Salus at Drexel University
The Eye Institute

Pennsylvania College of Optometry

The Focal Point

October 2024 Edition

Elle Cornman

Scholars Class of 2025

Hometown: Richmond, Virginia
Undergrad: Virginia Tech (Go Hokies!)
Major: Neuroscience; Minor Sustainable Agriculture
Favorite Animal: doggos
Optometry Goal: spell “fluorescein” right on the first try
Favorite instrument: harmonica
Hobby: road biking
Last Show I binged: SPRINT

Christian Nemeh

Scholars Class of 2025

Hometown: Allentown, Pennsylvania
Undergrad: Muhlenberg College
Majors: Neuroscience & Theatre Arts
Favorite Animal: my dog, Benny
Optometry Goal: good work-life balance
Favorite instrument: tuning fork
Hobbies: volleyball, video games
Last Show I binged: Fleabag

Christin DeMoss, OD, FAAO

Pennsylvania College of Optometry 2017; Pennsylvania College of Optometry Residency in Low Vision Rehabilitation 2018
Hometown: Mohnton, Pennsylvania
Undergrad: Pennsylvania State University
Major: Biology; Minor Psychology
Favorite Diagnostic Instrument: Clarus Fundus Autofluorescence
Hates: suspenseful movies
Hobby: water polo & running

A New Look:

The evolution of vision following plastic surgery

Salus at Drexel University
The Eye Institute

Pending accreditor and third party approvals

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

37 y/o white transgender female

Chief complaint: reduced vision secondary to achromatopsia & degenerative myopia

- Wears **Kontur** tinted contact lenses, current lenses are 2 years old, needs update
- Notes increased brightness and greater ease of upgaze since recent facial feminization surgery

History of present illness

Signs/symptoms: diffuse blur
Location: OU
Nature of onset: since birth/infancy
Accompanying signs/symptoms: photophobia, lack of color vision, nystagmus

Patient ocular history

Achromatopsia
Degenerative myopia
Nystagmus
(-) glaucoma, (-) ocular injuries/surgeries

Family ocular history

Sister: Achromatopsia
(-) Family history of glaucoma

Patient medical history

Trans woman; undergoing hormone therapy

- Facial feminization plastic surgery

Borderline Personality Disorder, Anxiety
(-) HTN, Diabetes, Hyperlipidemia

Family medical history

Mother: vascular disease
Father: heart disease

Medications taken by patient

Emtricitabine 200mg- tenofovir disoproxil fumarate 300mg — for PrEP therapy
Estradiol 1mg, 3 tablets daily — for gender affirming care
Medroxyprogesterone 10mg — for gender affirming care
Finasteride 5mg — for gender affirming care
Spironolactone — for gender affirming care
Lorazepam 0.5mg — for anxiety

Patient allergy history

NKDA

Review of systems

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

Mental status

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

Clinical findings

BVA:

	Distance	Near
OD:	20/125 -	0.18/1.25M
OS:	20/125	0.16/1.0M

Pupils: PERRLA (-) APD OU

EOMs: full with no restrictions OU

Nystagmus: pendular nystagmus of medium amplitude and frequency OU

Confrontation fields: FTFC OD and OS

Hirschberg: Asymmetric, CAXT, OS out most frequently

Subjective refraction:

	VA Distance
OD: -10.00 sph	20/125
OS: -10.00 sph	20/125

Contact Lens Fit:

CL RX 1

	Rx	BC	Diam	Brand	Type	Color
OD	-8.50 sph	8.3	14.5	Kontur	Tinted, 11mm	Dark magenta
OS	-8.50 sph	8.3	14.5	Kontur	Tinted, 11mm	Dark magenta

CL RX 2

	Rx	BC	Diam	Brand	Type	Color
OD	-8.50 sph	8.3	14.5	Kontur	Tinted, 11mm	Chromagin
OS	-8.50 sph	8.3	14.5	Kontur	Tinted, 11mm	Chromagin

Slit lamp:

Lids/lashes/adnexa: lids and lashes normal OU
Conjunctiva: white and quiet bulbar, pink and quiet palpebral OU
Cornea: neovascularization extending 0.5mm onto cornea 360 OU
Anterior chamber: AC deep and quiet OU
Iris: flat and intact OU, iris nevus OD
Lens: clear lens capsule, cortex and nucleus OU
Vitreous: vitreous clear OU

IOP: 18/17 mmHg via Goldmann

Fundus OD: - *see image 1*

C/D: 0.15/0.15
macula: flat, no hemorrhages, exudates, pigmentary changes, or macular edema, no foveal reflex
posterior pole: ⅔ AV, normal course and caliber
Periphery: nonpigmented lattice 360

