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From eye to diagnosis: Imaging as a key tool in pediatric choroidal melanoma

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Abstract
Uveal melanomas, particularly those involving the choroid, represent the most common form of intraocular malignancy in adults, yet they are extremely rare in the pediatric population. This report details the case of a 3-year-old male patient diagnosed with mixed choroidal melanoma, a highly unusual finding in this age group. The patient initially presented with peripheral ocular redness without other associated symptoms. A series of imaging studies, including ultrasound (US), magnetic resonance imaging (MRI), and PET-MRI, were instrumental in staging the disease. Advanced imaging modalities, alongside personalized diagnostic strategies, are crucial for characterizing the tumor and ensuring comprehensive patient care. This case highlights the importance of an interdisciplinary approach between ophthalmologists and radiologists for the early detection and management of ocular neoplasms.

Keywords: Choroidal melanoma, intraocular neoplasms, pediatric melanoma, pediatric ophthalmology, pediatric radiology, pediatric PET

Introduction
The choroid, in conjunction with other structures such as the ciliary body, constitutes a continuous layer within the eye, collectively referred to as the uvea [1]. Among the established risk factors for the development of both uveal and choroidal melanoma are fair skin, light-colored irises, congenital ocular melanocytosis, melanocytoma, and BAP1 cancer predisposition syndrome, which is linked to renal carcinoma, mesothelioma, and melanocytic tumors [2].

This condition is exceedingly rare, particularly in younger populations [3]. The incidence has remained relatively constant over the past several decades, with an estimated 5 to 7 cases per million individuals annually in Europe and 6 cases per million annually in the United States [4].

While some reports suggest a marginally higher incidence in females (55.6%) [3], other studies indicate an equal distribution between genders [2], rendering any definitive gender-based predisposition unclear.

Despite its rarity in routine clinical practice, it is imperative to ensure a robust, multidisciplinary approach involving both ophthalmology and diagnostic imaging teams. Such collaboration is essential not only for optimizing patient care but also for facilitating the early detection and management of both local and systemic complications, ultimately enhancing overall patient quality of life.

Case Presentation
A 3-year-old male patient, with a phototype of 2, presented with peripheral ocular redness for approximately 5 months, without any apparent associated symptoms. This prompted his parents to seek an ophthalmology consultation, where an evaluation of the anterior segment revealed prominence of the vascular pattern of the sclera in the left eye. Additionally, an examination of the fundus revealed a lesion suggestive of an intraocular neoplasm, likely a melanoma. Based on these findings, complementary studies were ordered for a complete evaluation, with particular emphasis on the possible local extension of the lesion.

Initially, the patient underwent ocular ultrasonography (Fig. 1), which confirmed the presence of a tumor in the choroid of the left eye. The diagnostic work-up was further supplemented with magnetic resonance imaging (MRI), which additionally identified a lesion at the temporal margin of the left eye (Fig. 2 and 3).

Given these findings, the ophthalmology team decided to proceed with enucleation of the left eye. The biopsy of the excised tissue revealed a solid tumor measuring 18x16 mm, with a basal diameter of 16 mm, and a histological diagnosis of mixed-type choroidal melanoma (epithelial and spindle cells), with all surgical margins, including the optic nerve margin, found to be negative. A month and a half after enucleation, the patient underwent a PET/MRI scan, which yielded negative results in the search for any signs of tumor activity (Fig.4).

