



Valsalva retinopathy – a patient with a Leiden mutation in the coagulation factor V gene and increased blood clotting

Aleksander Lamkowski, Martyna Kałużna, Tomasz Pabin

Department of Ophthalmology, Voivodeship Hospital in Koszalin, Poland

ABSTRACT

Valsalva retinopathy is a rare disease in which the pressure in the eye increases, leading to the development of a pre-retinal hemorrhage. It frequently occurs in completely healthy patients and leads to sudden and painless vision loss. It may arise due to vomiting, coughing, constipation, pregnancy, childbirth, and intense physical or sexual activity. In this

article, we describe the case of a young man who developed Valsalva retinopathy. During diagnostics, increased blood clotting and V Leiden mutations were detected. We used Nd:YAG laser membranotomy. After treatment, the patient's vision improved.

KEY WORDS: Leiden mutation, Valsalva retinopathy, Nd:Yag laser, preretinal hemorrhage, increased blood clotting.

INTRODUCTION

Valsalva retinopathy is a rare condition in which, due to a sudden increase in blood pressure in the retina's veins, the superficial capillaries of the retina rupture and form a pre-macular hemorrhage. As a consequence, there is a sudden and painless loss of vision.

This effect is observed, among other causes, due to severe vomiting, coughing, constipation, pregnancy, childbirth, and intense physical or sexual activity. However, there are cases where the direct cause of the hemorrhage cannot be determined. The available literature has not yet described a thrombophilic patient who developed Valsalva retinopathy.

CASE REPORT

A 40-year-old patient showed up to the hospital on Monday morning due to a sudden, painless visual impairment in his right eye after a Saturday night rest. No other ailments accompanied the loss of vision. The interview showed that the patient had not been chronically ill so far and had not received chronic treatment, and the only risk factor for eye diseases was long-term smoking. Although the man worked physically in a sawmill daily, he denied carrying heavy objects the day before the onset of the disease and could not provide any tangible reasons for the condition.

In the ophthalmological examination on the day of admission, in the right eye there was light perception, in the left eye

there was 20/20 vision, and intraocular pressure was within the normal range. Examination of the fundus of the right eye revealed a large posterior preretinal hemorrhage covering the entire macular area (Figure. 1); the left eye had no pathological changes. The spectral optical tomography (SOCT) examination of the right eye showed that the hemorrhage was located under the internal limiting membrane (ILM), making it impossible to see into the deeper layers of the retina.

Based on the clinical picture and the tests performed, Valsalva retinopathy was suspected. For this reason, we decided to hospitalize the patient to extend the diagnosis.

Laboratory tests showed mild anemia and shortening of the prothrombin time to 8.54 s (range: 10.4-13.0 s) and APTT 23.76 s (range: 25.3-33.8 s). In additionally performed tests, the following were found: increased concentration of coagulation factor VIII 180.34% (range: 70-150%), factor V 121.5% (range: 70.0-120.0), and the resistance coefficient to the active protein C 1.428 (< 1 factor V Leiden resistance to activated protein C) – indicating hypercoagulability. We ordered genetic tests for the factor V Leiden mutation and the G-A 20210 mutation in the prothrombin gene. The study showed the presence of a Leiden mutation in one of the alleles of the coagulation factor V gene.

Based on the above image, 3 days after the onset of symptoms, we decided to try membranotomy treatment with a Nd:YAG laser in the lower part of the hemorrhage. The laser was set to single pulse with the focus at the zero position.

