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CASE REPORT

Atypical Form of Congenital Excavated Anomaly of the Optic Disc With Characteristics of Peripapillary Staphyloma and Morning Glory Anomaly

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ABSTRACT
We present a 51-year-old female with a unilateral congenital excavated optic disc anomaly. After clinical examination and appropriate diagnostic procedures we were unable to determine with certainty whether it is a morning glory anomaly or a peripapillary staphyloma. The atypical finding was an optic disc with characteristics of both states. The affected eye had almost normal visual acuity (0.9 Snellen chart), which is a rare finding in congenital anomaly of the optic disc. Confocal scanning laser ophthalmoscopy (Heidelberg Retina Tomograph, HRT 3.0) was not of diagnostic value in comparison with optical coherence tomography (OCT).

KEY WORDS
Peripapillary staphyloma, morning glory, optic disc anomaly, optical coherence tomography, confocal scanning laser ophthalmoscopy (HRT 3.0).

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A 51- year-old Caucasian woman with no visual complaints went to her local ophthalmologist for an eye examination after developing peripheral paresis of the left facial nerve. Cranial CT findings were within the normal range, and an ENT specialist found no abnormality. With the exception of peripheral facial nerve paralysis, neurological findings were normal.

The patient's best-corrected visual acuity was OD: 0.9 and OS: 1.0 distance (Snellen chart) and Jaeger 1 near for both eyes. Her color vision, external examination, slit lamp biomicroscopy, intraocular pressures, and motility were all normal in both eyes. Funduscopic examination was normal in the left eye with an optic nerve cup to disc ratio of 0.2-0.3. Dilated funduscopic examination of the right eye showed significant excavation on the posterior globe, in which the optic disc was hardly recognizable. It had a poorly visible temporal border and the appearance of hyperpigmentation at the 12 o'clock position of the excavation. Blood vessels radiating from the papillary region seemed to be increased in number and appeared tortuous in the center of the excavation. The macula and periphery were normal (Figure 1).

Figure 1. Patient's right optic disc

A deep excavation was noted on an ultrasonography B-scan of the right eye. Automated perimetry showed an enlarged blind spot and a relative superior altitudinal defect on the right and a full field on the left. Optical coherence tomography (OCT) revealed a deeply seated right optic nerve and the depth of staphyloma was measured 1,2 mm. Retinal pigment epithelium showed significant atrophic changes (Figure 2). OCT of the left optic disc showed it to be normal.

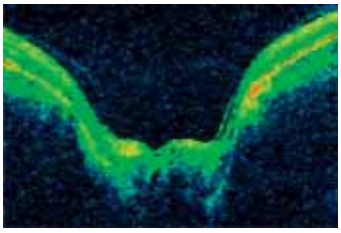

Figure 2. OCT showed a deeply seated right optic nerve

An examination of both eyes was also performed with HRT 3.0, and analysis of the left eye was completely normal. There was a problem in setting the contour line on the right eye due to the decreased visibility of the optic disc. Using the funduscopic picture of the right eye, we placed the contour line as precisely as we could. Stereometric analysis showed an enlarged disk area (3.20 mm^2) as well as an enlarged rim area (3.20 mm^2), so that the Cup/Disc area ratio was 0.00. Moorfields Regression Classification was within normal limits for all segments (Figure 3). Glaucoma probability score classification (GPS) was not classified, because the GPS model was not compatible with the shape of this optic nerve head.

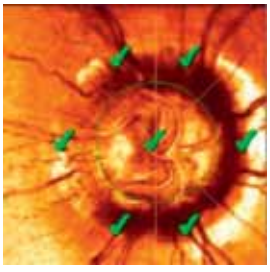

Figure 3. Moorfields Regression Classification on HRTwas within normal limits globally and in all segments

After completion of all diagnostic procedures, we concluded that this atypical optic disc is a congenital anomaly, most likely mild form. Its adequate function was confirmed by the fact that the patient had almost normal visual acuity. However, we were unable to determine with certainty whether the anomaly is a peripapillary staphyloma or a morning glory anomaly, because the atypical optic disc had characteristics of both conditions.

Discussion

Congenital excavated optic disc anomalies include optic disc coloboma, morning glory disc anomaly, peripapillary staphyloma, megalopapilla, and optic pit. These are all extremely rare conditions, which are most commonly are found in early childhood when they cause decreased vision, strabismus and nystagmus. In both morning glory disc anomaly and peripapillary staphyloma, an excavation of the posterior globe surrounds and incorporates the optic disc. Usually, these conditions are associated with unilateral appearing. Visual acuity in the involved eye may be minimally or severely affected, depending on the extent of lesion. These disc anomalies may be associated with other congenital disorders of the eye; often they accompany central nervous system malformations[1] or renal hypodysplasia, where they are part of an autosomal dominant condition called renal coloboma syndrome (RCS) or papillorenal syndrome[2]. In addition, these optic disc anomalies may be associated with retinal detachment, retinochisis, macular edema, choroidal neovascularisation and lipid exudation. Rarely are they associated with the optic disc contractility[3].

