SALUS UNIVERSITY
The Eye Institute

Pennsylvania College of Optometry
The Focal Point
December 2023 Edition

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Traditional Class of 2025

Hometown: Willow Street, PA
Undergrad: Lebanon Valley College
Major: BCMB + Neuroscience
Favorite Animal: Pigs or Hedgehogs
Optometry Goal: To pass boards
Favorite instrument: Fundus Camera
Hobby: Baking! (especially cinnamon rolls)
Last Show I binged: Love on the Spectrum

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Class of 2019, Pennsylvania College of Optometry

Hometown: Virginia Beach, VA
Undergrad: Old Dominion University
Major: Biological Sciences
Favorite Diagnostic Instrument: Optical Coherence Tomography
Hates: cold weather and peanut butter
Hobby: Weightlifting, Attending the opera

Setting Expectations: Primary care considerations for patients with ocular albinism

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Some details of the case have been changed to protect patient confidentiality. Though the NBEO PAM-Style format is utilized for the case write-up, there is no affiliation with NBEO.

Demographics

32 yo Hispanic Female; Former Kindergarten Teacher (bless her) who switched to more administrative role

Chief complaint: Recently switched jobs to one that requires more computer work. Has difficulties unless wearing +3.00 OTC readers. She wants to update her glasses and additionally wants to pursue a driver's license for Pennsylvania.

History of present illness

Character/signs/symptoms: eye strain on computer
Location: computer/near work
Severity: moderate
Nature of onset: switching jobs
Duration: working on computer
Frequency: anytime she's on the computer
Exacerbations/remissions: +3.00 OTC readers
Relationship to activity or function: her job is exclusively related to computer work
Accompanying signs/symptoms: N/A

Patient ocular history: (+) oculocutaneous albinism secondary to Hermansky Pudlak Syndrome Type, nystagmus (-) eye surgery (-) glaucoma

Family ocular history

N/A

Patient medical history: (+)Hermansky Pudlak syndrome (bleeding disorder) and Irritable Bowel Syndrome; (-) DM (-) HTN

Medications taken by patient: Flonase Allergy Relief (50mcg)

Patient allergy history: NKA

Family medical history

(+) diabetes - maternal/paternal grandfather
(+) ovarian cancer
(-) HTN (-)DM

Review of systems

Constitutional/general health: denies
Ear/nose/throat: Cardiovascular: denies
Pulmonary: Endocrine: denies
Dermatological: denies
Gastrointestinal: denies
Genitourinary: denies
Musculoskeletal: denies
Neurologic: denies
Psychiatric: denies
Immunologic: denies
Hematologic: denies

Mental status

Orientation: oriented to person, place, and time
Mood/Affect: normal

Clinical findings

BVA: Distance Near

OD: 20/60- PH: NI 0.4/0.6M

OS: 20/80+ PH: NI 0.4/0.6M

Pupils: PERRL, +3 reaction to light, (-) APD OU, 3 bright/6 dim OU

EOMs: full/no restrictions OU; left beating nystagmus - worse in left gaze

Confrontation fields: FTFC OU

Esterman field: 140 degrees across the horizon **(See Image 5)**

Hirschberg: Grossly symmetric

Stereopsis: (+) FORMS; 200 sec circles

Subjective refraction: VA Distance

OD: +4.25 -4.50 x177 20/60-1

OS: +4.00 -4.25 x003 20/70-2

20/60- OU

*Note - BCVAs are stable to previous encounters

Slit lamp:

lids/lashes/adnexa: normal OU

conjunctiva: trace injection OU

Cornea: normal endothelium, epithelium, stroma, and tear film OU

anterior chamber: deep and quiet OU

Iris: flat and intact OU

lens: clear lens capsule, cortex, and nucleus OU

Vitreous: clear OU

IOPs/method: 15/13 - iCare

Fundus OD:

C/D: 0.15/0.15

Optic nerve: Flat, well perfused, hypoplastic in appearance, stable to photo on file **(See Image 1&2)**