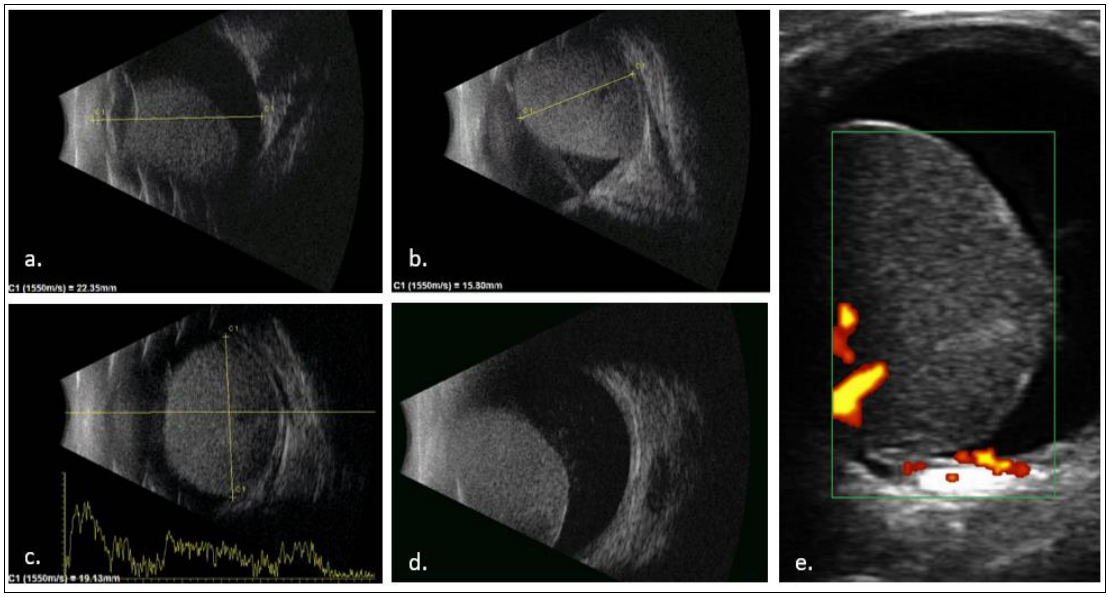

Fig 1: US. Left eyeball ultrasound. a., b., and c. Phakic eyeball with a coroideal lesion in the temporal inferior border that extends from M3 to M6, elevated, domed, homogeneous ecogenicity with coroideal excavation. d. Vitreous humor with some puntiform echoes suggestive of haematic content. e. Power Doppler ultrasound with evidence of intralesional vessel

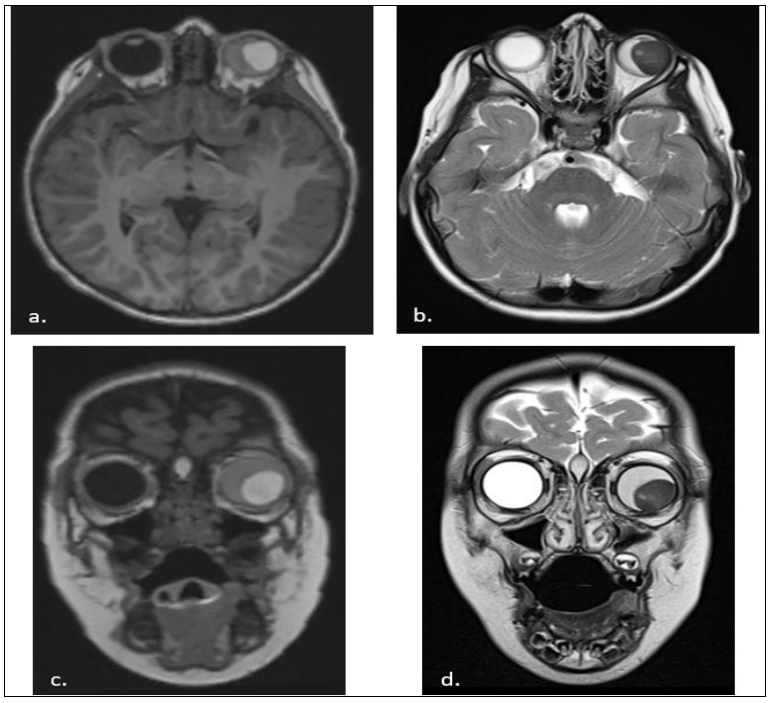

Fig 2: MRI. Left eyeball with evidence of an oval defined lesion at its temporal margin in close contact with the lens, that flattens the posterior sclera and has a subretineal appearance, hyperintense in T1WI, predominantly hypointense in T2WI. Additionally, the vitrious humor in the same eyeball is diffusely hyperintense in T1 and slightly hypointense in T2 highly suggestive of vitrious hemorrhage. a. T1WI axial image. b. T2WI axial image. c. T1WI coronal image. d. T2WI coronal image

