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# Optical coherence tomography angiography of astrocytic hamartoma demonstrates intrinsic vascularity

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ABSTRACT

*Purpose:* To evaluate the findings of astrocytic hamartoma in the setting of gyrate atrophy, including details of optical coherence tomography angiography (OCTA).

*Observations:* Multimodal imaging was obtained on a 20-year-old woman with genetically-confirmed gyrate atrophy. Dilated fundus exam was performed, followed by ultra-widefield color and green autofluorescence imaging and OCTA of bilateral peripapillary and optic disc lesions. Clinical and imaging findings were consistent with gyrate atrophy. The bilateral peripapillary and optic disc lesions had a glistening, translucent, and mulberry-like appearance. OCTA imaging of these lesions clearly demonstrated an intrinsic vascular network and hyporeflective spaces within the lesion, which could not be seen on routine examination.

*Conclusions and importance:* OCTA was used to noninvasively diagnose astrocytic hamartoma in this patient with gyrate atrophy by showing the intrinsic vasculature and hyporeflective spaces of the lesion. This imaging modality can help differentiate astrocytic hamartoma from other lesions that typically lack intrinsic vascularity, such as optic disc drusen.

## 1. Introduction

Astrocytic hamartomas of the retina or optic disc are benign tumors that are typically seen in association with tuberous sclerosis but that have also been rarely reported in association with gyrate atrophy.[1,2] These tumors usually have yellow-gray coloration, intrinsic blood vessels, and appearances that vary from translucent and flat to calcified and mulberry-like. Optical coherence tomography angiography (OCTA) may be useful for evaluating astrocytic hamartoma because it non-invasively provides both structural and angiographic information. Here we describe a case wherein OCTA imaging was used to identify a vascular network within fundus lesions, thereby confirming the diagnosis of astrocytic hamartoma, rather than optic disc drusen, in the setting of gyrate atrophy.

## 2. Case report

A 20-year-old woman with a diagnosis of gyrate atrophy was referred for evaluation of bilateral fundus lesions. She had experienced nyctalopia and peripheral visual field constriction since childhood. Her systemic diagnosis had been confirmed by multiple ancillary tests, including markedly elevated plasma ornithine at 732 μmol/L (normal reference 30.5–131.4 μmol/L), fibroblast enzyme studies, and genetic analysis showing a homozygous splice site mutation in *OAT1*. Goldmann perimetry showed constricted isopters bilaterally. Full-field electroretinography was consistent with severe generalized rod and cone dysfunction in both eyes.

Visual acuity measured 20/32 in both eyes. She had mild anterior subcapsular cataracts bilaterally. Posterior segment examination showed bilateral cystoid macular edema, retinal vascular attenuation, and widespread well-demarcated areas of peripheral chorioretinal atrophy. There were glistening, translucent, mulberry-like pre-retinal lesions in the peripapillary region of the right eye and over the optic disc of the left eye. These lesions exhibited hyperautofluorescence (Fig. 1) but did not appear to contain intrinsic blood vessels on biomicroscopy. However, OCTA imaging demonstrated obvious blood vessels, as well as hyporeflective spaces within the lesions (Fig. 2).

Because of the definitive presence of intrinsic blood vessels, together with the typical ophthalmoscopic and imaging features, the patient was diagnosed with bilateral astrocytic hamartomas, for which observation was recommended. She continued follow-up with her clinical geneticist, who recommended an arginine-restricted diet until more definitive

