MELANOCITOM GLAVE OPTIČKOG NERVA UZ JUKSTAPAPILARNI NEVUS HORIOIDEJE: PRIKAZ SLUČAJA I PREGLED LITERATURE

PRIKAZ SLUČAJA

CASE REPORT

MELANOCYTOMA OF THE OPTIC NERVE HEAD COEXISTING WITH JUXTAPAPILLARY CHOROIDAL NEVUS: CASE REPORT AND LITERATURE REVIEW

Dolika D. Vasović[1], Mirjana Mitrović[2], Ivan Marjanović[1,3], Tanja Kalezić[1,3]

[1] Univerzitetski klinički centar Srbije, Klinika za očne bolesti, Beograd, Srbija

[2] Ordinacija Mitrović, Beograd, Srbija

[3] Univerzitet u Beogradu, Medicinski fakultet, Beograd, Srbija

[1] *University Clinical Center of Serbia, Eye Hospital, Belgrade, Serbia*

[2] *Mitrović Eye Clinic, Belgrade, Serbia*

[3] *University of Belgrade, Faculty of Medicine, Belgrade, Serbia*

SAŽETAK

Uvod: Melanocitom glave optičkog nerva predstavlja retku, benignu pigmentovanu leziju koja nastaje od melanocita u području glave optičkog nerva. Iako je benigne prirode, melanocitom može predstavljati dijagnostički izazov zbog moguće sličnosti sa melanomom, kao i zbog potencijalnih komplikacija, uključujući defekte vidnog polja i, u retkim slučajevima, malignu transformaciju. Tačna dijagnoza se postavlja primenom pomoćnih dijagnostičkih metoda, poput fluoresceinske angiografije, optičke koherentne tomografije (OCT) i OCT-a sa povećanom dubinskom rezolucijom (EDI-OCT).

Prikaz slučaja: Četrdesetogodišnja pacijentkinja dolazi zbog pigmentovane promene na glavi levog optičkog nerva, koja je unazad 10 godina praćena u drugoj ustanovi. Oftalmološkim pregledom uočena je jasno ograničena, uzdignuta, sivo-crna lezija tipičnih kliničkih karakteristika melanocitoma. Uz melanocitom je primećen i jukstapapilarni horoidalni nevus. Dijagnostička obrada, koja je uključivala fluoresceinsku angiografiju i B-scan ultrasonografiju, potvrdila je benignu prirodu lezije. Pacijentkinji je preporučeno redovno praćenje kako bi se na vreme uočila eventualna progresija ili maligna transformacija.

Zaključak: Ovaj slučaj ističe važnost primene pomoćnih dijagnostičkih metoda i dugoročnog praćenja melanocitoma glave optičkog nerva. Koegzistencija jukstapapilarnog horoidalnog nevusa dodatno otežava dijagnostički proces, čime se naglašava potreba za redovnim kontrolama radi pravovremenog uočavanja mogućih komplikacija.

Ključne reči: melanocitom, horioidalni nevus, ultrazvučna dijagnostika, fluoresceinska angiografija

ABSTRACT

Introduction: Melanocytoma of the optic nerve head (melanocytoma papillae) is a rare, benign pigmented lesion originating from melanocytes within the optic nerve head. Despite its benign nature, melanocytoma can present diagnostic challenges due to its potential mimicry of melanomas and associated complications, including visual field defects and, in rare cases, malignant transformation. Accurate diagnosis relies on multimodal imaging, including fluorescein angiography, optical coherence tomography (OCT), and enhanced depth imaging-OCT (EDI-OCT).

Case report: A 40-year-old female presented with a pigmented lesion on the left optic nerve head, which had been monitored at another institution for the past 10 years. Ophthalmologic examination revealed a sharply demarcated, elevated, gray-black lesion with clinical features typical of melanocytoma. Adjacent to the melanocytoma, a juxtapapillary choroidal nevus was observed. The diagnostic workup, including fluorescein angiography and B-scan ultrasonography, confirmed the benign nature of the lesion. The patient was advised to undergo regular follow-up to monitor for potential progression or malignant transformation.

