



ASTROCYTOMA OF THE OPTIC NERVE HEAD

© E.V. Goltsman², V.V. Potemkin^{1, 2}, Yu.S. Astakhov¹, O.A. Marchenko²,
V.A. Antonov¹, A.E. Kel'ina³

¹ Pavlov First Saint Petersburg State Medical University, Saint Petersburg, Russia;

² City Multi-Field Hospital No. 2, Saint Petersburg, Russia;

³ North-Western State Medical University named after I.I. Mechnikov, Saint Petersburg, Russia

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✧ In this article, a clinical case of astrocytoma of optic nerve head in patient with neurofibromatosis type 1 is presented. The main feature of this clinical case is a difficulty in differential diagnosis with amelanotic choroidal melanoma.

✧ **Keywords:** optic nerve head astrocytoma; neurofibromatosis type 1; amelanotic choroidal melanoma.

АСТРОЦИТОМА ГОЛОВКИ ЗРИТЕЛЬНОГО НЕРВА

© Е.В. Гольцман², В.В. Потёмкин^{1, 2}, Ю.С. Астахов¹, О.А. Марченко²,
В.А. Антонов¹, А.Е. Кельина³

¹ ФГБОУ ВО «Первый Санкт-Петербургский государственный медицинский университет им. академика И.П. Павлова» Минздрава России, Санкт-Петербург;

² СПбГУЗ «Городская многопрофильная больница № 2», Санкт-Петербург;

³ ГАУЗ АО ФГБОУ ВО «Северо-Западный государственный медицинский университет им. И.И. Мечникова» Минздрава России, Санкт-Петербург

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✧ В рамках данной статьи представлен клинический случай астроцитомы головки зрительного нерва у пациента с нейрофиброматозом 1-го типа. Особенностью случая является сложность дифференциальной диагностики с беспигментной меланомой хориоидеи.

✧ **Ключевые слова:** астроцитомы головки зрительного нерва; нейрофиброматоз; беспигментная меланома.

In January 2019, a 72-year-old patient visited the Ophthalmology Department No. 5 of the City Multi-field Hospital No. 2 with the complaint of a gradual painless reduction in the vision of his left eye for 10 years. Initially, he noted a slight visual impairment of the left eye in 2009. He was then diagnosed with retinal cyst in a primary healthcare facility, and a case follow-up was recommended. Over the next 9 years, the patient regularly visited the ophthalmologist on an annual basis, and grad-

ual negative changes in visual acuity were noted. In 2018, there was rapid deterioration in the vision of the left eye, prompting the patient to visit another ophthalmological institution, wherein diagnosis of a choroidal neoplasm of the left eye and secondary retinal detachment was made, and enucleation was recommended.

An important aspect of the case history was the presence of type 1 neurofibromatosis, which was established when the patient was 43 years old.



Fig. 1. Coffee with milk spots (arms and head) and dermal neurofibroma in patient with neurofibromatosis type 1

Рис. 1. Пятна цвета кофе с молоком на коже рук и головы и подкожные нейрофибромы при нейрофиброматозе 1-го типа

Upon examination, manifestations of the disease such as multiple café au lait macules of different size on the scalp and body, some reaching 1.5 cm, and multiple subcutaneous neurofibromas were observed (Fig. 1).

Ophthalmological examination data included examination of the visual acuity of the right eye of 0.8, which was incorrigible and that of the left eye was hand motion. The tonometry result was 18/17 mmHg. Biomicroscopy revealed no abnormalities in the anterior segment of both eyes. Particular attention was paid to the search for Lisch nodules in the iris, which were absent. At ophthalmoscopy, no focal pathologic change was detected in the right eye, while there was a protruding multinodular pink–orange neoplasm in the optic nerve head of the left eye, with clear boundaries, at the base of which, a retinal detachment was visualized in the lower fundus segments (Fig. 2). At the B-scan ultrasound examination, the optic

nerve head had a protruding fungiform neoplasm (with height of up to 6.7 mm and diameter of up to 13.0 mm), medium echogenicity, and inhomogeneous density, at the base of which, retinal detachment was observed in the lower fundus segments (Fig. 3). Magnetic resonance imaging of the orbits also confirmed the presence of left eye neoplasm (Fig. 4).

The described clinical case is of interest because of the complexity of the differential diagnosis between choroid non-pigmented melanoma and astrocytoma of the optic nerve head. The fungiform shape of the neoplasm, secondary retinal detachment, degree of prominence in the vitreous, and age of the patient were associated with amelanotic melanoma. However, the presence of type 1 neurofibromatosis in the patient's history, a 10-year history of intraocular neoplasm ("retinal cyst"), multilobar structure, and neoplasm localization directly above the optic nerve head suggested the diagnosis of astrocytoma.

