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Porencephaly as a rare cause of nystagmus

Porencefalia jako rzadka przyczyna oczopląsu

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Abstract

Nystagmus is a condition of involuntary oscillatory movement of the eyes that may present in various patterns. These different types of movement and direction of nystagmus along with associated signs and symptoms can give a clue as to the location of the site of lesion, and indicate possible aetiologies. In paediatric patients, most of the causes of nystagmus are infantile or congenital, and only a small percentage may be acquired. We describe the case of a boy with spontaneous nystagmus noted since he was 3 years old and having a learning disability without other otological or neurological problems. Magnetic resonance imaging of his brain showed a porencephalic cyst which is a very rare occurrence.

Keywords: nystagmus, porencephalic cyst, spontaneous nystagmus, learning disability

Streszczenie

Oczopląs jest schorzeniem polegającym na mimowolnych, rytmicznych ruchach gałek ocznych, które mogą przyjmować różną postać. Rodzaj ruchu gałek ocznych i kierunku oczopląsu, a także obecność objawów towarzyszących mogą dawać klinicystom wskazówkę dotyczącą umiejscowienia zaburzenia wywołującego oczopląs i jego etiologii. U pacjentów pediatrycznych schorzenie jest zwykle wadą wrodzoną lub pojawia się w okresie niemowlęcym – jedynie w niewielkim odsetku ma charakter nabyty. W pracy przedstawiono przypadek chłopca z samoistnym oczopląsem występującym od 3. roku życia. U pacjenta nie stwierdzono żadnych innych schorzeń otologicznych ani neurologicznych. Odnotowano natomiast trudności w uczeniu się. W rezonansie magnetycznym mózgu chłopca wykazano obecność bardzo rzadko występującej zmiany w postaci torbieli porencefalicznej.

Słowa kluczowe: oczopląs, torbiel porencefaliczna, oczopląs samoistny, trudności w uczeniu się

INTRODUCTION

Nystagmus is a condition of involuntary oscillatory movement of the eyes which may be pendular or jerky in character. The direction of nystagmus may be horizontal, vertical, or rotatory[1]. These different types of movement and direction of nystagmus along with associated signs and symptoms can give a clue as to the location of the site of lesion, and indicate possible aetiologies.

The aetiology of nystagmus can be classified into three main categories: physiological, infantile and acquired. Most of the cases found in paediatric patients are infantile or congenital, while only 17% of nystagmus cases may be acquired[2].

Porencephaly is an extremely rare disease of the central nervous system characterised by a solitary or multiple cavity in the brain parenchyma, filled with cerebrospinal fluid[3].

CASE REPORT

A 13-year-old boy was referred from a primary care physician with bilateral spontaneous nystagmus noted since he was 3 years old. The condition was persistent, with no specific aggravating or relieving factors. There were associated visual disturbances evidenced by a history of frequently hitting things in front of him while walking, and he preferred to sit close while watching television. There was no history of headaches, no nausea or vomiting, and no history of seizures. The child did not complain of impaired hearing, and there was no tinnitus or vertigo, and no otalgia.

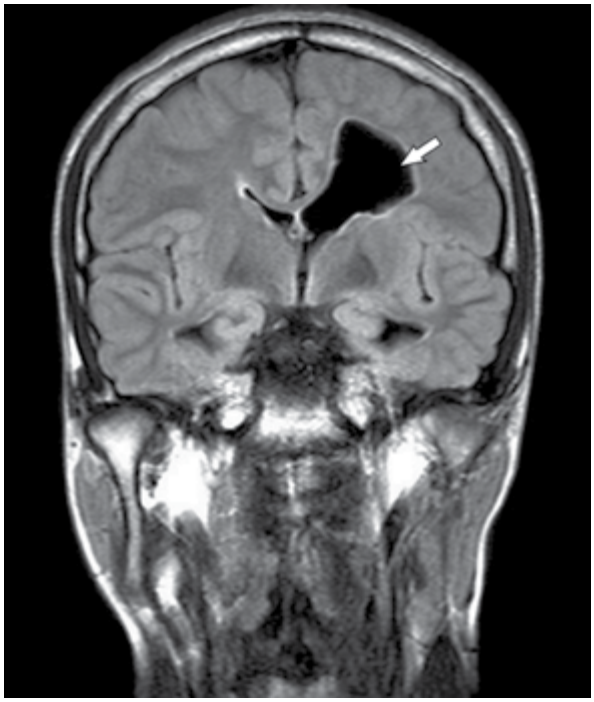

Fig. 1. A well-defined left frontal cystic lesion (arrow) measuring 2.7 × 2.4 × 3.0 cm, directly communicating with the body of the left lateral ventricle

He was a slow learner, attending a special school and at the time able to recognise alphabets. He was independent at home, and able to communicate with others. As for birth history, he was the second twin born at 28 weeks and incubated for four months. His twin brother passed away at 2 weeks of life. He was just currently brought for medical assessment because of symptom exacerbation.

The boy's physical examination revealed he had spontaneous horizontal pendular nystagmus which was not suppressed by visual fixation. His facial nerve function was intact. The gait was normal, and there were no cerebellar signs. An otoscopic examination revealed clear bilateral external ear canals. Both tympanic membranes were intact and normal. Pure tone audiometry showed that bilateral hearing was within normal limits. He also had bilateral type A tympanogram and absence of acoustic reflex bilaterally. Since the history and examination findings pointed towards a central cause of nystagmus, we referred the patient to the neurosurgery and ophthalmology team.

