EUROPEAN OPHTHALMIC PATHOLOGY SOCIETY

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Titre : Circumscribed choroidal hemangioma with *GNAQ* Q209 mutation

Clinical history :

A 53-year-old patient consulted with left eye pain associated with ocular hypertonia. In his history, he reports a trauma to the left eye in 2014, complicated by retinal detachment and intraocular hemorrhage. On clinical examination, he had 10/10 vision in his right eye and « can see the hand moving » in his left eye. Left intraocular pressure is 50 mmHg. The left eye fundus was inaccessible due to a cataract and corneal oedema.

Ultrasound revealed a temporal choroidal mass in the left eye. Orbital MRI revealed a posterior tissue formation (9 mm by 3 mm thick) with a T2 hyposignal, T1 hyper signal and homogeneous enhancement after injection.

Because of the difficulty of monitoring the tumour, the loss of vision in the eye and the hypothesis of a choroidal melanoma, enucleation was performed.

Ophthalmic pathology :

Examination disclosed a benign vascular tumor in choroid, well demarcated. The tumor was composed of large thin-walled vessels with little intervening stroma. Choroidal adjacent melanocytes appeared compressed around.

Diagnosis :

Circumscribed choroidal hemangioma with *GNAQ* Q209 mutation

Discussion :

Because of its benign nature and rarity, circumscribed choroidal hemangioma (CCH) often receives limited attention, leading to a high rate of misdiagnosis.

CCH is a rare benign intraocular tumor predominantly characterized by vascular proliferation. The highest incidence is among individuals aged 30-50 years (1). Clinically, it often manifests as a well-circumscribed lesion that can lead to various symptoms such as blurred vision or visual field defects, depending on its location and size. Histologically, CCH is characterized by thin-walled vascular channels lined with endothelial cells and scant stroma cells (2). This abnormal vascular architecture can increase hydrostatic pressure and cause subsequent leakage.

Despite being a benign condition, CCH poses significant clinical challenges. First, its clinical manifestations are highly variable and nonspecific, making diagnosis difficult and often leading to confusion with other conditions such as choroidal nevus or melanoma (3). It has been reported that only a third of patients in large series were referred with the correct diagnosis, leading to inadequate treatment (4). One report found that 5% of clinicaly « possible posterior uveal melanoma » were CCH (5).

Choroidal hemangioma may be diffuse (as in Sturge Webeer syndrome) or circumscribed. Contemporary imaging techniques have reduced the likehood of incorrect clinical diagnoses and pathologists seldom encounter these lesions in enuceated eyes exept as incidental findings.

Based on the size of their vascular channels, choroidal hemangioma have been classified as cavernous (most frequent), capillary or mixed. In contrast to cavernous hemangiomas in the orbit, choroidal hemangiomas have relatively little stroma and lack the thick fibrous septa found in orbital lesions. The individual vascular channels almost appear to abut each other (6). Solitary tumors have clearly demarcated pushing margins that compress adjacent melanocytes and choroidal lamellae. Patients with Sturge Webeer syndrome have diffuse angiomatosis that may involve more than half of the choroid and shows intermixture of engorged preexisting vessels with the vascular tumor (6).

GNAQ mutations occur in both diffuse and solitary hemangiomas, although at distinct codons. An R183 codon is mutant in diffuse choroidal hemangiomas, consistent with other Sturge-Weber vascular malformations. By contrast, CCH have mutations in the Q209 codon, similar to other intraocular melanocytic neoplasms like choroidal naevi and uveal melanoma (7). But *GNAQ* mutation spectrum in CCH is possibly restricted to p.Q209R, and p.Q209R are very rare in uveal melanoma (8).

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