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Case Report

A Brief Review of Optic Nerve Head Abnormality and Report Three Cases

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Abstract:

Congenital Optic disc anomaly have conflicting feature that sometimes it will be difficult to differentiate acquired or congenital entity of each other. This anomaly in spite of rarity but diagnosis of many congenital or acquired optic disc derangement is a diagnostic dilemma. So it need clinical and paraclinical study which in this report discuss the variety of clinical and paraclinical finding of them.

Key words: abnormalities; ophthalmology; optical coherent tomography

Introduction

One of the rare abnormalities of the optic disc is congenital optic nerve head pit. This lesion, if accompanied by macula involvement, causes progressive vision loss due to failure of the embryonic cleft to close during the developmental period in the optic nerve head.

Only 15% of cases are bilateral, and its incidence is one in 11,000 people, with no difference in the two sexes. Unilateral cases are hereditary and are seen as autosomal dominant. [1]

There is no consensus on the cause of its embryonic origin, and its physiopathology in the formation of intraretinal or sub retinal fluid is not completely clear. [2] Although the majority believe that the origin of this edema is either from the vitreous fluid or from the cerebrospinal fluid. Since a normal eye has a closed system and its different parts have a slight pressure difference between them, the pitting of the optic nerve head may cause the transfer of cerebrospinal fluid to the retina or vice versa. [2] In this report, we present three cases of optic nerve head abnormalities with clinical and pathological manifestations conflict, two of which were accompanied by maculopathy and obvious optic disc anomalies and one case with peripapillary atrophy indeed of macular edema in the macula-papilla area. So, there is diagnostic conflict which not only the clinical examination but also Para clinical examination could be done for diagnostic purpose.

Case Report

The first case is a 12-year-old boy with decreased vision. On examination, the visual acuity of the right eye was 10/10 without correction and the left eye was 10/5. On fundus examination, as seen in Figure 1, serous retinal detachment is evident as a balloon appearance, and cystic edema is also seen on optical coherent tomography (OCT). The retina of the left eye is normal, and other examinations of both eyes are normal in terms of eye examination.

