Pseudoglaucomatous Cupping in A Young Adult with Cerebral Palsy/Periventricular Leukomalacia: A Brief Review of the Literature

Serebral Palsili (Periventriküler Lökomalazi) Genç Erişkin Bir Hastada Psödoglokomatöz Çanaklaşma

Murat KÜÇÜKEVCİLİOĞLU[1], Atilla BAYER[2], Tarkan MUMCUOĞLU[3], Yusuf UYSAL[3]

Case Report *Olgu Sunumu*

ABSTRACT

Pseudoglaucomatous cupping of the optic disc can be the result of many ocular or nonocular conditions. Herein we report a young adult with a rare etiology of optic disc cupping. He was referred to our clinic for bilateral low vision. Ophthalmic examination revealed bilateral peripapillary atrophy, temporal pallor and excavation of the optic nerve head in his right eye. Intraocular pressure measurements at initial presentation and diurnal followup were within normal limits. Gonioscopy revealed bilateral open angles. Optic coherence tomography of optic discs revealed bilateral thinning of the retinal nerve fiber layer in the upper and upper temporal quadrants. On Heidelberg Retinal Tomography examination, topographic measurements were abnormal in his right eye. After performing standard automated perimetry we noticed a lower hemispheric visual field defect crossing the horizontal line and advancing to right upper quadrant in his both eyes. This type of a visual field defect evoked our suspicion about some kind of a central nervous system pathology. Neuroimaging revealed left periventricular (parietooccipital) and right occipital chronic infarction field consistent with the term "periventricular leukomalacia".

Key Words: Cerebral palsy, periventricular leukomalacia, pseudo-glaucomatous cupping, retrograde transsynaptic degeneration, temporal pallor.

ÖZ

Optik diskin psödoglokomatöz çanaklaşması göze ait olan veya olmayan birçok nedenden dolayı olabilir. Biz bu yazıda çok nadir bir etyolojiye bağlı optik disk çukurlaşması olan genç bir erkek olguyu bildirmek istedik. Olgu kliniğimize her iki gözde görme azlığı nedeniyle sevk edilmişti. Oftalmik muayenede bilateral peripapiller atrofi ve sağ gözde optik disk temporal solukluğu ve optik sinir başı çanaklaşması mevcuttu. Başlangıç ve günlük takiplerdeki göz içi basınç ölçümleri normal sınırlar içerisindeydi. Açı muayenesi her iki gözde doğaldı. Optik diskin optik koherens tomografi incelemesinde her iki gözde üst ve üst temporal kadranlarda retina sinir lifi tabakasında incelme mevcuttu. Heidelberg retinal tomografi incelemesinde ise topografik ölçümler sağ gözde patolojikti. Standart otomatize perimetri incelemesi sonrası her iki gözde ağırlıklı olarak alt hemisferi tutan ve horizontal hattı geçerek sağ üst kadrana uzanan görme alanı defekti izlendi. Bu tip bir görme alanı defekti olası bir santral sinir sistemi patolojisi açısından bizi şüphelendirdi. Sonrasında yapılan santral sinir sistemi radyolojik görüntülemesi periventriküler lökomalazi ile uyumlu sol periventriküler (parietooksipital) ve sağ oksipital kronik enfarkt alanını ortaya çıkardı.

Anahtar Kelimeler: Periventriküler lökomalazi, psödoglokomatöz çanaklaşma, retrograd transsinaptik dejenerasyon, serebral palsi, temporal solukluk.

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1- GATA Askeri Hastanesi, Göz Hastalıkları Anabilim Dalı Ankara, Uz. Dr.
2- GATA Askeri Hastanesi, Göz Hastalıkları Anabilim Dalı Ankara, Prof. Dr.
3- GATA Askeri Hastanesi, Göz Hastalıkları Anabilim Dalı Ankara, Doç. Dr.

