Mesectodermal leiomyoma. Unusual tumor of the ciliary body

ABSTRACT

Background: Mesectodermal leiomyoma is a benign, smooth muscle tumor of the ciliary body, which is derived from the neural crest.

Clinical case: We report the case of a 35-year-old visually impaired Mexican female with blurred vision of the right eye of 2 months duration. Clinical and imaging presumptual diagnosis was adenoma of the nonpigmented epithelium of the ciliary body and surgical resection was done. Microscopically, the tumor was composed of cells with round nuclei and scant cytoplasm without atypia or mitosis, arranged in a fibrillar background. Immunohistochemical markers for vimentin, muscle specific actin, smooth muscle actin and calponin were strongly positive in the cytoplasm of the neoplastic cells, whereas glial fibrillary acidic protein and S-100 protein were negative in the same cell population.

Conclusions: Mesectodermal leiomyoma of the ciliary body is a benign tumor of smooth muscle and is extremely rare in this location. Up until this time, only 25 previous cases have been reported in the literature. The main differential diagnosis is uveal malignant melanoma; therefore, some eyes were enucleated. Ultrabiomicroscopy and A- and B- mode imaging studies are useful in the evaluation; however, microscopic examination with routine and histochemical staining are mandatory as well as the use of immunohistochemical markers such as vimentin, muscle specific actin, smooth muscle actin and calponin to establish the smooth muscle origin of this neoplasm and to rule out other malignant neoplasms such as malignant melanoma.

Key words: mesectodermal leiomyoma, immunohistochemistry, ciliary body.
INTRODUCTION

Ciliary body mesectodermal leiomyoma is a rare tumor that is benign in nature, originating in the smooth muscle of the ciliary body. Characteristically, it has a double differentiation (muscular and neural crest), which explains the neural appearance of the ovoid and spindle cells of this tumor by light microscopy. Immunohistochemistry markers play a fundamental role in the diagnosis of smooth muscle tumors. The specific markers for these neoplasms are vimentin, h-caldesmon, muscle specific actin, smooth muscle actin and calponin, which are positive in the cytoplasm of neoplastic cells. However, with regard to the neural origin, only two cases positive for the CD56 antigen have been reported in the medical literature, a neural marker.

This rare tumor could clinically be confused with a melanoma, which may result in radical therapeutic behavior such as enucleation. Of the 25 cases reported in the literature, enucleation was carried out in 12 of the cases. We report the case of a mesectodermal leiomyoma of the ciliary body, which was clinically diagnosed as an adenoma of the non-pigmented epithelium of the ciliary body. However, using immunohistochemical markers, the final diagnosis was able to be established.

CLINICAL CASE

We report the case of a 35-year-old female without prior clinically significant medical history. The patient presented for consultation due to progressive decrease in visual acuity, red eye and blurred vision of the right eye of 1-month evolution.

On ophthalmological examination, visual acuity in the right eye was 20/50 and 20/25 in the left eye. Biomicroscopy showed the right eye with a clear cornea, formed anterior chamber, central pupil, and irregular iris due to a pigmented lesion of neoplastic appearance in the posterior iris of the VII to the XI meridians, which protruded towards the pupil. There was partial involvement of the visual axis and the lens was clear (Figure 1).

During ultrabiomicroscopy, a lesion was reported in the ciliary body affecting the VII to the XI meridians with displacement of the iris anteriorly in contact with the corneal endothelium in the periphery, measuring 10.7 x 2.48 mm (Figure 2). On gonioscopy, the angle was open and the intraocular pressure was 15 mmHg. It was possible to assess the fundus under mydriasis, which revealed no abnormalities. An A-mode ultrasound study showed medium to high (78-82%) internal reflectivity data without vascularization (Figure 3).

