

Retina Case: *“Spared Fovea”*



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Patient Presentation

CC

“I think I need a new glasses prescription”

HPI

- 55 yo WM with hx of undisclosed retinal disorder OU, p/w gradually worsening cloudiness over his vision OD and is requesting a new MRx.



History (Hx)

Past Ocular Hx: “Retinal Disorder” OU

Ocular Meds: none

PMH: HTN, Depression/Anxiety, chronic pain

Fam Hx: Glaucoma, Diabetes

Meds: Wellbutrin, Ativan, Percocet

Allergies: NKDA

Social Hx: Never smoker, no EtOH or drugs

RoS: chronic back pain



Physical Exam

	OD		OS
CC BCVA	20/25-2 (-1.75+1.25x020) 20/20 (-2.25+1.50x020)		20/20-2 (-1.75+0.75x175) 20/20 (-1.75+1.25x175)
Pupils	5mm. Round. Brisk.	No rAPD	5mm. Round. Brisk.
IOP	17 mmHg		17 mmHg
EOM	Full		full
CVF	Full, with central scotoma		Full, with central scotoma
Adnexa	Small amplitude nystagmus		Small amplitude nystagmus



Anterior Exam

SLE	OD		OS
External/Lids	WNL		WNL
Conj/Sclera	White, Quiet		White, Quiet
Cornea	Epithelium intact		Epithelium intact
Ant Chamber	Normal depth, Quiet		Normal depth, Quiet
Iris	Flat, WNL		Flat, WNL
Lens	2+ NS		1-2+ NS
Vitreous	Clear		Clear



Posterior Exam

Fundus	OD		OS
Optic Nerve	Pink/sharp		Pink/sharp
Macula	Yellow pisciform flecks throughout the macula		Yellow pisciform flecks throughout the macula
Vessels	Normal caliber		Normal caliber
Periphery	No tears		No tears



