SALUS UNIVERSITY
The Eye Institute

Pennsylvania College of Optometry
The Focal Point
April 2023 Edition

Ayah Abdelgawad
Traditional Class of 2024

Hometown: Astoria, Queens
Undergrad: Rutgers University
Major: Kinesiology and Health
Favorite Subject: Dry Eye
Optometry Goal: Open a private practice with my best friend
Favorite food: Italian
Hobby: Playing beat saber on VR
Last Show I binged: YOU

Laine S. Higa
Class of 2014, Illinois College of Optometry

Hometown: Kāneʻohe, Hawai'i
Undergrad: Pacific University
Major: Cellular & Molecular Biology
Ocular Anomaly: Deuteranomalous trichromat
Interesting Fact: I'm able to identify the make and model of commercial jetliners
Current Libation of Choice: A glass (or bottle) of Chianti Classico Riserva D.O.C.G.

Don't be Fooled! Walking the Talk and Fixating on the Important Stuff in a Case of Oculocutaneous 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

80 yo retired caucasian female

Chief complaint: Reports difficulty reading small print at the supermarket and seeing cards when playing card game, “Bridge”

History of present illness

Character/signs/symptoms: Blurry vision at near
Location: both eyes
Severity: moderate
Nature of onset: longstanding
Duration: Frequency: constant
Exacerbations/remissions: congenital nystagmus; relief with gaze in left head tilt (null point)
Relationship to activity or function: tasks at near
Accompanying signs/symptoms: significantly reduced vision**;** patient presents with a mobility cane but reports difficulty navigating environment.

Patient ocular history

Oculocutaneous albinism, Congenital nystagmus, s/p CE c PCIOL OD, OS 2 year ago

Family ocular history

(-) Glaucoma (-) Retinal disease

Patient medical history

Osteoarthritis, Depression, Overactive Bladder/bladder infections, Muscle pain

Medications taken by patient

Meloxicam 7.5 mg, Lexapro 10 mg, Clonazepam 1mg, oxybutynin chloride ER 5 mg, baclofen 10 mg, nitrofurantoin, monohydrate/macrocrystals 100 mg capsule

Patient allergy history

NKDA

Family medical history

No known family history

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: appropriate

Clinical findings

BVA:

	Distance	Near
OD:	20/80-1 PHNI	0.4/0.5M
OS:	20/80-1 PHNI	0.4/0.5M

Pupils:

OD: PERRL Round/ 1+ Direct/ (-) APD
Bright illumination: 3mm
Dark illumination: 4.5mm
OS: PERRL Round/ 1+ Direct/ (-) APD
Bright illumination: 3mm
Dark illumination: 4.5mm

EOMs:

OD: Full; right beat nystagmus; null point- 20 degree left head tilt
OS: Full; right beat nystagmus; null point- 20 degree left head tilt

Confrontation fields: Grossly Full with some difficulty fixating OD, OS
Hirschberg: symmetric

Subjective refraction:	VA Distance	VA Near
OD: +1.00-1.50x110	20/80-1	0.4/0.5M
OS: +1.00-1.50x063	20/70+2	0.4/0.5M

ADD: +3.50

Slit lamp:

Adnexa: normal; lids and lashes normal OU
Conjunctiva: white and quiet bulbar conjunctiva; pink and quiet palpebral conjunctiva OU
Cornea: normal endothelium, epithelium, stroma and tear film OU
Anterior Chamber: deep and quiet OU; VH: 4 T and N OU
Iris: (+) iris translucency with retroillumination; flat OU
Lens: PCIOL in place, trace PCO OU
Vitreous: clear OU

IOPs/method: 12/12mmHg Goldmann

Dilated Fundus Exam:

Optic Nerve: distinct margins with pseudo pallor, C/D ratio 0.1/0.1, anomalous trifurcation of vessels exiting cup OU
Maculae: flat and intact (-) FR (+) foveal hypoplasia OU
Retina: blonde fundus OU
Vessels: normal course and caliber OU
Periphery: flat and intact 360 (-) breaks or detachments OU

Case Images:

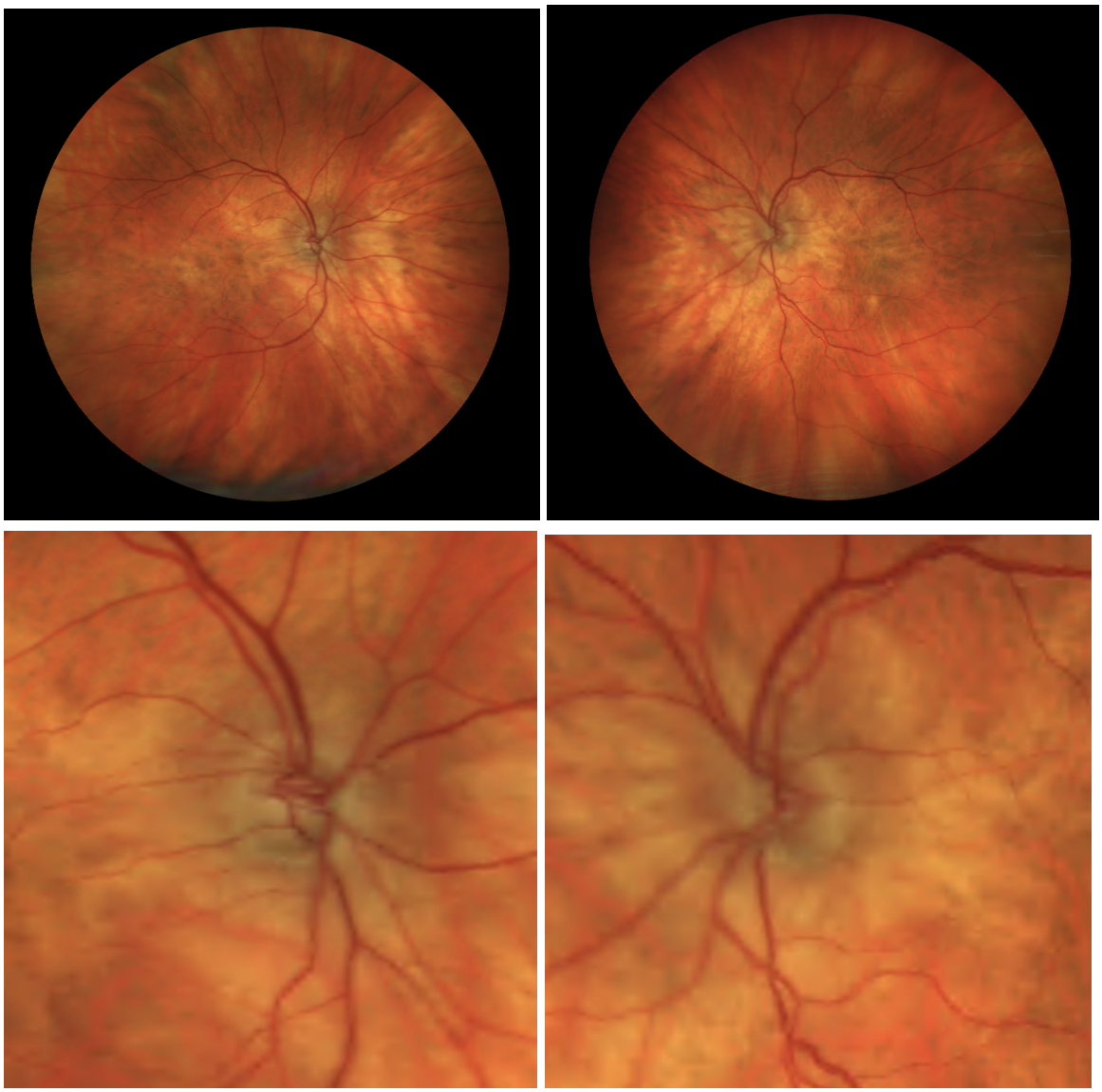

Image 1: Widefield colored fundus photos OD and OS, respectively and 20 degree colored fundus photo OD and OS, respectively demonstrating pseudopallor and trifurcation of the vessels of the optic nerves bilaterally and blonde fundi OU