1- Gülhane Military Medical Academy, Department of Ophthalmology, Ankara/TURKEY
KÜÇÜKEVCİLİOĞLU M., doctorminik@yahoo.com
2- Gülhane Military Medical Academy, Department of Ophthalmology, Ankara/TURKEY
BAYER A., atillabayer@hotmail.com
3- Gülhane Military Medical Academy, Department of Ophthalmology, Ankara/TURKEY
MUMCUOĞLU T., tarkanmumcuoglu@yahoo.com
UYSAL Y., yuysal002@yahoo.com
Correspondence: M.D., Murat KÜÇÜKEVCİLİOĞLU
Gülhane Military Medical Academy, Department of Ophthalmology, Ankara/TURKEY

INTRODUCTION

Glaucoma is the leading cause for acquired excavation of the optic disc.[1] Cupping related to other optic neuropathies is not very common. Only a very limited portion of these entities are considered to have an appearance that may resemble glaucomatous excavation.

Some examples of optic disc cupping (ODC) unrelated to intraocular pressure (IOP) elevation include; congenital or physiological cupping, low-tension glaucoma, congenital optic-disc anomalies, arteritic anterior ischemic optic neuropathy and rarely posterior ischemic optic neuropathy, 'shock' optic neuropathy, traumatic optic neuropathy, hereditary (Leber's and autosomal dominant optic neuropathies), compression from fusiform aneurysms of the intracranial carotid arteries or tumors compressing the anterior visual pathway, radiation optic neuropathy and methanol poisoning.[1-12]

As a variant of optic nerve hypoplasia in children, periventricular leukomalacia (PVL) is another anomaly that can produce cupping.[7] Clinical distinction between glaucomatous and nonglaucomatous cupping is not always straightforward. But some fine details make physcians to consider that the cause is glaucoma rather than other entities.

These include vertical-total-asymmetrical cupping, thinning of the neororetinal rim, backward bowing of the lamina cribrosa, residual neuroretinal rim that is not pale, disc hemorrhage and peripapillary atrophy.[1,13]

On the other hand, findings that favor nonglaucomatous etiology are generalized or sectorial disc pallor, retinal arteriolar narrowing, retinal vascular sheathing and posterior pole exudates.[1,13] But all these findings are not 100% sensitive or spesific for these scenarios. Hence it is crucial to consider the patient history, visual field and fundoscopic findings all together before making a diagnosis. We herein report a young adult with periventricular leukomalacia causing optic disc excavation.

CASE REPORT

A twenty one year old male presented with low vision in his both eyes that had been present since his childhood. Medical history was unremarkable. Corrected visual acuity was 0.3 (snellen equivalent) in the right and 0.6 in the left eye. He had 30 prism diopters alternating exotropia, latent horizontal nystagmus and minimal dissociate vertical deviation (DVD). Pupillary light reactions were normal in his both eyes. Anterior segment and vitreous examinations were normal on slit lamp biomicroscopy. Fundoscopy revealed bilateral peripapillary atrophy and temporal pallor. Additionally excavation of the optic disc was present in his right eye (Figure 1). Gonioscopy revealed bilateral open angles. Intraocular pressure measurements at initial presentation and diurnal followup were within normal limits. Optic coherence tomograhy of peripapillary nerve fiber layer (RNFL) revealed bilateral thinning in the upper and upper temporal quadrants. On HRT (Heidelberg Retinal Tomography/Heidelberg Engineering Dossenheim-Germany) examination topographic measurements were abnormal in his right eye (Figure 2).

Visual evoked potentials were abnormal with low amplitude and impaired morphology. After performing standard automated perimetry (SAP) (Humphrey 24-2) we noticed a lower hemispheric visual field defect crossing the horizontal line and advancing to right upper quadrant congruously in both eyes (Figure 3). Along with the neuroretinal rim pallor, this type of a visual field defect was inconsistent with glaucomatous cupping and evoked our suspicion about some kind of a central nervous system (CNS) pathology.

Neuroimaging (Magnetic Resonance Imaging-MRI) revealed left parietooccipital and right occipital parasagittal chronic infarction fields associated with surrounding reactive gliosis. Also cortical atrophy on the left occipital lobe was noticed. Additionally atrium of the left lateral ventricule was dilated secondary to infarction and an arachnoid cyst (size:24-42-32 mm) was located anterior to temporal lobe in the left middle cerebral fossa (Figure 4). Neurology consultation resulted in the diagnosis of cerebral palsy.