Macula: flat, (-)hemorrhages,(-) exudates, (-)pigmentary changes, (-) edema

Posterior pole: Blonde fundus

Periphery: flat x 360 degrees, no RD, no holes, telangiectasia

Fundus OS:

C/D: 0.15/0.15

Optic nerve: Flat, well perfused, hypoplastic in appearance, stable to photo on file **(See Image 1&2)**

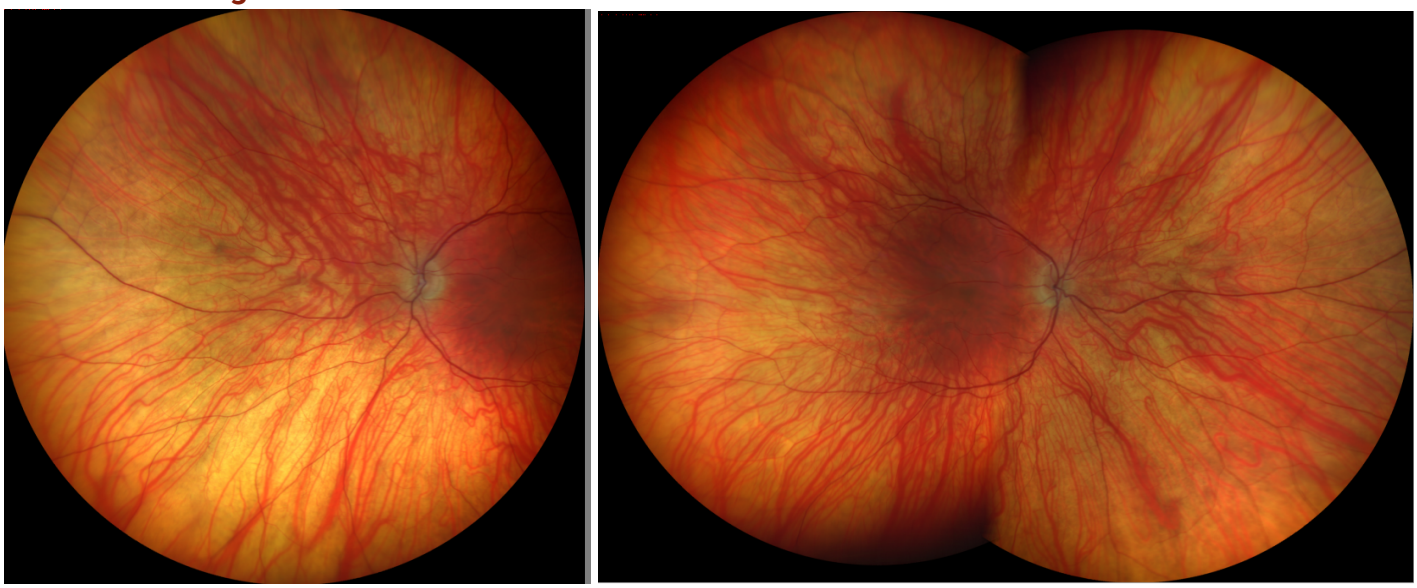
Macula: flat, (-)hemorrhages,(-) exudates, (-)pigmentary changes, (-) edema

