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Primary CNS Melanoma

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Patient Presentation

- 49-year-old female referred for bilateral disc edema
- Bilateral disc edema noted by an optometrist in April of 2018, associated with a headache
 - Referred to an ophthalmologist
- Has gained 20-30 lbs. in the last 4 years
- Saw an outside ophthalmologist, visual field showed some constriction in the right eye
- Did not start diamox at that time, referred to neurology • Neurology ordered an MRI and LP:
 - MRI/MRV Mild bilateral disc edema, no intracranial mass, no venous thrombosis
 - LP opening pressure of 27
 - Started her on Diamox 1000 mg BID, then decreased to 1000 mg AM and 500 mg PM due to not tolerating the diamox
- Returned to optometrist 2 weeks later with continued decreased peripheral vision
- Discussed with neurology and increased diamox back to 1000 mg BID
- Referred to neuro-ophthalmology

Diagnostic Considerations

Elevated disc margins

- Pseudopapilledema
- Disc Drusen
- Malignant hypertension
- Diabetic papillopathy
- Hyperviscosity syndromes
- Hypotension/blood loss
- Early in the course of toxic optic neuropathies

Elevated Intracranial Pressure

- Idiopathic intracranial hypertension
- Intracranial mass lesions
- Obstruction of venous outflow
- Venous sinus thrombosis • Jugular vein compression
- Obstructive hydrocephalus
- Decreased CSF absorption
- Arachnoid granulation adhesions after bacterial or other infectious meningitis
- Subarachnoid hemorrhage
- Increased cerebrospinal fluid (CSF) production
- Choroid plexus papilloma
- Malignant systemic hypertension

Workup for Bilateral Optic Disc Edema

- Full ophthalmologic exam including:⁷
 - Visual acuity
- Visual field testing
- Evaluation for afferent pupillary defect
- Color vision testing
- Dilated fundoscopic exam
- Optical coherence tomography (OCT) to quantify the elevation of the retinal nerve fiber layer
- MRI/MRV and lumbar puncture⁶

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Initial Imaging







Figure 1: (1) Optic disc pallor with surrounding watershed area, (2) Optic disc edema with hyperemia and vessel obscuration (arrow), (3) OCT nerve showing diffuse optic atrophy in the right eye and elevated retinal nerve fiber layer in the left eye, (4) Ganglion cell layer section of OCT showing significant atrophy in the right eye and full ganglion cell layer in the left eye, (5) Humphrey visual field of the left eye showing a full visual field (6) Humphrey visual filed of the right eye showing diffuse constriction with a small infra-temporal island remaining

Follow Up Visits

Date	Interval History	Exam	Testing	Plan
1/14/19	No changes in vision Lost 8-10 lbs. Taking Diamox 1000 mg BID	3+ APD OD Full color plates HVF unchanged Optic atrophy OD with possible refractile bodies Persistent edema OS	Stable RNFL and GCL on OCT	FAF and B-scan: Negative Decrease Diamox to 1000 mg AM and 500 mg PM
3/28/19	No changes in vision Headaches have improved Lost 15 lbs. Taking Diamox 1000 mg BID	3+ APD OD Full color plates HVF unchanged Optic atrophy OD Persistent edema OS	Stable RNFL and GCL on OCT	Increase Diamox to 1500 AM and 1000 mg PM Consider surgical intervention if no improvement at next visit
6/20/19	Nausea and vomiting Gait abnormalities Taking Diamox 1000 mg BID	3+ APD OD Full color plates HVF early depression in the left eye Optic atrophy OD Persistent edema OS	Increased elevation of the RNFL on OCT in the left eye	Repeat MRI Brain and lumbar puncture Continue Diamox 1000 mg BID



MD -26.31 dB P < 0.5 PSD 9.63 dB P < 0.55



hyperintense, intramedullary mass centered at T2-T3 (circle)

- Laminectomy from C7 to T3 was performed on 07/19/2019 • Pathology confirmed metastatic melanoma
- Gait improved after debulking of tumor
- Full dermatologic exam revealed no other sites of melanoma Supports presumptive diagnosis of primary CNS melanoma
- Started on systemic chemotherapy per medical oncology • Received one infusion
- Had a rapid decline and died on 8/25/19

- Melanocytes exists in the uvea, cerebral parenchyma, leptomeninges, mucous membranes, and skin³
- Uncommon and constitute approximately 1% of all melanoma cases and 0.07% of all brain tumors^{1,2}
- uncommonly metastasize to systemic organs
- Hyperintense on T1 weighted images and hypointense on T2 weighted images due to presence of melanin⁴
- 10 weeks⁵

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Follow Up Imaging



Figure 2: (1) MRI brain showing leptimeningeal enhancement (arrows) and nodularity demonstrating some increased signaling on T1 without contrast, (2) Intrinsically T1

Subsequent Course

Primary CNS Melanoma

- Primary cerebral melanomas often develop in patients under 50 years of age and
- Once the cancer spreads to leptomeninges, the overall median survival is generally only

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