TUBEROUS SCLEROSIS - TYPE III ASTROCYSTIC HAMARTOMA OF RETINA: CASE REPORT

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PURPOSE

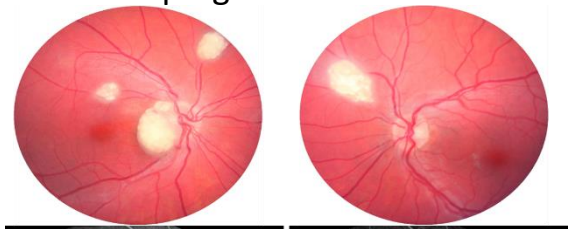
Report a case of type III astrocystic hamartoma secondary to tuberous sclerosis in a patient with epilepsy and without ophthalmologic complaints.

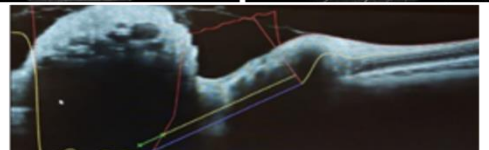
METHODS

Information was obtained through review of medical records and literature review.

RESULTS

NLT, female, 20 years old, with tuberous sclerosis and epilepsy. Visual acuity is 1.0. Fundoscopy: circumscribed lesions with a yellowish appearance and raised in peripapillary region, affecting the papillomacular bundle, in superior temporal and nasal arcades OR and in superior nasal arcade OS suggestive of astrocystic hamartomas. OCT: raised lesion with optically empty internal spaces and presence of intratumoral hyperreflective dots (lesion type III). AGF:circumscribed lesions with early hyperfluorescence and late impregnation.





DISCUSSION

Tuberous sclerosis or Pringle-Bourneville phakomatosis is an autosomal dominant syndrome and courses with systemic hamartomas. Prevalence is 1/6000 people and 60% of cases are sporadic. Astrocystic hamartomas are the main ocular manifestations, they emerge at the level of the retinal nerve fiber layer or around the optic nerve, and may manifest as yellow-gray, sessile or raised lesions. Retinal astrocytomas occur in approximately 50% of patients and 50% have bilateral retinal involvement. Hamartoma is classified by OCT into types I, II, III and IV, and there is a higher prevalence of skin alterations in type II, brain disorders in type III and pulmonary diseases in type IV.