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Letter to Editor

Multimodal Imaging of Retinal Astrocytic Hamartoma Undergoing Bourneville's Disease

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Abstract

Bourneville's disease also called tuberous sclerosis complex, is an uncommon genetic disorder that causes benign tumors. It is a rare multisystem autosomal dominant genetic disease growing in the brain and on other vital organs such as the kidneys, heart, liver, eyes, lungs, and skin Signs and symptoms vary widely, depending on where the growths develop and how severely a person is affected.

This disorder is usually identified in infants and children based on characteristic skin lesions, seizures, and cellular overgrowth or hamartomas in the heart, brain, and kidneys. Tuberous sclerosis complex is a genetic disorder caused by a mutation in either the TSC1 or TSC2 gene leading to dysfunction of hamartin or tuberin, respectively [1]. Although there is no cure for tuberous sclerosis, and the course or severity of the disorder can't be predicted, treatments are available to manage symptoms.

Keywords: *Multimodal Imaging; Retinal Astrocytic Hamartoma; Bourneville's Disease*

Tuberous sclerosis complex (TSC) is a genetic disorder characterized by the growth of tumors in many organs; most often in the brain, heart, kidneys, and skin [1]. It was first described in 1880 by D.M. Bourneville. The diagnosis is based on revised clinical criteria approved at a consensus conference in 1998.TSC is an autosomal dominant inherited syndrome. Mutations in tumor suppressor genes on chromosomes 9(TSC1:) or 16(TSC2:) give rise to different phenotypic expressions. The common denominator is a tendency towards increased cell growth due to the inactivation of the gene products hamartin (chromosome 9) or tuberin (chromosome 16) [2].

Retinal astrocytic hamartoma (RAH)is a benign retinal tumor that is composed of glial cells, predominantly astrocytes [3].

The evaluation of retinal manifestations of RAH includes the clinical exam and fundus fluorescein angiography (FFA), Indocyanine green angiography (ICGA), Spectral-Domain optical coherence tomography (SD-OCT) and fundus photography. However, the findings of retinal lesions can be equivocal, deciding for treatment challenging.

Multimodal imaging is useful for the diagnosis and monitoring of RAH. We present a case of a 35-year-old male, who has referred to us Department of Ophthalmology. Pitié-Salpêtrière Hospital, Sorbonne University. Paris, France) with a diagnosis of the intraocular retinal tumor, which was detected on a routine eye examination. The patient was asymptomatic. There was, however, a history of recurrent

epilepsy in childhood, stop at 10 years old and completely resolved after treatment with antiepileptics. He presented claustrophobia and social phobia. On examination, his best-corrected visual acuity was 20/20, P2. The anterior segment in both eyes was within normal limits. We used the SPECTRALIS Heidelberg (Heidelberg-Engineering, Germany) widefield imaging module which provides a 55-degree field of view for all fundus imaging including Multicolor, IR, FFA, ICGA and SD-OCT. Capturing areas beyond the vessel arcades in a single image. Fundus photography in the left eye showed a 4 × 1.7 × 1 mm retinal lesion, about 6-disc diameters from the optic disc along the supratemporal arcade at the peripheral of the retina. The lesion had a glistening yellowish spherule of calcification that was surrounded circumferentially by a grayish transparent sessile and fairly flat (Figure A). FFA showed a well-defined hyperfluorescent lesion, filling up with a strong diffusion of fluorescein in the late phase (Figure B). ICGA showed a hypo fluorescent lesion on the earlier and late time (Figure C). SD-OCT revealed an inhomogeneous mass arising from the retinal nerve fiber layer with overlying vitreous adhesions, hyperreflective dots, and optically empty spaces at all depths of the tumor (Figure D). Macula OCT was normal (Figure D1). The left eye fundus was normal. Facial examination revealed the presence of subtle but characteristic cutaneous angiofibromas or adenoma sebaceous (Figure E). Hypomelanotic macules of the skin were also observed. They are caused by a complex process, involving the synthesis and maturation of melanosomes [4] (Figure F). However, there were no, shagreen patches, or periungual fibromas. Cerebral MRI revealed subependymal nodule, subependymal giant cell astrocytoma. Renal ultrasound showed multiple angiomyolipoma's (echogenic masses with shadow, max 8.5 cm). Rectal polyps were also found by colonoscopy (Figure G). Annual monitoring with SD-OCT and fundus photography has been decided.

