

CE Credit - Case Report(s) & Topic Review

Peripapillary Hyperreflective Ovoid Mass-Like Structure (PHOMS): The Ovoids We Keep Missing

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ABSTRACT

Background: Peripapillary hyperreflective ovoid mass-like structure (PHOMS) is a relatively new phenomenon, most recently defined as an independent entity between 2015 and 2018.

Histopathologically, PHOMS is described as a lateral protrusion of dilated axons into the peripapillary region as they exit the lamina cribrosa. These fibers are at the level of Bruch's membrane opening as the optic nerve enters the optic globe proper. The identification of PHOMS is dependent upon optical coherence tomography (OCT) and its effects may be seen on a Humphrey visual field (HVF).

Case Report: The following case report reviews an incidental finding of PHOMS in a 43-year-old male whose optic nerve appeared small and crowded, with slightly blurred disc margins on dilated fundus examination.

Conclusion: Eye care providers should be aware of PHOMS as a newly distinguished entity and its association with other disease entities. Dilated eye exams are important even when patients are asymptomatic. Such incidental findings should be monitored for progression or complications and baseline imaging should be captured.

Keywords: Peripapillary Hyperreflective Ovoid Mass-Like Structure, Optic Disc Drusen, Small and Crowded Disc Size, Papilledema, Tilted Disc

INTRODUCTION

In 2018, the Optic Disc Drusen Studies Consortium established peripapillary hyperreflective ovoid mass-like structure (PHOMS) as its own entity¹. PHOMS is described as a lateral protrusion of dilated axons into the peripapillary region as they exit the lamina cribrosa and enter the optic

globe proper¹. The herniation of the swollen axons into the peripapillary region is caused by axonal stasis. Proper axoplasmic flow is essential for maintaining the health and structural integrity of the optic nerve. Without such flow, axonal swelling is a common result².

Morphologically, PHOMS presents as a doughnut-shaped structure with a convex, semi-circular enclosure². PHOMS can also appear as oval with smooth edges, and is further recognized as one continuous structure as opposed to a multi-dispersed structure³. Located in the peripapillary region, PHOMS are either partial or fully present circumferentially, extending as far as the outer nuclear layer with its base at Bruch's membrane (Figure 1)^{1,3,4}. PHOMS is not known to cause a break in Bruch's membrane. Studies have reported that PHOMS appears most commonly within the nasal region of the optic nerve head and has a space-occupying effect, causing overlying tissue to shift outward and upward, directly contributing to its elevated appearance on funduscopy^{3,5}.

PHOMS are mainly associated with small optic discs and not associated with any structural damage (Figure 2) nor any major systemic diagnoses^{2,6}. PHOMS is idiopathic, but can be seen as a consequence of other ocular ailments⁶. This includes optic disc drusen, papilledema, non-arteritic anterior ischemic optic neuropathy, acute demyelinating optic neuritis and tilted myopic discs^{1,6,7}.

CASE REPORT

Initial Examination: Comprehensive Eye Exam

A 43-year-old Hispanic male presented for a baseline comprehensive eye exam. The patient reported he used to wear glasses in elementary school, but currently did not wear any spectacle correction. The patient could only

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recall the last eye exam being from elementary school and did not recall being told of any ailment. The patient complained of intermittent blurry vision, with his eyes needing to “come in and out of focus,” and was unsure if it was at distance or near. The patient reported usage of olopatadine hydrochloride 0.2% as needed in both eyes to help with ocular irritation. Medical history was positive for hypertension, stage 3 chronic kidney disease, and migraines with aura, for which the patient was being followed by neurology. The patient denied any eye injury, eye surgery, eye diseases, or eye pain. The last blood pressure reading per the patient’s chart was 132/90 mmHg. The patient denied any family history of ocular conditions such as glaucoma, macular degeneration, or blindness. Medications included amlodipine, fluticasone nasal spray, propranolol HCL, riboflavin, magnesium oxide, losartan, and rizatriptan benzoate. The patient denied any illicit or recreational drug use and reported social alcohol consumption.

Entering vision without correction was 20/20 -1 in each eye at distance and near, measured with Snellen/Reduced Snellen chart, respectively. Cover test revealed a small exophoria at near without correction, measuring 4-6 prism diopters. Cover test was ortho at distance.