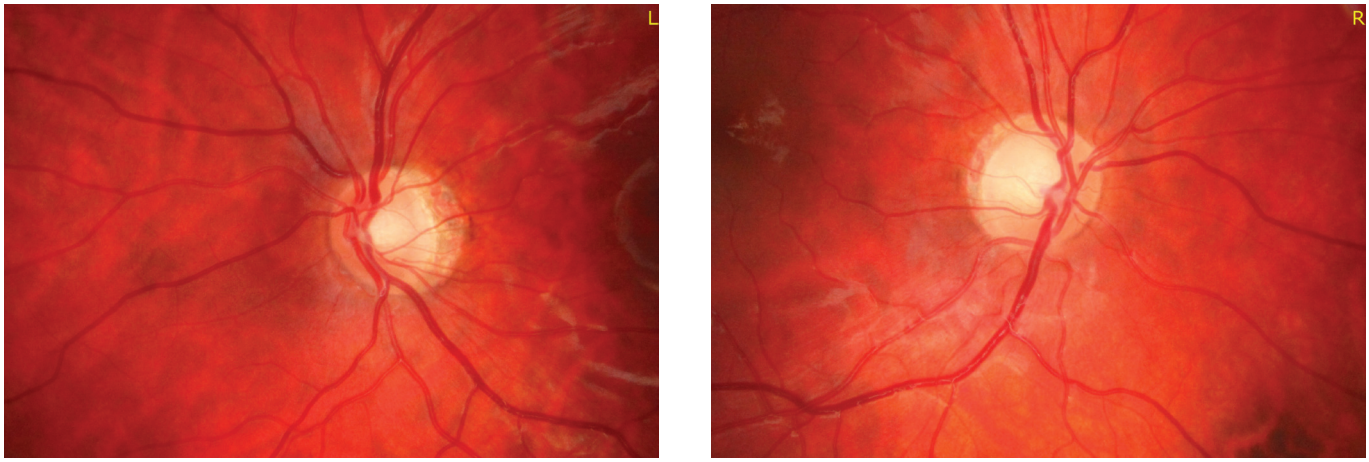

Figure 1: A fundus image of the right eye showing peripapillary atrophy, temporal pallor and thinning of upper temporal rim (left). A fundus image of the left eye showing peripapillary atrophy and temporal pallor (right).