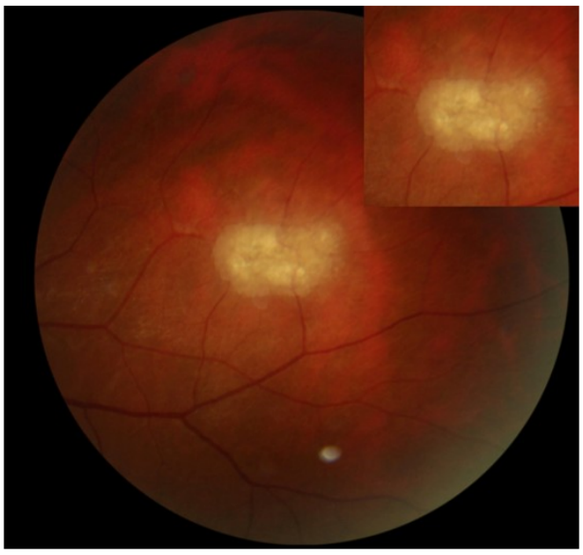

Figure A: *The photograph of Fundus in the left eye showed a retinal lesion of 4 mm in diameter, located about 6 disk diameters of the optical disk along the supratemporal arcade at the peripheral of the retina. The lesion had a sparkling yellowish, spherule of calcification.*

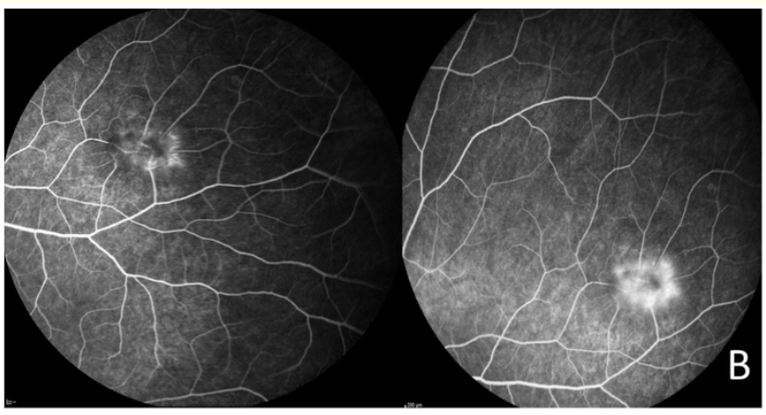

Figure B: *FFA of RHA showed a hyperfluorescent lesion with an increase of the staining in the late phase.*