Extraocular movements were smooth, accurate, full, and equal without pain and confrontation fields were full via finger counting for each eye. Pupils were equal and reactive to light and no afferent pupillary defect was noted. Pachymetry measurements revealed 560 micrometers in the right eye and 535 micrometers in the left eye. Subjective refraction revealed a small amount of hyperopia at distance with a small correction for near (+0.50 sph for right and left eye, +1.00 ADD) with best-corrected visual acuities of 20/20 OD and 20/20 OS at both distance and near. Slit-lamp biomicroscopy showed mild meibomian gland dysfunction in both eyes. All other anterior segment and lenticular findings were within normal limits. Intraocular pressure measured via Goldman applanation tonometry was 14 mmHg in the right eye and the left eye.

With dilated fundus examination, both optic nerves appeared small and crowded with a graded cup-to-disc ratio of 0.15 round in both eyes. The patient’s left optic nerve head appeared slightly elevated superiorly and nasally, contributing to mildly indistinct borders around that region. The left optic nerve head also had a tilted appearance. No other significant findings were noted on dilated fundus examination. The appearance of the left optic nerve head prompted an inquiry about pain on eye movements, performance of red cap desaturation testing, and baseline optic nerve head imaging. The patient reported no pain on eye movements and no red cap desaturation difference between the right or left eye. On Heidelberg Spectralis optical coherence tomography (OCT), the retinal nerve fiber layer (RNFL) thickness map showed elevation in the superior temporal and inferior temporal regions of the left eye (Figure 2). An OCT raster pattern through the

left optic nerve showed a large, ovoid, nodular structure superior-nasally; this lesion appeared considerably larger than typical optic disc drusen (Figure 1). There was no choroidal neovascular membrane or RNFL thinning noted.

Due to the questionable appearance of this lesion, a neuro-ophthalmologist was consulted regarding this incidental finding via curbside (in-person) consultation. He agreed that the lesion did not look like optic disc drusen and confirmed the diagnosis of peripapillary hyperreflective ovoid mass-like structure of the left eye. He further explained that this diagnosis was common in crowded, tilted nerves and the condition was likely congenital and benign, without clinical consequence.

Follow-Up Visit - One Week Later: Additional Testing

The patient returned a week later to capture Zeiss Clarus disc photos, OPTOS fundus autofluorescence (FAF) photography, and a baseline Humphrey visual field 30-2 SITA Fast (Figures 3,4,5 respectively). Results of the Humphrey Visual Field 30-2 Sita Fast were full in the right eye. However, in the left eye, there were shallow inferior nasal points, which correlated to the PHOMS location clinically. The patient was reassured that this finding was incidental and was likely to have been present for his entire life. Annual monitoring with comprehensive dilated eye exams and OCT was recommended to detect any potential choroidal neovascular membrane.



Figure 1: Maximum width (558um) and height (375um) of PHOMS in the left eye, respectively. Notice the diffuse, solid nature of the lesion along with the uneven hyperreflectivity.