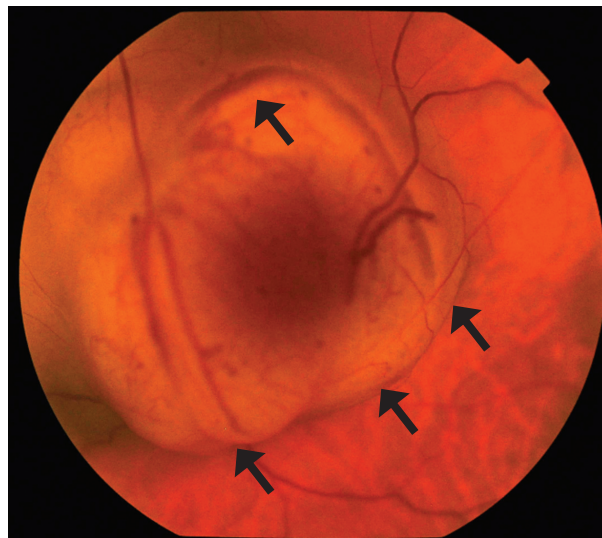


Fig. 2. Astrocytoma of the optic nerve head. Black arrows demonstrate a lobular structure of the tumor

Рис. 2. Астроцитомы головки зрительного нерва. Чёрными стрелками указаны выступающие части опухоли

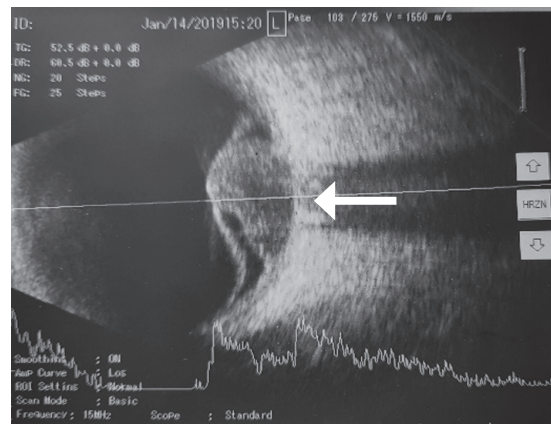


Fig. 3. B-scan ultrasound of left eye. Tumor grows from optic nerve (white arrow) causing secondary retinal detachment

Рис. 3. Ультразвуковое В-сканирование левого глаза. Опухоль располагается непосредственно над зрительным нервом (белая стрелка), вызывая вторичную отслойку сетчатки

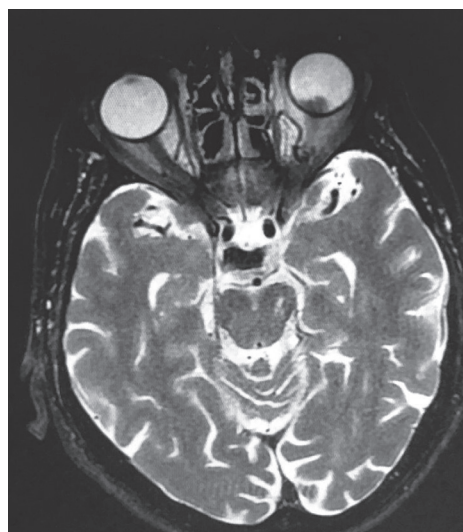


Fig. 4. MRI of orbits. Intraocular tumor over the optic nerve

Рис. 4. Магнитно-резонансная томография орбит. Видна внутриглазная опухоль в проекции зрительного нерва

Figure 5 shows a typical astrocytoma of the optic nerve head, and Figure 6 shows amelanotic melanoma of the juxtapapillary localization. The most characteristic symptoms of astrocytoma are clear contours; yellow, orange, and pink color or a combination thereof; and multilobar structure (often having a mulberry shape). Retinal and optic head astrocytomas are most often observed in tuberous sclerosis complex but can also develop with type 1 neurofibromatosis or without any underlying medical condition [1, 2, 9, 14]. Typical astrocytoma of the optic nerve head often has a mulberry shape due to multiple calcification nodules. However, the fact that astrocytomas may not be accompanied by calcification sites [1, 2] should be remembered. Both tumors may be accompanied by secondary retinal detachment [2, 9]. Amelanotic melanoma with juxtapapillary localization can spread to the optic nerve head but does not directly originate from it, as in the clinical case presented. In addition to the typical lenticular form, melanoma can be fungiform (or take the form of a cufflink) due to the penetration of the Bruch's membrane and infringement of the neoplasm apex in the defect [1]. Choroidal melanoma can often have two humps, indicating the presence of two growth epicenters, usually while developing from the previous nevus. However, we have never observed a multilobate structure of the choroidal melanoma. At the B-scan ultrasound imaging for vascular membrane melanoma, echogenicity is sub-average, sometimes with acoustic cavities and choroidal excavation [1, 2]. Melanoma can penetrate into the optic nerve head with circular growth, but the tumor prominence above it is not the highest.