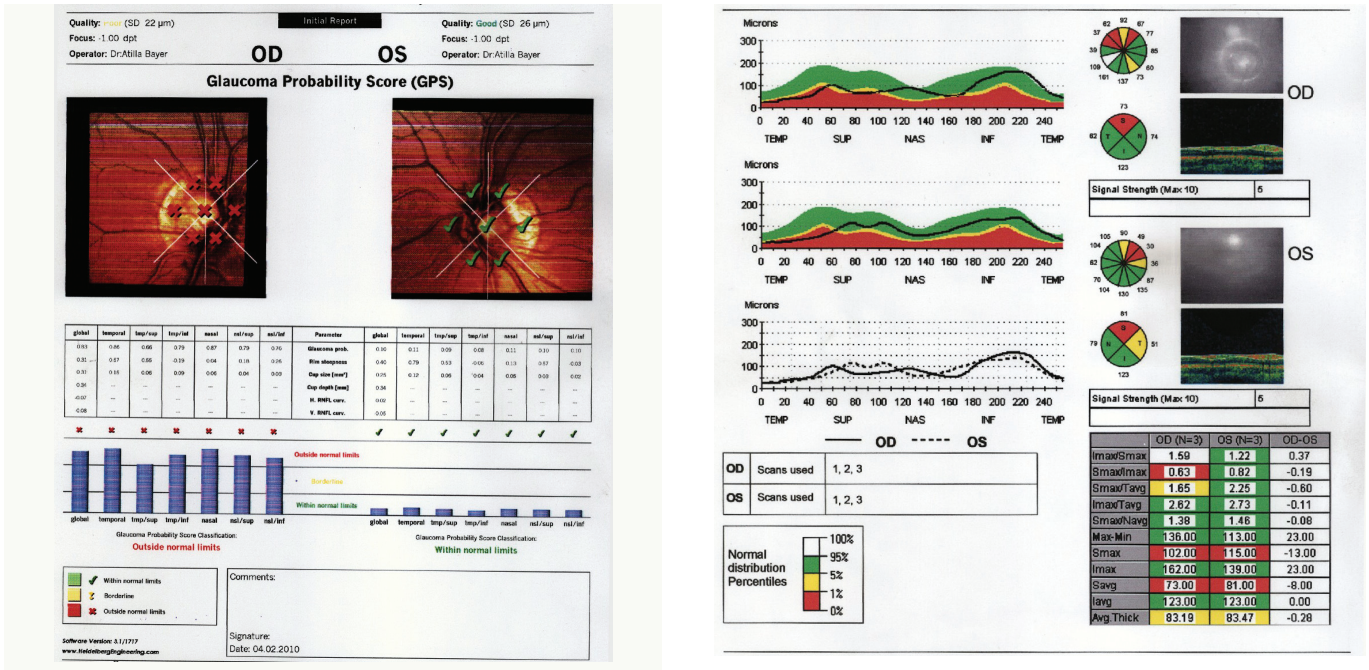

Figure 2: Both eyes showed upper temporal thinning on RNFL analysis (left). Glaucoma probability score map of HRT revealed generalized thinning of all quadrants in the right eye (right).

DISCUSSION

Cerebral palsy (CP) is a term used to describe a sum of motor syndromes caused by early brain damage. Various developmental, metabolic, genetic, infectious, ischemic and other acquired etiologies may give rise to a typical phenotype of CP.[14-16] Periventricular leukomalacia is one of the major etiologies for CP. It was firstly described by Banker and Larroche in 1962.[17] It represents softening of the white matter around the ventricles that results from extensive necrosis of white matter. The periventricular white matter includes two pathways of high importance; pyramidal tracts and the optic radiations. The impairment of these pathways results in;

1) motor symptoms (CP)

2) visual symptoms; low vision, strabismus (mostly esotropia), nystagmus (latent-manifest), crowding phenomenon, transient upgaze holding insufficiency

3) impaired cognitive function.[14-16]

Transsynaptic degeneration is the term used to describe the morphologic alterations of neurons caused by the loss of synaptic input coming from afferent neurons.[18]

