curative. Histopathological examination of all lesions showed upper dermis–proliferating fibroblasts and leukocytes within mucinous background that was positive with Alcian blue stain and negative with periodic acid–Schiff stain. The fibroblastic cells expressed the S-100 protein.[7]

The second case of eyelid focal mucinosis was reported in 2007, when a 19-year-old Hispanic female presented with asymptomatic multiple pink papules on the face involving the right cheek, right temple, left lower eyelid, and neck area. The lesions appeared several years before her presentation and remained stable in size over the following years. On examination, the lesions were well-demarcated, slightly raised pink papules and plaques with no scales. Punch biopsy was performed and showed superficial perivascular dermatitis with a slight increase in the number of fibroblasts. Colloidal iron staining was diffusely positive with replacement of collagen by mucinous material in the dermal layer.[4]

Our patient was older than the previously reported eyelid cases but with similar histopathological findings. Our case was also unique because the eyelid lesion mimicked a nevus. After an extensive review of the literature, we found a trunk lesion that mimicked a nevus clinically and was similar to our case histopathologically; the authors there proposed the term "follicular mucinous nevus" as a final diagnosis.[8] A recent review by Arora et al.[9] has described connective tissue nevi (CTN) hamartomas of the dermis involving the presence of 3 main components in these lesions: collagen, elastin, and proteoglycans (either singly or in combination). CTN are considered benign and can be either isolated or represent a manifestation of a syndrome. Our lesion fits in their classified acquired proteoglycan CTN group, which includes focal cutaneous mucinosis; however, the authors did not present histopathological photographs in their review.[9]

Management of these lesions includes surgical excision, laser treatment, dermabrasion, and intralesional steroids.[9] The lesion in our patient was surgically excised with no evidence of recurrence after a follow-up period of 3 years.

Focal mucinosis is a rare entity that has been recognized recently as one of the entities in the group of skin hamartomas named "connective tissue nevi." These lesions have different forms of presentation, which makes diagnosis based on clinical examination alone challenging. They also can occur as isolated lesions or in association with syndromic conditions and thus require full evaluation of patients and monitoring of the lesions. We aim to bring the attention of ophthalmologists to this rare entity. We also recommend tissue diagnosis by experienced pathologists to reach the correct diagnosis and manage patients accordingly.

Saleh Hamad Alrashed, [*] **Hind Manaa Alkatan,** [†] **Adel Ayman Helmi,** [‡,§] **Azza M.Y. Maktabi** [¶]

[*]King Saud University, Riyadh, Saudi Arabia; [†]King Saud University Medical City, Riyadh, Saudi Arabia; [‡]Dalhousie University, Halifax, N.S; [§]King Fahad Medical City, Riyadh, Saudi Arabia; [¶]King Khaled Eye Specialist Hospital, Riyadh, Saudi Arabia.

Correspondence to:
Hind Manaa Alkatan, MD: hkatan@ksu.edu.sa

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Optic nerve giant cell astrocytoma in tuberous sclerosis complex

The retinal astrocytic hamartoma is a hallmark ophthalmic feature of tuberous sclerosis complex (TSC).[1] The typical course of such tumours is a relatively slow-growing nonaggressive lesion in the retina that can be managed expectantly.[2] Nonetheless, there have been several reports in the literature of aggressive astrocytic hamartomas that do not demonstrate this benign course.[3] We present here a rare case of an aggressive tumour in a patient with TSC that has cell types more typical of the subependymal giant cell astrocytoma (SEGA) that is seen in the brains of such patients.