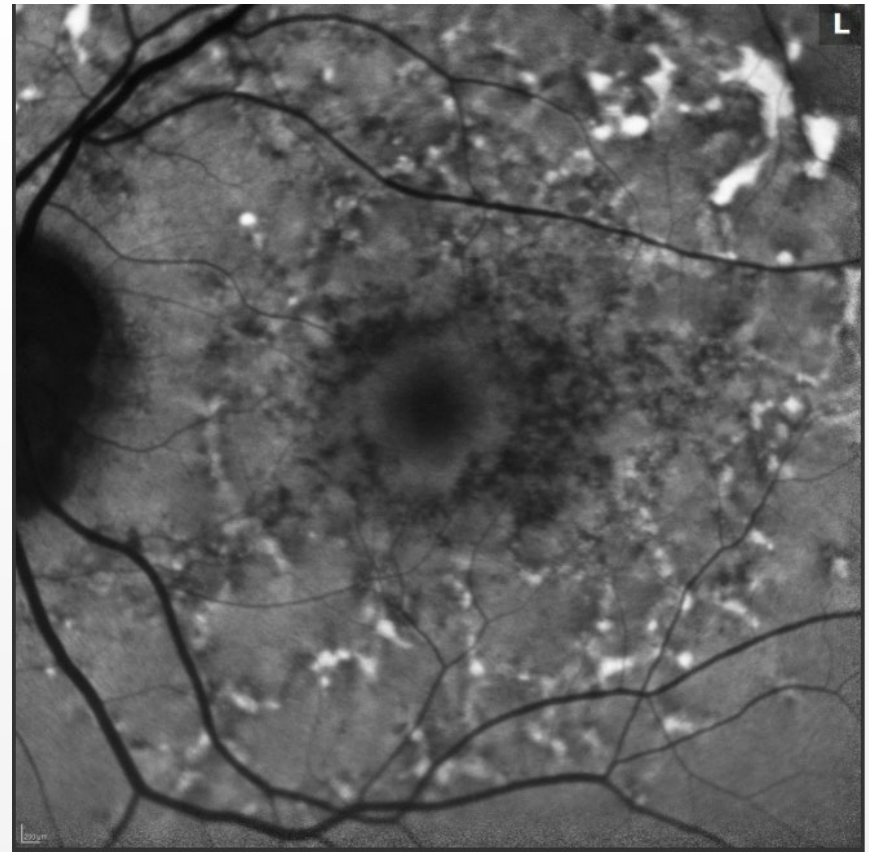
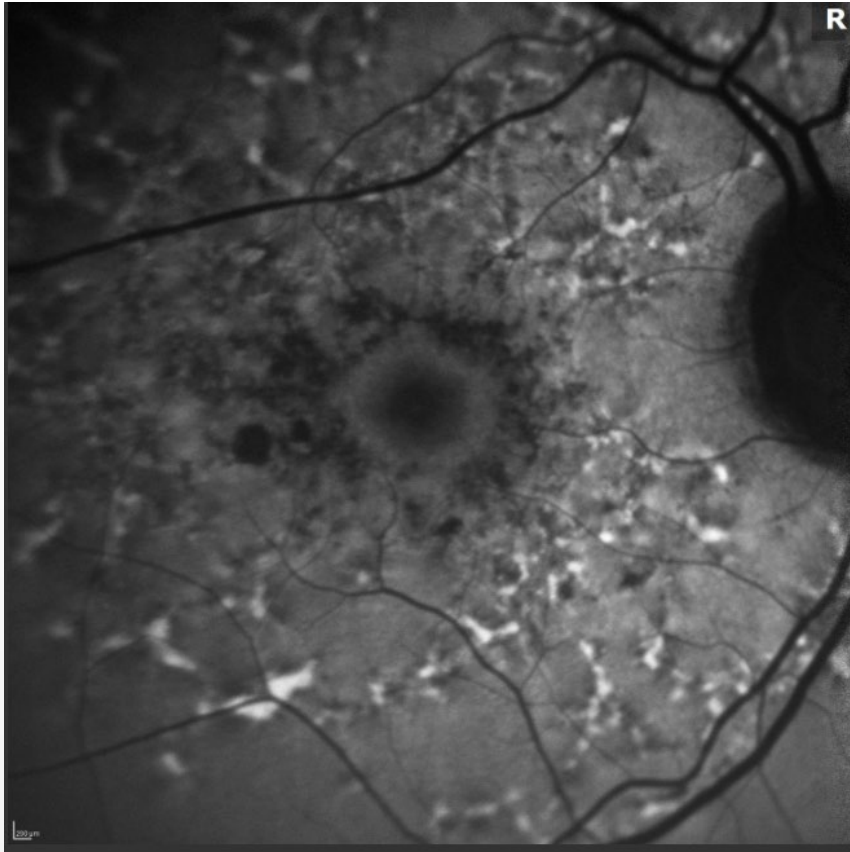
(Representative) Fundus Photos