Conclusion: This case underscores the importance of auxiliary diagnostic methods and long-term surveillance in managing melanocytoma of the optic nerve head. The coexistence of a juxtapapillary choroidal nevus further complicates the diagnostic process, emphasizing the need for regular follow-up to enable the timely detection of potential complications.

Keywords: melanocytoma, choroidal nevus, ultrasonography, fluorescein angiography

Autor za korespondenciju:
Dolika D. Vasović
Klinika za očne bolesti, Univerzitetski klinički centar Srbije
Pasterova 2, 11000 Beograd, Srbija
Elektronska adresa: dolika.vasovic@gmail.com

Corresponding author:
Dolika D. Vasović
Eye Hospital, University Clinical Centre of Serbia
2 Pasterova Street, 11000 Belgrade, Serbia
E-mail: dolika.vasovic@gmail.com

Primljeno • *Received*: January 10, 2025; **Revidirano • *Revised*:** February 12, 2025; **Prihvaćeno • *Accepted*:** February 26, 2025; ***Online first*:** March 15, 2025

DOI: 10.5937/smclk6-55918

UVOD

Melanocitom glave optičkog nerva (melanocytoma papillae) je retka, benigna melanocitna lezija koja potiče od melanocita unutar glave optičkog nerva. Prvi su je opisali Limas i Ulrich 1962. godine. Melanocitom je histološki različit od melanoma, ali deli sličnu tamnu pigmentaciju zbog prisustva jako pigmentisanih, okruglih ili poligonalnih ćelija koje sadrže obilne granule melanina. Iako je obično asimptomatski, njegov klinički značaj ogleda se u mogućnosti izazivanja defekata vidnog polja, otoka optičkog diska i, u retkim slučajevima, maligne transformacije [1].

Ova lezija često koegzistira sa jukstapapilarnim horoidalnim nevusima, koji su pigmentisane, ravne lezije locirane u blizini optičkog diska. Studije sugerišu da je do 50% slučajeva melanocitoma povezano sa takvim nevusima, što dodatno komplikuje kliničku diferencijaciju od agresivnijih patologija, poput melanoma [2,3].

Napredak u tehnikama snimanja, kao što su fluoresceinska angiografija, optička koherentna tomografija (OCT), OCT sa poboljšanim prikazom dubine (EDI-OCT) i B-sken ultrasonografija, značajno je poboljšao sposobnost razlikovanja melanocitoma od lezija [4].

Iako su obično benigni, melanocitomi mogu pokazivati lokalno agresivno ponašanje, što može dovesti do kompresije optičkog nerva ili zahvatanja susednih struktura, poput retine i horioideje. Najčešće se dijagnostikuju kod osoba srednjih godina, bez značajne polne predispozicije [5,6]. Dugoročno praćenje je ključno kako bi se pratile moguće komplikacije, poput oštećenja vida, optičke neuropatije ili retkog, ali ozbiljnog rizika od maligne transformacije [7].

Ovaj prikaz slučaja predstavlja redak slučaj melanocitoma glave optičkog nerva sa koegzistirajućim jukstapapilarnim horoidalnim nevusom, naglašavajući dijagnostičke i terapijske izazove povezane sa ovom bolešću. Sveobuhvatan pregled literature pruža dodatni uvid u njegove kliničke, radiološke i histopatološke karakteristike, ističući značaj redovnog praćenja [8-10].

PRIKAZ SLUČAJA

Pacijentkinja, stara 40 godina, javila se našoj klinici sa poznatom anamnezom pigmentovane lezije na levom očnom dnu, koja je prvi put primećena pre više od 10 godina tokom rutinskog oftalmološkog pregleda. Pacijentkinja nije prijavila nikakve vizuelne simptome; njena medicinska anamneza bila je uredna, bez sistemskih oboljenja ili porodične istorije očnih bolesti. Oštrina vida bila je 1,0 na oba oka bez korekcije.

