

WYBURN-MASON SYNDROME

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✧ In the article, a description of a clinical case of an extremely rare isolated ophthalmic form of the Wyburn-Mason syndrome is presented. Etiology, pathogenesis, clinical picture, and diagnostic criteria of this disease are also described.

✧ **Keywords:** Wyburn-Mason syndrome; arteriovenous malformations; phacomatosis; Bonnet-Dechaume-Blanc syndrome; neuroretinal angiomatosis.

СИНДРОМ УАЙБЕРН-МЕЙСОНА

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✧ В статье представлено описание клинического случая чрезвычайно редкой изолированной глазной формы синдрома Уайберн-Мейсона. Также описаны этиология, патогенез, клинические проявления и критерии диагностики данного заболевания.

✧ **Ключевые слова:** синдром Уайберн-Мейсона; артериовенозные мальформации; факотомоз; синдром Бонне – Дешома – Бланка; нейроретинальный ангиоматоз.

Wyburn–Mason syndrome, also known as Bonnet–Dechaume–Blanc syndrome, is a rare phacomatosis characterized by the development of arteriovenous malformations (AVMs) in the central nervous system, skin, and retina.

Retinal arteriovenous anastomoses were first described by H. Magnus in 1874 [17]. In 1932, S. Brock, C. Dyke, E. Krug, and B. Samuels reported a case of retinal arteriovenous anastomoses, brain AVMs, and facial vascular lesions in a 21-year-old male [6, 15]. J. Dechaume and

L. Blanc first suggested an association between retinal and intracranial AVMs (1937) [4]. Six years later, R. Wyburn–Mason published his observations of nine patients with AVMs in the midbrain, retina, and face [30].

Wyburn–Mason syndrome is an extremely rare congenital disorder with approximately 150 reported cases. The condition's heritability and gender and racial predispositions remain poorly understood. It is believed that the development of vascular disorders initiates at the seventh week

of gestation, leading to AVMs in the eyeball and midbrain [27].

The following variants of Wyburn–Mason syndrome were previously described:

- classic form (AVMs in the central nervous system, skin, and retina) [4, 9, 26, 30];
- incomplete classic form (no pathological changes in the skin) [3, 23, 26];
- isolated ocular form (retinal arteriovenous anastomoses) [8, 22, 26]; and
- isolated cerebral form (AVMs in the central nervous system) [26].

The disease may remain asymptomatic for a prolonged period of time. Neurological manifestations are diverse and depend on the location, shape, and size of the AVMs. Patients may exhibit cognitive disorders, headaches, epileptic seizures, or cranial nerve lesions [2, 7, 25, 29]. Compression of the surrounding structures can lead to perifocal edema of proximal brain matter or to subarachnoid hemorrhages [30]. Cerebral AVMs are predominantly located in the midbrain [30], but they can also be found in the orbit [7, 13]. The orbital AVMs usually manifest as partial ptosis. In cases of aneurysm emergence, the patient can develop pulsating or intermittent proptosis, pronounced dilatation of conjunctival vessels, and impaired eyeball mobility [19, 20].

Skin AVMs are located primarily on the face (cheeks, upper eyelid, and forehead) and appear as dilated, convoluted, purple vascular “stars” [1, 4, 9, 10, 16]. However, a number of authors report such changes on the lips, ears, and upper extremities [4, 5, 19, 24]. There is some evidence of moderate temperature increase, thickening, and swelling of the skin near the AVM [29]. Muthukumar et al. described pulsation of the affected skin on patient’s forehead [19].

Ocular manifestations are typically unilateral. Visual acuity values typically vary between 0.01 and 1.0. Decreased visual acuity is usually associated with AVMs located at the posterior pole. The visual acuity may also decrease due to retinal hemorrhages, thrombosis, retinal neuroepithelium edema in the macular area, neovascularization, and development of neovascular glaucoma [12, 18, 21, 26, 28].

Ophthalmoscopy reveals significantly dilated, tortuous, and twisted (similar to a ball of worms)

retinal vessels with multiple arteriovenous shunts. Diffuse retinal edema and intraretinal hemorrhages may be detected in this area. Several cases of optic nerve atrophy were described in patients with AVMs [14, 19, 29].

Few cases of spontaneous AVM involution with obliteration and sclerosis of abnormal retinal vessels were also reported [7, 11].