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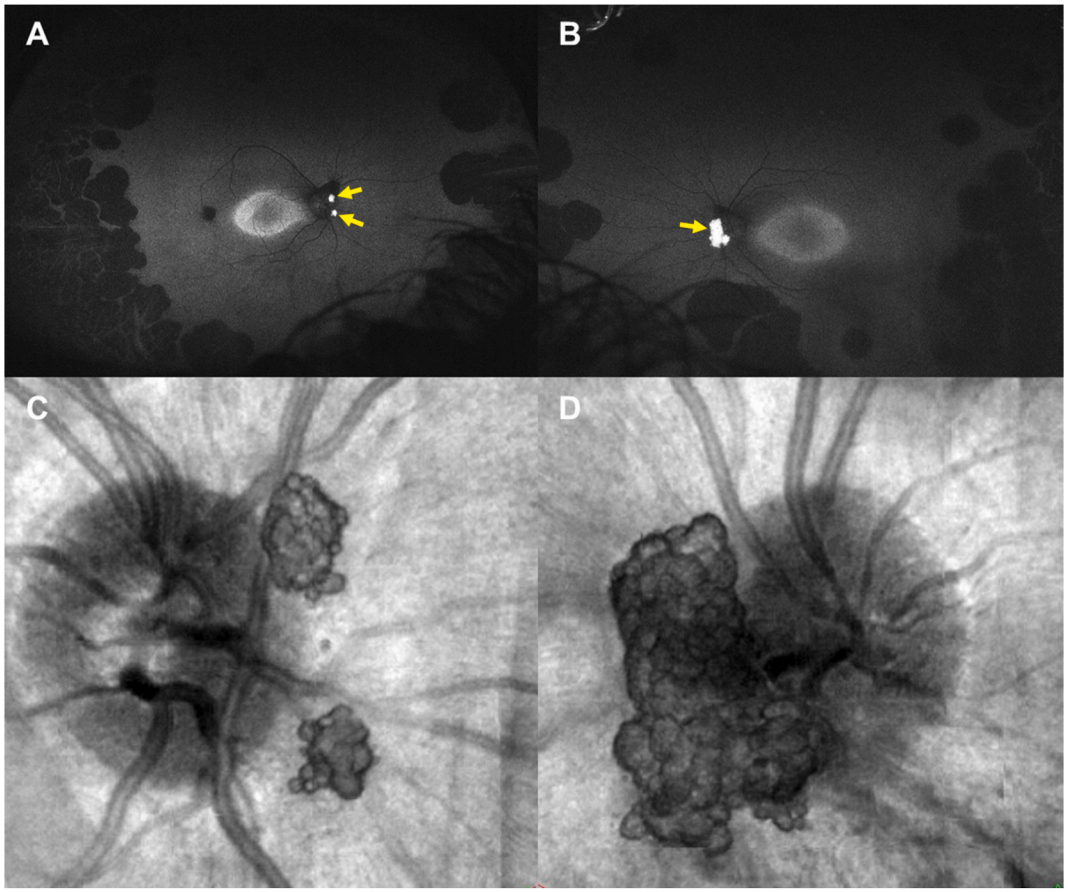


**Fig. 1.** Green autofluorescence imaging (200Tx, Optos, Inc., Marlborough, USA) of the right (A) and left (B) eye showing areas of hyperautofluorescence in the macula and hypoautofluorescence in the periphery, with the latter corresponding to areas of chorioretinal atrophy seen on fundus examination. The hyperautofluorescent peripapillary lesions (arrows) have a mulberry-like appearance that is best demonstrated on confocal scanning laser ophthalmoscopy (Avanti RTVue XR, Optovue, Inc., Fremont, USA) of the right (C) and left (D) optic disc. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