INTRODUCTION

Melanocytoma of the optic nerve head (melanocytoma papillae) is a rare, benign melanocytic lesion originating from melanocytes within the optic nerve head. Initially described by Limas and Ulrich in 1962, melanocytoma is histologically distinct from melanomas but shares a similar dark pigmentation due to heavily pigmented, round, or polygonal cells containing abundant melanin granules. While typically asymptomatic, its clinical significance lies in its potential to cause visual field defects, optic disc swelling, and, in rare cases, malignant transformation [1].

This lesion frequently coexists with juxtapapillary choroidal nevi, which are pigmented, flat lesions located near the optic disc. Studies suggest that up to 50% of melanocytoma cases are associated with such nevi, further complicating clinical differentiation from more aggressive pathologies such as melanoma [2,3].

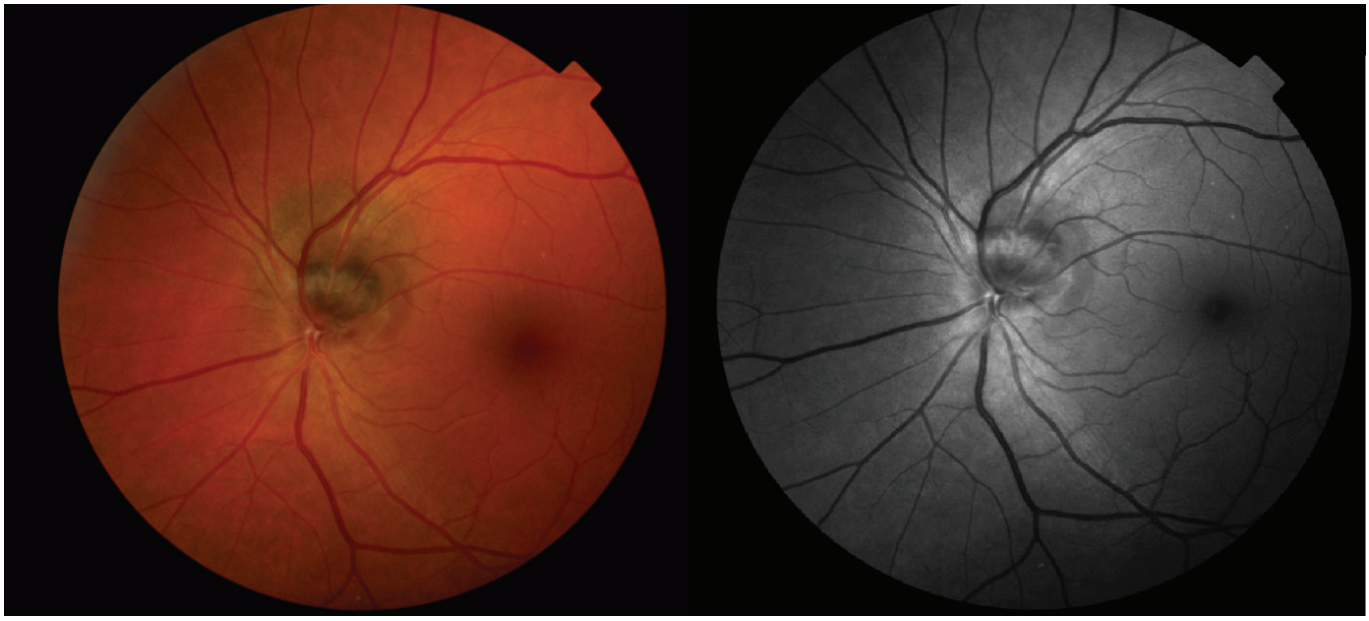
Advancements in imaging techniques, such as fluorescein angiography, optical coherence tomography (OCT), enhanced depth imaging-OCT (EDI-OCT), and B-scan ultrasonography have significantly improved the ability to distinguish melanocytoma from malignant lesions [4].