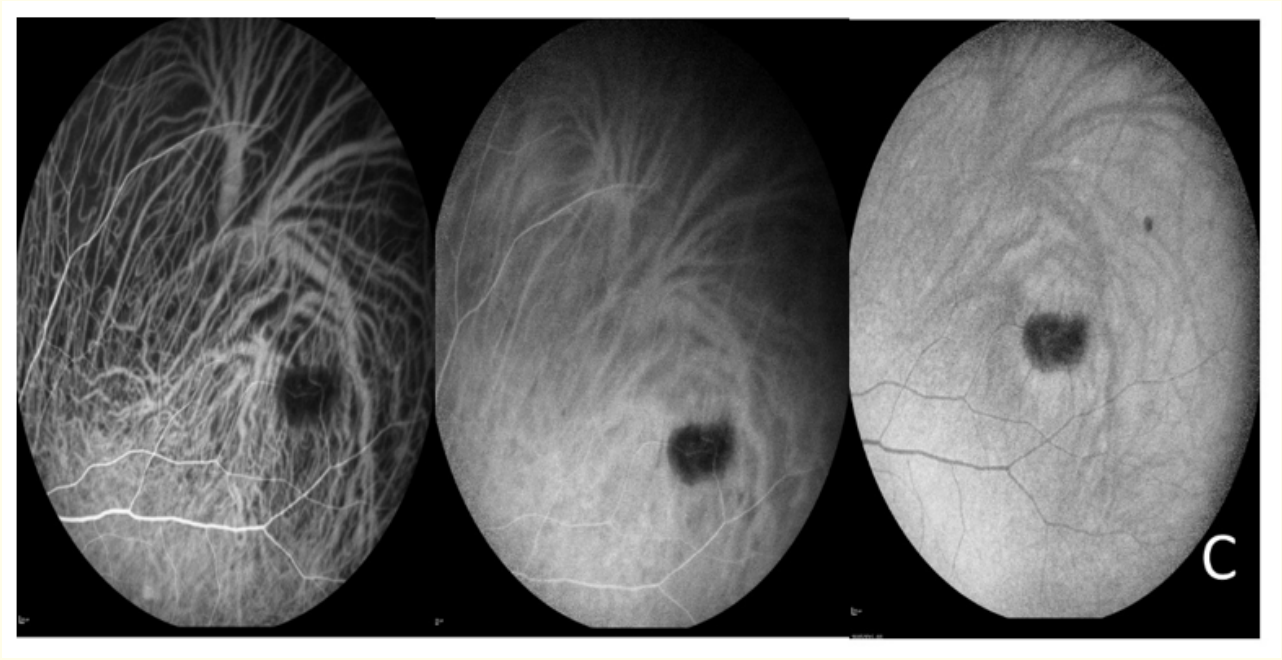

Figure C: *ICGA: A hypo fluorescent lesion in the earlier and late phase.*

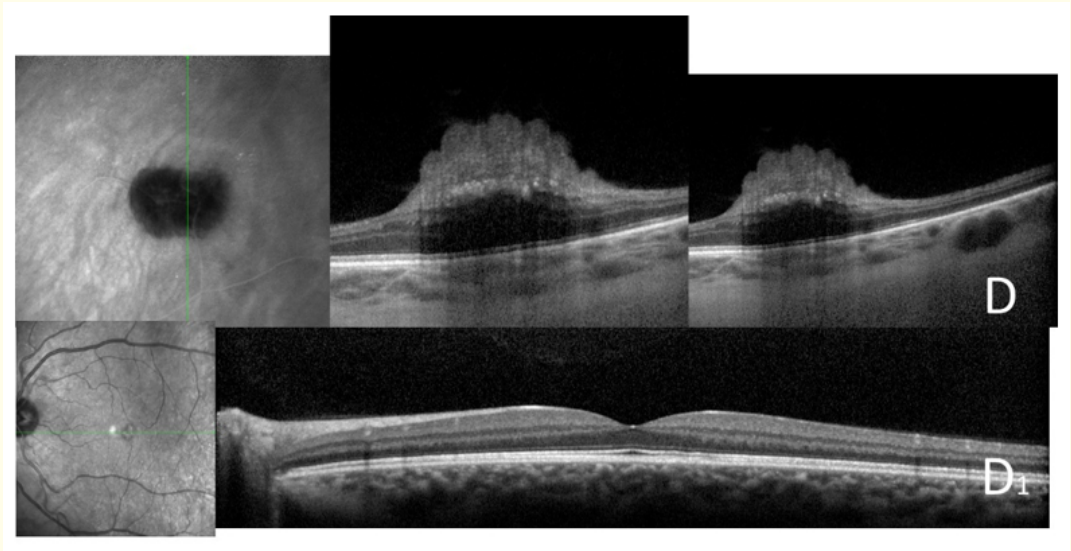

Figure D and D1: *D: SD-OCT showed an inhomogeneous lesion arising from the retinal nerve fiber layer with optically empty spaces at all depths of the tumor and hyperreflective dots. D1: Normal SD-OCT on the macula area.*

