SHORT REPORT

Arnold Chiari type 1 malformation presenting with sleep disordered breathing in well children

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Healthy children, aged 3, 9, and 13 years referred with sleep disordered breathing had marked central apnoea and bradypnoea on polysomnography, necessitating the use of non-invasive bilevel ventilation in two cases. Each had normal neurological examinations and an Arnold Chiari type 1 malformation was confirmed on magnetic resonance imaging. All underwent urgent posterior fossa decompression which normalised their sleep breathing.

The Arnold Chiari type I malformation is a congenital brainstem abnormality characterised by caudal herniation of the cerebellar tonsils through the foramen magnum, resulting in crowding at the cranio-cervical junction^{1,2} (fig 1). It seldom presents in childhood with symptoms and a normal neurological examination. More typically, Arnold Chiari type I malformation presents in adults with neurological symptoms including headache, neck pain, ataxia, lower cranial nerve palsies, and sensory deficit.^{1,2}

We describe three well children with an Arnold Chiari type I malformation who underwent urgent posterior fossa decompression which normalised their sleep breathing. Repeat sleep studies documented normal sleep breathing.

CASE 1

A 3 year old girl presented with a four month history of intermittent snoring and frequent nocturnal waking, the pattern of which was not typical of commonly seen causes of waking in this age group (behavioural wakings and partial sleep arousals). The tonsils were of average size. Polysomnography (PSG) revealed central apnoeic episodes of up to 15 seconds duration and marked sleep related bradypnoea, with a respiratory rate of 4–7 breaths per minute in sleep (fig 2). There was no evidence of obstructive sleep apnoea (OSA). Bi-level non-invasive ventilation (NIV) was initiated to normalise gas exchange prior to urgent neurosurgery. Following posterior fossa decompression, sleep breathing returned to normal and NIV was ceased.

CASE 2

A 9 year old girl was referred with a 12 month history of variable snoring and brief witnessed apnoeas. She had gained excessive weight (8 kg) in the preceding 15 months. A PSG was conducted, revealing central apnoea occurring approximately 20 times per hour, associated with a bradypnoea of 7–8 breaths per minute in stable non-REM sleep. OSA was not shown. There were no significant effects on gas exchange (Min SpO₂ 90% and no CO₂ retention). Posterior fossa decompression followed within weeks without the need for NIV.

CASE 3

A 13 year old girl was referred after failing to exhibit appropriate levels of respiratory drive during general anaesthesia for a minor orthopaedic procedure. Postoperatively she showed somnolence and lethargy for three days which improved spontaneously, suggestive of amplification of an underlying disorder of sleep breathing secondary to the general anaesthetic. Respiratory problems had not been noted during two prior peri-operative periods. Enquiry revealed a three year history of episodic, paroxysmal headache with no obvious precipitating features, and mild difficulty swallowing fluids. The neurological examination was normal. The tonsils were not enlarged and the oropharynx appeared normal. A PSG revealed marked bradypnoea with a respiratory rate of 4–5 breaths per minute, interspersed with long central apnoeas and frequent nocturnal arousal. There was no evidence of OSA. NIV was instituted. A foramen magnum decompression and cerebellar tonsil resection was performed, with no complications; NIV was ceased.

DISCUSSION

Type I Chiari malformation is characterised by caudal herniation of the cerebellar tonsils through the foramen magnum, resulting in crowding at the cranio-cervical junction.² It is commonly associated with cervical syringomyelia (syrinx), typically presents in adulthood, and is assumed to rarely present clinically before adolescence.^{1,2}

Figure 1 A sagittal T1 weighted image of an MRI scan showing herniation of the cerebellar tonsils more than 1 cm below the foramen magnum into the cervical spinal cord.

Figure 2 Sixty second epoch of stable REM sleep showing bradypnoea (respiratory rate of 9 breaths per minute) or hypopnoea (<50% of normal breath) with interspersed pure central apnoea.

Thus, its incidence is rare, particularly in the “asymptomatic” paediatric population with intermittent to mild snoring.² In comparison, in a young child with a history of snoring, OSA attributable to enlarged tonsils and adenoids is the most common abnormality seen on polysomnography.

The most common presenting symptom of the type I Chiari malformation is headache in adults.¹⁻³ Other common presentations are limb weakness and paraesthesia, vertigo, visual and hearing disturbances, and swallowing difficulties.¹⁻³ Physical signs may include motor deficits, sensory loss, nystagmus, lower cranial nerve palsy, trophic changes, and ataxia.¹⁻³ Adolescents and older children may present with headache and neck pain, swallowing difficulty, scoliosis and motor weakness, impaired oropharyngeal function, and developmental delay.³ Apnoea and snoring may also occur in some children, and cases of sudden death have been reported.¹⁻⁴ However, most patients present with multiple symptoms and signs, not sleep disordered breathing without neurological abnormalities on physical examination.²

Cases of sleep apnoea in adults and children with type I Chiari malformation have been reported, most of which involved additional neurological symptoms.³⁻⁵ Apnoeas in conjunction with the type 1 malformation have been described as being predominantly obstructive; however mixed and central apnoeas have also been reported. Few studies have emphasised pure central apnoea and bradypnoea on polysomnography as the primary presenting symptom.³⁻⁵

It has been postulated that central apnoea may occur because of compression or ischaemia of the respiratory centre, dysfunction of the ascending medullary reticular

activating system, abnormal ventilatory chemosensitivity, and paralysis of movement in the upper respiratory wall.¹⁻⁵ Depending on the degree or site of the compression, patients may manifest central apnoea with or without further neurological abnormalities.⁵ In our three cases, although intermittent snoring occurred, there was no evidence of OSA on polysomnography and on this basis the snoring was considered benign. In all cases, sleep breathing normalised rapidly following surgery and was confirmed with polysomnography.

The role of surgery in children is difficult to assess. Reports have described patients with abnormal neurological signs with a diagnosis of type I Chiari malformation who have benefited from surgical intervention with improved long term neurological outcomes.²⁻⁵ Treatment has centred on posterior fossa decompression, either alone or in combination with resection of cerebellar tonsillar tissue.²⁻⁵ It has been suggested that surgery should be undertaken in adults with sleep disordered breathing in order to prevent progression and onset of neurological symptoms.⁵

In conclusion, few cases have been identified in which benign snoring and life threatening central apnoea is the sole presenting symptom of the type I Chiari malformation. This highlights the notion that the presence of marked central apnoea and bradypnoea on polysomnography may be the primary presentation of brainstem compression, and its presence should prompt consideration of the diagnosis of a type I Chiari malformation.

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