Wide, open surgical resection of the tumor was decided upon, encompassing the iris and ciliary body from the lower meridian of the VI to the 1 meridian. Surgical specimen was sent to the Ophthalmic Pathology Service. The tissue was fixed in 10% formalin and was embedded in paraffin. Four-µm sections were made and were stained with hematoxylin and eosin, periodic
In addition to the tissue fixed in 10% formalin and embedded in paraffin, histological section of 4 μm were done, which were placed on slides previously treated with poly-L-lysine as tissue adhesive. A battery of primary antibodies were used such as vimentin, smooth muscle actin, calponin, gliofibrillary acid protein and S-100 protein, including negative and positive controls. Markers for vimentin, SMA and calponin resulted strongly positive in the cytoplasm of the neoplastic cells and not so for the gliofibrillary acid protein and S-100 protein, which supported its smooth muscle origin and ruled out other neoplasms of glial and peripheral nerve origin, respectively (Figures 5A and 5B).

**DISCUSSION**

The first cases of mesectodermal leiomyoma of the ciliary body were reported in 1970 by Jabobiec et al. who determined its probable neural crest origin. However, Blodi, in 1950, reported...
a ciliary body tumor that supposedly was a leiomyoma. To date there have been 25 cases reported of this rare tumor with preference for the female gender, as in the case reported here. Only five cases have been males. In the most recent cases the markers for estrogen and progesterone receptors have been investigated; however, all resulted in be negative. The most common clinical presentation is blurry vision or progressive decrease in visual acuity. Although it may extend posteriorly to the choroid, it generally manifests itself as a tumor dependent on the iris or ciliary body that is visible on biomicroscopic examination. There are no reports of bilaterality of this tumor.

Melanoma is the most frequent differential diagnosis, although it is a smooth muscle tumor with cells that lack melanin pigment. Its pigmentation can be provided by the pigmented epithelium of the iris and ciliary body that could clinically simulate a melanoma. This is supported by the fact that in 12/25 cases of leiomyoma, enucleation was done as the treatment of choice.

For diagnosis of tumor, a comprehensive evaluation is required that includes imaging with ultrabimicroscopy that helps to determine the extension or invasion to other structures by the tumor, A- and B- mode ultrasound to search the manner and internal vascularization to determine characteristic of the melanoma and, in some cases, also magnetic resonance imaging (MRI) and computed tomography (CT) of the orbits were done, which did not contribute to the diagnosis. In the case reported here, the characteristics demonstrated by ultrabimicroscopy and the location and absence of internal vascularity demonstrated by A-mode ultrasound point to the consideration of a benign neoplasm. A systemic examination of the patient was carried out with MRI and CT of the skull that did not reveal any alterations. In the eyes that were not enucleated, conservative resection via iridocyclectomy is the most common surgical treatment for this tumor, although in one case, sclerotomy was done with tumor biopsy.

One of the aspects of greatest interest in this tumor is its double differentiation. From 1989 to date, immunohistochemical markers have been used in 19/25 cases reported. In the case presented here, these markers revealed an evident smooth muscle origin because they resulted positive for smooth muscle actin and calponin.
These markers were negative in melanomas. The negativity in markers such as the gliofibrillary acid protein such as S-100 protein ruled out glial neoplasms (gliomas) and peripheral nerve sheath tumors (neurofibromas or Schwannomas), respectively. The latter may also be positive in melanoma. According to what was reported in the medical literature, all cases have proved positive for vimentin, actin smooth muscle, h-caldesmon and calponin markers, thus confirming its smooth muscle origin. Conversely, neural markers of the S-100 protein, gliofibrillary acidic protein, neurofilaments, chromogranin and synaptophysin have been negative in this neoplasm. Neuron-specific enolase markers (positive in the two cases), S-100 protein (positive in one case), and the CD56 antigen, a neural adhesion molecule (positive in three cases) are the only ones that so far support the neural origin of this tumor.

In conclusion, mesectodermal leiomyoma is a rare benign tumor of the ciliary body, which is clinically confused with melanoma. This tumor should be considered in the differential clinical diagnosis of females between the second and fourth decades as a tumor localized in the iris or ciliary body. Conservative treatment should be attempted by means of an excisional biopsy or surgical resection via an iridocyclectomy by an expert surgeon. Definitive diagnosis is established with the histopathological study with routine staining and with immunohistochemistry markers, which will determine the smooth muscle origin of this neoplasm.

REFERENCES