We rarely resort to biopsy of the intraocular neoplasms because diagnosis can be made based on the clinical presentation in a vast majority of cases. However, considering the difficulties described above in establishing the final diagnosis, which influenced not only the further approach regarding eye preservation but also the prognosis, a decision was made to perform fine-needle aspiration biopsy. The patient underwent barrier laser coagulation of the retina around the neoplasm. Then, fine-needle aspiration biopsy using a 27G needle was performed under intravenous anesthesia under direct visual control without vitrectomy, followed by cytological examination of the material. The cytological examination revealed fragments of the tumor tissue, comprising small, relatively uniform, round oval, and elongated cells with short, unexpressed cytoplasmatic processes of the, that are tightly

packed and lack pigmentation. No mitosis and necrosis were detected. The neoplasm was benign, presumably from the nervous tunic. However, we could not definitively confirm it because there are no data on melanoma.

Thus, with fine-needle aspiration biopsy, the final diagnosis of astrocytoma of the optic nerve head was made. Enucleation in this case was not recommended. Considering that visual acuity during the patient's visit already was hand motion with incorrect light projection, without any ophthalmic hypertension and pain, delimitation of retinal detachment by laser coagulation made, and a slow growth during the past 10 years as per the medical history, attempts of any type of radiation or laser treatment were decided to be avoided in favor of the case follow-up with monitoring of the B-scan ultrasound results in 3 months.

The term "intraocular astrocytoma" indicates a low-grade neoplasm originating from the retina or optic nerve to the lamina cribrosa (optic nerve head). Astrocytomas with this localization are fundamentally different from astrocytic hamartomas and infiltrative astrocytomas (so-called massive gliosis) with progressive autonomous growth [9]. In clinical practice, the differences among astrocytomas, hamartomas, and reactive astrocytoma are unclear. Intraocular astrocytomas are benign neoplasms; however, they can cause vision loss [9].

The age of patients with newly diagnosed intraocular astrocytoma varies from 1 month to 45 years. The size of astrocytomas can also significantly differ. Tumors of only a few millimeters in size and those involving the entire eyeball have been described. Intraocular astrocytoma can be unilateral or bilateral, single or multiple, and calcified or noncalcified. Astrocytoma can often simulate retinoblastoma in children and amelanotic melanoma or choroidal metastasis in adults [4, 7, 8, 13]. The retina is often the source of intraocular astrocytoma. According to one study, among 1278 intraocular astrocytomas, only 1.6% were localized in the optic nerve head [10].

The presence of retinal and optic nerve astrocytomas is often associated with tuberous sclerosis and less commonly with neurofibromatosis. Thus, according to one study, of 42 intraocular astrocytomas, 57% were associated with tuberous sclerosis, 14% were associated with neurofibromatosis, and 29% were sporadic [12].

Type 1 neurofibromatosis (Recklinghausen's disease) is the most common disease from the group



Fig. 5. Astrocytoma of the optic nerve head in 56 year old man. Nodules of calcification are marked with black arrows, better visualization in red-free light

Рис. 5. Астроцитомы головки зрительного нерва у мужчины 56 лет. Узлы кальцификации показаны чёрными стрелками, они лучше визуализируются в бескрасном свете

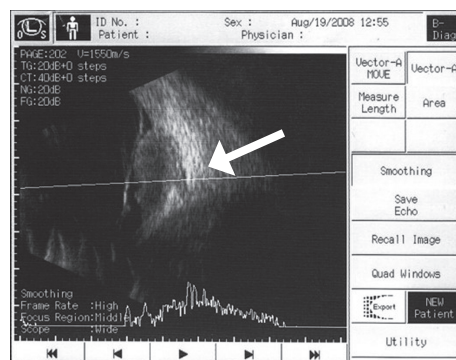
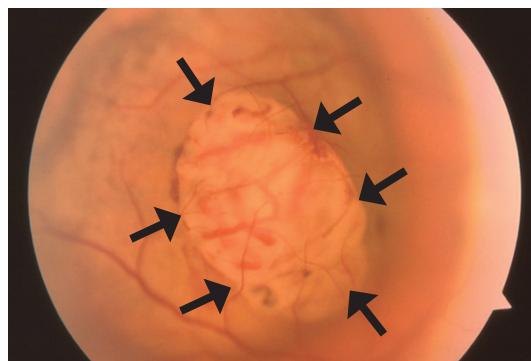


Fig. 6. Amelanotic juxta-papillary choroidal melanoma penetrating Bruch's membrane (black arrows). B-scan demonstrates deep choroidal excavation (white arrow)

Рис. 6. Беспигментная меланома хориоидеи юстапапиллярной локализации с зоной прорастания мембраны Бруха (чёрные стрелки). При В-сканировании видна глубокая экскавация хориоидеи (белая стрелка)

of phakomatoses. The disease was first described by the German physician Friedrich Daniel von Recklinghausen in 1882. It is characterized by a wide range of clinical manifestations involving different organs and systems [10].

