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Congenital optic disc cyst concomitant with persistent hyaloid artery – a case report

Wrodzona torbiel tarczy nerwu wzrokowego współistniejąca z przetrwałą tętnicą ciała szklistego – opis pacjenta

Monika Modrzejewska[1], Ewelina Lachowicz[1], Ewa Tokarz-Sawińska[1], Wojciech Lubiński[1], Adam Koryzma[2], Anna Walecka[2], Tomasz Urasiński[3], Stanisław Zajączek[4]

[1] Department of Ophthalmology, Pomeranian Medical University in Szczecin, Poland
Head: Wojciech Lubiński, MD, PhD, Professor PUM, FEBO
[2] Department of Diagnostic Imaging and Interventional Radiology, Pomeranian Medical University in Szczecin, Poland
Head: Professor Anna Walecka, MD, PhD
[3] Clinic of Paediatrics, Haematology and Oncology, Pomeranian Medical University in Szczecin, Poland
Head: Tomasz Urasiński, MD, PhD
[4] Department Genetics and Pathomorphology, Pomeranian Medical University in Szczecin, Poland
Head: Professor Jan Lubiński, MD, PhD

Summary: The aim is to present a rare case of solitary malformation in the form of a congenital optic disc cyst concomitant with the persistent hyaloid artery. The intrabulbar congenital cyst of the optic disc partially covering the medial part of the disc was found in a 3-month old infant. B-San ultrasound confirmed the presence of the intrabulbar heterogeneous mass (7.0 x 2.5 x 5.4 mm) within the vitreous cavity and the concomitant persistent hyaloid artery was shown in Colour Doppler Imaging. The axial length of the involved eye was shorter than of the healthy one (16.68 mm vs. 18.42 mm). The magnetic resonance imaging of the head and orbits performed in the fast spin echo, spin echo and gradient echo sequences in T_1 and T_2-weighted scans revealed the intrabulbar cyst (7.0 x 2.5 x 6.4 mm), with sharp margins, whose lower part showed intense contrast enhancement. The pericerebral fluid spaces within the frontal and temporal lobes were dilated. Intrauterine toxoplasmosis, cytomegaly, protozoan and helminth infections as well as metabolic diseases were excluded. Patient leukocyte DNA *RB1* gene sequencing and negative results of mutation searching excluded retinoblastoma. In a 2-year follow-up period, regression of the mass with the absence of ophthalmic complications was noted. An important reason for the authors to present the discussed case is possible permanent impairment of visual function in patients with similar presentation of congenital peripapillary lesions.

Key words: congenital optic disc cyst, developmental anomalies of the optic disc, congenital ocular malformations.

Streszczenie: celem pracy jest przedstawienie izolowanej wady w postaci wrodzonej torbieli tarczy nerwu wzrokowego występującej z przetrwałą tętnicą ciała szklistego. W badaniu okulistycznym dna oka prawego, wykonanym w 3. miesiącu życia dziecka, stwierdzono obecność wewnątrzgałkowej cysty zasłaniającej przyśrodkową część tarczy nerwu wzrokowego. W badaniu kolorowego USG-Doppler potwierdzono obecność heterogenicznej zmiany (7,0 x 2,5 x 5,4 mm) w komorze ciała szklistego z współistniejącą przetrwałą tętnica ciała szklistego. Badanie biometryczne wykazało mniejszą długość gałki ocznej w porównaniu do wymiaru zdrowego oka (16.68 mm *vs* 18,42 mm). Wyniki badań rezonansu magnetycznego głowy i oczodołu w sekwencjach FSE, SE, GE w czasie T_1 i T_2-zależnym ujawniły dobrze odgraniczoną wewnątrzgałkową cystę (7,0 x 2,5 x 6,4 mm) ze znacznie kontrastującą się dolną częścią zmiany oraz poszerzenie przestrzeni płynowych okołomózgowych w płatach czołowym i skroniowym. Wykluczono infekcje wewnątrzmaciczne toksoplazmozą, cytomegalią, pasożytniczą i pierwotniakową oraz choroby metaboliczne. Sekwencjonowanie genu *RB1* leukocytów pacjenta oraz negatywne wyniki oceny mutacji pozwoliły na wykluczenie retinoblastomy. Podczas 2-letniego okresu obserwacji stwierdzono regresję cysty bez powikłań okulistycznych. Istotnym powodem przedstawienia zmian w regionie tarczy nerwu wzrokowego jest możliwość trwałej utraty funkcji narządu wzroku u pacjentów z podobnymi wadami wrodzonymi okołotarczowymi.