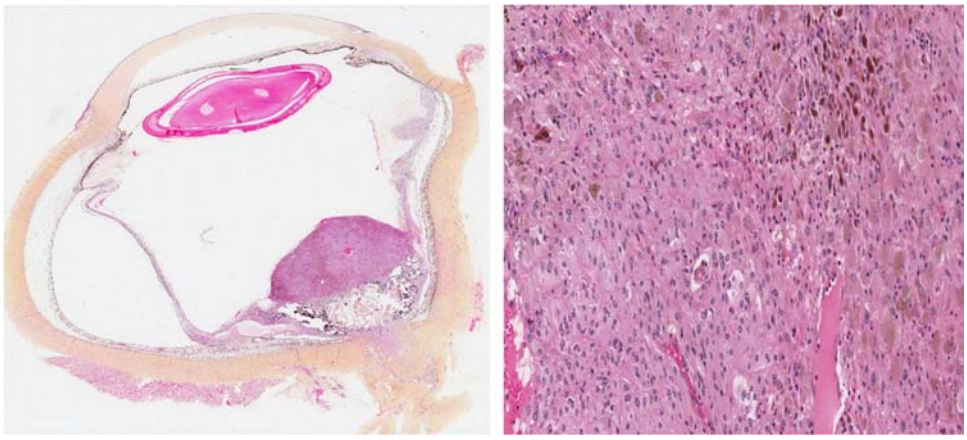

Fig. 1—*Left:* Low-power scanning view of tumour arising from optic nerve head and subtotal retinal detachment. *Right:* The tumour is composed of large-sized astrocytes with focal accumulation of hemosiderin (brown pigment) (hematoxylin, phloxine and saffron (HPS)).

J.M. was a patient with known TSC who was first seen in our pediatric ophthalmology clinic at the age of 5 years, at which point he was diagnosed with a small right optic nerve head hamartoma not affecting vision. Over the years, he was subsequently followed at multiple centres, and records indicate that he developed a chronic right retinal detachment. An examination under anaesthesia (EUA) at the age of 13 years revealed a disorganized retina and a large astrocytic tumour emanating from the right optic nerve head with underlying exudative retinal detachment, all of which appeared chronic in nature and was confirmed on B-scan ultrasound. The left retinal examination was unremarkable.

At the age of 14 years, the patient was reassessed in our clinic for new-onset redness and photophobia and discomfort of the right eye. EUA revealed an intraocular pressure of 48 mm Hg in the right eye. Anterior segment examination revealed frank neovascularization of the iris, a shallow anterior chamber, and a secluded pupil, consistent with neovascular glaucoma. Transscleral diode cyclophotocoagulation was performed, and intravitreal bevacizumab (Avastin (R), Genentech, California, 0.1 mL of 25 mg/mL).

Two weeks later, the patient's pain had somewhat subsided, and the iris vessels had partially regressed. One month later, however, the right eye was still causing discomfort despite maximal intraocular pressure–lowering drops and difluprednate eye drops. Given that this was a blind and painful eye, the family decided to proceed with enucleation.

Pathologic evaluation of the enucleated eye revealed the following findings:

1. Optic nerve head giant cell astrocytoma: The tumour emanated directly from the optic nerve head and demonstrated osseous metaplasia, calcification, large-sized astrocytes, and significant hemorrhage and was associated with a subtotal retinal detachment (Fig. 1, top). The tumour can be seen extending into the substance of the optic nerve at the level of the lamina cribrosa (Fig. 2, top). A central area of dystrophic calcification can be seen in the centre of the optic nerve substance, distal to the globe (Fig. 2, middle).
2. Optic nerve atrophy: There was significant atrophy of the optic nerve itself with loss of myelin and axons. A neurofilament immunostain highlights the atrophy of axons (Fig. 2, bottom).
3. Retinal astrocytic hamartoma: In comparison to the optic nerve head astrocytoma, this tumour showed a proliferation of smaller glial cells and less vasculature.
4. Combined retinal pigment epithelial and vascular hamartoma: This lesion consisted of large blood vessels, a disorganized retinal pigment epithelial proliferation, and a component of glial tissue.

A case series by Shields et al. in 2004 reported 4 cases of atypically aggressive astrocytic hamartomas in patients with TSC.[2] The tumour growth was extensive in all 4 patients, and all cases resulted in enucleation as a result of total retinal detachment and neovascular glaucoma. Histopathologically, the tumours consisted of cell types that are usually seen in the SEGA tumour of tuberous sclerosis rather than the typical cell types more commonly seen in the classic retinal astrocytic hamartoma of TSC.

