CASE REPORT

Unilateral Optic Nerve Hypoplasia with Contralateral Optic Pathway Hypoplasia: A Case Report

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ABSTRACT

Optic nerve hypoplasia is diagnosed by the ophthalmoscopic appearance of the fundus of the eye and by standard magnetic resonance imaging of the brain. The ability to study eyes with optic nerve hypoplasia by magnetic resonance diffusion tensor imaging has improved the evaluation of the optic pathways. The authors report a case of unilateral optic nerve hypoplasia with hypoplasia of the contralateral optic pathway. The entire visual pathway of this patient was examined by magnetic resonance and magnetic resonance diffusion tensor imaging. The images show a decrease of the volume of the optic radiation contralateral to the optic nerve abnormality and also pre- and post-chiasmal abnormalities.

Keywords: Magnetic resonance diffusion tensor imaging, optic pathway hypoplasia, unilateral optic nerve hypoplasia

INTRODUCTION

Optic nerve hypoplasia is traditionally diagnosed by the ophthalmoscopic appearance of the fundus of the eye and by standard magnetic resonance imaging (MRI) of the brain.^{1,2} The ability to study eyes with optic nerve hypoplasia in greater detail by magnetic resonance diffusion tensor imaging (MRDTI) has improved the evaluation of the optic pathway.³ MRDTI is an emerging noninvasive imaging technique that allows for an in vivo, quantitative assessment of the microstructural integrity of white matter fibre tracts. MRDTI is a useful adjunct to conventional MRI for the evaluation of the optic pathways.

We report a case of unilateral optic nerve hypoplasia with hypoplasia of the contralateral optic pathway. The visual pathways from the eye to the visual cortex were examined in detail by MRI and MRDTI.

CASE REPORT

The parents of a 3-year-old boy reported that the child appeared to have difficulty in vision with his left eye

and brought him to our hospital for a visual evaluation. The child was born by Caesarean delivery at a gestational age of 38 weeks with a birth weight of 2642 g. Echography found that he had foetal cerebral ventriculomegaly but he had no neurological symptoms or developmental delay. He was followed without any treatment.

Our initial examination showed that his visual acuity was 20/20 in his right eye and light perception in his left eye. The intraocular pressure was 16 mm Hg OD and 17 mm Hg OS measured by a noncontact tonometer (CT-80; Topcon, Tokyo, Japan). In dim illumination, the diameters of both pupils were 3 mm. Both pupils were round and reacted normally to direct light but a relative afferent pupillary defect was present in the left eye. All of the examinations showed that the right eye was completely normal. Slit-lamp examination of the left eye showed that the anterior segment was normal; however, ophthalmoscopy showed optic disc dysplasia with the double ring sign (Figure 1a, b).

MRI showed that the left optic nerve was thinner than was the right, i.e., left optic nerve dysplasia (Figures 2 and 3a). The optic chiasma did not appear

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FIGURE 1 Fundus photographs of the optic discs of a 3-year-old boy. (a) Right optic disc is normal. (b) Left optic disc is small and shows the double-ring sign (*Note:* Figures 1, 4, and 5 of this article are available in colour online at www.informahealthcare.com/oph).

FIGURE 2 Axial T2-weighted magnetic resonance image (MRI) of the optic pathway of a 3-year-old boy. MRI shows that the left optic nerve is thinner than the right.

asymmetric in the MR images (Figure 3b) but optic tract dysplasia was detected with the contralateral (right) optic tract being thinner than the left optic tract (Figure 3c). In addition, MRDTI showed a decrease in the volume of the optic radiation on the contralateral side (Figures 4 and 5). MRDTI was performed according to the method previously described by one of the authors (T.T.).⁴ The region of interest (ROI) placement was performed according to a method described in detail.³ Tractography was performed to evaluate the tensor metrics of the optic radiations. Deterministic streamline fibre tracking, using the fibre assignment by the continuous tracking method, was used together with the multiple regions of interest method to track the optic radiations. Using T2-weighted anatomic images superimposed on a fractional anisotropy (FA) map, one ROI was placed on the white matter near the lateral geniculate nucleus and one or two ROIs were placed on the white matter coursing posterior to the occipital lobe, which is known to contain the optic radiations. We took special

care in the drawings by making the positary of the seed and target areas by a method that one of the authors has explained in detail.⁴ There were no signs of hypopituitarism, and the patient is still being followed without treatment.

