



# ASTROCYTARY HAMARTOMA OF THE RETINA DURING TUBEROUS SCLEROSIS OF BOURNEVILLE

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## Abstract

*We report the case of a 20-year-old patient followed in the dermatology department for tuberous sclerosis of Bourneville. Ophthalmologic examination found visual acuity of 10/10 P2 with appendages and an anterior segment without notable abnormalities in both eyes. The fundus of the right eye revealed a translucent, greyish-white, finely vascularized nasal para-papillary tumor formation evoking a type I astrocytic hamartoma, with two other small cottony infero-papillary lesions (Figure 1-A). In the left eye, there is a whitish, partially calcified tumor lesion with a papillary diameter and nasal para-papillary site (Figure 1-B). Fluorescein angiography revealed an aspect of early auto-fluorescent lesions becoming hyper-fluorescent late in both eyes (Figure 1-C, 1-D) with a peripheral lesion causing a window effect (Figure 1-E). General examination found diffuse angiofibromas on the face (Figure 1-F) and trunk with achromatic spots on the back (Figure 1-G). The neurological, cardiovascular and abdominal examinations were normal. Chest x-ray, kidney ultrasound, echocardiography, and brain computed tomography did not show other locations. The diagnosis of Bourneville tuberous sclerosis was retained due to the association of cutaneous angiofibromas with retinal astrocytic hamartomas.*

**Key words:** Astrocytary hamartoma, retina, tuberous sclerosis of bourneville

## I-Introduction :

Astrocytic retinal hamartomas are rare benign tumors. 50% of them are associated with phacomatosis, namely tuberous sclerosis of Bourneville. The latter is a genetic condition, characterized by the development of astrocytic hamartomas in different organs: skin, brain, kidney, heart and eye. This condition usually manifests before the age of 20, and few cases have been reported in the literature.

### III-Observation

The general examination showed:

- Diffuse angiofibromas on the face (figure D) and trunk with achromatic spots on the back (figure B).
- Neurological, cardiovascular and abdominal examinations were unremarkable.
- Chest x-ray, renal ultrasound, echocardiography and cerebral computed tomography having shown no other localizations. The diagnosis of tuberous sclerosis of Bourneville was retained in view of the association of cutaneous angiofibromas with retinal astrocytic hamartomas.

A 20-year-old patient followed in the dermatology department for tuberous sclerosis in Bourneville.

A The ophthalmological examination:

- AV at 10/10th P2 in ODG without optical correction. - Appendages and anterior segment without notable abnormalities in ODG.
- FOD: finely vascularized parapapillary nasal translucent grayish-white tumor formation, evoking a type I astrocytic hamartoma, with two other small cottony lesions inferopapillary (figure A).
- FOG: whitish, partially calcified tumoral lesion with a papillary diameter of nasal parapapillary seat (figure A).