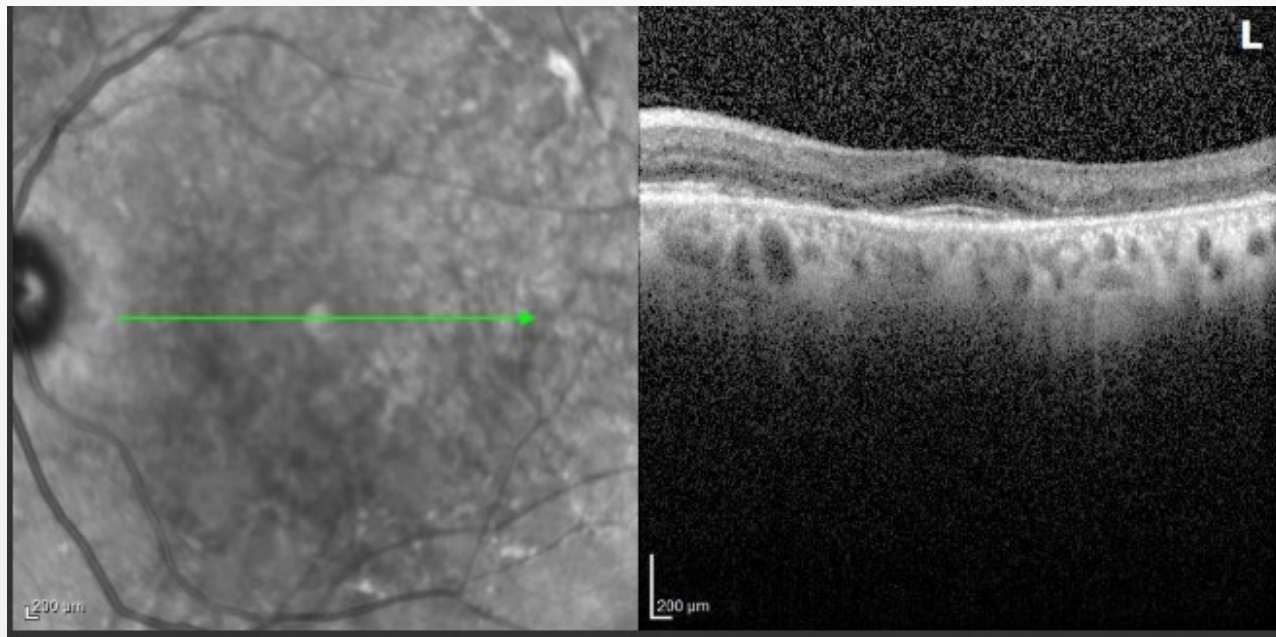
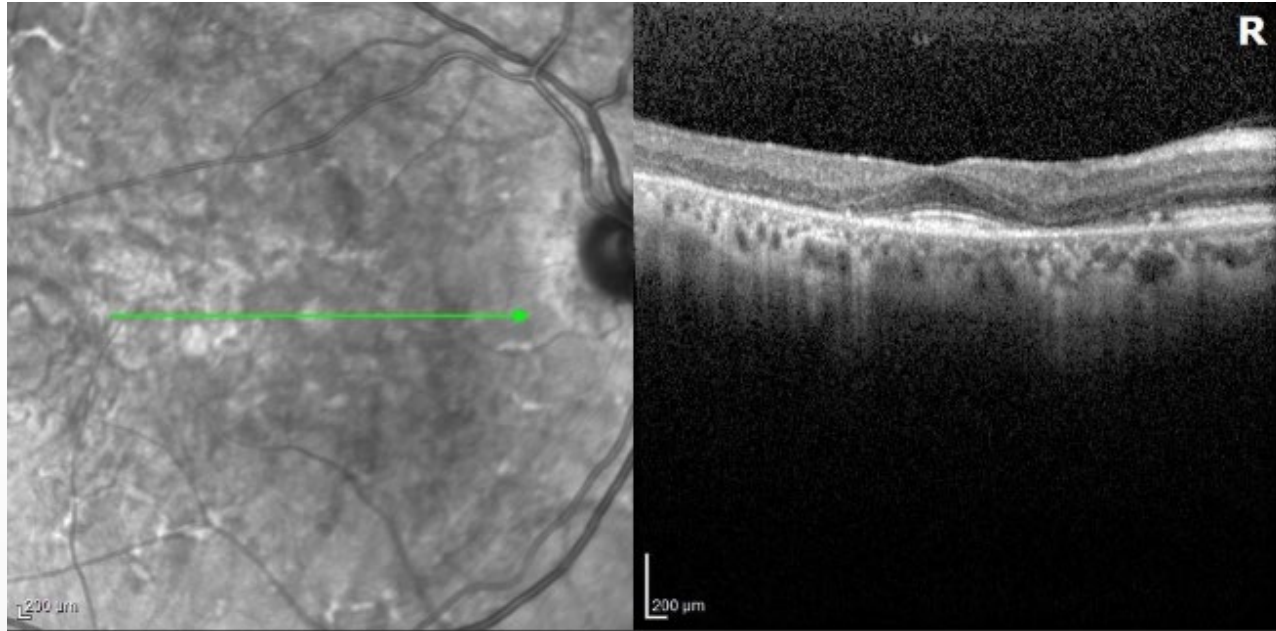
<https://webeye.ophth.uiowa.edu/eyeforum/atlas/pages/Stargardt-disease-13.htm>



Fundus Autofluorescence



OCT Macula



Assessment

55 yo WM with Stargardt's Disease

- Stable retinal disease
- BCVA 20/20 OU
- Decrease in vision OD due to myopic shift from cataract



Discussion

What is Stargardt Disease?