Magnetic resonance imaging (MRI) of the brain was performed and revealed a well-defined left frontal cystic lesion which communicated directly with the body of the left lateral ventricle (Fig. 1). It measured 2.7 × 2.4 × 3.0 cm, and followed cerebrospinal fluid signal intensity in all sequences (Fig. 2). The findings were consistent with a porencephalic cyst. There was no mass effect or midline shift. The child was treated conservatively and referred to the ophthalmology team for visual assessment, however he defaulted the follow-up.

DISCUSSION

Nystagmus is a condition of involuntary oscillatory movement of the eyes which may be pendular or jerky in character. The pendular type of nystagmus implies that the back and forth movements have the same pace, while the movement in jerky nystagmus has a different pace. The direction of nystagmus may be horizontal, vertical, or rotatory[1]. Spontaneous nystagmus signifies involuntary eye movements while the eyes are in the primary position without any evoking stimuli[4].

These different types of movement and direction of nystagmus along with associated signs and symptoms can give a clue as to the location of the site of lesion, and indicate possible aetiologies. Although nystagmus usually indicates a lesion involving the peripheral vestibular labyrinth, brainstem, or cerebellum, it also can be the result of lesions in the cerebral hemisphere[5]. The aetiology of nystagmus can be classified into three main categories: physiological, infantile and acquired. Most of the cases found in paediatric patients are infantile or congenital, while only 17% of nystagmus cases may be acquired[2].

Our patient had spontaneous horizontal pendular nystagmus which was not suppressed by visual fixation. It is pathognomonic of a central system pathology[6]. Considering this type of nystagmus, along with the patient's

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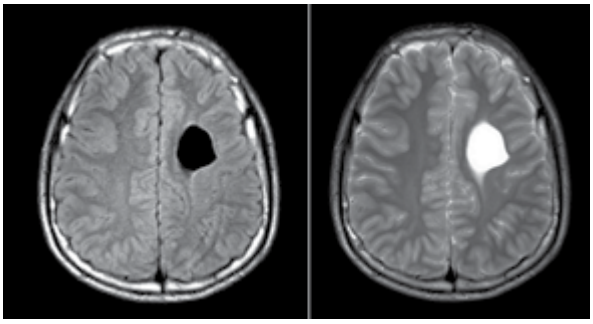

Fig. 2. Axial cut of brain MRI suggesting that the cyst contains cerebrospinal fluid; hypointense in T1WI (right) and hyperintense in T2WI (left)

learning disability and absence of other otological problems, we decided there was a strong indication to investigate possible central causes. It was then confirmed by the findings of the MRI of the brain which was consistent with a porencephalic cyst of the left frontal lobe.

Porencephaly is an extremely rare disease of the central nervous system characterised by a solitary or multiple cavity in the brain parenchyma, filled with cerebrospinal fluid[3]. The condition can be congenital or acquired after birth. Congenital porencephaly includes maldevelopment during the early prenatal period, and intracranial haemorrhage or ischaemia during the perinatal period[7]. In contrast, the causes of acquired porencephaly include cerebral haemorrhage or infarction, trauma, infection and idiopathic causes[8]. In view of the history of premature delivery and an eventful postnatal period, the patient most probably had an acquired porencephalic cyst after birth secondary to intracranial haemorrhage or ischaemia during the perinatal period.

A wide extent of clinical presentations associated with porencephaly have been reported. It may cause only minor neurological problems without any adverse impact on intelligence, while other affected patients may be severely disabled. Seizures, delayed growth and development and focal motor problems are among other common clinical findings[9]. Major brain parenchymal defects such as porencephaly, hydranencephaly, and anencephaly have been associated with secondary retrograde optic nerve axon degeneration resulting in optic nerve hypoplasia, which usually presents as mild to severe visual impairment and nystagmus[10].

The brain MRI findings obtained in this patient were consistent with the description of a porencephalic cyst; a well-defined cystic lesion lined by a thin layer of white matter and usually communicating with the body of the lateral ventricle. It also follows cerebrospinal fluid signal intensity in all sequences: hypointense in T1WI (weighted image), hyperintense in T2WI, suppressed in FLAIR, and non-enhancing post-contrast (Fig. 2).

In terms of treatment, our patient was treated conservatively, as there were no significant neurological symptoms. Other various treatment options include surgical removal of the cyst, shunts, antiepileptics, rehabilitation, and physical therapies[11]. Referral to the ophthalmology team for visual assessment was essential, since the patient showed evidence of visual disturbances.

CONCLUSION

Nystagmus can present in various patterns and types which may suggest different locations of the site of lesion and aetiology. MRI of the brain in different sequences is imperative for locating the lesion and making proper diagnosis. Mild neurological symptoms secondary to a porencephalic cyst can be managed conservatively, while more severe symptoms may need surgical intervention and rehabilitation.

Conflict of interest

The authors do not declare any financial or personal links with other persons or organisations that might adversely affect the content of the publication or claim any right to the publication.

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