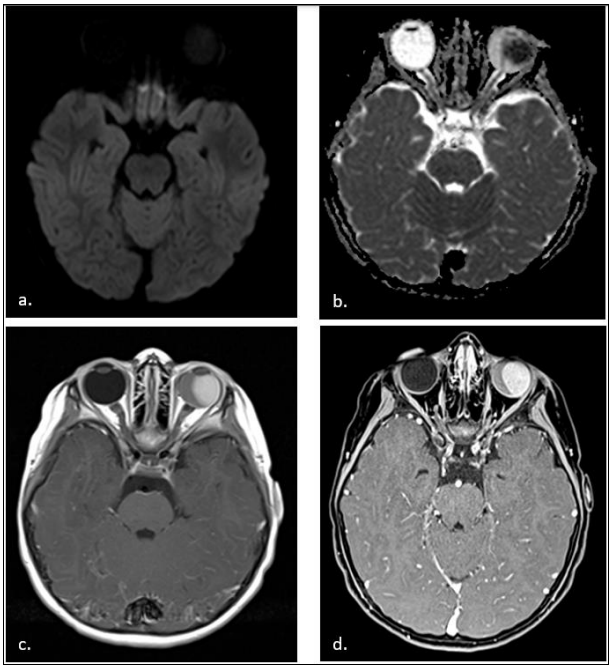

Fig 3: Contrast enhanced MRI. a. and b. DWI and ADC map with slight restriction, markedly hypointense in ADC map. c. T1WI axial image with contrast and d. T1WI FS axial image with contrast shows a rapidly enhancing and hypervascular lesion

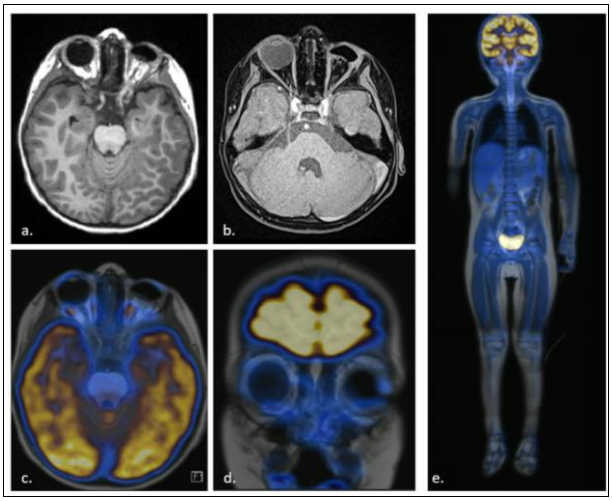

Fig 4: MRI and PET/MRI with 2-[^{18}F] FDG. Postoperative changes of left ocular enucleation. a. T1WI axial image shows fibroscarring changes in the intraconal fat, without alterations in the extraocular muscles. b. T1WI FS axial image with contrast with no evidence of abnormal enhancement after administration of intravenous contrast medium. c. axial, d. coronal, and e. coronal whole body PET/MRI without areas of abnormal focal increase in metabolism in the orbital fossa nor in the whole body

Discussion

Uveal melanomas are considered the most common cause of intraocular malignant tumors in adults. Any part of the uveal tract may be affected by this pathology [4]; however, in terms of frequency, the choroid is the most involved site, with approximately 90% of cases localized there, while the ciliary body (6%) and iris (4%) are affected less frequently [5].

Uveal melanoma, in general, is rare in children and young adults, with an estimated 1% of the diagnosed population being individuals under 21 years of age. Primary uveal melanoma in pediatric patients exhibits a highly variable incidence, with most cases manifesting between the ages of 13 and 30, that is, during or after puberty [3]. Moreover, studies on the prevalence of this condition across different age groups suggest that the diagnosis of ciliary body or choroidal melanoma increases significantly between the ages of 11 and 17, which has led to the association of its appearance with growth hormone levels [3].

