PHOTO ESSAY

Patulous Subarachnoid Space of the Optic Nerve Associated with X-Linked Hypophosphatemic Rickets

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ABSTRACT

Although the deficiency forms are the most common manifestations of rickets, there are other forms of rickets that are resistant to vitamin D. Of these, the most common is X-linked hypophosphatemic rickets. Rickets represents a group of multiple cranial bone disorders—craniosynostosis and the presence of Chari I malformation being the most notable—that explain the increase in intracranial pressure. We present a 4-year-old patient with an unusual association of X-linked hypophosphatemic rickets, bilateral proptosis, and prominent bilateral widening of the optic nerve sheaths. Although the association between intracranial hypertension and rickets is known, to the best of our knowledge, such a prominent distention of the subarachnoid space of the optic nerve without papilloedema has not been previously described.

Keywords: Craniosynostosis, intracranial hypertension, optic nerve sheath, proptosis, X-linked hypophosphatemic rickets

CLINICAL CASE

We present a 4-year-old patient with potential nutritional rickets. Due to the characteristic clinical findings (detailed below), a diagnosis of X-linked hypophosphatemic rickets (XLH) was suspected and confirmed by genetic testing (heterozygous mutation c.1735G→A in exon 17 of the PHEX gene). At the age of 3 years, the patient presented with bilateral proptosis and significant lower extremity deformity.

Upon ophthalmologic examination, the patient presented a visual acuity (VA) of 20/30 OU (both eyes). The fundus of the eye revealed an optic disc of normal colour with well-defined borders OU (Figure 1).

A plain radiograph of the head revealed lacunar lesions (Figure 2) and of the lower extremities showed a marked genu varum (Figure 3). Cranial and orbital magnetic resonance imaging (Figure 4A–C) detected tubular widenings of the subarachnoid spaces (SAS) of both optic nerves. There was a slight flattening of the base of the cranium with cerebellar tonsillar

descent. Computerised tomographic (CT) imaging (Figure 5) of the cranium and orbits showed the optic canals to be of normal dimension bilaterally.

After 2 years of follow-up, the patient maintained the same VA, without the occurrence of papilloedema.

DISCUSSION

There are forms of rickets that are characterized by normal levels of vitamin D, thus termed vitamin D-resistant rickets.^{1,2} Within this group, the most common form is X-linked hypophosphatemic rickets (XLH).³

Rickets may be associated with a variety of cranial bone disorders – craniosynostosis and the presence of cerebellar tonsillar descent being the most notable – that can result in an increase in intracranial pressure.^{4–10}

In our patient there was a suspicion of intracranial hypertension associated with XLH. Unfortunately, a lumbar puncture with measurement of the opening

FIGURE 1 (A, B) Fundus of the eye revealed an optic disc of normal colour with well-defined borders OU (A, OD; B, OS) (Note: Figure 1 of this article is available in colour online at www.informahealthcare.com/oph).

FIGURE 2 Head X-ray showing disseminated lacunar lesions ("copper beaten skull").

pressure could not be performed, as this procedure is considered to be dangerous in patients with an existing cerebellar tonsillar descent.

What is most apparent in our patient is the presence of a sharp increase in the diameter of the SAS of the optic nerve in the absence of papilloedema. It is generally accepted that an increase in volume of the SAS of the optic nerve is associated with the presence of intracranial hypertension.^{11–13}

FIGURE 3 X-ray of the lower extremities showing marked genu varum.

However, distention of the SAS of the optic nerve has also been described idiopathically in patients without an apparent increase in intracranial pressure.¹⁴ Garrity et al.¹⁵ coined the term "optic nerve meningocele". However, according to some the term "patulous subarachnoid space" is to be preferred as a true meningocele does not exist.¹⁶ Distention of the SAS around the optic nerve has also been described as secondarily associated with cases of inflammation

FIGURE 4 (A, B, C) Head magnetic resonance with T1 sagittal sequences, coronal and axial CISS (constructive interference in steady state), detecting tubular widening of the subarachnoid spaces of both optic nerves leading to repositioning along their courses and an empty sella. There are also herniations of both cerebellar tonsils compatible with a cerebellar tonsillar descent.

FIGURE 5 Axial CT of the head and orbits showing optic canals of normal dimensions.

(perioptic neuritis) and infiltration by metastases or glioma (arachnoid gliomatosis).¹⁷

In the case of our patient, although the association between intracranial hypertension and rickets is known, to the best of our knowledge, such a prominent distention of the subarachnoid space (SAS) of the optic nerve without papilloedema has not been previously described.

How could the distention of the SAS of the optic nerve in our patient without papilloedema be explained?

We propose that the absence of papilloedema and the visual stability in our patient could be correlated with this unusual distensibility of the optic nerve sheath. This distention would somehow attenuate the transmission of the intracranial pressure to the optic nerve head, thus avoiding its progressive damage.

Finally the authors recognise that there might be other reasons for the optic nerve sheath dilation other than intracranial pressure (e.g. congenital dilation of the optic nerve sheath, lack or dysfunction of the collagen component in the sheath).

Declaration of interest: The authors report no conflicts of interest. The authors alone are responsible for the content and writing of the paper.

Note: Figure 1 of this article is available in colour online at www.informahealthcare.com/oph.

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