The diagnosis of neurofibromatosis is based on the diagnostic criteria defined by the US National Institutes of Health in 1987 [5]. If the patient has at least two of the following symptoms, the diagnosis of neurofibromatosis is definite [5]:

- At least five café au lait macules with a diameter >5 mm in children of prepubertal age and at least six macules with a diameter >15 mm in the post-puberty period
- Two or more neurofibromas of any type or one plexiform neurofibroma
- Multiple small lentigo-like pigmented spots localized in large skin folds (axillary or inguinal)
- Optic nerve glioma
- Two or more Lisch nodules on the iris
- Bone abnormalities (thinning of the cortical layer of the tubular bones, often leading to the formation of false joints)
- Presence of type 1 neurofibromatosis in first-degree relatives according to the above criteria [12]

Neurofibromatosis has several ophthalmic manifestations. The most common manifestation is Lisch nodules [11, 12]. The development of plexiform neurofibroma in the eyelid can cause S-shaped palpebral fissure. Neurofibromatosis is often accompanied by the development of optic nerve glioma. According to the literature, the latter is found in approximately 15%–25% of patients with type 1 neurofibromatosis [6].

Generally, astrocytomas only require follow-up. However, in the case of massive exudation and retinal detachment, laser retinal coagulation, cryotherapy, photodynamic therapy, or brachytherapy can be performed [1, 6].

Thus, diagnosis of retinal and optic head astrocytoma is not always evident because of the wide range of clinical manifestations. One of the most difficult tasks is the differential diagnosis between non-pigmented melanoma and choroidal metastasis because the further approach and prognosis depend on it.

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Information about the authors

Elena V. Goltzman — Ophthalmologist. City Hospital No. 2, Saint Petersburg, Russia. E-mail: ageeva_elena@inbox.ru.

Сведения об авторах

Елена Владимировна Гольцман — врач-офтальмолог. СПбГБУЗ «Городская многопрофильная больница № 2», Санкт-Петербург. E-mail: ageeva_elena@inbox.ru.

Information about the authors

Vitaly V. Potyomkin — PhD, Assistant Professor. Department of Ophthalmology. Academician I.P. Pavlov First Saint Petersburg State Medical University; ophthalmologist. City Hospital No. 2, Saint Petersburg, Russia. E-mail: potem@inbox.ru.

Yuriy S. Astakhov — MD, Doctor of Medical Science, Professor, Department of Ophthalmology. I.P. Pavlov State Medical University of St. Petersburg, Saint Petersburg, Russia. E-mail: astakhov73@mail.ru.

Olga A. Marchenko — Ophthalmologist. City Multi-Field Hospital No. 2, Saint Petersburg, Russia. E-mail: oamarchenko@yandex.ru.

Vladimir A. Antonov — Resident, Department of Ophthalmology. I.P. Pavlov State Medical University of St. Petersburg, Saint Petersburg, Russia. E-mail: antonov@alborada.fi.

Anastasiya E. Kel'ina — Student. North-Western State Medical University named after I.I. Mechnikov, Saint Petersburg, Russia. E-mail: stokecity@bk.ru.

Сведения об авторах

Виталий Витальевич Потёмкин — канд. мед. наук, доцент кафедры офтальмологии. ФГБОУ ВО «ПСПбГМУ им. И.П. Павлова» Минздрава России; врач-офтальмолог. СПбГБУЗ «Городская многопрофильная больница № 2», Санкт-Петербург. E-mail: potem@inbox.ru.

Юрий Сергеевич Астахов — д-р мед. наук, профессор кафедры офтальмологии, кафедра офтальмологии. ПСПбГМУ им. акад. И.П. Павлова Минздрава России, Санкт-Петербург. E-mail: astakhov73@mail.ru.

Ольга Анатольевна Марченко — врач-офтальмолог. СПбГБУЗ «Городская многопрофильная больница № 2», Санкт-Петербург. E-mail: oamarchenko@yandex.ru.

Владимир Александрович Антонов — ординатор кафедры офтальмологии. ПСПбГМУ им. акад. И.П. Павлова Минздрава России, Санкт-Петербург. E-mail: antonov@alborada.fi.

Анастасия Евгеньевна Кельина — студентка. ФГБОУ ВО «Северо-Западный государственный медицинский университет им. И.И. Мечникова», Санкт-Петербург. E-mail: stokecity@bk.ru.