Evidence for structural abnormality in the optic radiations in children with Optic Nerve Hypoplasia

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Introduction

Optic nerve hypoplasia (ONH) is a congenital abnormality characterised by an underdevelopment of the optic nerve and is the third most common cause of severe visual impairment in children [1, 2]. It may present as a unilateral or bilateral lesion in association with a variable degree of visual impairment. However, the etiology of ONH is still not understood. It has been described in association with absence of the septum pellucidum (Morsier Syndrome) [3]. ONH is a clinical diagnosis and conventional MRI alone does not diagnose all the suspected cases. This study examined whether diffusion tensor imaging (DTI) could identify abnormal development of the visual pathways in children with ONH.

Methods

Eleven children with ONH aged 2 to 11 and twenty two controls aged 2 to 11 years were enrolled in the study. Imaging data was acquired using a Siemens Avanto 1.5T clinical MRI scanner. Echo-planar diffusion-weighted images were acquired along 20 non-collinear gradient directions at \( b=1000 \text{ s mm}^{-2} \). This was repeated 3 times to improve signal to noise ratio. The voxel dimensions were 2.5 × 2.5 × 2.5 mm. Other acquisition settings: TR=6400 ms, TE=89 ms, gradient strength=40 mT m\(^{-1}\). T2- and T1-weighted images were also acquired in the same session. Analysis was performed using tract based spatial statistics (TBSS) (FSL 4.2) [4]. Diagnosis and ophthalmology examination was performed using clinical protocols at Great Ormond Street Hospital for Children. The study had local ethics committee approval and assent and consent was obtained from participants and parents.

Results

Eight patients had bilateral ONH, in two patients affecting the left optic nerve and in one patient affecting the right optic nerve. Most of the patients were referred to an ophthalmologist aged between 3 weeks and 12 months. The main presenting complaint was convergent squint in 60%, followed by nystagmus (40% of the cases). No abnormalities in the optic radiations were found on conventional T1- and T2-weighted images reported by consultant neuroradiologists. The TBSS analysis revealed that fractional anisotropy (FA) within the optic radiations was lower bilaterally in patients with ONH compared to controls (\( p < 0.05 \)) as shown in Figure 1.

![Figure 1 – TBSS analysis of FA in children with ONH. Red voxels correspond to those where FA is significantly lower in ONH patients compared to controls (\( p < 0.05 \), corrected).](#)

Conclusions

TBSS analysis provided evidence for structural abnormality in the optic radiations in paediatric patients with ONH compared to an age matched control group. This raises the possibility that DTI may be used to play a role in the diagnosis of ONH in children. Further research is now required to determine the relationship between the observed FA differences and physiological measures of visual function such as those obtained from visual evoked potentials.

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References