Case Report

Intermittent and Unilateral Chorioretinal Folds due to Combined Chiari 1 Malformation and Basilar Invagination

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Keywords
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Abstract
We report the case of a 35-year-old female with combined Chiari 1 malformation and basilar invagination, who presented with intermittent conjunctival chemosis and unilateral chorioretinal folds that were temporally correlated. She denied any flashes, floaters, eye redness, or pain. She also denied nausea or vomiting. Clinical exam and optical coherence tomography imaging revealed conjunctival chemosis and chorioretinal folds in the left eye. Subsequent magnetic resonance imaging of the brain and the orbits were consistent with combined Chiari 1 malformation and basilar invagination. The unilateral and intermittent chorioretinal folds and conjunctival chemosis presentation of combined Chiari 1 malformation and basilar invagination is unusual. To the best of our knowledge, this is the first case to be reported with this unique clinical presentation. It is most important to be aware that unilateral and intermittent chorioretinal folds associated with conjunctival chemosis may be signs of intracranial disease.
Introduction

Chorioretinal folds (CRFs) are undulations of the choroid, Bruch's membrane, retinal pigment epithelium (RPE), and the overlying neurosensory retina [1]. Friberg provided a biomechanical analysis that explained the development of CRFs as a result of the stress-and-strain relationship that occurs between the sclera and choroid, with a reduction in the area of the inner lining of the sclera, resulting in a buckling force affecting the choroid from either scleral thickening or shrinkage [2].

On clinical examination, CRFs have a characteristic appearance seen as alternating light and dark bands on ophthalmoscopy. The crests of the folds appear hypopigmented, corresponding to the areas of thinned RPE, whereas the troughs appear dark, corresponding to the areas of compressed RPE [3].

Many diseases have been ascribed to cause CRFs that can be categorized as ocular or extraocular. These include choroidal tumors, hypotony, inflammatory conditions of the orbit, neovascular membrane, retrobulbar mass, papilledema, and extraocular hardware [4].

Basilar invagination (BI) is an occipitocervical malformation characterized by odontoid apophysis displacement of the axis inwards towards the foramen magnum [5]. Chiari 1 malformation (CM1) is a common associate of BI and is the soft tissue component of dysgenesis [5]. CM1 is a rare congenital disorder recognized by caudal displacement of the cerebellar tonsils through the foramen magnum into the cervical canal [6]. Studies have shown that both CM1 and BI share common pathomechanisms involving compromised cerebrospinal fluid (CSF) dynamics at the craniocervical junction [7].

The complex symptom patterns for CM1 isolated or combined with BI are usually late-onset and are related to increased intracranial pressure, or the presence of an associated syrinx and/or the compression of neural structures at the cervicomedullary junction by the herniated tonsils [7]. The most common presenting symptom of CM1 is suboccipital headache, which has been reported in 81% of patients and is classically exacerbated by Valsalva maneuvers [8]. Symptoms involving ocular function consist of pseudotumor-like episodes such as blurred vision, photophobia, diplopia, retro-orbital pain, and visual field cuts that occur in 78% of patients [9].

Case Report/Case Presentation

A 35-year-old female with a history of occasional retro-orbital headache for many years presented with intermittent conjunctival swelling, photophobia, and blurring of vision of the left eye for 4 months. She has had systemic evaluation at an internal medicine clinic that included thyroid function tests, complete blood count, erythrocyte sedimentation rate (ESR), Fasting blood sugar, and the results were reported normal. Before she was referred to us, she had also visited another eye clinic where a nonspecific diagnosis (orbital pseudotumor) was entertained for which she had been treated with systemic steroids to relieve symptoms during episodic attacks.

Her best corrected visual acuity was 20/20 in the right eye and 20/100 in the left eye. Intraocular pressures using applanation tonometry were 12 mm Hg in the right and 10 mm Hg in the left eye. Anterior segment exam revealed slight lid edema and conjunctival chemosis on the left eye. Pupils were equally round and reactive to light and accommodation. Extraocular movements were normal with motility.

Dilated funduscopic evaluation of the right eye showed pink disc, blurred and slightly raised disc margin, normal foveal light reflex, and normal calibers of optic nerve head and retinal vessels (shown in Fig. 1). Dilated funduscopic evaluation of the left eye showed pink
disc, blurred and slightly raised disc margin. In the posterior pole retina, the macula was notable for fine striae of choroidal folds that gradually became wider and more coarse alternating light and dark spiral bands towards the equator and mid periphery (shown in Fig. 2, 3). OCT of the macula and disc showed pink optic disc, blurred and raised neuroretinal rims (nasal) in both eyes suggestive of Fresnel grade I papilledema (shown in Fig. 4).

Because of the association of CRFs with systemic and intracranial diseases, the patient was referred to an internist and a neurologist for evaluation. After we referred her, MRI of the brain and the orbits were done and the result was obtained which revealed cerebellar tonsillar herniation 2 cm below the foramen magnum and tip of the odontoid process projecting towards the foramen magnum suggestive of CM1 and BI (shown in Fig. 5).

