Chiari 1 malformation with optic disc drusen

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ABSTRACT

A 32 year old female patient with CM 1 diagnosis was referred for the management of papilledema. Ophthalmologic examination revealed normal visual acuity (20/20 in both eyes), normal optic nerve function tests and normal slit-lamp biomicroscopic findings.

Funduscopy revealed bilateral irregular optic nerve heads with blurred margins. B scan ultrasonography (USG) and Spectral domain optical coherence tomography were performed and bilateral optic nerve heads were diagnosed as ODD. We concluded that the pseudopapilledema must be taken into account before making papilledema diagnosis in patients with CM 1 to protect the patients from redundant interventional procedures.

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Background

Chiari 1 malformation (CM 1) is caused by the caudal herniation of the cerebellar tonsils through the foramen magnum (FM). The symptoms of CM 1 are suboccipital headaches, dizziness, disequilibrium, vertigo, tinnitus, dysphagia, sleep apne, tremors, palpitations, paresthesia, fatigue, and neck pain.¹ Patients with CM 1 may also have ocular symptoms like retro-orbital pressure and/or pain, diplopia, photophobia, impaired visual acuity, nistagmus, strabismus, and papilledema.¹ Cerebrospinal fluid (CSF) flow is abnormal in patients with CM 1 and papilledema secondary to increased intracranial pressure (ICP) has been reported in some patients with this disorder.^{1–5} It has been revealed that ICP in CM 1 is nonresponsive to medical treatment and decompression surgery is more likely to be successful.^{6,7} Therefore, in order to avoid the redundant surgical treatment accurate diagnosis of papilledema is essential in these patients.

Optic disc drusen (ODD) is one of the most common causes of pseudopapilledema and can be easily misdiagnosed as papilledema in ophthalmology practice.^{8,9} Ultrasonography (USG) and optical

coherence tomography (OCT) were claimed as reliable devices in the differential diagnosis of these two disorders.^{10,11} In this report we aimed to present a case with CM 1 and pseudopapilledema caused by ODD.

Case presentation

A 32-year-old female was referred to the neurosurgery department with the diagnosis of CM 1 and papilledema. She was suffering from CM1 (Figure 1) and syringomyelia for 8 years, and she had been diagnosed with papilledema for 1 month. She was consulted to the ophthalmology department for papilledema. She had a previous ophthalmology consultation because of bilateral blurred vision where papilledema was noted. After papilledema diagnosis, she was referred to the neurosurgery clinic as she already had CM1 diagnosis. On examination, the patient's best-corrected visual acuity was 20/20 in the right eye and 20/20 in the left eye. Spherical refractive error of the patient was –1.00 D in both eyes. Optic nerve function tests, including color vision, 30–2 Humphrey threshold visual field test (Humphrey Instruments, San Leandro, CA, USA) (Figure 2), and pupillary light reactions were

Figure 1. Sagittal T-2 weighted brain MRI image of the case.

Figure 2. 30-2 Humphrey threshold visual field test images of the right (a) and left (b) eyes.

all normal in both eyes. Biomicroscopic anterior segment examination revealed normal. Dilated fundus examination showed an irregular elevation of the optic nerve head with blurry margins in both eyes (Figure 3). Peripapillary retinal vessel tortuosity was present on the right eye. Spontaneous venous pulsations were seen on both optic nerve heads. Peripapillary retinal hemorrhages, cotton-wool spots or blurring of the nerve fiber layer was not present. B-scan USG and Spectral domain OCT (SD-OCT, Optovue, Inc, Fremont, California) were performed (Figures 4 and 5). The diagnosis of ODD was established in her both eyes.

Conclusions

Papilledema has been reported rarely with CM 1, and if it exists, it indicates the presence of intracranial hypertension^{1,4,5,12–15} Milhorat and associates reported the clinical findings of 364 symptomatic CM 1 patients. Papilledema was present in nine (2%).¹ Seven of the nine patients had normal-sized ventricles without any evidence of hydrocephalus.¹ Vrabec and associates reported a case with CM 1 who developed papilledema associated with intermittent obstructive hydrocephalus.¹⁶ ODD can accompany with some ocular and systemic pathologies and it is the first entity to be considered in the differential

Figure 3. Colour fundus photographs of the right (a) and left (b) eyes. Blurred and elevated optic disc margins in both eyes.

Figure 4. B-mode ultrasound images of the right (a) and left (b) eyes. Bright echogenic area at the papilla with acoustic shadowing is seen in both eyes.

Figure 5. OCT images of the right (a) and left (b) eyes. Elevated optic nerve head with a lumpy internal contour and shadowing in the OCT signal.

diagnosis of pseudopapilledema. During patient evaluation, differentiation of optic disc edema from ODD is crucial. Examination techniques commonly performed in the diagnosis of ODD are B-scan USG, fluorescein angiography, and CT.^{10,11,17} USG is superior to other methods due to its practical and highly diagnostic characteristic. Application of OCT, a non-invasive imaging technique that creates images closely resembling histologic sections, has achieved increasing popularity in differentiating optic disc edema from ODD.¹⁸ OCT is an important tool in patients with CM1 both for the differential diagnosis of papilledema and pseudopapilledema and for the screening of the probable retinal nerve fiber layer thickness changes.¹⁹

The CSF flow has been demonstrated as abnormal in patients with CM1.^{1–3} Patients with CM1 has diminished anterior-posterior diameter of the CSF space at the FM, decreased CSF flow rates at the FM, and increased cervical CSF and pulse pressures. This altered CSF flow may lead to an increase in ICP by mechanical obstruction of outflow at the FM region. This obstruction can be welded from the compression of draining veins in the crowded posterior fossa leading to an increase in the brain turgor or from the sagging of ectopic cerebellar tonsils.^{20,21} It is thought that the disruption of normal CSF flow leads to a pressure gradient between the intracranial and spinal spaces, and causes cerebellar herniation and syringomyelia.⁷ Thus, lumbar puncture can worsen herniation and also the results are not accurate in patients with CM1. Vaphiades et al. presented a series of four patients with papilledema, increased ICP, normal ventricles, and CM1 (prelumbar puncture), all of whom failed to respond to pseudotumor cerebri medical therapy but responded to CM1 surgical treatment.⁶ As these patients require surgical treatment, the differential diagnosis of pseudopapilledema including ODD should be made before making the diagnosis of papilledema in patients with CM1.

In this article, we aimed to draw attention to ODD in patients with CM1. The diagnosis of pseudopapilledema should be considered when bilateral optic disc edema is present in patients with CM1 in order to prevent these patients from unnecessary surgical procedures. There is no reported association between the embryologic origins of CM1 and ODD in the literature. Therefore, the association

between these two disorders may be coincidental or there may be complex molecular mechanisms that need to be defined with further studies.

Declaration of interest

The authors declare that they have no competing interests.

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Consent for publication

Written informed consents were obtained from the patient for publication of this Case Report and any accompanying images. A copy of the written consent is available for review by the editor of this journal.

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