Fundus OS: - *see image 1*

C/D: 0.2/0.2
macula: flat, no hemorrhages, exudates, pigmentary changes, or macular edema, no foveal reflex
posterior pole: ⅔ AV, normal course and caliber
Periphery: nonpigmented lattice 360

Case Images

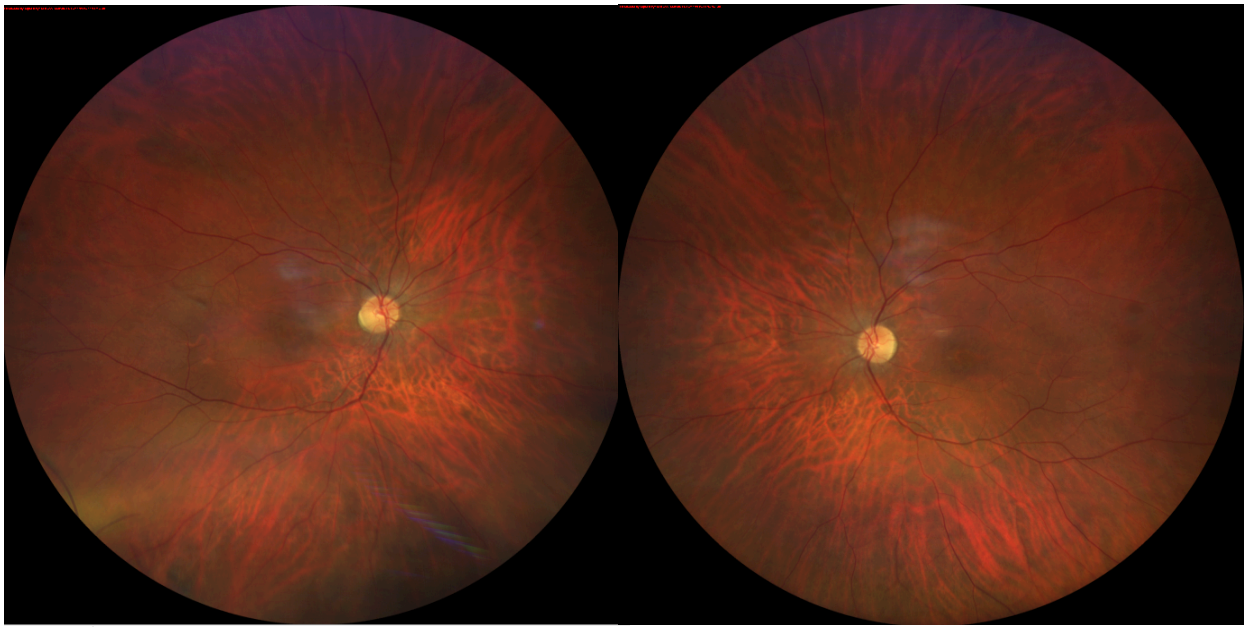

Image 1: **Clarus fundus photography of posterior pole OD and OS.** Of note: Questionable temporal pallor OD and OS. Loss of foveal reflex with mild macular pigment mottling OU. Blood vessel attenuation OU. Lens artifacts along superior arcades OU and inferior OD. Findings are consistent with the diagnosis of congenital Achromatopsia.