Choroidal melanoma typically presents with painless loss of visual acuity or distortion of vision. Larger tumors are associated with retinal detachment, causing photopsia, although there are cases of asymptomatic patients [6]. In our specific case, it is noteworthy that the condition appeared at a very young age, as well as the association with an evident prominence of the vascular pattern during the ocular evaluation, which made the eye's appearance particularly striking to both the parents and the ophthalmology team.

The comprehensive management of this pathology is multidisciplinary, requiring the evaluation of both local and systemic structures. Among the diagnostic imaging techniques, one of the initial and crucial ones due to its effectiveness and lower risk of complications owing to its minimal invasiveness is ocular ultrasound, which aids in identifying characteristic and distinctive features of choroidal melanoma [5]. Ultrasonographic findings typically include the appearance of a dome or cup-shaped mass, which is the most definitive presentation of choroidal melanoma [6]. Fine-needle aspiration biopsy (FNAB) is not ruled out during the diagnostic process as a histological and scientific origin [7].

Positron emission tomography-computed tomography or MRI is utilized as a diagnostic and confirmatory method for metastasis, particularly in patients exhibiting abnormalities such as extraocular dissemination found during patient evaluation. The liver is the primary organ affected via hematogenous spread in 90% of cases, followed by the lungs in 24%, with other regions like lymph nodes also potentially affected. This justifies the performance of extension studies to assess the organ and systemic function of the patient [8].

Fundus examination, as a physical and semiological assessment of the ocular organ, is critical for diagnosis. With the advent of new non-invasive imaging techniques like autofluorescence imaging of the fundus, high-resolution digital images can be used to indicate the metabolic state of the retina and its pigmentary epithelium, regardless of the timing of the therapeutic intervention for choroidal melanoma [9].

For evaluating tumor extent, magnetic resonance imaging (MRI) plays a crucial role, enabling timely treatment initiation and patient monitoring. Uveal melanoma prognosis depends on melanin content, i.e., the areas with pigmentation or secondary necrosis due to the disease [4].

The primary objective of early ocular cancer detection is the preservation of the eye and its functional lifespan, with adequate visual acuity. Ocular therapeutic interventions include enucleation and various forms of radiotherapy, such as brachytherapy, laser therapy, and tumor resection, all aimed at preventing metastasis and improving survival rates. Mortality rates stand at 31% at 5 years and 45% at 15 years from the initial diagnosis [5, 6].

In light of these findings, it is essential that clinicians remain vigilant in the early detection of choroidal melanoma, especially in pediatric patients, where the condition is exceedingly rare. Employing advanced

diagnostic tools and a multidisciplinary approach ensures optimal patient management, ultimately improving both prognosis and quality of life. Early intervention remains the cornerstone of treatment, underscoring the importance of timely diagnosis and personalized care strategies.

Conclusion

The interdisciplinary collaboration between ophthalmologists and radiologists is pivotal for the early detection of ocular neoplasms involving both intraocular and extraocular structures. This integrated approach not only enhances the patient's clinical outcome but also plays a critical role in preserving their visual function. A comprehensive clinical assessment, including a meticulous anamnesis, thorough physical examination, and the utilization of advanced diagnostic techniques such as ocular ultrasound, magnetic resonance imaging (MRI), and PET-CT/MRI, is indispensable to the diagnostic process. Moreover, the importance of continuous follow-up with imaging studies is underscored, not only to monitor the primary lesion but also to ensure holistic and long-term patient care.

Patient Consent

Informed consent was obtained from both patient's parents for the publication and sharing of this case, including the associated images. We ensure that the confidentiality and privacy of the patient are maintained in full compliance with ethical standards and regulations. All identifying information has been carefully excluded to protect the identity of the patient, and the case has been presented in accordance with applicable ethical guidelines for medical publications.

Conflict of Interest

The authors declare no conflict of interest in the publication of this article.

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