The diagnosis of Wyburn–Mason syndrome is based on brain and orbital magnetic resonance angiography (MRA) findings and ophthalmoscopy. Differential diagnosis should exclude Sturge–Weber syndrome, Von Hippel–Lindau disease, and other phacomatoses. There is no cohesive approach to the treatment of Wyburn–Mason syndrome. The treatment of ocular manifestations is usually symptomatic. The patients should be followed up by a neurosurgeon and an ophthalmologist.

CLINICAL CASE DESCRIPTION

A 29-year-old female patient presented with complaints of decreased visual acuity and object distortion and was admitted to the Ophthalmology Clinic of the Pavlov First Saint Petersburg State Medical University (Saint-Petersburg, Russia) in March 2017. She first noticed these symptoms at the beginning of 2017. Upon examination, her best corrected visual acuity value was 0.5 in the right eye and 1.0 in the left eye. Her intraocular pressure (measured using the Icare tonometer TAOli, Finland) was 11 and 10 mm Hg in the right and left eyes, respectively. Biomicrophthalmoscopy revealed severe vascular tortuosity in the right eye and a large number of thick and twisted vessels in the area of the optic disc and inferior macula. The biomicrophthalmoscopy of the left eye showed no pathological changes. Some vessels were white and sclerosed (Figure 1).

The patient underwent automated perimetry, optical coherence tomography, fluorescein angiography, and indocyanine green angiography. The automated perimetry revealed blind spot enlargement and absolute scotomas in the inferior part of the central visual field. Optical coherence tomography was used to obtain multiple cross-sectional images of the vessels in the inner layers of the retinal neuroepithelium, to demonstrate the areas of destruction in the retinal pigment epithelium,



Fig. 1. Fundus photo: a lot of enlarged convoluted blood vessels resembling tangled worms

Рис. 1. Фотография глазного дна: большое количество расширенных, извитых, напоминающих «клубок червей» сосудов

and an increase in macular volume (Figure 2). Fluorescein angiography of the macular area showed pathologically dilated, tortuous retinal vessels with extremely poor or absent blood circulation (Figure 3a). It also revealed arteriovenous anastomoses and an area of capillary non-perfusion in the upper outer quadrant (Figure 3c). Slight dye leakage in the macular area was detected during the late phase (Figure 3b). Indocyanine green angiography showed no pathological changes in the choroidal blood flow (Figure 4). The patient was referred for brain and orbital MRA, which revealed no pathological changes. No AVMs were detected in the skin. Based on the examination results, we diagnosed the patient with an isolated ocular form of the Wyburn—Mason syndrome. The patient underwent retinal la-

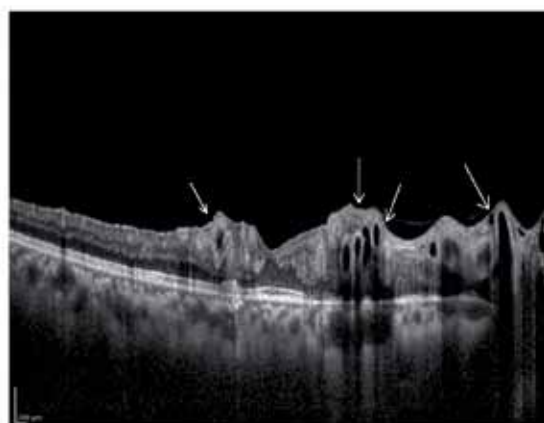
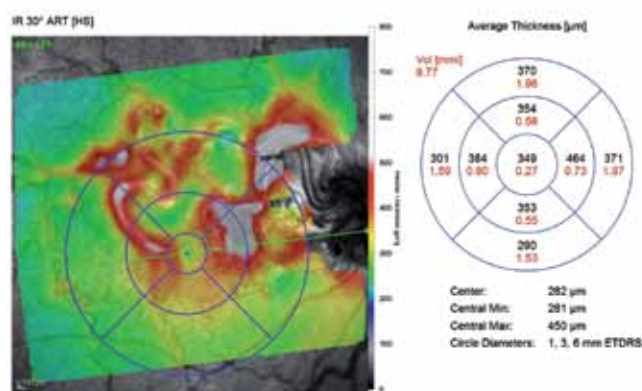
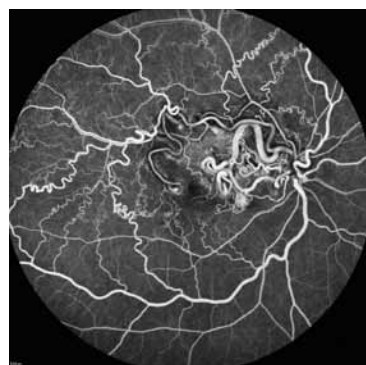
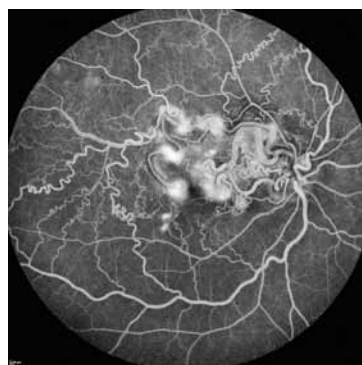