DISCUSSION

We were able to investigate the entire course of the optic pathways in a patient with reduced vision in the left eye. MRI showed that there was a unilateral left optic nerve hypoplasia, and MRDTI showed that the contralateral optic tract and radiation were thinner than that of the ipsilateral optic tract and radiation. This was unexpected because even if the optic nerve is atrophic unilaterally, the optic tracts and the optic radiations on both hemispheres should be equal in size because approximately 50% of the optic nerve fibres decussate at the optic chiasm with the fibres from the temporal retina remaining ipsilateral and the fibres from the nasal retina decussating in the chiasma.

Two possibilities exist for our findings in this patient. The first explanation is that the nasal retina was affected more than the temporal retina. Unfortunately, perimetry, even confrontational, could not be performed on this child. The second explanation for this asymmetry is that the temporal retinal fibres that usually pass to the ipsilateral visual cortex may cross anomalously at the chiasm and terminate in the contralateral visual cortex. Abnormalities in temporal retinal projections have been reported in albinos,⁵ Chédiak-Higashi syndrome,⁶ chiasmal lesions,⁷ congenital nystagmus,⁸ and in dissociated vertical deviation (DVD).⁹ The bilaterally asymmetric, photically evoked potentials after monocular illumination in human albinos suggest that there is a

FIGURE 3 Coronal T2-weighted MRI of the optic pathway. (a) Optic nerve of his left eye is thinner than that of the right. (b) No asymmetry can be seen in the chiasma. (c) The optic tract of the right (contralateral) side is thinner.

FIGURE 4 Fractional anisotropy (FA) map of a 3-year-old boy. FA of the right optic radiation shows a lower value than that of the left side. Arrows point to the thinner optic radiation on the right side (contralateral to the optic nerve hypoplasia).

disorganization of the nondecussated optic fibres similar to that reported for albino mammals. As yet there is no histological confirmation of abnormal retinostriate projections in humans, and there are no reports on asymmetry by MRI and MRDTI in human albinos. However, several investigators have reported that dysplasia of the ipsilateral optic tract and radiation was present in cases of optic nerve hypoplasia.^{10–13} These cases differ from our case because the optic tract dysplasia was present only on the contralateral side in our case.

Our patient also had a complete absence of the septum pellucidum, hypoplasia of the contralateral optic tract and radiation, and no schizencephaly. As best we know, there are no reports of cases with a unilateral reduction of the optic tract and radiations on the contralateral side in cases with optic nerve hypoplasia.

FIGURE 5 Diffusion tensor visualizer image of a 3-year-old boy. MRDTI shows a reduction of the optic radiation of the right side. Arrow points to the right optic radiation.

We suggest that there was a failure in the decussation pattern in our patient because of a midline prosencephalic dysgenesis at the optic chiasma. Thus, instead of 50% of the optic nerve fibres crossing at the chiasma, more than 50% or a complete breakdown and 100% of the optic nerve fibres crossed at the chiasma. Thus, all of the fibres from the dysplastic left optic nerve would have decussated to form the right optic tract and radiation. Similarly, all of the right optic nerve fibres would cross to form the left optic tract and radiation. This would then result in a dysplasia of the contralateral optic tract and radiation (Figure 6).

CONCLUSIONS

A 3-year-old patient presented with reduced vision in his left eye. MRI and MRDTI showed hypoplasia of

FIGURE 6 Diagram of our hypothesis. Septo-optic dysplasia (SOD) with unilateral optic tract dysplasia, all of the optic nerve fibres decussate to the contralateral optic tract at the chiasm. This would result in reduced volume of the contralateral optic tract and radiation.

the left optic nerve, whereas the optic chiasma was symmetrical. The contralateral optic radiation was reduced in size, and MRI and MRDTI were used to evaluate the optic pathways and to demonstrate the pre- and post-chiasmal abnormalities. Further investigations of this patient as he grows older should help us understand the aetiology of this complex disorder.

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

Note: Figures 1, 4, and 5 of this article are available in colour online at www.informahealthcare.com/oph.

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