Obesity hypoventilation syndrome (OHS) is defined as obesity (body mass index, >30 kg/m²) with daytime hypoventilation (PCO₂ >45 mmHg) that is not secondary to cardiopulmonary or neurologic disease. Around 80–90% of the patients with OHS have obstructive sleep apnea and severe arterial oxygen desaturation. Obstructive sleep apnea has been found to be associated with a variety of eye diseases, including floppy eyelid syndrome, glaucoma, papilledema, nonarteritic anterior ischemic optic neuropathy, and retinal vein occlusion (RVO). While the association between OHS and ophthalmologic disorders is not well-known, our case report sheds light on this association.

A 27-year-old obese male patient presented for ophthalmic evaluation in August 2013. His chief complaint was a blurred vision for 7 months, combined with intermittent bulbar conjunctiva hyperemia that would spontaneously heal. His blood pressure was 130/110 mmHg, the light reflex of both pupils was normal, the corrected visual acuity was 0.6 (left) and 0.8 (right), and the intraocular pressure was 12 mmHg (left) and 13 mmHg (right). Blood routine test results were as follows: Hemoglobin 219 g/L, red blood cell count 9.01 × 10¹²/L, and hematocrit 70.7%. The reticulocyte absolute value was 0.1364 × 10¹²/L (normal <0.084 × 10¹²/L) and erythropoietin was 7.17 ng/ml (normal <3.28 ng/ml). D-dimer and coagulation function tests were normal. Polymerase chain reaction showed that JAK2V617F was negative. Bone marrow biopsy showed myeloproliferative active. Funduscopic exam revealed bilateral papilledema, hemorrhages around the optic discs, multiple patch hemorrhages in both retinas, and macular edema [Figure 1]. Abdominal ultrasound showed the mild fatty liver disease and a normal spleen. Because of the polycythemia vera and RVO, he was treated with laser, avastin injections in both vitreous bodies, oral hydroxycarbamide, enteric-coated aspirin, allopurinol, and bloodletting. However, these were invalid, and he became completely blind in his right eye while his left eye retained only light perception. Upon inquiry, the patient reported that he had a nocturnal snore for 4 years, complicated with mild daytime sleepiness and dry mouth. Referral of the patient to the department of sleep medicine was suggested.

After hospitalization, physical examination revealed the following: Weight, 115 kg; body mass index, 37.1 kg/m²; resting blood pressure, 140/100 mmHg; and SpO₂, 95%. The soft palate, uvula, and tonsils were normal, Mallampati class III, and mild ocular proptosis. The thyroid gland was normal. Lung and heart examinations found no abnormalities. The extremities showed no cyanosis, clubbing, or edema. Arterial blood gas analysis on room air revealed the following: pH, 7.35; PaO₂, 56 mmHg; a PaCO₂, 59 mmHg; and bicarbonate (HCO₃⁻), 32.6 mmol/L. The fibroblast growth factor receptor 2 was negative. The electrocardiogram, chest radiographs, cephalometric examination, and brain magnetic resonance venography were normal. A pulmonary function test showed mild restrictive ventilatory dysfunction. Lumbar puncture demonstrated that the intracranial pressure was high at 20 cmH₂O, and the cerebrospinal fluid was normal. Full-night polysomnography showed that the patient had obstructive sleep apnea with severe hypoxemia. A full-night polysomnography showed the patient had OHS.

Key words: Obesity Hypoventilation Syndrome; Papilledema; Polycythemia; Retinal Venous Occlusion

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Access this article online

Quick Response Code:  
Website: www.cmj.org
DOI: 10.4103/0366-6999.167364

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Received: 16-05-2015 Edited by: Yi Cui
How to cite this article: Wang YY, Han F. Polycythemia, Ablepsia, and Obesity Hypoventilation Syndrome: A Case Report. Chin Med J 2015;128:2815-7.
The apnea-hypopnea index was 88/h, the lowest oxygen saturation was 30%, the highest transcutaneous PCO$_2$ was 77 mmHg, and the total time of transcutaneous PCO$_2$ >55 mmHg was 175 min (Radiometer TCM4, Denmark). After the patient was prescribed with acetazolamide tablets 500 mg once daily and underwent bilevel positive airway pressure titration, the inspiratory pressure was 25 cmH$_2$O and the end-expiratory pressure was 15 cmH$_2$O. After 8 weeks of continuing treatment, arterial blood gas analysis was as follows: pH, 7.39; PaO$_2$, 88 mmHg; PaCO$_2$, 44 mmHg; and HCO$_3$, 26.1 mmol/L. Routine blood test results were restored to normal. The apnea-hypoxia index decreased to 3.8/h and the lowest oxygen saturation increased to 90%. An eye examination showed that the papilledema had resolved, so the acetazolamide was discontinued.

As we could exclude cardiopulmonary and neurological diseases based on this patient’s history and auxiliary examinations, the diagnosis of OHS could be established. Because the cephalometric examination and fibroblast growth factor receptor 2 were normal, we could exclude the diagnosis of Crouzon syndrome. The diagnoses of brain tumor and pseudotumor cerebri as causes of papilledema could be ruled out because the patient’s intracranial pressure and brain magnetic resonance venography were normal. We could also exclude the other possible etiologies for blood hypercoagulable states. We speculated that OHS was the cause of polycythemia, papilledema, and RVO in this patient, and effective experimental treatment further confirmed our conjecture.