Fig. 2. Optical coherence tomography: increased neuroepithelium thickness in the vascular loops area, vessels at the neuroepithelium inner layers level (white arrows), areas of the retinal pigment epithelium destruction

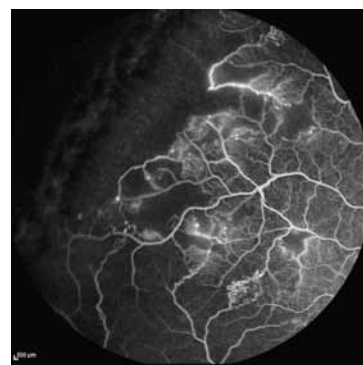
Рис. 2. Данные оптической когерентной томографии: увеличение толщины нейрорэпителлия в области сосудистых петель, срезы сосудов на уровне внутренних слоёв нейрорэпителлия (белые стрелки), участки деструкции пигментного эпителия сетчатки



a



b



c

Fig. 3. Fluorescein angiography: *a* — early venous phase (28 seconds); *b* — recirculation phase (12 minutes); *c* — capillary non-perfusion area in the late phase at the far periphery in the upper-outer quadrant. Description in the text

Рис. 3. Данные флюоресцентной ангиографии: *a* — ранняя венозная фаза (28 секунд); *b* — фаза рециркуляции (12 минут); *c* — в позднюю фазу на крайней периферии в верхненаружном квадранте зона капиллярной неперфузии

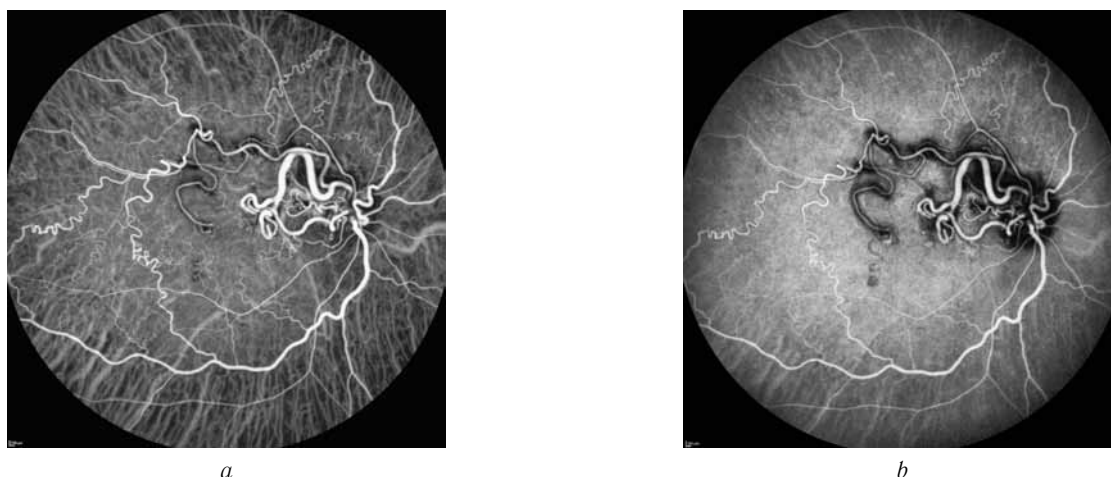


Fig. 4. Indocyanine green angiography: *a* — early phase (38 seconds); *b* — late phase (12 minutes)

Рис. 4. Ангиография с индоцианином зелёным: *a* — ранняя фаза (38-я секунда); *b* — поздняя фаза (12 минут)

ser photocoagulation in the ischemic area and is currently followed up by a neurosurgeon and an ophthalmologist.

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