Słowa kluczowe: torbiel wrodzona tarczy nerwu wzrokowego, rozwojowe anomalie tarczy nerwu wzrokowego, wrodzone malformacje gałki ocznej.

Background

According to the available literature, congenital ocular anomalies occur in 2–3% of live births and may result from the disturbed differentiation of ocular structures during both the embryonic and foetal stages of development (1). The origin of those pathologies might be due to genetic factors and irregularities in differentiation of ophthalmic cup just after closing of foetal fissure or neural crest at 6–7 gestational weeks (0–7/10000 live births) (2, 3). In etiopathogenesis of developmental malformations, teratogenic and environmental factors are enumerated, including: infections, pharmacological, physical and chemical compounds, toxins, electromagnetic radiation, medicaments,

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alcohol and mother's diabetes (4). In 50% of cases, etiological factor remains unknown (2, 5). Orbital cysts occurring in association with optic disc colobomata and microphthalmic eyes have rarely been reported in the literature so far (2, 3). The first review of congenital optic disc cyst incidence in fissure region was noted in 1884 by Dimmer in "Dr. Knapp's Archives" and by Risley in 1896 (6). However, morphological and main histopathological characteristics similar to the ones described in this paper, were reported only by Christopher Pedler in 1961 (3, 7, 8).

Due to a very low prevalence of congenital developmental pathology of the optic disc region, as well as a limited number of publications addressing this issue, the following report describes a cyst localized at the optic nerve head entrance concomitant with persistent hyaloid artery.

Case presentation

The ophthalmic examination of a 3-month old, full term male infant born at 38 gestational weeks with birth weight of 2680 g, Apgar Scale 10 revealed the congenital optic disc malformation in right eye fundus. The prenatal history was normal – gravida I, para I, no perinatal complications were observed. A detailed assessment with the Ret-Cam II device showed the presence of a spherical cyst covering the nasal part of the optic disc, and adjacent retina, with venous beading on the surface. The widespread hyperpigmented areas and single vitreo-retinal connective bands were observed in the peripheral portion of the lesion (Fig. 1.).

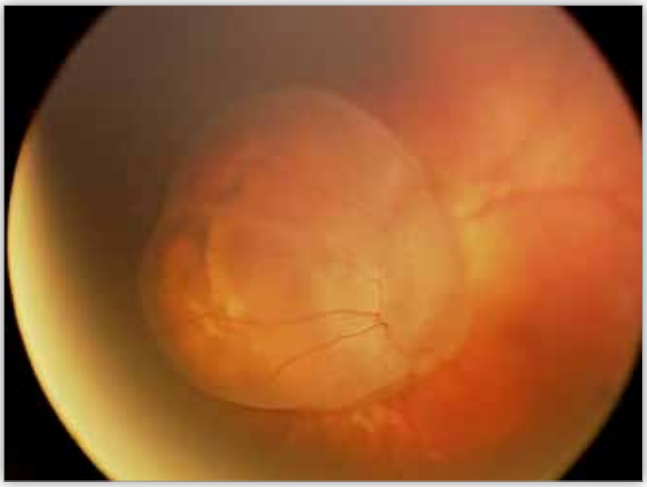

Fig. 1. Congenital cyst of optic disc in a 3-month old infant in Ret-Cam II visualisation.
Ryc. 1. Wrodzona torbiel tarczy n. II w badaniu Ret-Cam II u 3-miesięcznego niemowlęcia.

After a two-year ophthalmic follow-up, cyst regression and volume reduction were observed in Ret-Cam II images, leaving the hyperpigmented patches in those retinal areas, where the lesion resolved (Fig. 2.).

Hyper- and hypoechogenic mass sized approx. 7.0 mm x 2.5 mm x 5.4 mm was seen in the ultrasound examination (B scanUSG-B; Eyecubed apparatus, 10.0 MHz frequency probe). Additional abnormal echogenic structure continuing from the surface of the cyst towards vitreous cavity was disclosed. It moved vigorously during ocular movement and arterial blood flow in CDI (Voluson apparatus, 10.5 MHz frequency probe) was noted. Moreover, obliteration of that vessel occurred followed by its gradual involution (lack of Doppler flow) (Fig. 3.).