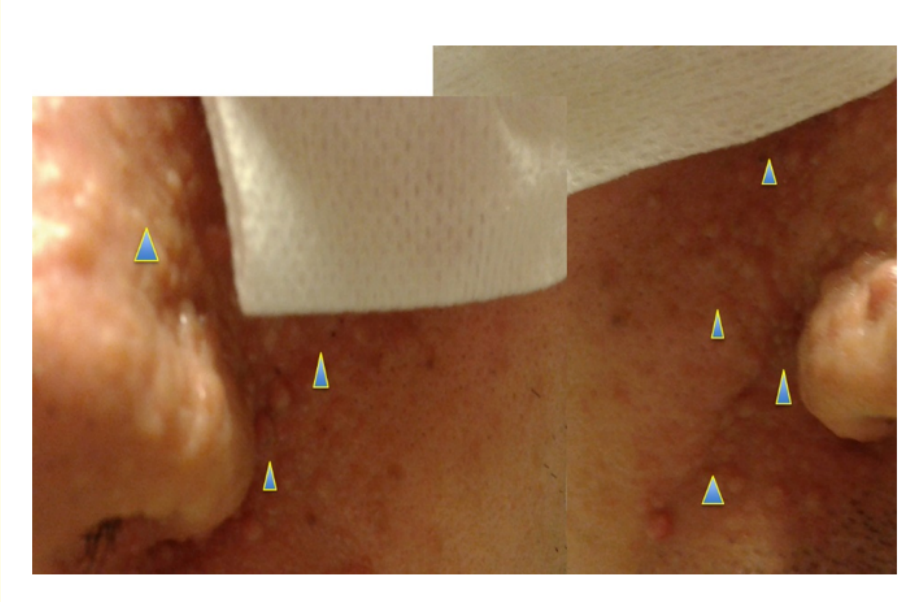

Figure E: *Skin abnormalities, in the form of acne-like facial growth present on the nose and the cheeks. Cutaneous Angiofibroma (Arrowhead).*