In peripapillary staphyloma the area around the disc is deeply excavated, with atrophic changes in the retinal pigment epithelium. The disc remains well-defined, relatively normal in appearance with an absence of glial and vascular anomalies[1]. As opposed to the morning glory disc anomaly, the blood vessels in the peripapillary staphyloma lesion have a normal pattern[4]. Unlike other excavated optic disc anomalies, peripapillary staphyloma is rarely associated with other congenital defects or systemic diseases[5].

Morning glory disc anomaly has a less deep funnel-shaped excavation, along with a grossly anomalous, poorly defined optic disc, including a white tuft of glial tissue that covers the central portion of the cup. Blood vessels appear to be increased in number and emanate from the edge of the disc. After arising from the disc, the vessels turn sharply at the edge of the cup and follow an abnormally straight pattern within the peripapillary region[1].

In our case, optic disc was poorly defined, deeply seated at the bottom of the excavation, without central tuft of glial tissue and with blood vessels abnormal according to the number and arrangement. Due to the atypical form of this optic disc which has the same qualities of peripapillary staphyloma and morning glory anomaly, problem appeared in establishing the diagnosis between these two conditions.

We used two important diagnostic tools for structural analysis of the optic disc: optical coherence tomography (OCT) and confocal scanning laser ophthalmoscopy (Heidelberg Retina Tomograph, HRT, 3.0)[6].We were interested in establishing their ability to distinguish optic disc head, as in our case, from a normal optic disc. OCT showed significant atrophic changes in retinal pigment epithelium and structural changes most similar to those in peripapillary staphyloma. In contrast, HRT 3.0 showed poorly ability to detect structural changes on this atypical disc.

In summary, this case presents an atypical optic disc with characteristics of both peripapillary staphyloma and morning glory anomaly. An accurate diagnosis is still unclear. Because we found no other ocular or systemic congenital disorders usually associated with morning glory disc anomaly, we believe that the malformed optic disc presented in our case is likely a variant of peripapillary staphyloma. The retention of good visual acuity by our patient is rare in cases like this, so we believe that it is a mild

form . Our findings of visual field, coupled with results from ultrasonography and OCT are important for understanding the structure and function of the optic nerve like this as well as for further monitoring. Finally, we assert that HRT 3.0 is not useful for differentiation of abnormal from normal optic discs in cases like this.

Contributors

BM has performed examination of anterior segment, ultrasound, examination of color vision, analysis of visual field test and HRT examination. MM has performed OCT examination and was consultant on writing this article. EI has performed dilated funduscopic examination and made fundus photography.

Conflict of interest

We declare that we have no conflicts of interest.

References

1. Brodsky MC. Congenital optic disc anomalies. In: Yanoff M, Duker JS, eds. Ophthalmology. 3rd ed. Philadelphia: Mosby; 2009:956-9.
2. Schimmenti LA. Renal coloboma syndrome. Eur J Hum Genet 2011;19:1207-12.
3. Sawada Y, Fujiwara T, Yoshitomi T. Morning glory disc anomaly with contractile movements. Graefes Arch Clin Exp Ophthalmol 2012; 250:1693-5.
4. Kim SH, Choi MY, Yu YS, et al. Peripapillary Staphyloma: Clinical Features and Visual Outcome in 19 cases. Arch Ophthalmol 2005; 123:1371-6.
5. Blair M P, Blair N P, Rheinstrom S D. *et al*. A case of peripapillary staphyloma. Arch Ophthalmol 2000;118:1138–9.
6. Yang B, Ye C, Yu M, et al. Optic disc imaging with spectral-domain optical coherence tomography: variability and agreement study with Heidelberg retinal tomography. Ophthalmology 2012; 119:1852-7.

Atipični oblik kongenitalne ekskavacije optičkog diska s karakteristikom peripapilarnog stafiloma i *morning glory* anomalije

APSTRAKT

Prikazan je slučaj 51-godišnje pacijentice sa unilateralnom kongenitalnom anomalijom optičkog diska. Kliničkim pregledom i učinjenim dijagnostičkim pretragama nije se moglo sa sigurnošću utvrditi da li se radi o peripapilarnom stafilomu ili o morning glory anomaliji optičkog diska zbog atipičnog javljanja papile sa karakteristikama oba stanja. Ustanovljena je gotovo normalna vidna oštrina zahvaćenog oka (0,9 kuke po Snellen-u), što je izuzetno rijedak nalaz u slučaju sa prisutnom kongenitalnom anomalijom optičkog diska poput ove. Konfokalna skening laser oftalmoskopija (Heidelberg Retina Tomograph, HRT 3.0) se nije pokazala od dijagnostičkog značaja u ovom slučaju, za razliku od optičke koherentne tomografije (OCT).

KLJUČNE REČI

Peripapilarni stafilom, morning glory anomalija, optička koherentna tomografija, konfokalna skening laser oftalmoskopija (HRT 3.0)