CORRESPONDING AUTHOR

Aleksander Lamkowski, MD, PhD, Department of Ophthalmology, Voivodeship Hospital in Koszalin, 7 Chalubinskiego St., 75-581 Koszalin, Poland, e-mail: lamkowski@gmail.com

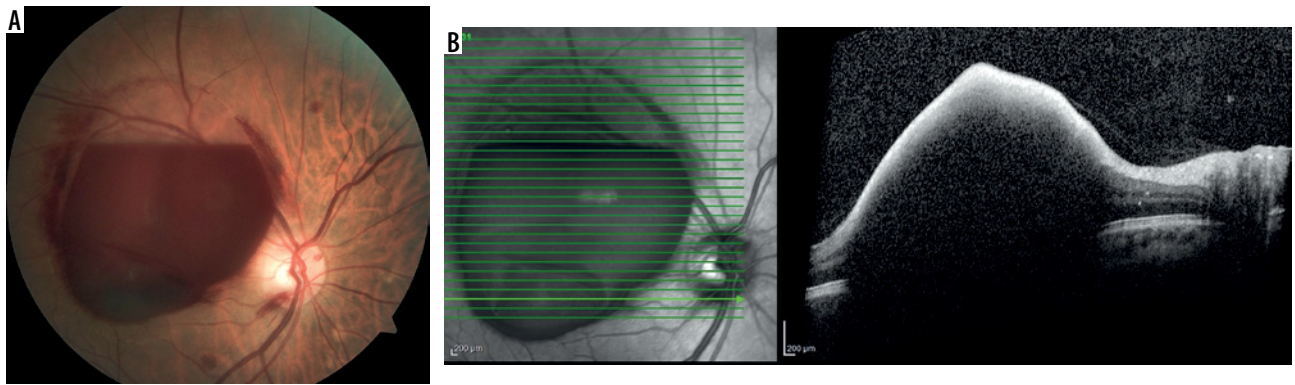


Figure 1. Preretinal hemorrhage on admission to the department: A) Fundus photography; B) SOCT photography