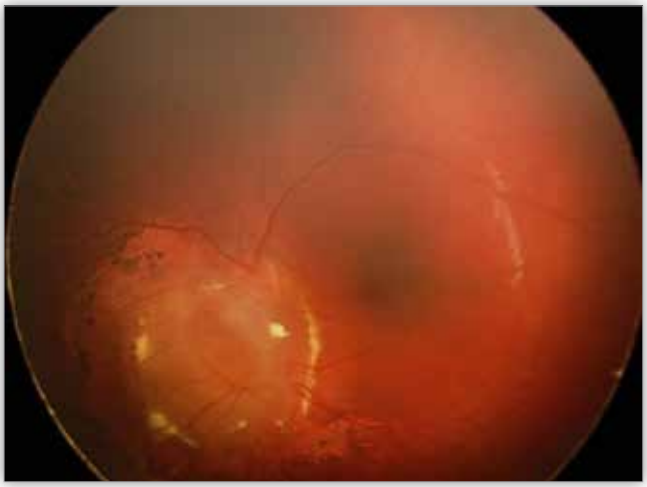

Fig. 2. Congenital optic disc cyst in Ret-Cam II visualization after a 2-year follow-up.
Ryc. 2. Wrodzona torbiel tarczy n. II w badaniu Ret-Cam II po 2 latach obserwacji.

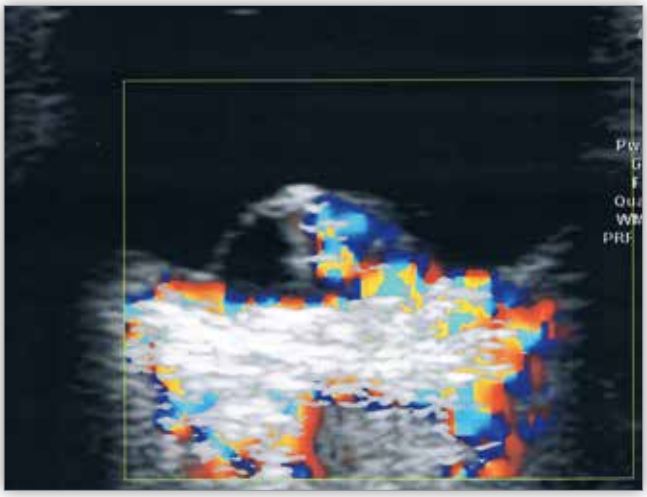

Fig. 3. Congenital optic disc cyst concomitant with persistent hyaloid artery in color Doppler ultrasonography.
Ryc. 3. Wrodzona torbiel tarczy n. II współistniejąca z przetrwałą tętnicą ciała szklistego w badaniu kolorowego USG Doppler.

The axial length of the affected eyeball was shorter than of another one as well shorter than normal length for this age (16.68 mm vs. 18.42 mm). Hypermetropia and astigmatism (+7.0 Dsph, -2.0 Dcyl) were observed in basic ophthalmic examination, with no detected abnormalities in the anterior segment and intraocular pressure measurement (I-Care inductive tonometer, 10.2 mmHg). Direct and indirect reactions of the pupils to light were correct. MRI of the head and orbits performed in FSE, SE and GE sequences in T_1 and T_2-weighted scans after administering a contrast agent showed intrabulbar solid cyst of the right eye of the following size: 7.0 mm wide, 2.5 mm deep, 6.4 mm high, whose lower part depicted strong contrast enhancement. Furthermore, medium widening of pericerebral fluid spaces in the frontal and temporal lobes was present. Other pathological changes including focal lesions in white and gray matter of the brain were not detected (Fig. 4.).

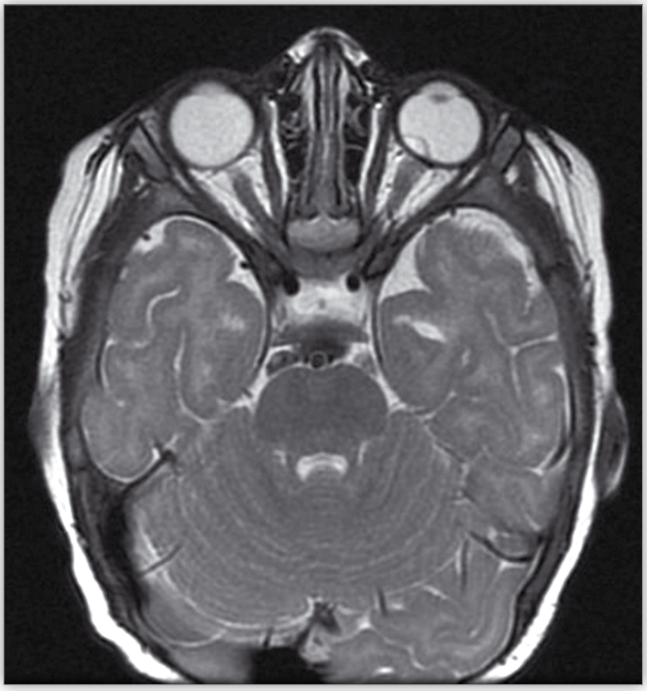

Fig. 4. Optic disc cyst as seen in contrast enhanced MRI.
Ryc. 4. Torbiel tarczy n. II w badaniu RM.