Although typically benign, melanocytomas can exhibit locally aggressive behavior, potentially leading to optic nerve compression or involvement of adjacent structures, such as the retina and choroid. They are most commonly diagnosed in middle-aged individuals with no significant gender predilection [5,6]. Long-term follow-up is crucial to monitor for complications, such as visual impairment, optic neuropathy, or the rare but serious risk of malignant transformation [7].

This report presents a rare case of melanocytoma of the optic nerve head with a coexisting juxtapapillary choroidal nevus, emphasizing the diagnostic and management challenges associated with this condition. A comprehensive literature review provides additional insights into its clinical, imaging, and histopathological features, highlighting the importance of multidisciplinary care in ensuring optimal outcomes for affected patients [8-10].

CASE REPORT

A 40-year-old female patient presented to our clinic with a known history of a pigmented lesion on the left optic nerve, first identified over 10 years ago during a routine ophthalmologic examination. The patient reported no visual symptoms; her medical history was unremarkable, with no systemic illnesses or family history of ocular diseases. Visual acuity was 20/20 bilaterally without correction.



Slika 1. Ova slika prikazuje fundus fotografiju melanocitoma lociranog na glavi optičkog nerva, praćenog jukstapapilarnim horoidalnim nevusom. Melanocitom je vidljiv kao jasno ograničena, uzdignuta, sivo-crna lezija na optičkom disku, što je u skladu sa njegovom karakterističnom pigmentacijom. Pored melanocitoma, horoidalni nevus izgleda kao ravno zelenkasto-braon polje sa jasnim granicama

Figure 1. The image presents a fundus photograph of a melanocytoma located on the optic nerve head, accompanied by a juxtapapillary choroidal nevus. The melanocytoma is visible as a sharply demarcated, elevated, dark gray-black lesion on the optic disc, consistent with its characteristic pigmentation. Adjacent to the melanocytoma, the choroidal nevus appears as a flat, greenish-brown area with distinct borders

Oftalmoskopski pregled otkrio je oštro ograničenu, prominentnu, sivo-crnu leziju na glavi optičkog nerva levog oka. Pored melanocitoma, uočen je jukstapapilarni horoidalni nevus, koji se prezentovao kao ravna, zelenkasto-smeđa lezija sa jasnim granicama. Nije bilo znakova edema optičkog diska, a retinalni sudovi su izgledali normalno (Slika 1). Automatska perimetrija pokazala je prisustvo malog, neprogresivnog centralnog skotoma.