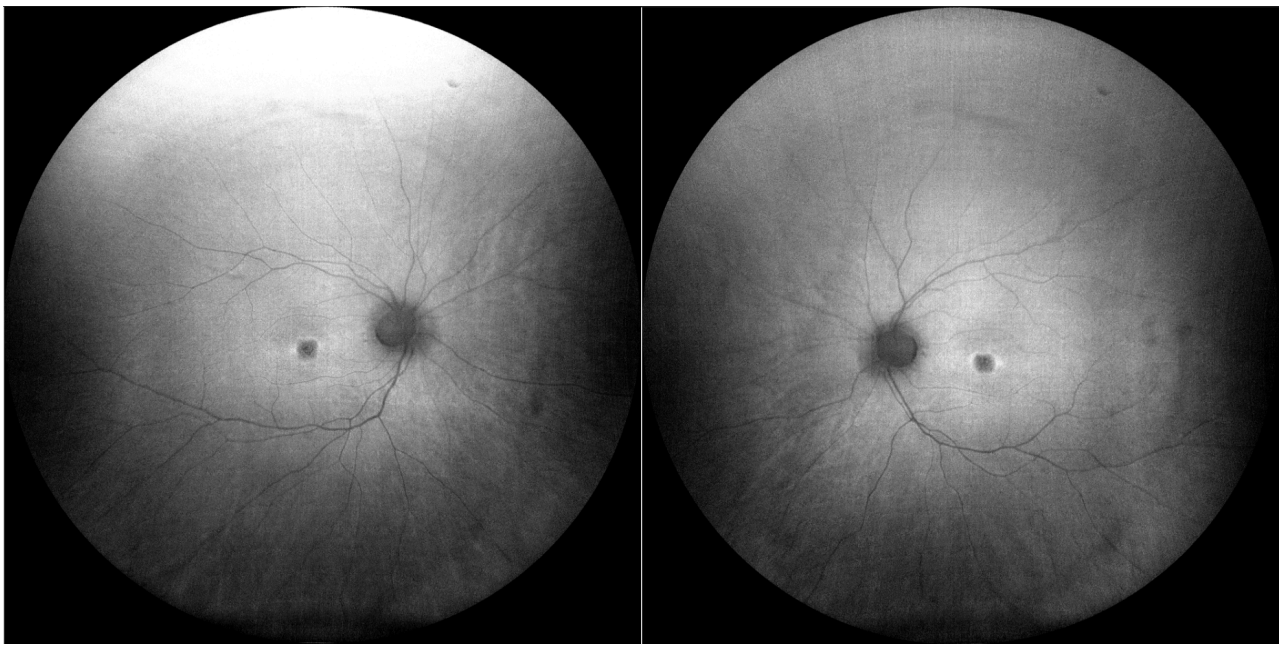

Image 2: **Fundus Autofluorescence (FAF) OD and OS.** Of note: generalized iso autofluorescence throughout posterior pole OU. Vessel attenuation OU. Well defined central area of hypo autofluorescence (which correlates to RPE atrophy) with small surrounding hyper autofluorescent ring (active breakdown of the RPE). Findings are consistent with Stage 4 Achromatopsia, which is marked by RPE destruction.

Case Management Summary

Assessment 1: Achromatopsia (H53.51)

- Symptomatic longstanding hx of Achromatopsia, previously monitored in NY
- Unknown hx of genetic testing, findings consistent with diagnosis:
    - (+) reduced VA
    - (+) nystagmus
    - (-) foveal reflex
    - (+) disruption/loss of foveal outer segments on OCT
    - (+) reduced color vision
- (+) Sister with Achromatopsia

Plan: Achromatopsia

- Patient was educated on exam findings. Color photos, FAF and OCT (mac/ONH) performed for baseline. Consider genetic testing at the next visit to confirm gene mutation. Recommend annual LV exam. RTC x 1 year or sooner PRN.

Assessment 2: Legal Blindness as defined in USA (H54.8)

- BCVA OD: 20/125, OS: 20/125
- BCNVA OU: 0.13/0.8M with +4.00 Add over old magenta CLs
- VF: full OD and OS
- Contrast: 1.52 Log with and w/o magenta CLs = normal contrast
- Goals: ocular health exam, updated CL rx

Plan: Legal Blindness as defined in USA

1. Patient was educated on the findings of today's exam and the services available.
2. **Glasses:** Provided updated script for DVO glasses. Discussed the importance of having a back-up pair of glasses to CLs.
3. **Low Vision Services:** Offered to re-refer her to Occupational Vocational Rehab through BBVS in the future should she need any other assistive devices or training. Patient is currently happy with accessibility features of her new computer.
4. **Tinted Contact Lenses:** Patient is happy in current CLs (-8.50 sph) in both dark magenta for brighter days and chromagin (orange) for darker days. Submitted to insurance and was approved for medical necessity. Reordered lenses through Kontour. Patient educated on the importance of continued CL hygiene by not swimming/sleeping/showering in lenses. Patient has been reducing the amount of time she wears the lenses given the peripheral neovascularization noted on both eyes. Recommended Clear Care at least weekly and Biotrue Solution nightly.
5. **Facial Feminization Surgery:** Patient recently had facial feminization surgery with a significant brow bone reduction. She subjectively notes an overall increase in brightness (but no additional glare) which is helpful for her vision. She also reports greater ease with looking up which has also been helpful.

Other Assessments: Degenerative Myopia (H44.23), Nystagmus (H55.00), Glare Sensitivity (H53.71), Unspecified Retinal disorder (H35.9)

- See H53.51 and H35.9

Case Pearls

Achromatopsia is a rare inherited retinal disorder that affects all 3 cone types.[1]
Also known as Rod Monochromacy, this genetic disorder causes all cones in the retina to be non-functional since infancy, leaving the patient with only rods for their visual needs. Because rods are only intended for low spatial frequency perception (such as sensing a dim light in a dark room), patients with achromatopsia have reduced visual acuity and no color vision. In bright lighting, rods become overstimulated and unable to repolarize, which leaves the patient with worse vision during the day. Lack of a clear retinal image as a child leads to the development of nystagmus. The most common gene mutations that lead to achromatopsia are of the Cyclic Nucleotide Gated (CNG) channels located on the outer segments of cones. Without these functioning channels, cones are unable to fire an electrical signal and slowly deteriorate throughout the patient's life.