Discussion

Here, we report an unusual case of a 35-year-old female with intermittent unilateral conjunctival chemosis and intermittent choroidal folds as the presenting signs of CM1 and BI. Although CM1 alone rarely causes papilledema, some patients with congenital CM1 progress to secondary intracranial hypertension (ICH) without hydrocephalus [10, 11]. Both CM1 and
BI share common pathomechanisms involving compromised CSF dynamics at the craniocervical junction [7]. The proposed mechanism is intermittent craniospinal dissociation of the CSF fluid pressure brought on by primary stagnation, not complete obstruction, of CSF flow at the foramen magnum [8, 11]. The ICH, in turn, results in elevated pressure in the optic nerve sheath and leads to papilledema due to axonal swelling [12]. Choroidal folds arise from globe compression from the distended optic nerve sheath leading to globe distortion [13]. There are no previous reports referring to the association between intermittent and unilateral CRFs and conjunctival chemosis with either isolated CM1 or combined CM1 and BI.

One or more of the following mechanisms could explain how the unilateral and intermittent CRFs are developed in the setting of raised intracranial pressure (shown in Fig. 2–5). Based on these known pieces of evidence, we think that the asymmetric findings in our case could be explained by one/or a combination of the anatomophysiologic differences described here.

1. A difference in the optic nerve sheath distention response between the two eyes to increased intracranial pressure. It has been reported that choroidal folds may precede the formation of retrolaminar hypoperfusion and papilledema [14, 15].
2. Intrinsic variations of elastic properties of the sclera that may determine how increased pressure is transmitted through the globe.

Fig. 2. Color fundus photos (a, c) of the left eye: alternating yellow and dark bands, coarse and radially oriented striations (chorioretinal folds), mostly pronounced on the temporal macula and mid-periphery. Corresponding SD-OCT fovea B-scan image (b) showing broad full-thickness chorioretinal undulations.
3. Variations of rigidity and insertion of the dural sheath may also explain choroidal folds associated or not with disc swelling [16].

4. Asymmetrical presentation could be explained by the differences in intraocular pressure between the two eyes. The relatively higher intraocular pressure in one eye may allow the globe to withstand greater scleral compression, while the relatively lower intraocular pressure in the other may predispose it to greater scleral compression (choroidal folds) as a result of the disc swelling [17]. However, in our case, the intraocular pressure difference between the two eyes was not significant.

Interestingly, the CRFs in our case were located mostly on the temporal macula and the mid-periphery which has not been previously reported. A study of retinal and choroidal folds in papilledema due to idiopathic ICH has shown that most of the retinal folds were seen in the papillo-macular bundle, and choroidal folds were usually located above and temporal to the nerve head in the posterior pole [18]. The temporal macular and mid-peripheral location of the CRFs could be related to the variation of the insertion of and structural geometry of the optic nerve sheath.

We also observed that the intermittent and unilateral conjunctival chemosis was temporally associated with CRFs. There was no evidence of allergy or infection as a cause of conjunctival edema. CSF was previously thought to only drain through the arachnoid villi and granulations, however, recent evidence suggests that CSF drains through other pathways [19]. Koh et al. [20] in their review of the literature concluded that the majority of flow of CSF is along the cranial nerves and into extracranial lymphatics. In CM1 and BI, if there is a transient increase of ICP during pressure pulses; there could be a higher flow of CSF through the subarachnoid space of the optic nerve head to the sclera and orbital cavity. This will, in turn, overwhelm the conjunctival lymphatics to drain this away and eventually develop conjunctival chemosis.

Overall, the intermittent clinical features of this case could be related to the transient elevation of ICP as a result of Valsalva maneuvers and its subsequent effect on optic nerve...
Fig. 4. Optic nerve head photo of the right (a) and left eye (c). Corresponding optical coherence tomography B-scan of right (b) and left (d) eyes: pink optic discs, blurred and raised neuroretinal rims (nasal) in both eyes suggestive of Fresnel grade I papilledema. No vascular changes are seen.

Fig. 5. MRI of brain report: Peg-like cerebellar tonsillar herniation 2 cm below the foramen magnum (CM1) as indicated by red arrow, associated with BI of the tip of odontoid process projecting 5 mm (green line) above the Chamberlain’s line (yellow line). Otherwise no sign of hydrocephalus or intracranial mass.
sheath. The absence of localizing cranial nerve deficits may indicate the absence of both chronically-elevated ICP and the absence of compression of neuronal structures. This is consistent with a report by Milhorat where 7 of 9 patients with Chiari 1 malformation and papilledema had no evidence of ventricular enlargement via MRI [8].

The waxing and waning features in our case could also be a result of the variable clinical manifestations of CM1 and BI. Whether or not some symptomatic CM1 and BI have a benign course without surgical intervention is an essential question that should be addressed for optimal management. The unilateral and intermittent CRFs associated with concurrence of conjunctival chemosis due to combined CM1 and BI are unusual. To the best of our knowledge, this is the first case to be reported with a unique and atypical clinical presentation.

**Conclusion**

It is most important to be aware that unilateral and intermittent CRFs associated with conjunctival chemosis may be signs of intracranial disease. When the etiology is unknown, brain and orbital imaging should be warranted to rule out serious conditions.

**Statement of Ethics**

This study protocol was reviewed and approved by the Ethics Review Committee of Roha Specialized Eye Clinic, approval number Roha/IRB/05/2021. Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the informed consent is available for review upon request.

**Conflict of Interest Statement**

The authors have no conflicts of interest to declare.

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**Author Contributions**

Alemu Kerie Tesfaw was responsible for analysis of the case findings, drafting the manuscript, review and editing. Nikhil N. Batra was involved in review and editing. Cong T. Phan was also involved in editing of the manuscript. Workayehu Kebede Woldegiorgis and Mulusew Asferaw Melesse have contributed in the intellectual content of this case report and were involved in the final review.

**Data Availability Statement**

All data analyzed during this case report are included in the manuscript. Further inquiries can be directed to the corresponding author.
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