Posterior pole: Blonde fundus

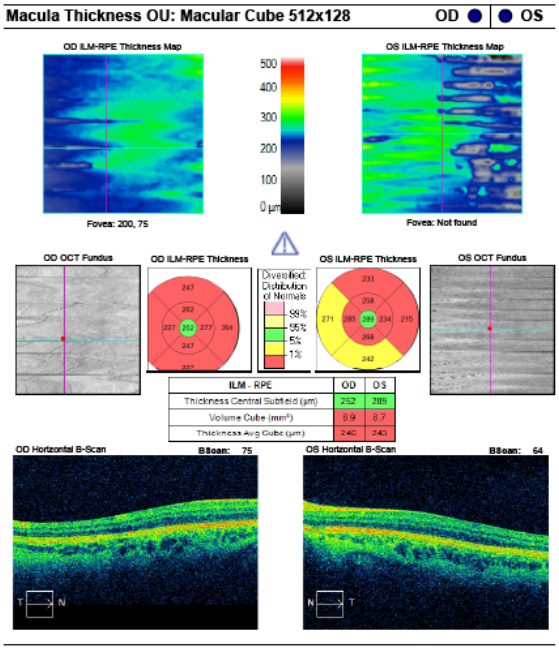
Periphery: flat x 360 degrees, no RD, no holes, telangiectasia

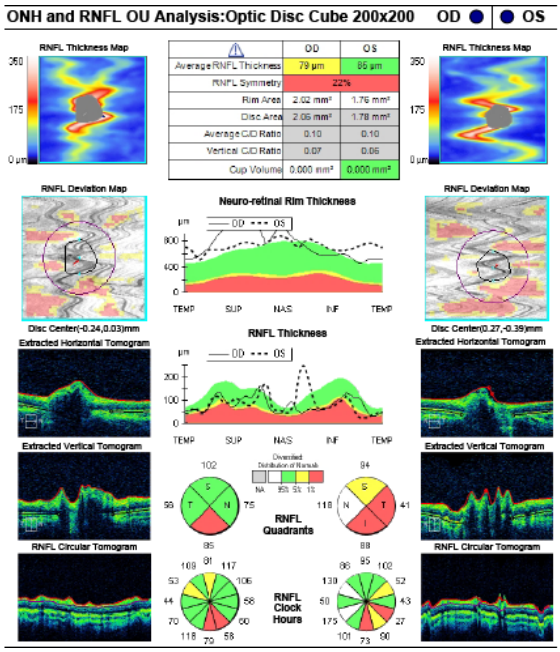
Blood pressure: 110/82 mmHg

Case Images:



Images 1 and 2: Colored Fundus Photos OD, OS respectively. Note the blonde fundus with prominent underlying choroidal vessels and hypoplastic appearance of the optic nerves OU.





Images 3 and 4: Cirrus Macular Thickness OCT OU (left) and Cirrus ONH and RNFL Analysis OU (right). Note the signal interference secondary to nystagmus in both images apparent in the thickness and deviation maps. The Macular OCT indicates generalized retinal thinning OU with a lack of foveal development consistent with foveal hypoplasia in ocular albinism. The ONH OCT demonstrates small disc area with small cupping OU in addition to elevated appearance of the ONH seen on the tomograms which is consistent with hypoplastic optic nerves.