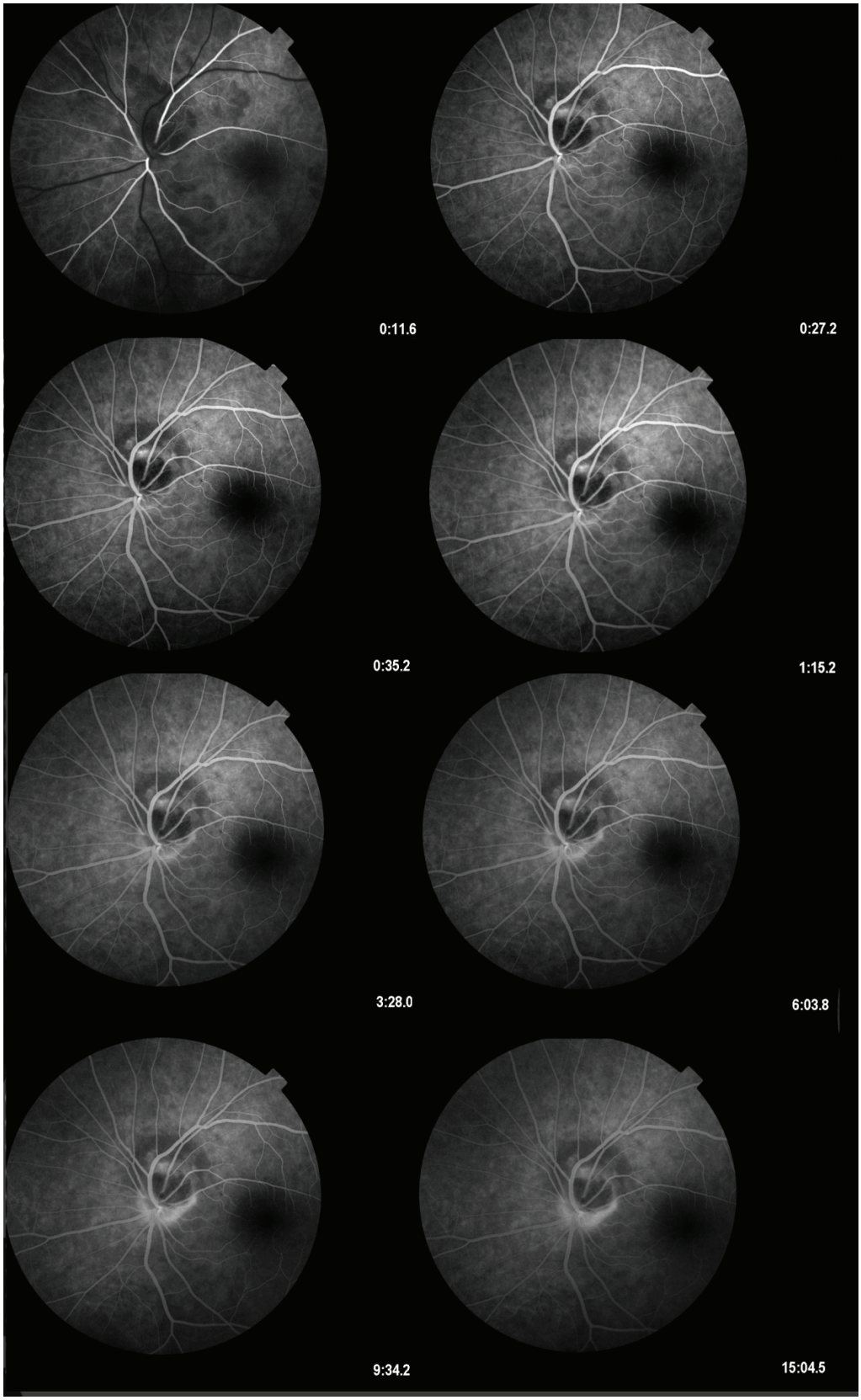
Fluoresceinska angiografija pokazala je potpunu blokadu fluorescencije koja odgovara melanocitomu, bez dokaza o curenju ili neovaskularizaciji (Slika 2). Jukstapapilarni nevus je tokom svih angiografskih faza ispoljavao postojanu hipofluorescenciju. Optička koherentna tomografija potvrdila je prisustvo izdignute lezije bez disrupcije slojeva retine. B-sken ultrasonografija otkrila je solidnu leziju srednje reflektivnosti.

S obzirom na benignu prirodu lezije i odsustvo vizuelnih simptoma ili komplikacija, pacijentkinji je savetovano redovno praćenje svakih šest meseci radi nadzora mogućeg napredovanja, uključujući povećanje lezije, gubitak vidnog polja ili znakove maligne transformacije. Pacijentkinja je edukovana o simptomima mogućih komplikacija, uključujući iznenadne promene vida, koje bi zahtevale hitnu medicinsku pomoć. Hirurška intervencija u ovoj fazi nije bila neophodna.

The ophthalmoscopic evaluation revealed a sharply demarcated, elevated, gray-black lesion on the optic nerve head of the left eye. Adjacent to the melanocytoma, a juxtapapillary choroidal nevus was noted, presenting as a flat, greenish-brown lesion with distinct borders. There were no signs of optic disc edema, and retinal vessels appeared normal (Figure 1). Automated perimetry revealed a small, non-progressive central scotoma.