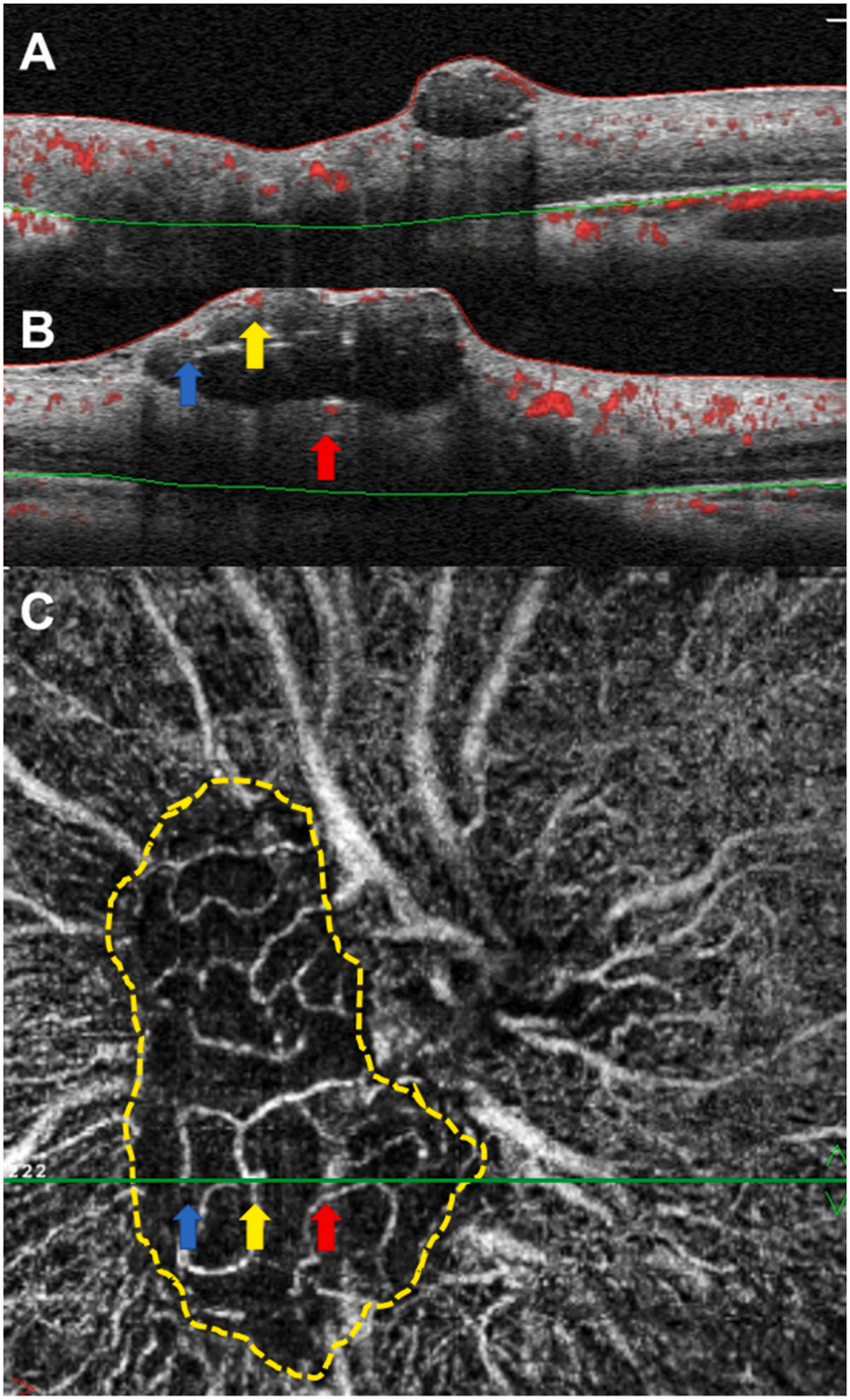


**Fig. 2.** Optical coherence tomography angiography (OCTA) (AngioVue, Avanti RTVue XR, Optovue, Inc., Fremont, USA) line scans through the astrocytic hamartomas at the right (A) and left (B) peripapillary retina showing flow (red) in the masses. En face OCTA image of the left eye (C) shows a distinct vascular network within the lesion (yellow dashed outline). The en face image was acquired using the green and red segmentation lines seen in the line scan (B). Colored arrows (blue, yellow and red) show different loci of flow that are seen in both the line scan and en face image. (For interpretation of the references to color in this figure legend, the reader is referred to the Web version of this article.)

treatment for gyrate atrophy becomes available.

## 3. Discussion

Astrocytic hamartoma in patients with gyrate atrophy has only rarely been described.[1,2] The far more common fundus lesions found in gyrate atrophy are optic disc drusen, which were seen in 14% of patients with gyrate atrophy in one series.[3] In some cases, the appearance of exposed calcified optic disc drusen can mimic that of calcified mulberry-like astrocytic hamartomas. Both types of lesions can exhibit hyperautofluorescence and staining on fluorescein angiography. Previous reports of structural OCT imaging have shown hyporeflective spaces with hyperreflective surfaces in both astrocytic hamartomas[4] and optic disc drusen.[5]

OCTA can rapidly and non-invasively detect both hyporeflective spaces and the intrinsic vascularization in astrocytic hamartoma.[6] This

imaging modality helped confirm the diagnosis of astrocytic hamartoma, rather than optic disc drusen, in the setting of gyrate atrophy. Future studies with OCTA with larger sample sizes may provide insight into the true incidence of astrocytic hamartoma in gyrate atrophy.

## Patient consent

Written consent to publish this case has not been obtained. This report does not contain any personal identifying information.

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## Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

## Declaration of competing interest

The following authors have no financial disclosures: RV, TL, MS, MG.

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