CM1 malformation occurs which is associated with a smaller posterior fossa volume. The herniation of the cerebellar tonsils results in a pressure effect on the central cord canal. In addition to increased pressure from the CM1, the systolic CSF pressure is transiently increased on coughing and with increased pulse pressure. This promotes movement of the CSF from the subarachnoid space around the cord through the Virchow-Robin spaces into the cord parenchyma and central canal. The systolic pulsations are exaggerated so a net movement across the cord occurs which exceeds the rate at which the cord parenchyma can absorb the fluid. Consequently, the central canal becomes dilated with CSF and a syrinx is formed.

Apart from increased intracranial pressure, it is unknown why the CM1 occurs with a syrinx although it is clear that the malformation occurs before the syrinx so thus must be related.

Conclusion

In conclusion, this case is one of a 15 year old girl who presented with a 6 month history of paresthesia and numbness in her right arm and a one year history of lower back pain which was exacerbated by coughing and sneezing. Following imaging, she was diagnosed as having a Chiari 1 malformation associated with an extensive syrinx, which was treated with surgical decompression of the foramen magnum. Although the aetiology was unclear in her case, the various syndromes and acquired problems which can be associated with the condition have been discussed.

References:

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