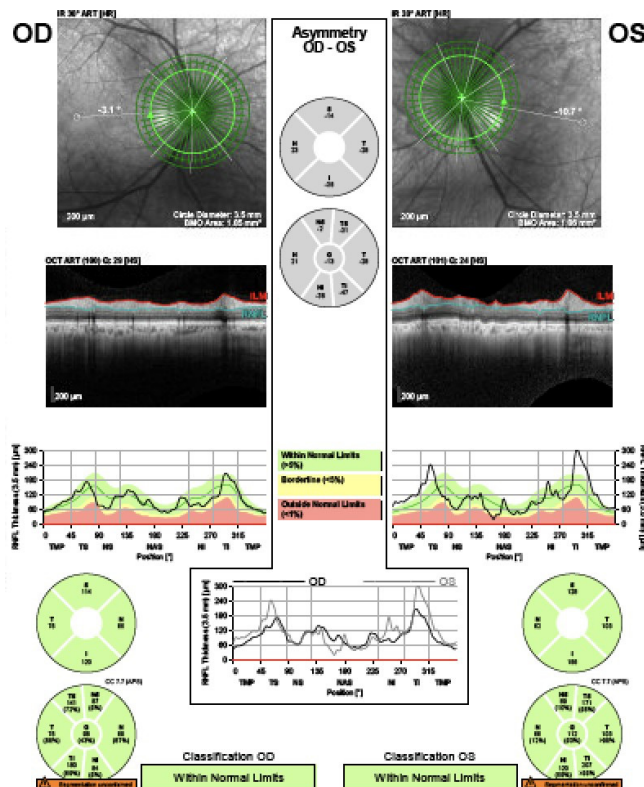


Figure 2: Baseline RNF OCT. Notice the small Bruch's membrane opening (BMO) correlating with a small crowded disc for both eyes, reading "1.85 mm²" for the right eye and "1.95 mm²" for the left eye. No quadrants flagged for RNFL thinning in either eye. Note elevation of RNFL in thickness map, interior temporal and superior temporal for the left eye.

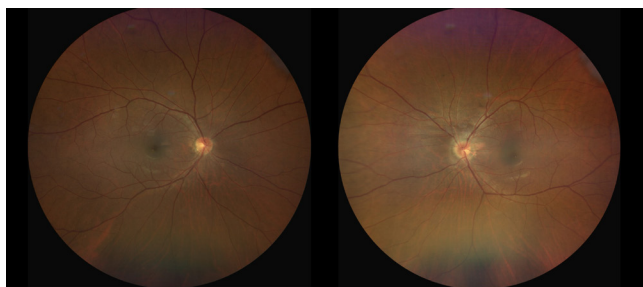


Figure 3: Zeiss Clarus photography of the right and left optic nerves, respectively. Notice an elevated appearance superior/nasal on the left optic nerve head, correlating with the location of PHOMS.

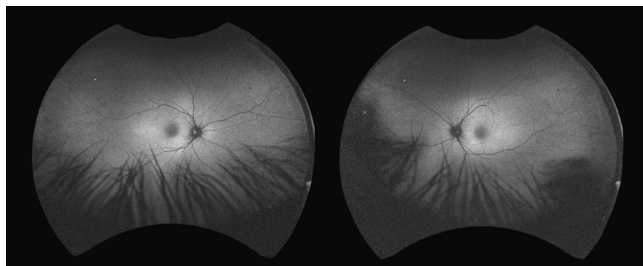


Figure 4: OPTOS photography of the right and left fundus, respectively, with FAF. Notice the lack of hyperreflectivity in the optic nerve head of the left eye, which was used to rule out optic nerve head drusen.

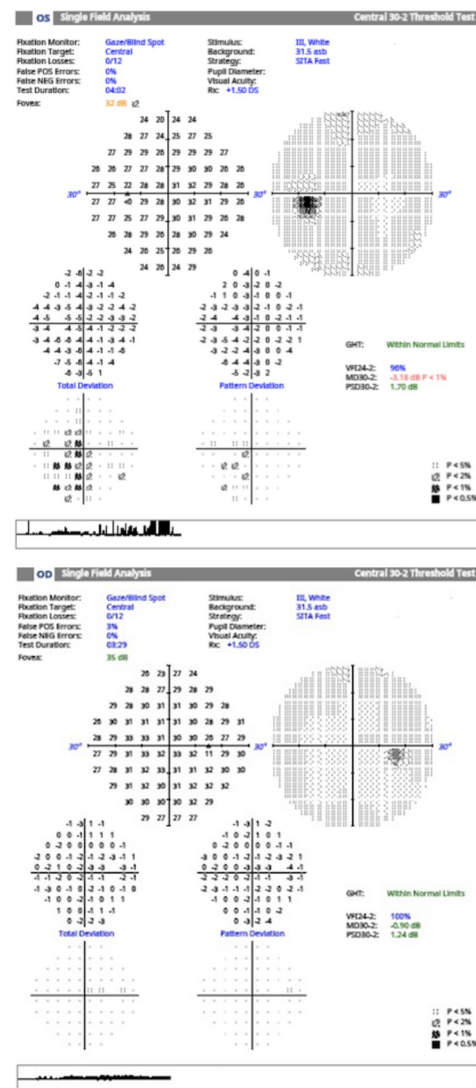


Figure 5: Baseline HVF 30-2 Sita Fast. Essentially clear field for the right eye. The left eye shows shallow defects inferior temporally, which correlate with the presence of PHOMS superior nasal. The right eye field was quite reliable, while the left eye field was borderline reliable, with high gaze tracking towards the end of the test, likely due to patient fatigue.