Fluorescein angiography demonstrated a complete blockage of fluorescence corresponding to the melanocytoma, with no evidence of leakage or neovascularization (Figure 2). The juxtapapillary nevus exhibited persistent hypofluorescence throughout all angiographic phases. Optical coherence tomography confirmed the presence of an elevated lesion with shadowing effects but without disruption of the retinal layers. B-scan ultrasonography revealed a solid lesion with moderate internal reflectivity.

Given the benign nature of the lesion and the absence of visual symptoms or complications, the patient was advised to undergo regular follow-up examinations every six months to monitor for potential progression, such as enlargement, visual field loss, or signs of malignant transformation. The patient was educated about the symptoms of possible complications, including sudden vision changes, which would necessitate immediate medical attention. Surgical intervention was deemed unnecessary at this stage.



Slika 2. Ova slika prikazuje seriju snimaka fluoresceinske angiografije (FA) melanocitoma glave optičkog nerva, praćenog jukstapapilarnim horoidalnim nevusom. Angiografski niz ilustruje odsustvo curenja ili prebojavanja melanocitoma, što je u skladu sa njegovom benignom prirodom. Lezija se vidi kao tamna, jasno ograničena oblast na optičkom disku, koja blokira fluorescenciju. Pored melanocitoma, horoidalni nevus se prikazuje kao hipofluorescentna oblast

Figure 2. This image depicts a series of frames from fluorescein angiography (FA) of a melanocytoma of the optic nerve head, accompanied by a juxtapapillary choroidal nevus. The angiographic sequence illustrates the absence of leakage or staining over the melanocytoma, consistent with its benign nature. The lesion appears as a dark, sharply demarcated area on the optic disc, causing blockage of fluorescence. Adjacent to the melanocytoma, the choroidal nevus is visible as a hypofluorescent area

DISKUSIJA

Melanocitom glave optičkog nerva je retka, benigna lezija koja može oponašati agresivnija stanja, poput melanoma, što naglašava važnost tačne dijagnoze. Shields i saradnici su pokazali da se melanocitom javlja kod približno 1% opšte populacije, sa većom prevalencijom kod osoba tamnije pigmentacije [1,4]. Ove lezije obično ostaju stabilne, iako su u retkim slučajevima dokumentovani rast i maligna transformacija, sa procenjenom stopom transformacije od 1% [7].

Nekoliko prikaza slučajeva u literaturi ističe kliničku varijabilnost i dijagnostičke izazove melanocitoma. Burgos-Blasco i saradnici su predstavili slučaj pacijenta sa melanocitomom optičkog nerva, naglašavajući upotrebu multimodalnog snimanja, uključujući OCT i OCT angiografiju, za potvrdu dijagnoze i praćenje stabilnosti lezije tokom vremena [5]. U drugom slučaju, Kita i saradnici su opisali 38-godišnjeg pacijenta sa defektima vidnog polja uzrokovanim kompresijom optičkog nerva; praćenjem u periodu od pet godina utvrđeno je da lezija ostaje stabilna bez potrebe za intervencijom [9].

Mannat i saradnici su opisali redak slučaj melanocitoma koji se širio u peripapilarnu retinu, uzrokujući nakupljanje subretinalne tečnosti i blage vizuelne smetnje. Lezija je ostala stabilna bez znakova maligne transformacije tokom trogodišnjeg praćenja [6]. Takođe, Garza-Garza i saradnici su izvestili o seriji slučajeva u kojima su napredne tehnike snimanja, poput EDI-OCT, pružile neprocenjiv uvid u morfologiju i dubinu lezije, pomažući u diferencijaciji benignih od malignih promena [8]. Kao što se vidi u ovom slučaju, koegzistencija jukstapapilarnog horoidalnog nevusa komplikuje kliničku sliku.