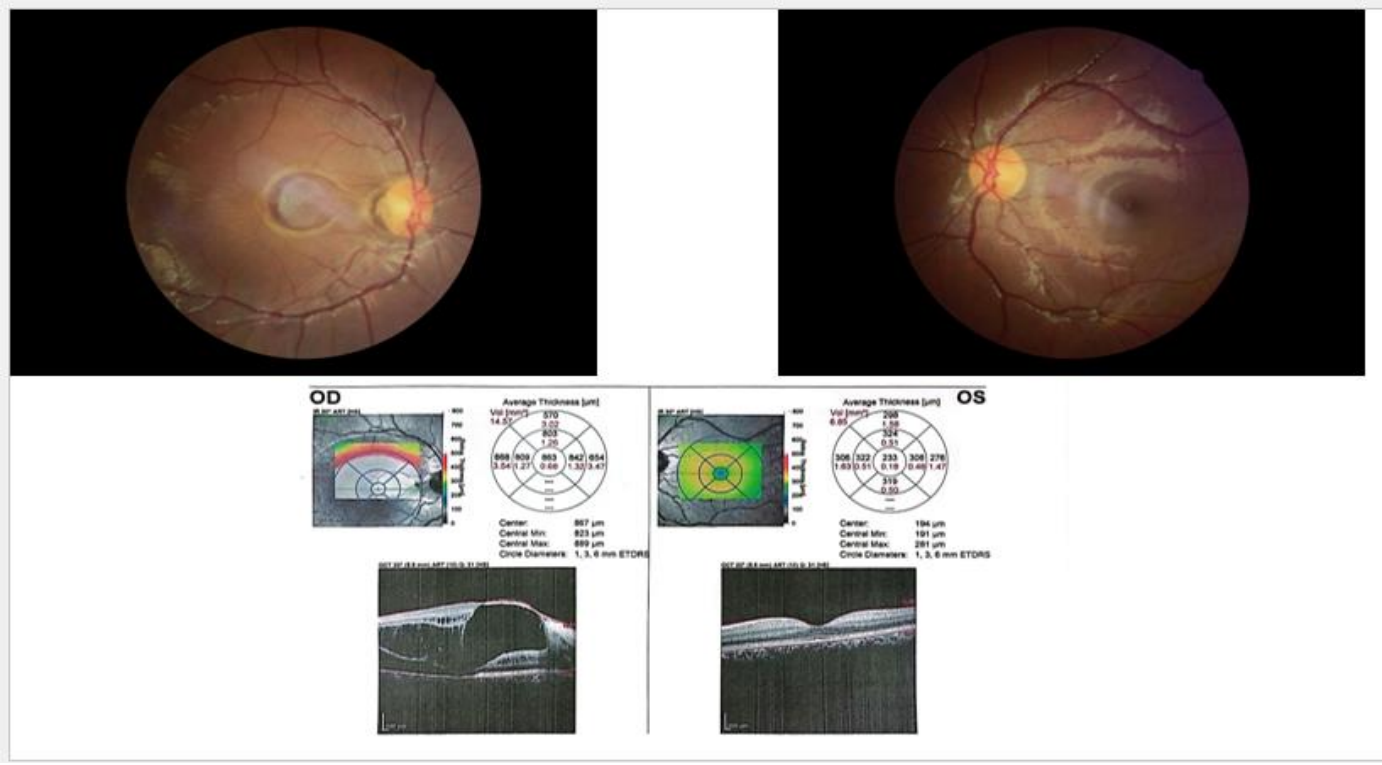

Figure 1: Fundus photo and OCT of both eye

The second case is a 10-year-old boy. The visual acuity of the right eye is 10/3 and the right eye is 10/10. The anterior segment examinations of both eyes are normal. On fundus examination, the nerve head appearance in the right eye is sunflower-shaped and the left eye also has a coloboma-like appearance.