DISCUSSION

Epidemiology and Risk Factors

In a Beijing-based study, it was found that PHOMS is relatively rare in the general adult population⁶. However, PHOMS has a high incidence in patients with optic disc drusen (ODD), with a prevalence of about 2% of the adult population of those who have optic disc drusen³. There are few published studies that specifically measure the prevalence of PHOMS in adults, only when associated with ODD. The majority of published studies reflect PHOMS prevalence in the context of children. In a population-based, observational study, PHOMS was found to be prevalent among 8.9% of healthy children who are myopic with tilted optic nerve heads⁹. In this study, PHOMS was found to be

predominantly located superonasally, as seen in the patient in this case study. A clinical pearl from the study reflected that PHOMS does not always indicate an optic nerve ailment.

Signs and Symptoms

As previously discussed, PHOMS will appear in the context of small, tilted nerves^{1,4,9}. PHOMS will cause the optic nerve head to appear elevated and/or indistinct. On OCT, PHOMS will appear as seen in Figure 1: a large, diffuse lesion with uneven hyperreflectivity. There are no published studies correlating visual field loss and the presence of PHOMS. However, the patient in this case report did have a visual field defect correlating with the location of the PHOMS. It can be hypothesized that if the patient ever does develop RNFL loss, there might be consequential visual field defects. There is no plan to repeat HVF unless changes are noted on OCT or funduscopy, which is unlikely given the etiology of PHOMS. There are no studies specifying the symptoms of PHOMS. In the context of the patient in this case study, PHOMS is classified as asymptomatic and often an incidental finding when presenting in isolation⁵.

Pathophysiology

PHOMS is a representation of herniated, distended axons into the peripapillary region, causing axonal stasis. Proper axonal flow is necessary for maintaining the health and structural integrity of the optic nerve and the surrounding neuronal cells². PHOMSs are unique, separate structures versus being a subtype of optic disc drusen^{3,4}. Prior to 2018, PHOMS was considered interchangeable with buried optic disc drusen, especially when seen with papilledema and tilted discs^{4,9}. PHOMS is idiopathic, but can be seen as a consequence of other ocular ailments³. Such conditions include optic disc drusen, papilledema, non-arteritic anterior ischemic optic neuropathy, acute demyelinating optic neuritis and tilted myopic discs^{1,6,7}. PHOMS can occur concurrently with the above ailments due to the pathophysiology of axonal stasis. Therefore, PHOMS is not the cause of the previously listed conditions, but is often seen within their context. The patient for this case study did not have any of the associated optic disc conditions. The sole presentation of PHOMS gives this case a unique element.

Differential Diagnosis

The number one differential diagnosis for PHOMS is optic disc drusen as they can have a similar appearance with common risk factors. There are many differences, as well as similarities, between PHOMS and optic nerve head drusen. Optic nerve head drusen are shiny, singular crystalline forms within the optic nerve disc, while PHOMS are “C” or “O” shaped in the peripapillary region^{3,6}. Anterior optic disc drusen will hyperautofluorescence on FAF imaging while PHOMS will not³. B-scan ultrasound will reveal the presence of optic disc drusen - appearing as hyperreflective, while PHOMS will remain undetected³. The way optic disc drusen was ruled out in this case study was the appearance on OCT (Figure 1), showing a diffuse, solid lesion along with the lesion being peripapillary versus

within the optic disc. In addition, fundus auto-fluorescence OPTOS (Figure 4) showed no hyperreflectivity within the optic disc. The reason PHOMS is mistaken for optic disc drusen is that they are both associated with small, crowded nerves. Histopathologically, optic disc drusen are acellular, calcified deposits anterior to the lamina cribrosa, which can impede axoplasmic flow, thus causing PHOMS to exist concurrently^{3,4}.

Management, Prognosis, and Complications

Ocular diseases such as non-arteritic anterior ischemic optic neuropathy (NAAION), optic neuritis, tilted disc syndrome, and intracranial hypertension/papilledema can all cause concurrent PHOMS secondary to the impaired axoplasmic flow caused by a congested optic nerve head^{2,3,5,10,11,12}. In a retrospective cohort study that compared isolated PHOMS to those with PHOMS as a consequence of other ocular ailments, it was concluded that PHOMS found in isolation should be considered a distinct diagnosis, as with the patient in this case study⁵.