### III-DISCUSSION

#### **Classification of the different types of astrocytic retinal hamartomas:**

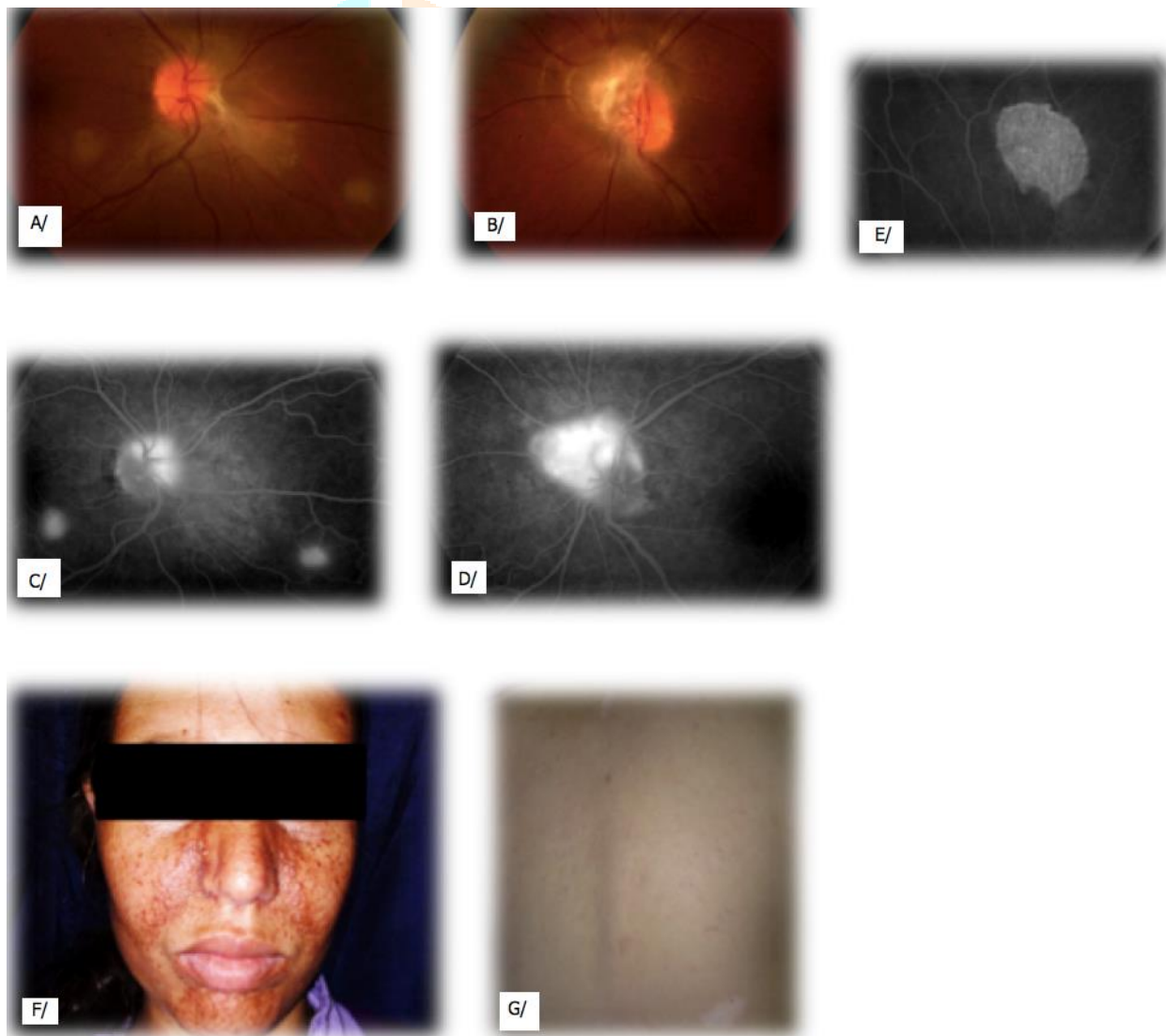
**Type I:** 57-70% of cases. is a cotton-wool, flat or slightly raised nodule from 1/4 to 2 papillary diameters, not calcified, greyish and translucent.

#### **Type II :**

50 to 55% of cases. It is a multi-nodular tumor, calcified, opaque, more posterior seat and 1/4 to 4 papillary diameters in size.

**Type III:** 9 to 12% of cases transitional between the two types I and II.

- Extra-retinal ocular manifestations, which are rarer, are mainly represented by angiofibromas of the eyelids, poliosis, non-paralytic strabismus, iris involvement (hamartomas, coloboma, depigmentation), cataracts, papilledema and refractive disorders.
- The other extra-ocular manifestations are in decreasing order: Cutaneous (cutaneous angiofibromas, areas of depigmentation), Neurological (intracerebral tubercles), Cardiac (rhabdomyomas, aortic aneurysms), Renal (multiple angiomyolipomas, renal cysts). Pulmonary (lymphangiomatosis).



**Figure 1:** A) Hamartomes astrocytaires réiniens de type I (OD); B) Hamartome astrocytaire partiellement calcifié de type II (OG); C) D) Aspect angiographique aux temps tardifs; E) atteinte périphérique de l'épithélium pigmentaire entraînant un effet fenêtre à l'angiographie à la fluorescéine; F) Angiofibromes diffus au niveau du visage; G) Tâches achromatiques au niveau du dos

#### IV- CONCLUSION

Retinal astrocytomas are rare tumors. Often integrated into the picture of tuberous sclerosis of Bourneville. Nevertheless, cutaneous and neurological lesions dominate the clinical picture of this phacomatosis. The ophthalmologist plays an important role in confirming the diagnosis. However, he must not miss a glioma of the optic nerve, which, although less frequent than in neurofibromatosis, can nevertheless affect the visual prognosis in this pathology. Like many genetic diseases, there is currently no specific treatment for uncomplicated astrocytic hamartomas