In the laboratory tests, intrauterine toxoplasmosis, cytomegaly and protozoan infections, as well as the presence of helminthiasis were excluded. Moreover, orthopaedic, neurologic, cardiologic and urologic consultation did not reveal any abnormalities. Screening towards genetic origin of metabolic diseases did not indicate any disorders. Normal constitutional karyotype from peripheral lymphocyte culture was detected. Patient leukocyte DNA *RB1* gene sequencing and negative results of mutation searching excluded retinoblastoma. Medical history revealed bleeding from vaginal canal at 9th and 14th week of pregnancy. However, other risk factors of a cyst formation were excluded. It was not possible to obtain histopathological evaluation of the cyst due to its localization.

Discussion

Developmental anomalies of the optic disc region are divided into two groups: one in which failure of coaptation occurs within the optic nerve sheath and the other in which lesions are present in the peripapillary choroid or sclera. The last mentioned are formed by imperfect closure of the fetal fissure with the result that they appear on a line running inferiorly from the rim of the optic disc to the equator, which contributes to the cyst formation (8). Another hypothesis of optic disc cyst formation is widening of primary nerve sheath as a result of congenital cavity in eyeball structure in the region of optic nerve entrance and disturbance of cerebrospinal fluid flow through abnormally differentiated tissue in the area of fovea or optic disc fissure (9, 10). Elevated intracranial and venous pressure may have an impact on the discussed pathology development, and their fluctuations are dependent on the change of body position, contrary to typical meningocele that mainly penetrate towards orbit (10, 11). Morphology evaluation of the described peripapillary lesion might suggest the presence of congenital cyst mentioned by Pedler, however the lack of possibility to perform of other examinations, including histological one, made it impossible to establish a detailed diagnosis (8). The remaining parts of the retina including macular region were normal, and the fact, that the cyst did not cover them, prevented development of squint.

In the areas where the cyst resolved, the generalized retinal atrophy with hyperpigmentation and hypovascularization may develop, which would be compliant with Pedler's findings. It should be noted that the persistent hyalorid artery was shown on the lower part of the cyst, which was confirmed both by the CDI as well as the MRI scans. Up to date, no explanations of those two pathologies can be found in the available literature. The cyst-like expansion of the involved vessel cannot, however, be excluded. There are reports describing glial debris seen within the vitreous cavity, anteriorly to the nerve head and the persistent hyaloid artery pulsating in harmony with the cardiac cycle (12, 13) or congenital optic pit associated with retrobulbar cyst and vitreous pulsations (14). We may speculate that the anomaly seen in our patient might be a part of an anomalous Cloquet's canal due to the presence of arterial flow in residual vessel within the lesion. A typical coloboma of the optic disc is a nasally situated congenital optic disc excavation. The optic disc pits are classically small and temporally located, but they appear to exist in line with a spectrum of congenital cavitary disc anomalies that are often referred to as a typical optic nerve colobomas (10). The concomitance of congenital optic disc anomalies and optic nerve or orbital cyst is extremely rare (3, 7). In patients with congenital optic disc pit and PHPVB (persistent hyperplastic primary vitreous body), malformations of other organs might coexist e.g. CHARGE or VATER association.

Lack of other pathological MRI findings excluded the diagnosis of optic nerve head meningocele (ONHM) and other lesions such as optic nerve sheath meningioma, empty sella syndrome, optic nerve glioma and neurofibromatosis. All these lesions may lead to the vision impairment or loss, as a result of nerve compression by the cyst (15). The natural history of this condition is not clearly known and it is difficult to predict in the early stages. In single cases of significant vision loss, surgical decompression of the optic nerve sheath should be considered. However,optimum treatment protocols have not been proposed yet (9, 10). In the presented case, the 2-year follow-up of the lesion suggests its regression, with the absence of ophthalmic complications probably due to its nasal location.

Conclusion

An important reason for the authors to present the discussed case is the possible permanent impairment of visual function in patients with similar presentation of congenital peripapillary lesions.

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Reprint requests to (Adres do korespondencji):
Monika Modrzejewska, MD, PhD,
Department of Ophthalmology, Pomeranian Medical University in Szczecin, Poland
al. Powstańców Wlkp. 72
70-111 Szczecin
e-mail: monika_modrzej@op.pl