The optic nerve head giant cell astrocytoma observed in this case also has strikingly similar pathology to the SEGA seen in the brains of patients with tuberous sclerosis. The astrocytes are larger, show glassy eosinophilic cytoplasm, and have a bland spindle-shaped and gemistocytic appearance. The substantial blood supply; dystrophic calcification; positive histochemistry stains for iron; and positive

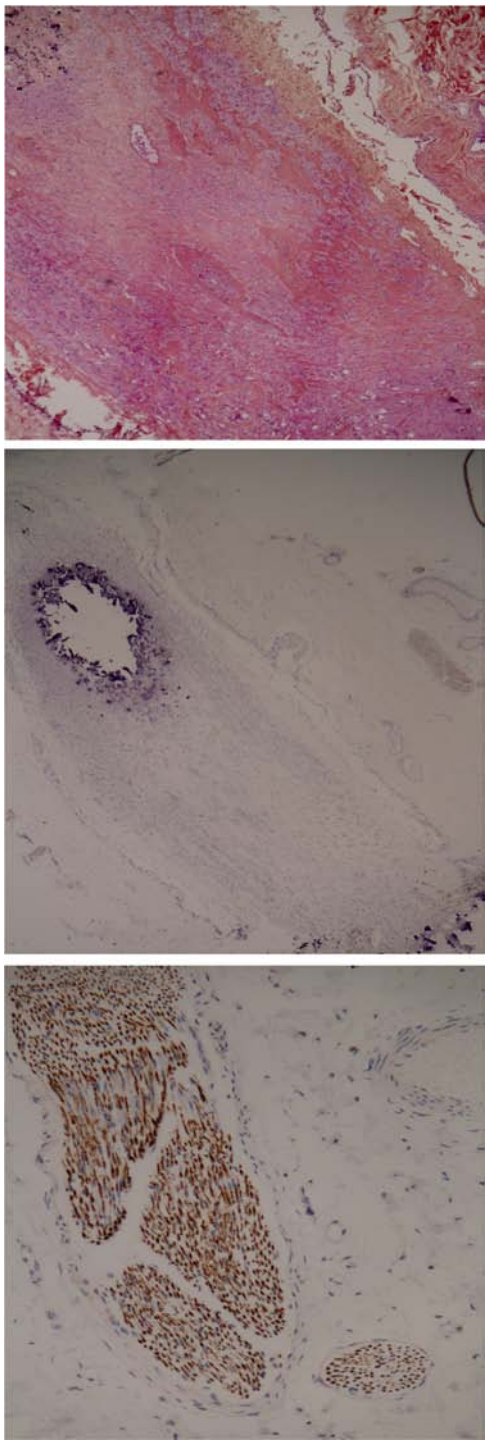

Fig. 2—*Top:* Tumour shown extending into optic nerve at level of lamina cribrosa (HPS). *Middle:* Von Kossa stain for calcium shows calcification of optic nerve distal to globe. *Bottom:* Neurofilament immunostain indicates axonal diminution in the optic nerve distal to globe. HPS = hematoxylin, phloxine and saffron.

immunohistochemistry stains for NSE, GFAP, and S100 provide further evidence for a SEGA as opposed to a retinal astrocytic hamartoma.

These findings are consistent with tumours outlined in several other reports regarding astrocytic hamartomas,[3–5] which described abundant necrosis and large cells with abundant eosinophilic cytoplasm, nuclear pleomorphism, and mitotic activity.

The present case adds to the emerging evidence that an aggressive form of an astrocytic hamartoma, more accurately defined as a giant cell retinal astrocytoma, is a rare but possible feature in TSC. In contrast to prior reports that cite peripapillary giant cell astrocytomas that have subsequently invaded the optic nerve, the tumour in this case actually originated from the optic nerve head itself and resulted in complete optic nerve atrophy. It remains to be determined whether there is a correlation between the presence of SEGAs and concomitant giant cell retinal astrocytomas and whether the histopathologic features of these giant cell retinal astrocytomas represent a distinct and more aggressive subtype on the TSC disease spectrum. The pathologic characteristics of giant cell astrocytomas may be significant in terms of treatment implications, as new therapies emerge to treat certain tumour subtypes.[6]

Zale Mednick,[*] James Farmer,[*,†] Gary Yau,[*] Vladimir Kratky,[*] Yi Ning J. Strube[*]

[*]Queen's University, Kingston, Ont; †Ottawa General Hospital, Ottawa, Ont.

Correspondence to:
Yi Ning J. Strube, MD: yining.strube@queensu.ca

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