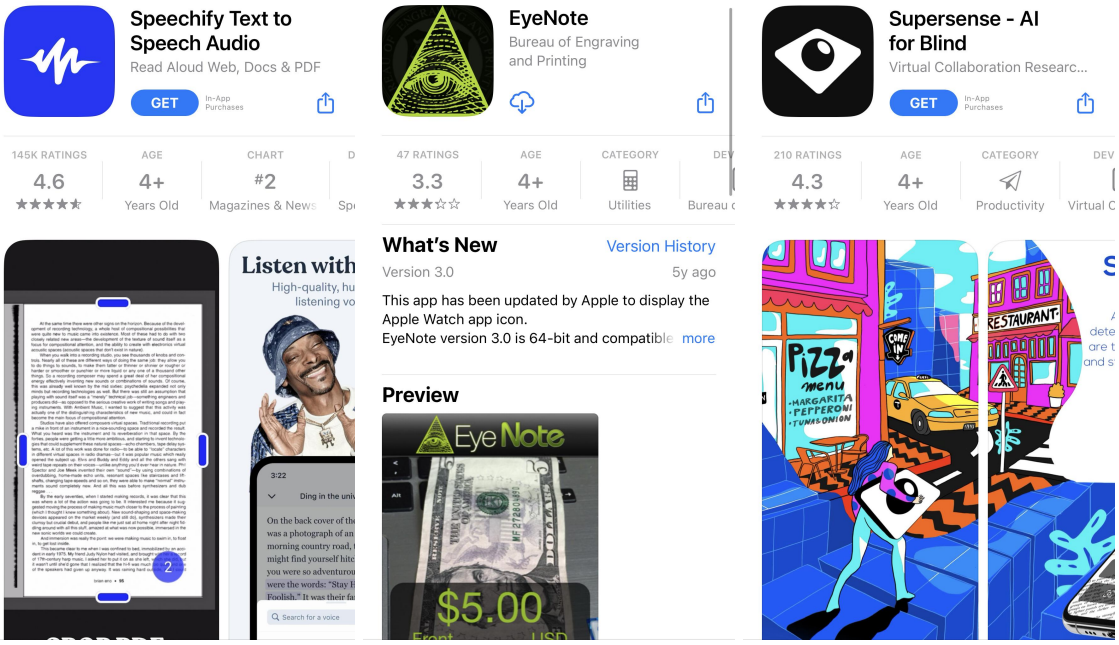

Image 2: Smartphone Apps shown to the patient in order to help her navigate her environment more effectively. *Speechify, EyeNote,* and *Supersense* from left to right.

Case Management Summary

1. Oculocutaneous Albinism (E70.329), OD, OS
Reduced acuity due to foveal hypoplasia and congenital nystagmus. Null point: 20 degree left head tilt. BCVA: 20/80 OD and 20/70 OS. Reported difficulty using mobility cane.

Patient educated about the condition. Patient scheduled with Vision Rehabilitation (VR) and Orientation and Mobility (OM) for evaluation of magnifying devices, glare control, and retraining with mobility cane. Given the patient's technology literacy, the patient was provided a list of applications for text-to-read, magnification, and money reading to use prior to consultation.

2. Congenital Nystagmus (H55.01), OD, OS
Secondary to Oculocutaneous Albinism. Null point: 20 degree left head tilt.

Patient educated about the condition. Will continue to monitor for changes at subsequent visits.

3. Secondary Cataract (H26.4), OD, OS
Not visually significant

Patient educated about the condition. YAG not indicated at this time. Will continue to monitor for changes at subsequent visits.

4. Presbyopia, (H54.2), OD, OS

DVO and NVO SRx released. Monitor yearly.

Case Pearls

Ocular Albinism vs. Oculocutaneous Albinism
Ocular albinism occurs in several forms and follows all the inheritance patterns for transmission. Ocular albinism is restricted to only the eyes. There are more than 15 genes currently identified as causing either absent or diminished tissue melanin. Oculocutaneous albinism occurs when both the skin and eyes are involved. Often associated with both these conditions are congenital nystagmus, iris transillumination defects, foveal hypoplasia, and blonde fundi.

Don't let the patient's age fool you
Although 80 years young, the patient reported ease of use with computers and technology. This allowed us to recommend phone and computer apps that could aid in reading, magnification, and money counting until her consultation with Low Vision/Vision Rehabilitation. Had we not asked, we might have assumed the patient could not use a computer and/or smartphone and not recommended these helpful apps.

Walk the talk
The patient presented with a mobility cane given her long standing diagnosis of oculocutaneous albinism. Upon further questioning, the patient reported continued issues with mobility even with her cane. She gratefully accepted a referral for Orientation and Mobility retraining. Just because a patient already has a mobility cane, don't assume they are comfortable with navigating their environment. Retraining is always a good option and allows the patient to continue to independently navigate safely and successfully.

Fixating on the important stuff
Congenital nystagmus is often associated with oculocutaneous albinism. As mentioned, one of the attributes of congenital nystagmus is latent nystagmus, or a worsening of the lateral eye movement when one eye is closed/covered. Don't panic! Expect difficulty with unilateral tests (VA, CVF, refraction etc.) and don't get too hung up on getting varied results. Consider binocular refraction in a trial frame while fogging the untested eye (as opposed to occlusion) in order to get more accurate results.