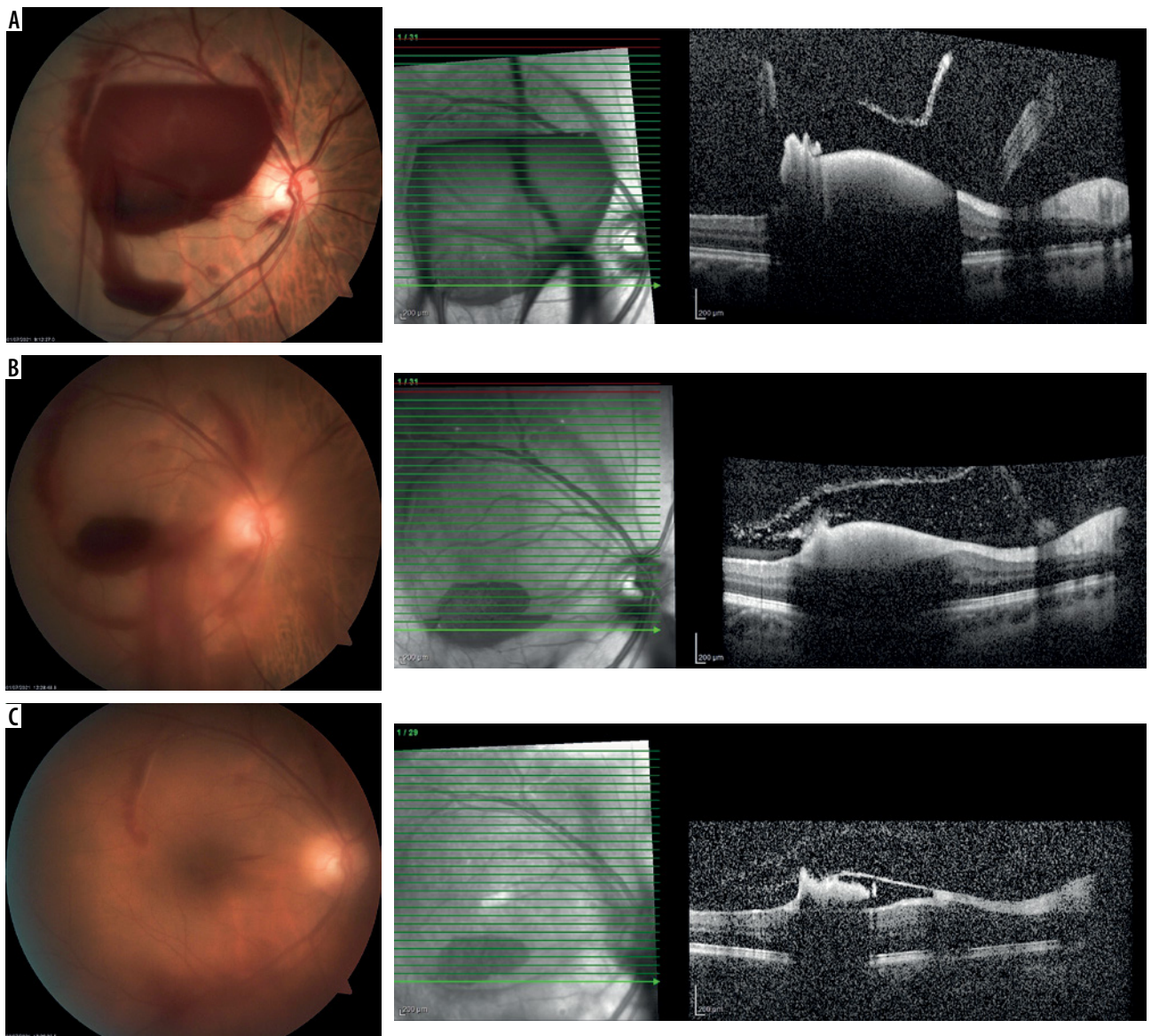


Figure 2. Color photography of the fundus and SOCT photography of the same area taken: A) immediately after membranotomy – visible reduction in the volume of pre-macular hemorrhage; B) 2 hours after treatment; C) after the first day of treatment, immediately before discharge from the department – reduction of transparency by blood extravasated into the vitreous body

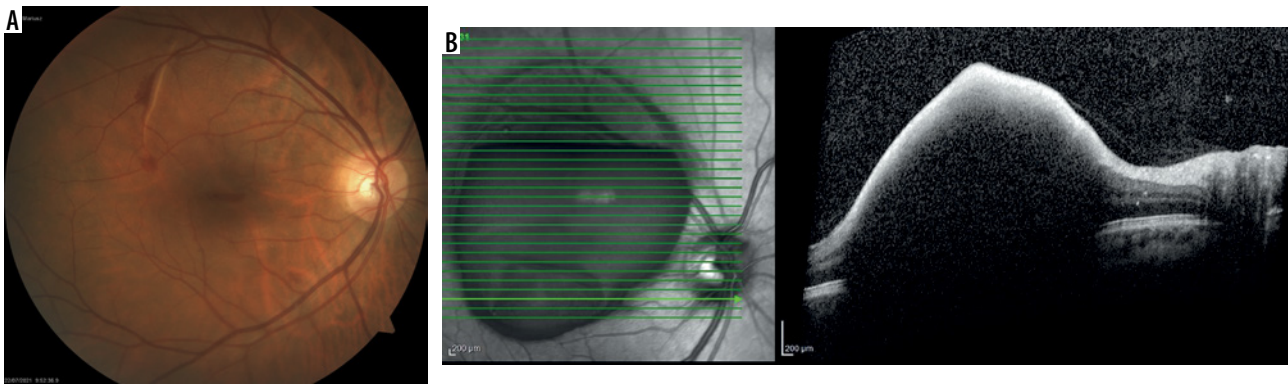


Figure 3. Follow-up after one week: A) fundus photography – almost complete regression of hemorrhage; B) SOCT scan of foveola – a small space of low reflectivity visible under the ILM; C) SOCT scan at the site of a previous laser pulse

Initially, an energy of 2 mJ was used, increasing it by one step with each impact to 4.3 mJ. The perforation effect occurred with the 4th laser impulse using a Volk Area Centralis lens. For comparison, in the available literature the authors used the Volk G-3 Goniofundus [1]. As a result, the hematoma evacuates, and the blood moves into the vitreous chamber. After the procedure, we immediately performed imaging tests (Figure 2).