Prema Gassu [2], prisustvo ovih nevusa povećava dijagnostički izazov, zahtevajući upotrebu multimodalnog snimanja za diferencijaciju. Fluoresceinska angiografija i OCT su ključni u prikazivanju karakteristične blokade fluorescencije i potvrđivanju benigne prirode lezije [3,5]. EDI-OCT dodatno poboljšava dijagnostičku preciznost pružajući slike visoke rezolucije glave optičkog nerva i okolnih struktura [6].

Histopatološke studije otkrivaju gusto raspoređene melanocite sa obilnim granulama melanina i niskom mitotskom aktivnošću, što je u skladu sa benignim tokom bolesti [10]. Međutim, faktori poput rasta lezije, kompresije optičkog nerva ili defekata vidnog polja zahtevaju pažljivije praćenje i potencijalnu intervenciju. Nedavne studije naglašavaju značaj genetskog i molekularnog profilisanja u diferencijaciji melanocitoma od melanoma u dvosmislenim slučajevima [8].

Lečenje melanocitoma se prvenstveno zasniva na redovnom praćenju radi otkrivanja promena u morfologiji ili funkciji lezije. Shields i saradnici preporučuju

DISCUSSION

Melanocytoma of the optic nerve head is an uncommon, benign lesion that can mimic more aggressive conditions such as melanoma, highlighting the importance of accurate diagnosis. Shields et al. reported that melanocytoma occurs in approximately 1% of the general population, with a higher prevalence among individuals of darker pigmentation [1,4]. These lesions typically remain stable, although growth and malignant transformation have been documented in rare cases, with an estimated transformation rate of 1% [7].

Several case reports in the literature emphasize the clinical variability and diagnostic challenges of melanocytoma. Burgos-Blasco et al. presented a case of a patient with optic nerve melanocytoma, highlighting the use of multimodal imaging, including OCT and OCT angiography, to confirm the diagnosis and monitor lesion stability over time [5]. Another case by Kita et al. detailed a 38-year-old patient who experienced visual field defects due to optic nerve compression; follow-up revealed lesion stability over five years without intervention [9].

Mannat et al. described a rare instance of melanocytoma extending into the peripapillary retina, causing subretinal fluid accumulation and mild visual disturbances. The lesion remained stable with no signs of malignant transformation during a three-year follow-up [6]. Additionally, Garza-Garza et al. reported a series of cases where advanced imaging techniques, such as EDI-OCT, provided invaluable insights into lesion morphology and depth, aiding in differentiating benign from malignant lesions [8]. As seen in this case, the coexistence of juxtapapillary choroidal nevi complicates the clinical picture.

According to Gass [2], the presence of such nevi increases the diagnostic challenge, necessitating the use of multimodal imaging for differentiation. Fluorescein angiography and OCT are pivotal in demonstrating characteristic blockage of fluorescence and confirming the lesion's benign nature [3,5]. EDI-OCT further enhances diagnostic accuracy by providing high-resolution images of the optic nerve head and surrounding structures [6].

Histopathological studies reveal densely packed melanocytes with abundant melanin granules and low mitotic activity, consistent with a benign course [10]. However, factors such as lesion growth, optic nerve compression, or visual field defects warrant closer surveillance and potential intervention. Recent studies have emphasized the importance of genetic and molecular profiling to differentiate melanocytoma from melanoma in ambiguous cases [8].

Management of melanocytoma primarily involves regular monitoring to detect changes in lesion morphology or function. Shields et al. recommend fol-

kontrolne preglede od šest meseci tokom prve godine, a zatim godišnje ako lezija ostane stabilna [10]. Edukacija pacijenata o simptomima optičke neuropatije ili maligne transformacije ključna je za pravovremenu intervenciju. U slučajevima značajnog rasta, progresivnog gubitka vidnog polja ili dijagnostičke neizvesnosti, može biti neophodna hirurška ekscizija i histopatološki pregled.

