Abstract: Retinal astrocytic hamartoma is a nonmalignant tumor, which may present as an isolated finding or in association with tuberous sclerosis or neurofibromatosis. This report presents clinical findings of astrocytic hamartomas and their related systemic diseases.

I. Case History
   A) Patient demographics: 56 year old Caucasian female
   B) Chief complaint: Patient presents with a chief complaint of blur at near, gradual in onset, OU. Patient states she was previously diagnosed with a tumor in the right eye in 2003.
   C) Ocular history: choroidal osteoma vs. melanoma per records in 2003
   D) Medical history: hyperlipidemia, tobacco use disorder, palpitations
   E) Current medications: Aspirin 81mg chew tab, calcium carbonate 650mg tab, magnesium oxide 400mg tab, Pyridoxine HCl 50mg tab

II. Pertinent findings
   A) Clinical
      i. Best corrected VA OD 20/40, OS 20/20, OU 20/20; pupils equal, round and reactive with no APD. Extraocular motilities full and smooth OU and confrontation visual fields are full. Anterior segment unremarkable with Intraocular pressures 21mmHg OU.
      ii. Dilated fundus exam unremarkable OS. Dilated fundus exam OD revealed an elevated intraretinal mass superior temporal to the optic nerve head extending along the arcade just superior to macula. The elevation had a mulberry-like appearance with glistening yellow spherules representing calcifications. The contiguous RPE exhibited disorganized atrophy and normal choroidal vascular anatomy was noted around the mass.

   B) Clinical tests
      i. B-scan reveals a highly reflective area centered superior and near optic nerve head with no apparent mass underlying the superficial lesion. Lesion on B-scan demonstrates retinal location with no choroidal involvement.
      ii. Fast macular OCT- Right eye had an atrophic appearance with a center macular thickness of 114 microns, signal strength 8. Left eye is unremarkable with a center macular thickness of 173 microns and signal strength of 6.
      iii. Raster Line OCT performed only on right eye reveals cystic spaces throughout the retinal mass. The lesion is contained within the retina showing variable signal hyperintensities and confirming elevation. The scan revealed a generalized thickened sensory retina within the tumor location.
      iii. Fluorescein angiogram shows leakage in late views indicating vascularization of the mass. There is no indication of invasive choroidal vascularization.
III. Differential Diagnosis
A) Primary/leading: Astrocytic hamartoma
B) Others: choroidal melanoma, choroidal osteoma, amelanotic choroidal nevus
   i. B-scan showed no hyper-reflectivity deeper than retinal tissue
   ii. Clinical testing rules out choroidal involvement.

IV. Diagnosis and discussion
A) Astrocytic hamartomas: are non-metastasizing retinal tumors, which may be isolate, multiple, unilateral, or bilateral. These congenital tumors may present variable ophthalmoscopic characteristics, but may be generalized into two subsets: type 1 and type 2 (Mennel, S). Type 1 is usually identified as a solitary lesion with an average size of 0.5DD. The tumor is flat with a soft appearance in the retinal nerve fiber layer without signs of calcification. The non-calcified variant is often attributed to younger age and typically described as a translucent, murky-white or yellow lesion, oval in shape and relatively smooth in contour. Larger lesions of this variety may also have a gray-yellow hue and cause traction. Type 2, also known as the calcified variant, is most often described as a yellow, multi-lobulated lesion with glistening spheres creating a mulberry appearance. These tumors are typically located in the posterior pole and have been attributed to an aged version of the smooth-type lesion. The extent of calcification can be variable, with some lesions showing both calcified and non-calcified components (Shields, Carol L.).
Clinical data obtained in this case confirmed a type 2 astrocytic hamartoma through ophthalmoscopic retinal characteristics, B-scan ultrasound, OCT, and fluorescein angiogram pattern. The retinal tumor had the characteristic mulberry appearance with calcification nodules. B-scan ultrasound revealed the highly reflective nature of the calcified lesion with OCT further confirming the tumors superficial location and lack of extension into the choroid. Vasculature description of astrocytic hamartomas can vary, but are typically characterized by late leakage by fluorescein pattern, as was the case in this study (Friedman, Neil J.).
B) Conditions related to Astrocytic Hamartoma:
   i. Tuberous sclerosis (TS): A genetic pathology that causes benign tumors to grow throughout the body. The condition may be characterized by a triad of epilepsy, mental retardation, and adenoma sebaceum. The tumors can grow in many vital organs, including the brain, heart, eyes, kidneys, lungs and skin (Kanski, Jack J.). TS is caused by a genetic mutation on the genes TSC1 and TSC2. When functioning normally, these genes account for tumor growth suppression (Ehninger, D)
   ii. Neurofibromatosis (NF): A genetic condition in which, tumors arise from neural tissue. These tumors may be of no harm or have the potential to cause serious compressive damage. All neural crest cells may be affected, including, melanocytes, astrocytes, Schwann cells, and fibroblasts. The proliferation of these cells may present as abnormal skin pigmentation, nodules throughout the skin, skeletal issues, or neurological issues due to compression on the spine (Kanski, Jack J.).
V. Treatment, management