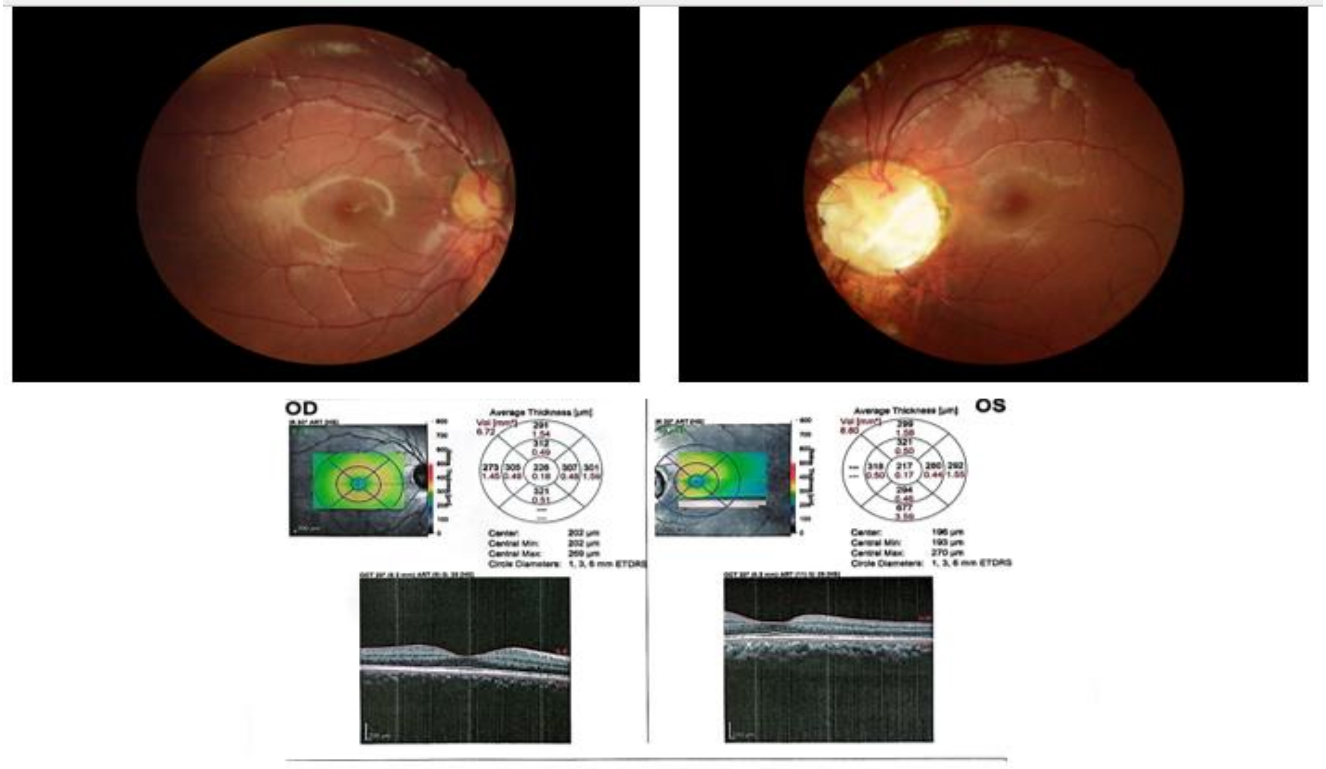

Figure 2: Fundus photo and OCT of both eye

Another case is a 45-year-old individual with peripapillary atrophy, as seen in Figure 3. There is evident prepapillary atrophy on the optic nerve head which apart from its cause has a similarity to congenital disorder resembling congenital optic disc anomaly. Although the maculopathy showed retinal edema, hemorrhage and prepapillary atrophy and cystic change in OCT which is obvious in figure 3.