ZAKLJUČAK

Melanocitom glave optičkog nerva je retka i obično benigna lezija, ali njegova mogućnost lokalne agresivnosti i retke maligne transformacije zahteva pažljivo i kontinuirano praćenje. Ovaj slučaj naglašava značaj multidisciplinarnog pristupa, koji uključuje napredne tehnike snimanja i redovne kontrole, kako bi se optimizovali ishodi lečenja i sprečile komplikacije. Temeljno razumevanje kliničkih i histopatoloških karakteristika lezije, potkrepljeno pregledom literature, ključno je za efikasno upravljanje ovim stanjem.

Sukob interesa: Nije prijavljen.

low-up intervals of six months for the first year and annual evaluations thereafter if the lesion remains stable [10]. Patient education regarding optic neuropathy or malignant transformation symptoms is crucial for timely intervention. In cases of significant growth, progressive visual field loss, or diagnostic uncertainty, surgical excision, and histopathological examination may be necessary.

CONCLUSION

Melanocytoma of the optic nerve head is a rare and typically benign lesion, yet its potential for local aggressiveness and rare malignant transformation necessitates careful and continuous monitoring. This case underscores the importance of a multidisciplinary approach, incorporating advanced imaging techniques and regular follow-up, to optimize patient outcomes and prevent complications. A thorough understanding of the lesion's clinical and histopathological features, supported by a literature review, is essential for effective management.

Conflict of interest: None declared.

LITERATURA / *REFERENCES* LITERATURA

1. Shields JA, Demirci H, Mashayekhi A, Shields CL. Melanocytoma of optic disc in 115 cases: the 2004 Samuel Johnson Memorial Lecture, part 1. Ophthalmology. 2004 Sep;111(9):1739-46. doi: 10.1016/j.ophtha.2004.02.016.
2. Gass JD. Stereoscopic atlas of macular disease: diagnosis and treatment. 4th ed. St. Louis: Mosby; 1997. 1061 p.
3. Miller NR. Primary tumours of the optic nerve and its sheath. Eye (Lond). 2004 Nov;18(11):1026-37. doi: 10.1038/sj.eye.6701592.
4. Raval V, Reddy R, Kaliki S, Das T, Singh AD. Optic nerve head melanocytoma: Optical coherence tomography/angiography features. Indian J Ophthalmol. 2021 Feb;69(2):332-6. doi: 10.4103/ijo.IJO_710_20.
5. Burgos-Blasco B, Ventura-Abreu N, Jimenez-Santos M, Narvaez-Palazon C, Saenz-Francés F, Santos-Bueso E. Multimodal imaging in optic nerve melanocytoma: Optical coherence tomography angiography and other findings. J Fr Ophtalmol. 2020 Dec;43(10):1039-46. doi: 10.1016/j.jfo.2020.01.032.
6. Mannat G, Meenakshi S, Navya NK, Subina N. Optic disc melanocytoma: a case report. Rom J Ophthalmol. 2021 Jan-Mar;65(1):89-92. doi: 10.22336/rjo.2021.18.
7. Zhou N, Xu X, Wei W. Optical coherence tomography angiography characteristics of optic disc melanocytoma. BMC Ophthalmol. 2020 Oct 27;20(1):429. doi: 10.1186/s12886-020-01676-7.
8. Garza-Garza LA, Ruiz-Lozano RE, Ancona-Lezama D, González-Godinez S, Garza-León M. Multimodal imaging assessment of a "micro" optic disk melanocytoma: a case report. Arch Soc Esp Oftalmol (Engl Ed). 2021 Dec;96(12):663-7. doi: 10.1016/j.oftale.2020.10.004.
9. Kita Y, Holló G, Murai A, Kita R, Hirakata A. Optical coherence tomography angiography findings of an optic disc melanocytoma in a glaucoma eye. Int Ophthalmol. 2019 Mar;39(3):677-82. doi: 10.1007/s10792-018-0839-9.
10. Shields JA, Demirci H, Mashayekhi A, Eagle RC Jr, Shields CL. Melanocytoma of the optic disk: a review. Indian J Ophthalmol. 2019 Dec;67(12):1949-58. doi: 10.4103/ijo.IJO_2039_19.