A) Astrocytic hamartoma
   i. Observation with yearly exams
   ii. Documentation with fundus photos
   iii. Rare complications
       a) Serous detachment—most commonly encountered with type 1 lesions
           i. spontaneous resolution may occur within 4 weeks, no treatment prior to this time frame
           ii. Argon laser photocoagulation may be performed, although treatment should be limited due to laser-induced choroidal neovascularization (Vrabec, TR)
           iii. Anti VEGF therapy (Mennel, S.)
       b) Macular detachment with retinoschisis due to vitreomacular traction
           i. pars plana vitrectomy with epiretinal membrane peel
           ii. glial tissue trim (Inoue, Makoto)
       c) Vitreous hemorrhage
           i. spontaneous resolution may occur
           ii. pars plana vitrectomy: longstanding or recurrent (Mennel, S.)

B) Tuberous sclerosis
   i. Cutaneous treatment: Dermabrasion
   ii. Neurological treatment
       a) Anti-epileptic medication
           i. Narrow spectrum
           ii. Broad spectrum
       b) Rapamycin study on reversal of learning deficits (Ehninger, D)

C) Neurofibromatosis
   i. Type 1
       a) Cosmetic surgery
       b) Chemotherapy for malignant peripheral nerve sheath tumors
       c) Promising results in medical targets against Ras pathway
       d) Surgical resection for compressive tumors
       e) Orthopedic support for scoliosis (Pletcher, Beth A.)
   ii. Type 2
       a) Stereotactic radiosurgery for vestibular schwannoma
       b) Surgical resection
       c) Early medical treatment studies for treatment of unresectable, progressive vestibular schwannomas
           i. Gleevac, tyrosine kinase inhibitor
           ii. Bevacizumab, an antivascular endothelial growth factor (Plotkin, SR, AO Stemmer-Rachamimov)
           iii. Erlotinib trials (Plotkin, SR, C Halpin)
VI. Conclusion
A) Retinal astrocytic hamartomas can be identified by characteristic clinical features
B) These retinal tumors are non-metastasizing and often observed as a stable condition
C) Vision threatening complications are rare. Initial monitoring would be recommended considering spontaneous resolution has been reported to occur within 4 weeks. Associated serous detachments or residual macular edema persisting greater than six weeks should be treated. Treatment would include argon laser photoagulation and anti-VEGF for exudative changes and macular swelling. Vitrectomy is indicated for longstanding vitreous hemorrhages and cases with vitreoretinal traction.
D) Clinical pearl: astrocytic hamartoma may be associated with two systemic conditions, tuberous sclerosis and neurofibromatosis. Appropriate referral may be warranted for their evaluation, especially in young or symptomatic patients.

VII. Bibliography