- MC juvenile macular dystrophy
 - (mostly AR inheritance: “st**AR**gardt”)
- Caused by mutations in the **ABCA4 gene**, which encodes an ATP-binding cassette (ABC) transporter protein expressed by rod outer segments.
- Leads to an accumulation of **lipofuscin** within the **RPE**



Discussion

What is Lipofuscin?

- An oxidative by-product within the RPE, formed by the repetitive phagocytosis of photoreceptor outer segments.
- The pigment within lipofuscin that autofluoresces is **A2E**, named for its derivation from 2 molecules of vitamin A aldehyde and 1 molecule of ethanolamine.

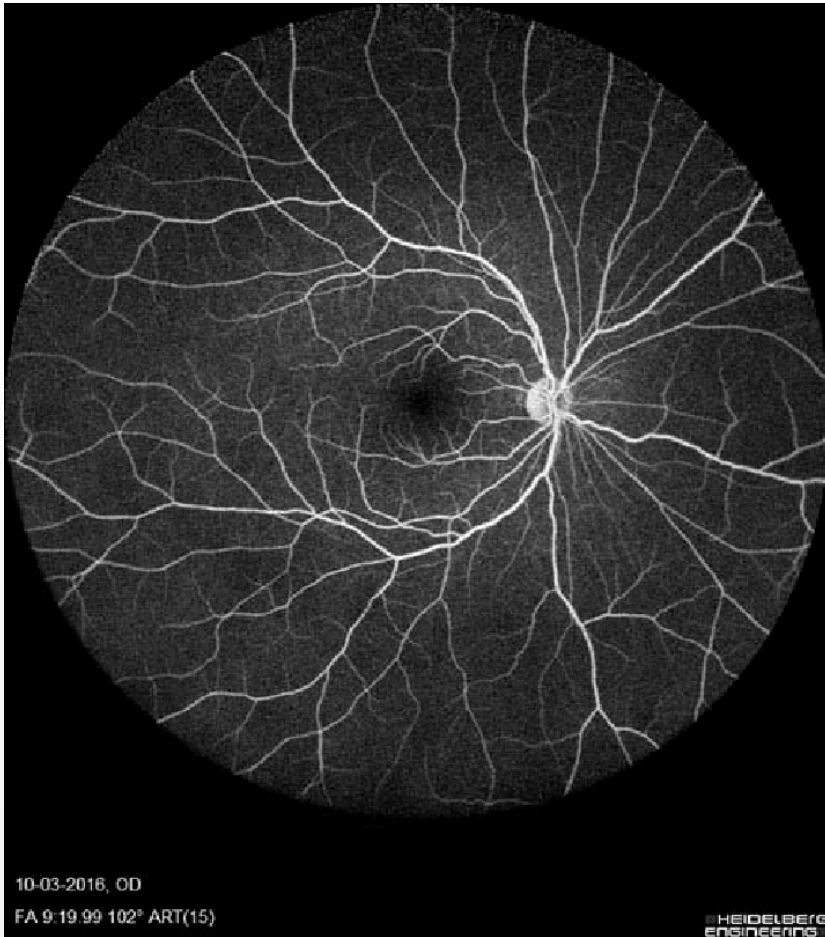


Clinical Features of Stargardt Disease

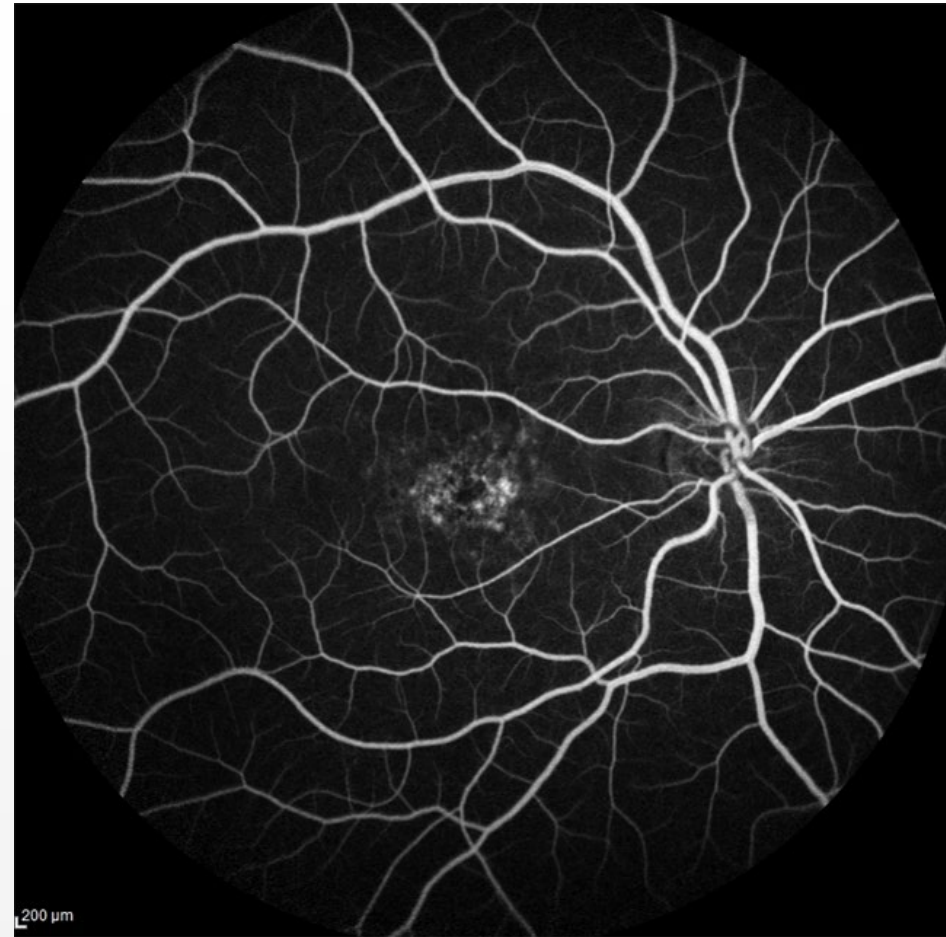
- Age of onset and presenting features varies considerably
- VA usually ranges from 20/50 to 20/200
- May cause a bull's-eye maculopathy
 - DDx includes: Cone and cone-rod dystrophies, Plaquenil toxicity, AMD, chronic macular hole, central areolar choroidal dystrophy, ceroid lipofuscinosis
- FA shows “dark choroid” (not specific)



Normal vs “Dark Choroid”



https://www.researchgate.net/figure/Normal-wide-field-fundus-fluorescein-angiography-with-Heidelberg-Spectralis-R-module_fig5_320724816



<https://imagebank.asrs.org/file/26818/stargardt-disease-dark-choroid>



Treatment?

- Nothing definitive yet ☹
 - Photoprotection and low vision aids
- Current treatments under investigation:
 - Supplementation with deuterium-enriched vitamin A (prevents dimerization in mouse models)
 - Isotretinoin use (dampens A2E deposition)
 - **Gene replacement therapy!**



References

Spaide RF. Fundus autofluorescence and age-related macular degeneration. *Ophthalmology*. 2003;110:392–399

2017-2018 Basic Clinical Sciences Course, Section 12: Retina and Vitreous. American Academy of Ophthalmology, 2017. Pages 226-7

<https://www.aao.org/bcscsnippetdetail.aspx?id=b05aa572-6de6-437c-a992-eee8c19b1604>

https://eyewiki.aao.org/Stargardt_disease/Fundus_flavimaculatus