During the follow-up visit one week after discharge from the hospital, visual acuity in the right eye increased to a score of 20/40. Imaging tests showed almost complete blood absorption, with no significant changes in the retina. The SD-OCT examination showed a normal retinal profile of the posterior pole, with a minimal pre-retinal space still visible (Figure 3).

DISCUSSION

Valsalva retinopathy is a rare condition caused by a sudden increase in intraocular venous pressure, which causes rupture of the capillaries in the retina, resulting in extravasation of the blood and the formation of pre-retinal hemorrhage. Usually, the disease manifests itself as a sudden vision loss in a healthy person [2].

The disorder has been reported to be related to exercise, including lifting weights, straining to defecate, coughing, and vomiting, in which the pressure in the chest and abdomen increases, following the Valsalva maneuver, i.e., forced exhalation with the glottis closed [2-4]. There are also known cases of the disease in pregnant women and during childbirth [5, 6].

Other causes of pre-macular hemorrhage include retinal aneurysms, proliferative diabetic retinopathy, retinal concussion, Terson's syndrome, and other less common disorders.

The exact location of pre-macular hemorrhage has been questioned in the literature. The authors describe the formation of hemorrhage under the ILM or under the posterior vitreous body, or both simultaneously [7-11]. Possible treatment options for Valsalva retinopathy include observation, vitrectomy, and laser membranotomy.

Valsalva retinopathy is a self-limiting disease, and hemorrhage usually resolves spontaneously without complications [12, 13]. However, the blood absorption process can take weeks or months and depends on the size of the hemorrhage.

Waiting for blood to be absorbed spontaneously may put a patient at risk of damaging his retina. In a few cases, patients developed poor vision, mainly attributed to the alleged toxic effects of thick pre-macular hemorrhage on the retinal pigment epithelium due to prolonged exposure to hemoglobin and iron, with or without epiretinal membrane formation [13].

Studies have shown that vitrectomy is the most effective treatment method in patients with severe pre-macular hemorrhage and insufficient spontaneous reabsorption [14]. However, it is associated with a higher risk of intra- and postoperative complications. In addition, there are reports of a possible hole in the macula and the formation of cataracts after the treatment of Valsalva retinopathy with vitrectomy [14, 15].

Laser membranotomy enables quick treatment and effective restoration of vision. Using the Nd:YAG laser in the lower part of the hematoma, a hole is made in the inner limiting membrane through which blood immediately enters the vitreous cavity. The effectiveness of Nd:YAG membranotomy has been proven in studies in most patients with Valsalva retinopathy undergoing this type of treatment. Possible complications include a macular hole, persistent pre-macular cavity [16, 17], retinal detachment [18], or epiretinal membrane formation [19]. Complications, however, are rare.

SUMMARY

Valsalva retinopathy is a rare pathology. Management of this condition includes observation or treatment with vitrectomy or laser membranotomy. Despite severe visual impairment in the early stages of the disease, visual acuity is usually restored to normal levels. Therefore, laser membranotomy should be considered the first-choice procedure in the treatment of pre-macular hemorrhages as an effective method that is minimally invasive and safe, especially in a young and professionally active patient, unwilling to wait several months for improvement of vision.

Factor V Leiden is a mutated form of human factor V and is one of the main inherited causes of an increased risk of thromboembolism in the human body.

According to previous studies, although the factor V Leiden mutation is an important risk factor for venous thrombosis, it does not seem to play an important role in the development of RVO [19] or RAO [20]. Currently, there are no studies in the literature on the risk of Valsalva retinopathy in patients with factor V Leiden mutation. It remains to be estab-

lished whether factor V mutation is a risk factor for Valsalva retinopathy.

DISCLOSURE

The authors declare no conflict of interest.

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