CASE REPORT

Peripapillary astrocytic hamartomas evolve from the optic nerve

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SUMMARY

Astrocytic hamartomas are rare benign tumours. Spectral domain optical coherence tomography (SD-OCT) with advanced visualisation (optic disc cube 200×200) was performed on a 27-year-old man with peripapillary astrocytic hamartomas in the right eye. En face (OCT scan) view (CT scan) of the optic nerve at the level of retinal pigment epithelium revealed optic nerve fibres along with three associated areas showing optical shadowing corresponding with astrocytic hamartomas in continuum. An area of optical shadowing was also observed superiorly in the retinal tissue corresponding with another astrocytic hamartoma. Astrocytes in diffusely vascularised rat retina have been reported to be immigrants from the optic nerve. En face SD-OCT-based in vivo histology for the first time provides strong evidence that peripapillary astrocytic hamartomas are derived from optic nerve astrocytes.

BACKGROUND

Astrocytic hamartomas are rare benign tumours and appear as white nodular masses with patchy calcification. Multifocal and bilateral lesions are usually associated with tuberous sclerosis and neurofibromatosis.^{1 2} However, solitary astrocytic hamartomas may be observed in normal individuals.³ Twenty-nine per cent of histologically proven astrocytic tumours of the retina have not been found to be associated with phacomatoses.² Tumour characteristics, using spectral domain optical coherence tomography, provide live in vivo histology of the retina and optic nerve, comparable to pathological features of the disease.

CASE PRESENTATION

A 27-year-old man presented for refraction. Best-corrected visual acuity was 6/12 OD and 6/9 OS. Slit lamp biomicroscopy of the anterior segment was unremarkable. Fundus examination of the left eye was unremarkable. The right eye examination revealed four juxtapapillary yellow-white glistening calcified lesions of various sizes, characteristic of astrocytic hamartomas (figure 1). A+B scan ultrasonography revealed multiple hyperechogenic lesions on the optic nerve head (figure 2). Autofluorescence imaging of the right optic nerve head showed increased autofluorescence of the nodular mass. Three-dimensional spectral domain optical coherence tomography (Carl Zeiss Meditec Inc, California, USA), optic disc cube 200×200 advanced visualisation was performed. En face view (CT-scan) of the optic nerve of the right eye, at the level of retinal pigment epithelium, revealed central retinal artery

and vein configuration (asterisk). Optic nerve fibres along with three associated areas corresponding with astrocytic hamartomas in continuum (arrowheads) showed optical shadowing. An area of optical shadowing was also observed superiorly in the retinal tissue (arrow) corresponding with another astrocytic hamartoma (figure 3). Comprehensive physical and dermatological examination did not reveal features of phacomatoses. Brain MRI, abdominal ultrasound and audiometry were unremarkable.

TREATMENT

In the absence of any known treatment, the patient was not subjected to any management.

DISCUSSION

Spectral domain optical coherence tomography of retinal astrocytic hamartoma shows characteristic features: a gradual transition from a normal retina into an optically hyper-reflective mass with retinal disorganisation, characteristic moth-eaten spaces and posterior shadowing.⁴ The retina in most mammals contains two types of macroglial cells: *Müller cells*, which span the entire thickness of the retina, and astrocytes, which are mainly confined

Figure 1 Fundus examination of the right eye shows three papillary (arrowheads) and one juxtapapillary (arrow) yellow-white glistening calcified lesions of various sizes, characteristic of astrocytic hamartoma.

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Figure 2 A+B scan ultrasonography shows multiple hyperechogenic lesions on the optic nerve head.

Figure 3 En face spectral domain optical coherence tomography of the optic nerve, at the level of retinal pigment epithelium, reveals central retinal artery and vein configuration (asterisk). Optic nerve fibres along with three associated papillary areas corresponding with retinal astrocytic hamartomas in continuum (arrowheads) show optical shadowing. An area of optical shadowing is also observed juxtapapillary in the retinal tissue (arrow) corresponding with another retinal astrocytic hamartoma.

to the nerve fibre layer.⁵ Astrocytes in diffusely vascularised rat retina have been found to be immigrants from the optic

nerve.⁶ Astrocytic hamartoma of the optic nerve and peripapillary retina was observed in the present case. On en face spectral domain optical coherence tomography, three areas corresponding with astrocytic hamartomas showed optical shadowing in continuation with the optic nerve. Similar optical shadowing was observed in a corresponding peripapillary area of the retinal tissue ascertaining these two lesions to be similar. These findings of astrocytic hamartoma of the optic nerve head and the peripapillary retina corroborate with an earlier rat retina model where it was demonstrated that retinal astrocytes immigrate from the optic nerve. Hence it can be concluded that peripapillary astrocytic hamartomas evolve from the astrocytes of the optic nerve.

En face spectral domain optical coherence tomography, for the first time, provides strong evidence that peripapillary astrocytic hamartomas are derived from the optic nerve astrocytes.

Learning points

- ▶ Astrocytic hamartomas are rare benign tumours usually associated with tuberous sclerosis and neurofibromatosis, however, solitary astrocytic hamartomas may be observed in normal individuals.
- ▶ On spectral domain optical coherence tomography, retinal astrocytic hamartoma shows characteristic features of moth-eaten spaces and posterior shadowing.
- ▶ En face spectral domain optical coherence tomography, for the first time, provides strong evidence that peripapillary astrocytic hamartomas are derived from optic nerve astrocytes.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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