During the past decades, several scholars have reported papilledema associated with Pickwickian syndrome and respiratory failure.\cite{1,2} Reeve et al.\cite{1} reported the case of a 58-year-old obese woman who presented with blurred vision. Ophthalmologic examination revealed gross bilateral papilledema with hemorrhages and venous congestion. Room air arterial blood gas analysis revealed a pH of 7.37, a PCO$_2$ of 62 mmHg, and a PO$_2$ of 54 mmHg. After treatment with oral medroxyprogesterone as a respiratory stimulant and a weight-reducing diet, her symptoms and eyesight were improved.

RVO is the second most frequently occurring retinal vascular disorder; most patients exhibit symptoms of vision loss on awakening. Risk factors include hypertension, diabetes mellitus, and cardiovascular diseases, which are closely related to sleep-disordered breathing. Turati et al.\cite{3} reported the case of a 37-year-old man who presented with progressive bilateral vision loss. He had morbid obesity, with a body mass index of 55 kg/m$^2$. His medical history included high blood pressure and deep venous insufficiency. Laboratory values were normal except for a hematocrit of 81%. Arterial blood gas measurements revealed a PaCO$_2$ of 52 mmHg and a blood oxygen saturation of 54%. Fundus examination revealed hyperemic optic discs with poorly defined edges, massive venous tortuosity and dilation, flame hemorrhages in the posterior pole, and mild macular edema in both eyes. Polysomnography demonstrated 133 obstructive hypopneas and 45 obstructive apneas. Treatment included a weight-loss diet and continuous positive airway pressure with supplementary oxygen via nasal mask; corrected visual acuity was 20/20 (right) and 20/25 (left) 2 years later.

The pathophysiological mechanisms of the association of papilledema and RVO with sleep disorder are not well-known. Mechanisms may include local and systemic effects of the hypoxemia and hypercapnia of OHS. First, hypoxemia-induced vasodilation of the central retinal artery may compress the central retinal vein and its tributaries because the central retinal artery and central retinal vein

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**Figure 1:** Funduscopic examination images. (a and b) Color image of the left and right eye, respectively, revealing a congestive and swelling optic disc with unclear boundaries, and hemorrhages around it; macular edema and the central retinal venous tortuosity. (c and d) Fluorescence angiography of left and right eye, respectively, demonstrating the capillaries of the surface of optic nerve dilated with high fluorescence; the venous tortuosity and dilation; the optic disc ill-defined, large fluorescence leaked by irregular peripheral retinal hemorrhage. (e and f) Color image of left and right eye, respectively, revealing optic disc papilledema and macular edema improved significantly and no hemorrhage, posterior pole veins tortuous after 2 months of continuous positive air pressure treatment.
lie within the same adventitial sheath. This may result in slowing of the retinal venous velocity. Hypercapnia could concurrently induce cerebral vasodilation leading to an increase of intracranial pressure, ultimately resulting in papilledema and increased venous pressure of the optic nerve head.\[^4\] Elevated venous pressure in the optic nerve head and papilledema may cause plasma leakage into the interstitial space, leading to hemoconcentration and local hyperviscosity. All of these factors could further slow the retinal circulation and initiate a vicious cycle, aggravating the development of RVO. Second, intermittent hypoxia may result in oxidative stress, abnormal activation of the sympathetic nervous system, and chronic systemic inflammation. These systemic effects may lead to endothelial cell dysfunction and produce inflammatory cytokines such as interleukin-6, tumor necrosis factor-\(\alpha\), matrix metalloproteinases, acute-phase proteins, intercellular adhesion molecule 1, and vascular cell adhesion molecules.\[^5\] These factors could trigger the extrinsic coagulation pathway. Third, respiratory efforts can lead to sleep fragmentation, which may activate the sympathetic nervous system and increase arterial blood pressure, leading to hemodynamic disturbances. These repetitive changes in the central retinal artery may contribute to occlusion of the fellow vein. In addition, sleep apnea may cause or aggravate risk factors of RVO such as hypertension, atherosclerosis, and diabetes. Together, these changes may explain papilledema and RVO development in patients with sleep disorders.

In conclusion, this case demonstrates that OHS is associated with ocular diseases. Early recognition of this association and proper treatment may prevent permanent optic nerve injury.

**Financial support and sponsorship**

Nil.

**Conflicts of interest**

There are no conflicts of interest.

**References**

1. Reeve P, Harvey G, Seaton D. Papilloedema and respiratory failure. Br Med J (Clin Res Ed) 1985;291:331-2.
2. Bloomfield RL, Felts JH, Burkart JM, Cashwell FL. Optic disc edema in a Pickwickian man mimicking hypertensive crisis. J Clin Hypertens 1987;3:27-30.
3. Turati M, Velez-Montoya R, Gonzalez-Mijares CC, Perez-Montesinos A, Quiroz-Mercado H, Garcia-Aguirre G. Bilateral central retina vein occlusion associated with obesity-hypoventilation syndrome (Pickwickian syndrome). Retin Cases Brief Rep 2009;3:140-3.
4. Purvin VA, Kawasaki A, Yee RD. Papilledema and obstructive sleep apnea syndrome. Arch Ophthalmol 2000;118:1626-30.
5. Kohler M, Stradling JR. Mechanisms of vascular damage in obstructive sleep apnea. Nat Rev Cardiol 2010;7:677-85.