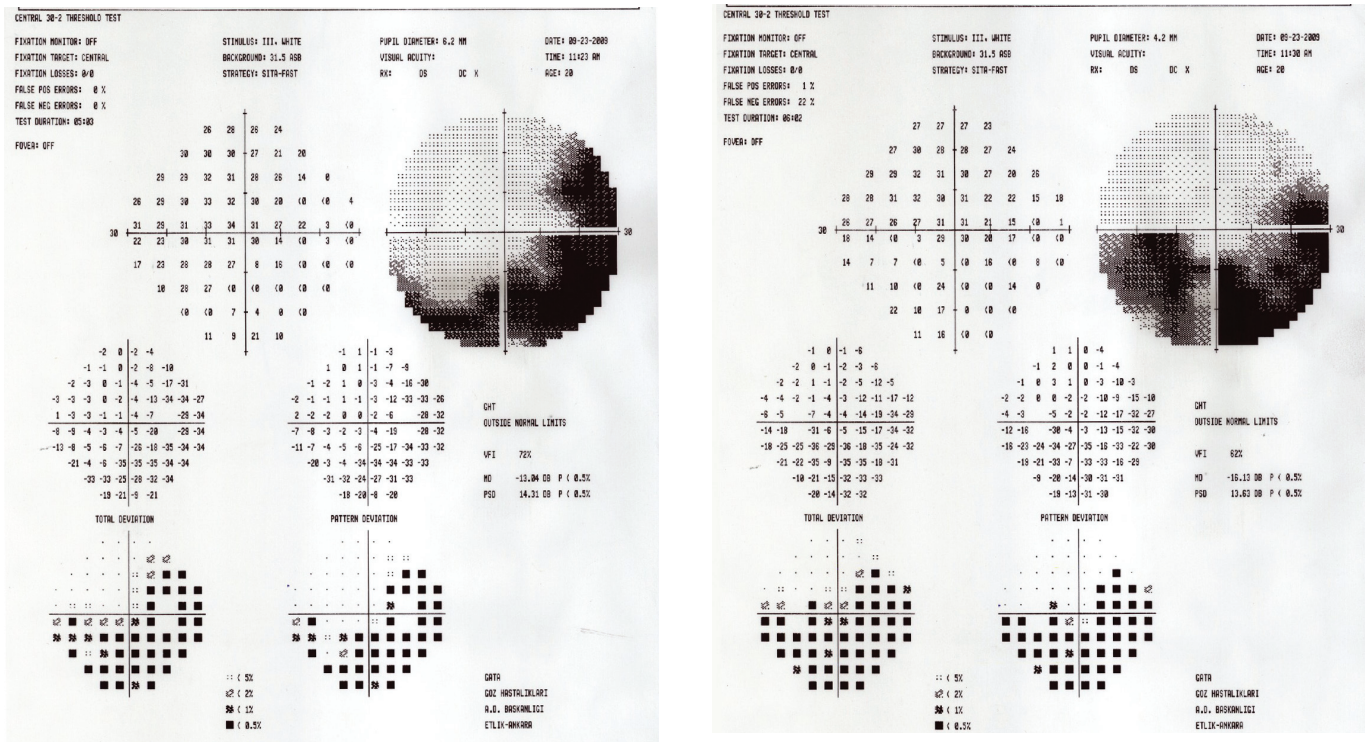

Figure 3: Predominant inferior visual field defect breaching the horizontal line in both eyes.