NAAION is a potentially blinding optic nerve ailment that is caused by ischemic infarction of the optic nerve head¹⁰. Risk factors include age 50 or over with small optic discs and certain vascular risk factors¹⁰. The presence of a small disc allows for PHOMS to be seen simultaneously with an NAAION. In tilted disc syndrome, optic nerve fibers are bent as they enter the lamina cribrosa, thus causing axoplasmic stasis. In one study conducted, it was found that PHOMS are found in 29.5% of high myopes with tilted discs^{3,6}. As seen in Figure 3, the patient appears to have a mildly tilted optic nerve head of the left eye; however, the elevated nasal/superior borders give a faux appearance of the nerve being tilted.

In papilledema, the optic nerve head becomes swollen due to elevated intracranial pressure¹¹. Acute demyelinating optic neuritis can be inflammatory in nature, and whether anterior or retrobulbar, the optic nerve can become congested¹². Said congestion of the optic nerve can give rise to PHOMS as a secondary outcome in both ailments. In the context of papilledema, PHOMS is also seen as larger in size⁵.

A 2021 study, looking at optical coherence tomography angiography (OCT-A), revealed that in patients with PHOMS, axoplasmic stasis can affect surrounding retinal vasculature. PHOMS can cause a rearrangement of retinal vasculature near the optic nerve head. This vasculature displacement can affect blood flow to the optic nerve head and thus could promote the production of vascular endothelial growth factor (VEGF), which is the precursor to neovascularization⁹. This can put PHOMS patients at risk for choroidal neovascular membrane (CNVM) development; however, PHOMS itself is not known to cause a break in Bruch's membrane as they occur above Bruch's and displace retina tissue upward and outward^{3,5}. A study published in the Journal of Neuro-Ophthalmology showed PHOMS progression in correlation

with myopic progression in children¹³. It is believed that the higher the myopia, the more severe the tilted disc, thus more axoplasmic stasis. The patient in this case report was not myopic and did not have a true tilted disc.

Blood flow and overall vascular density to the optic nerve can be reduced if PHOMS is >500um in height^{3,4}. This restricted blood flow can, in theory, lead to RNFL loss or optic nerve atrophy over time. In a 2024 study, a downward trend of the RNFL over time was found in patients with PHOMS. The study did not elaborate on the length of time needed to see RNFL thinning in the context of PHOMS¹⁴. In Figure 3, there is RNFL thickening rather than thinning noted on the thickness map in the inferior nasal/inferior temporal regions. Typically, there are no PHOMS effects on vasculature when its height is <300um-500um⁴. The average height and width of PHOMS in one study reflects 399um in height and 721um in width^{3,4}. As seen in Figure 1, the patient's maximum height is 375um and maximum width is 558um, making this patient less at risk for optic nerve damage over time. The patient being on the low end of average provides more assurance that the PHOMS was idiopathic in origin, requiring no intervention other than annual monitoring.

CONCLUSION

Eye care providers should be aware of PHOMS as a newly distinguished entity and its association with other disease entities. Dilated eye exams are important even when patients are asymptomatic. Furthermore, eye care providers need to understand the pathophysiology and mechanism of PHOMS when monitoring for its stability and when monitoring any other ocular ailments that can arise alongside PHOMS. Those secondary ailments, being separate entities, require treatment and monitoring.

When PHOMS is an isolated finding, there is no need for treatment. Although there is some debate in the literature, there is evidence that optic disc drusen may be prophylactically treated with brimonidine due to its neuroprotective properties¹⁵. It could be considered in the future that this treatment may be applied to PHOMS as well. In literature, PHOMS is mainly seen within the context of myopic children. More studies need to be done in an adult population, especially in distinguishing congenital versus de novo development.

In the future of optometric research, it will be interesting to follow the children in this population as they get older to see if PHOMS will be stable or cause future complications. It could be that PHOMS is rarely found in adult populations versus being more prominent in pediatric populations due to the specific patients being observed in these studies, since PHOMS is seen as congenital versus de novo. The future of eye care research and studies should promote improving the clinical application and database regarding PHOMS¹⁶.

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