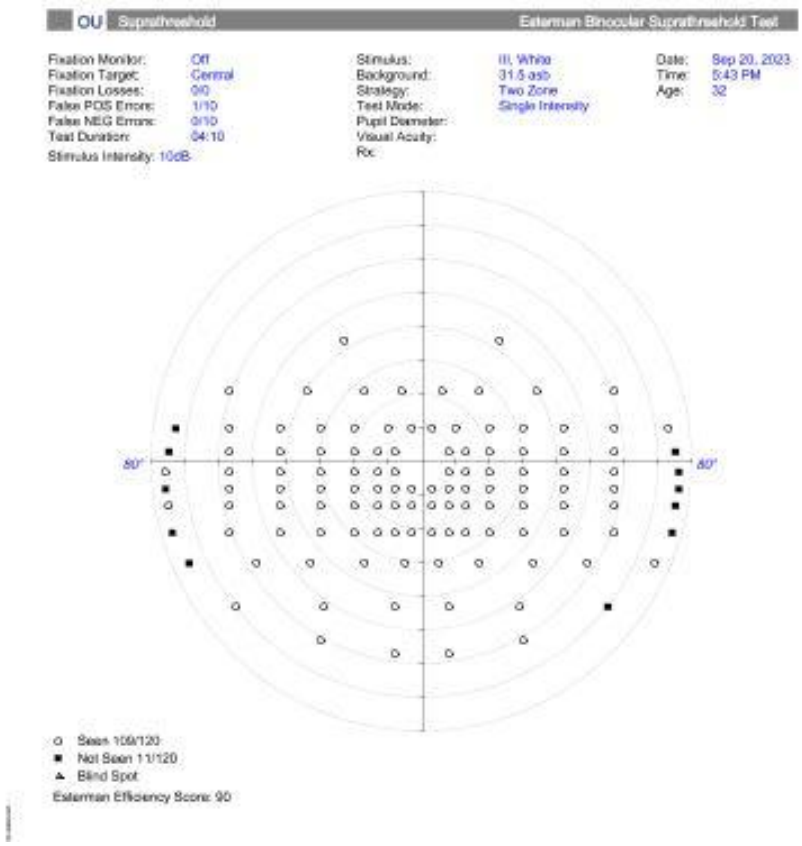


Image 5: Esterman Humphrey Suprathreshold Visual Field OU demonstrating intact horizontal visual fields of 140 degrees.

Case Management Summary

A1. Refractive Error
Bilateral. BCVA: OD: 20/70+ , OS: 20/60+
Esterman Visual field performed today: meets visual field qualifications for PA license

P1. Patient was educated on today's findings. Released updated glasses rx and rx sunglasses. Recommended two pairs, one for full time wear and another as sunglasses. DMV paperwork released to the patient for a restricted PA license. A copy was put on file for the patient. Pt to RTC in 1 year for CEE or if any visual symptoms arise.

A2/3. Oculocutaneous albinism secondary to Hermansky Pudlak Syndrome
Status: Symptomatic. Longstanding hx of OCA w/ HPS type 3.
(+)nystagmus & high WNL astigmatism
Dilated fundus evaluation performed today: demonstrates small, hypoplastic nerves and foveal hypoplasia OU

P2/3. The patient was educated on the findings. Advised to continue systemic

management with PCP and hematologist as directed for monitoring of HPS. Monitor annually or sooner if any new or worsening symptoms arise.

Case Pearls:

- **There are many etiologies for oculocutaneous albinism (and this is one of them!)**
  Hermansky Pudlak syndrome Type 3 is characterized by oculocutaneous albinism and a storage pool deficiency due to an absence of platelet dense bodies. Clinically, affected individuals have a bleeding diathesis, horizontal nystagmus, decreased vision and very mild pigment dilution of hair, skin, and iridesis characterized by oculocutaneous albinism, bleeding diathesis, and neutropenia.[1]

- **Don't aim for a perfect refraction (and be judicious with a Neuro consult)**
  The reduced VA in Hermansky Pudlak Type 3 is from nystagmus and decreased pigment and underdevelopment of certain anatomical structures, including the iris, macula, and optic nerve. 20/20 vision is unexpected, checking for stability is more important. And on that note, if the nystagmus is stable in characteristics (direction, amplitude, etc), there is no need to bring in the neuro team.

  TLDR; make sure the findings are stable!

- **Know the Driving Regulations/Limitations in Your Practicing State -** The patient's main goal during the examination was to obtain documentation for driving. This is a common goal that optometrists should be prepared to address. Our patient had at least 20/70 vision and more than 120 degrees on her Esterman visual field, so she qualified for a daylight driving license. Generally, optometrists should know (or be able to readily access) the vision requirements for various driver's licenses in their state be be prepared to provide documentation for their patients.

1. Anikster Y, Huizing M, White J, Shevchenko YO, Fitzpatrick DL, Touchman JW, Compton JG, Bale SJ, Swank RT, Gahl WA, Toro JR. Mutation of a new gene causes a unique form of Hermansky-Pudlak syndrome in a genetic isolate of central Puerto Rico. Nat Genet. 2001 Aug;28(4):376-80. doi: 10.1038/ng576. PMID: 11455388.