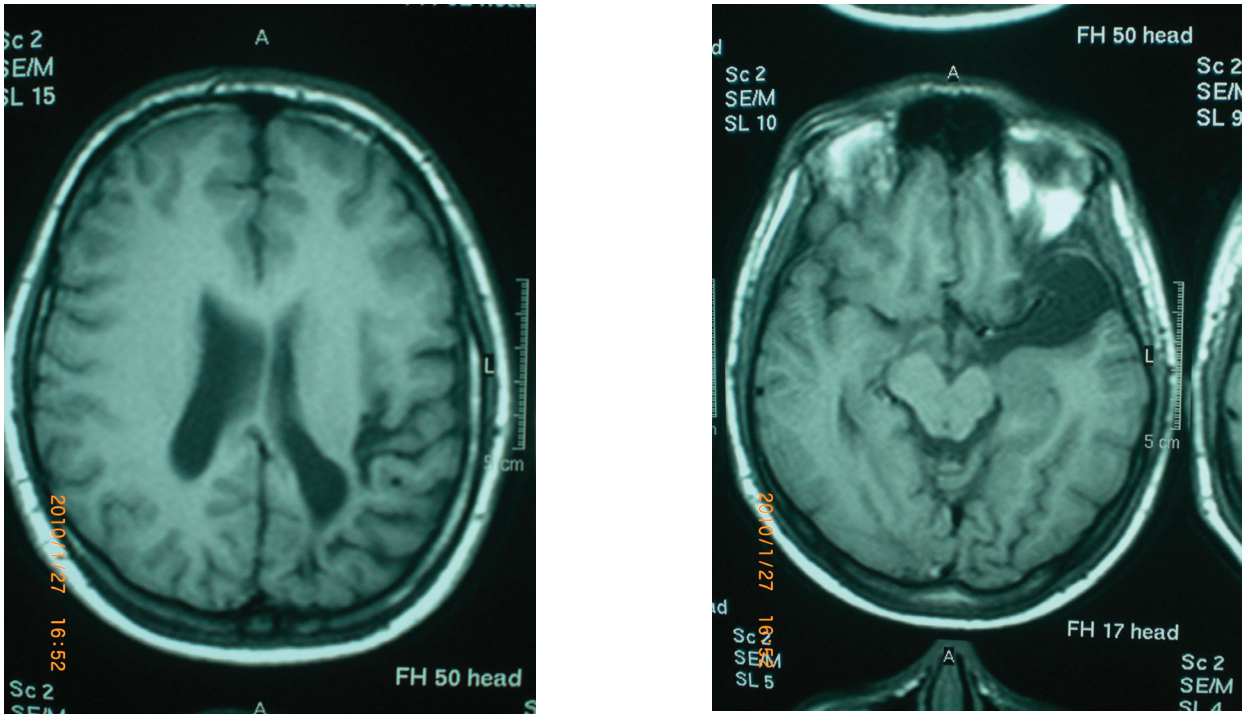

Figure 4: MRI (T1); left parietooccipital chronic infarction field and an araconhid cyst located anteriorly (left), cortical atrophy on left occipital lobe and dilated lateral ventricule posteriorly (right).

It is well demonstrated in the visual pathway, wherein cell layers of the lateral geniculate bodies are known to degenerate after loss of an eye, and in lesions of the retina, optic nerve, and optic tract.[19]

It is also known that transsynaptic degeneration may occur in a retrograde fashion like degeneration of retinal ganglion cells across the lateral geniculate body as a result of lesions of the optic radiation or calcarine cortex.[18-21] Recently this concept has been a center of interest for some researchers to explore the cerebral changes in glaucoma patients.[22,23] In preterm babies with anoxic brain damage, axonal disruption in the optic radiations may lead to transsynaptic retrograde degeneration across the geniculate body with corresponding visual field defects.[12]

The resulting loss of optic nerve axons may cause cupping. Cupping in this situation occurs when the anoxic insult occurs at a critical time between 29-34 weeks of gestation, prior to full development of the optic nerve and after the time when scleral plasticity leads to a small disc.[7,13] Most of these definitions were made for white matter injury of premature. But our patient did not have a premature or complicated labor history.

However, neuroimaging and autopsy studies showed that PVL also occurs in nearterm (late preterm) or term infants.[24] In both very preterm and late preterm infants, gray matter injury comes along with PVL. Similarly our patient had an additional cortical atrophy on the left occipital lobe on MRI. He had alternating exotropia, latent manifest nystagmus and minimal DVD. It is postulated that PVL related motility disorders, deviations and nystagmus are caused by disruption of the dorsal stream pathway that projects from occipital cortex to parietal and frontal cortices.[25] Nystagmus did not used to be considered as a common association in the very begining. But soon after, it was noted that it is almost invariably present as manifest or latent with this entity.[26,27]

Mostly esotropia is seen in white matter injury of preterm or late preterm.[28] Jacobson et al.,[29] reported six premature patients with white matter injury, of these one had alternating exotropia. This group consisted of small sized with normal cupping, normal sized with normal cupping and normal sized with large cupping optic discs. Like Dutton et al.'s report[30], visual field defects were predominantly affecting the inferior visual field. Our patient had a normal sized optic disc with a large cup in his right eye and an inferior visual field defect crossing the horizontal line was noticed in his both eyes. Sectoral rim pallor and this suspicious VF defect required neuroimaging. Jacobson et al.,[31] previously reported estimation of timing of insult in children with periventricular leukomalacia by optic disc morphology. They concluded that in children with small optic disc, brain lesion was estimated to have occurred before 28 weeks of gestation. Whereas in children with large cup and smaller neuroretinal rim area, it was estimated to have occured after 28 weeks of gestation.

Most of the articles concerning retrograde transsynaptic degeneration of ganglion cells in PVL/CP consist of children with premature labor. But despite having no premature or complicated labor history, our patient could be insulted by an ischemic/anoxic/infectious factor in preterm period.

From the optic disc appearance we assume that it took action between 28-34 weeks of gestation. In conclusion despite having no evidence of neurologic impairment, PVL related nonglaucomatous cupping must be considered in differential diagnosis when it's associated with suspicious optic disc appearance and VF defects. There are more mysteries embedded in the term "transsynaptic degeneration", but as the literature about this complex scenario enriches we would be able to determine the time of insult exactly in preterm period.

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