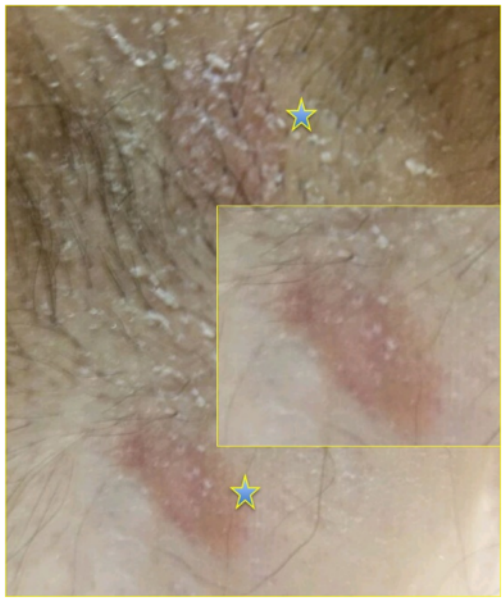

Figure F: *Hypomelanotic macules of the skin, inside the armpits (Star).*

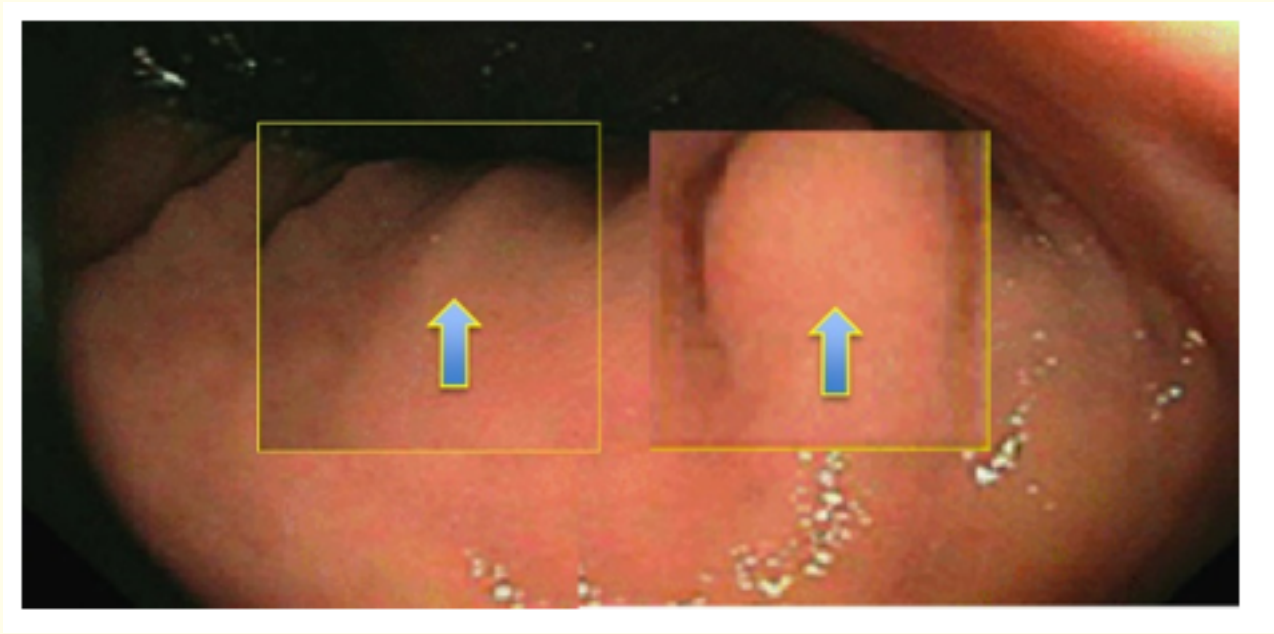

Figure G: *Multiples rectal polyps (blue arrow).*

Discussion

Tuberous sclerosis complex (TSC) is a rare multisystem autosomal dominant disease. Prognosis is highly variable and depends on the symptoms, but life expectancy is normal for many [5]. The prevalence of the disease is estimated to be 7 to 12 in 100,000 [6]. The disease is often abbreviated to tuberous sclerosis, which refers to the hard swellings in the brains of patients, first described by French neurologist Désiré-Magloire Bourneville in 1880 [7]. Symptoms can range from mild to severe, depending on the size or location of the overgrowth. Vision can be damaging if RAH impairment is located on the macula, this is uncommon. Thus, this letter to the editor aimed to show the use of retinal multimodal imaging in a patient with TSC. To identify the clinical and imaging characteristics of retinal astrocytic hamartomas, (RAH). A possible evolution and complications of the tumor such as Serous retinal detachment, vitreous retinal hemorrhage. Recently, Stacey AW., *et al.* [8] identified changed in Fundus autofluorescence in all eight cases. SD-OCT was able to document the tumor thickness and level of retinal invasion in all cases after a median follow-up of 59 months [9]. SD-OCT and FAF are sensitivity tools for identifying and can be used to follow the thickness and margins of these lesions. We have observed a RAH with systemic history. Although the signs and symptoms are unique for each person with tuberous sclerosis, they can include Skin abnormalities, Cognitive disabilities, Behavioral problems, Kidney problems, Heart issues, Lung problems, and Eye abnormalities. At the recent consensus conference on complex tuberous sclerosis, the clinical diagnostic criteria for the tuberous sclerosis complex were simplified and revised. The clinical and radiographic characteristics of the tuberous sclerosis complex have now been divided into major and minor categories. Traditionally, a clinical diagnosis of TSC is made by identifying major and minor features [10]. With the increased availability of genetic testing, the identification of pathogenic mutations in TSC1 or TSC2 is now sufficient to establish a diagnosis, regardless of the presence of clinical characteristics [10] and is particularly useful for confirming a suspect diagnosis, as many clinical manifestations of TSC are rare in young patients [11]. As demonstrated in the case report, the patient has a systemic disease with genetic test positive, four mayors' criteria brain, eye, kidneys, head and skin associated with one mayor criteria rectal polyps. Detection of the disease should be followed by genetic

counseling. patients can be treated symptomatically. Therefore, awareness of various TSC organ manifestations is important such as multimodal imaging for RHA diagnosis and annual follow-up by FAF, SD-OCT and fundus photography.

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