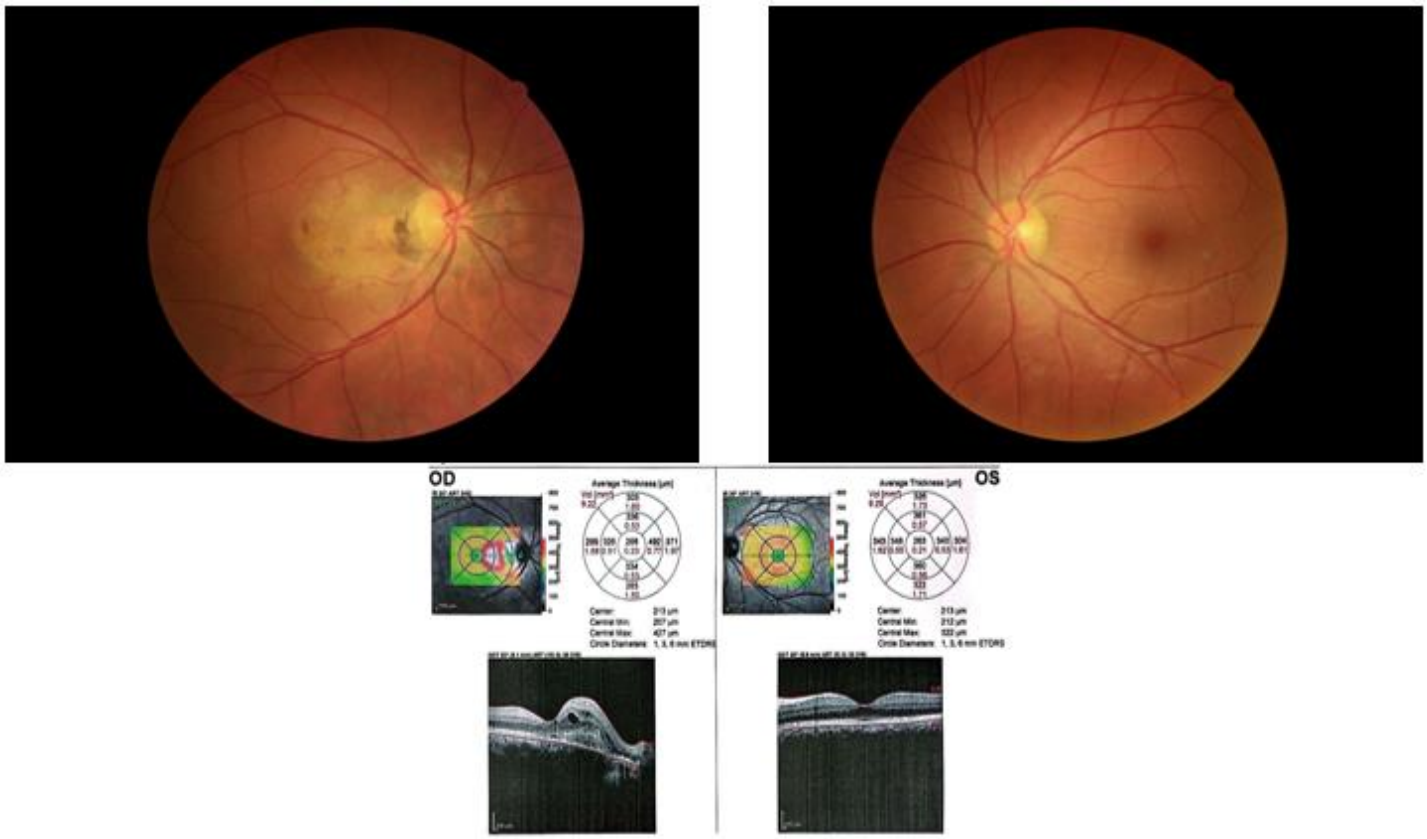

Figure 3: Fundus photo and OCT of both eye

Discussion:

Congenital optic nerve head disorders are rare and since they are most often discovered incidentally, this congenital anomaly must be distinguished from other similar lesions of the nerve head, whether acquired or congenital. Considering that retinal imaging, from fundus photography to OCT, is currently of great help in distinguishing them from each other, some of these types of anomalies have similar clinical manifestations.

In this review, we have illustrated 3 different cases of optic nerve head abnormality to be helpful when dealing with such lesion in differentiate them. So, separation and definitive diagnosis of each other plays a role in follow-up and treatment. The first case is located in the temporal part of optic disc and in the transverse line, its clinical appearance shows a transverse bell shape figure serous macula detachment clinically. The second case have a pit-like appearance which is located in the temporal region and has a view of the sunflower nerve head anomaly. The other eye has a similar view of the nerve head coloboma like abnormality clinically in fundus view. The extensive prepapillary atrophy in the third case could indicate repeated serous detachments that most probably be secondary atrophy and vascular changes. Although to find the definite diagnosis it needed more paraclinical evaluation. A study by Patel, Megha B et al supposed to differenciate such varity of optic disc anomaly. [3] Optic disc holes and morning glory disc anomaly both serve as differential diagnoses for each other. OCT showed subretinal fluid and intraretinal cystic spaces in our case. Various possibilities have been suggested such as vitreous infiltration through macular hole formation, cerebro spinal fluid infiltration from the subarachnoid space through the optic disc pit defect, leakage from blood vessels in the optic disc pit defect and fluid from the choroid, through Bruch's membrane, and peripapillary atrophy. Thus, the diagnostic dilemma between morning glory disc anomaly and optic disc pit was resolved using OCT angiography. [4]

Unfortunately, these aforementioned cases did not cooperate or did not refer despite requests for follow-up and imaging. This cases indicates the specific location of the pit is not diagnostic especially in all cases in the temporal region, unlike report 1, which was seen in the inferior temporal region. Therefore, congenital optic disc anomaly have broad spectrum that differentiation of each one needed clinical and paraclinical evaluation.

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