Mac OCT and Fundus Autofluorescence can be used to monitor achromatopsia.[2]
Achromatopsia was previously thought of as a stationary disease since visual acuity remains similarly reduced throughout the lifetime. However, research with OCT and FAF have shown that non-functioning cones are initially intact in earlier disease stages, but deteriorate with the progression. While knowing the staging of achromatopsia is inconsequential at this point, it may become critical as new genetic therapies emerge for the treatment of this disease. For example, patients with structurally intact cones may be more receptive to treatment than those with a disrupted ellipsoid zone and RPE atrophy. At this point, it is best to utilize OCT and FAF to visualize pathologic changes not otherwise appreciated on a dilated fundus exam (*See Image 2*).

Low Vision can help address the functional visual needs of people with Achromatopsia.[3]
The most common visual disturbances of achromatopsia include sensitivity to glare and reduced visual acuities, averaging 20/200 among these patients. Rod photoreceptors are bleached much more easily than cones in bright, white environments, leading to a high sensitivity to aberrations and glare. To prevent rod overstimulation, red-tinted glasses and contact lenses are used, as these only transmit long-wavelength, low-energy light. To improve acuity and reading fine print, patients also benefit from magnification at near. This form of aid can include any combination of high-powered reading glasses, magnifiers, tablets, and smart phones with magnifying apps.

The visual benefits of facial feminization.
Brow bone reduction, a form of forehead recontouring, is one of the core procedures in facial feminization surgery.[4] Recession of the superior orbital rim exposes the eyes to a wider superior visual field which can have a significant benefit in patients with deep-set eyes. Brow bone recession does not only expand visual fields, but it has been reported by many patients to have increased their perception of brightness from overhead light sources. While this may provide benefits to some, it should also be taken into consideration for patients with preexisting glare sensitivity complaints. This pattern of photophobia is especially common in conditions such as Glaucoma, Macular Degeneration, and Cataracts.[5]

Gender affirming treatment can improve self confidence, which can lead to better self care.
Cosmetic procedures in gender-affirming care notoriously improve patients' self confidence and overall quality of life. In one study published in 2022, both trans men and trans women reported significantly higher survey scores in global self worth after gender-affirming surgical treatment.[6] Improved self esteem has been shown to directly translate to better self-care habits, consistency with healthcare visits, and compliance with medical advice.[7] Although not thoroughly researched at this point, this case demonstrates how improved self worth can lead to improved contact lens compliance and better ocular health.

Providing healthcare of trans patients with awareness and compassion.[8]
Discussing gender identity is not taboo and is often appreciated by patients. It is crucial to listen to LGBTQ+ patients when talking about their identity and health history. Transgender and intersex individuals are particularly vulnerable to microaggressions in healthcare, so taking extra steps to ensure their comfort and safety in your office can leave a lasting impression.

REFERENCES

1) Cohen S, Bracha P, Simonette J, Lim J. Achromatopsia [Internet]. Harnett ME, editor. American Academy of Ophthalmology: EyeWiki. 2024 [cited 2024 Aug 1]. Available from: https://eyewiki.aao.org/Achromatopsia
2) Greenberg JP, Sherman J, Zweifel SA, Chen RW, Duncker T, Kohl S, Baumann B, Wissinger B, Yannuzzi LA, Tsang SH. Spectral-domain optical coherence tomography staging and autofluorescence imaging in achromatopsia. JAMA ophthalmology. 2014 Apr 1;132(4):437-45.
3) Chan C, Seitz B, Käsmann-Kellner B. Morphological and Functional Aspects and Quality of Life in Patients with Achromatopsia. Journal of Personalized Medicine. 2023 Jul 7;13(7):1106.
4) Facial Feminization Surgery - Brow bone reduction - Transforming a prominent male forehead into a feminine one - 2pass Clinic. (n.d.). 2pass Clinic. https://2pass.clinic/en/ffs/procedures/brow-bone-reduction
5) Light Sensitivity. (n.d.). UCLA Health. https://www.uclahealth.org/medical-services/ophthalmology/eye-signs-and-symptoms/light-sensitivity
6) Arnoldussen M, van der Miesen AI, Elzinga WS, Alberse AM, Popma A, Steensma TD, de Vries AL. Self-perception of transgender adolescents after gender-affirming treatment: a follow-up study into young adulthood. LGBT health. 2022 Jun 1;9(4):238-46.
7) Edmonds KA, Aspiras OG, Rose JP, Gratz KL, Pinkston MM, Naifeh JA, Konkle-Parker DJ, Tull MT. Cross-sectional evaluation of perceived health care provider engagement, self-efficacy, and ART adherence in people living with HIV/AIDS. Aids Care. 2021 Feb 1;33(2):154-8.
8) Bhatt N, Cannella J, Gentile JP. Gender-affirming care for transgender patients. Innovations in clinical neuroscience. 2022 Apr;19(4-6):23.