Retina Start

Clinical Case

Murilo Ubukata Polizelli
1st Year Retina Fellow

Medical History

- M.G.F.T, 57 years old, white, female, merchant, born and raised in São Paulo.

- CC: Lesion in the left eye.

- During a routine eye exam, the patient was told that she had a retinal scar in the left eye. She denies any low visual acuity or other symptoms.

Medical History

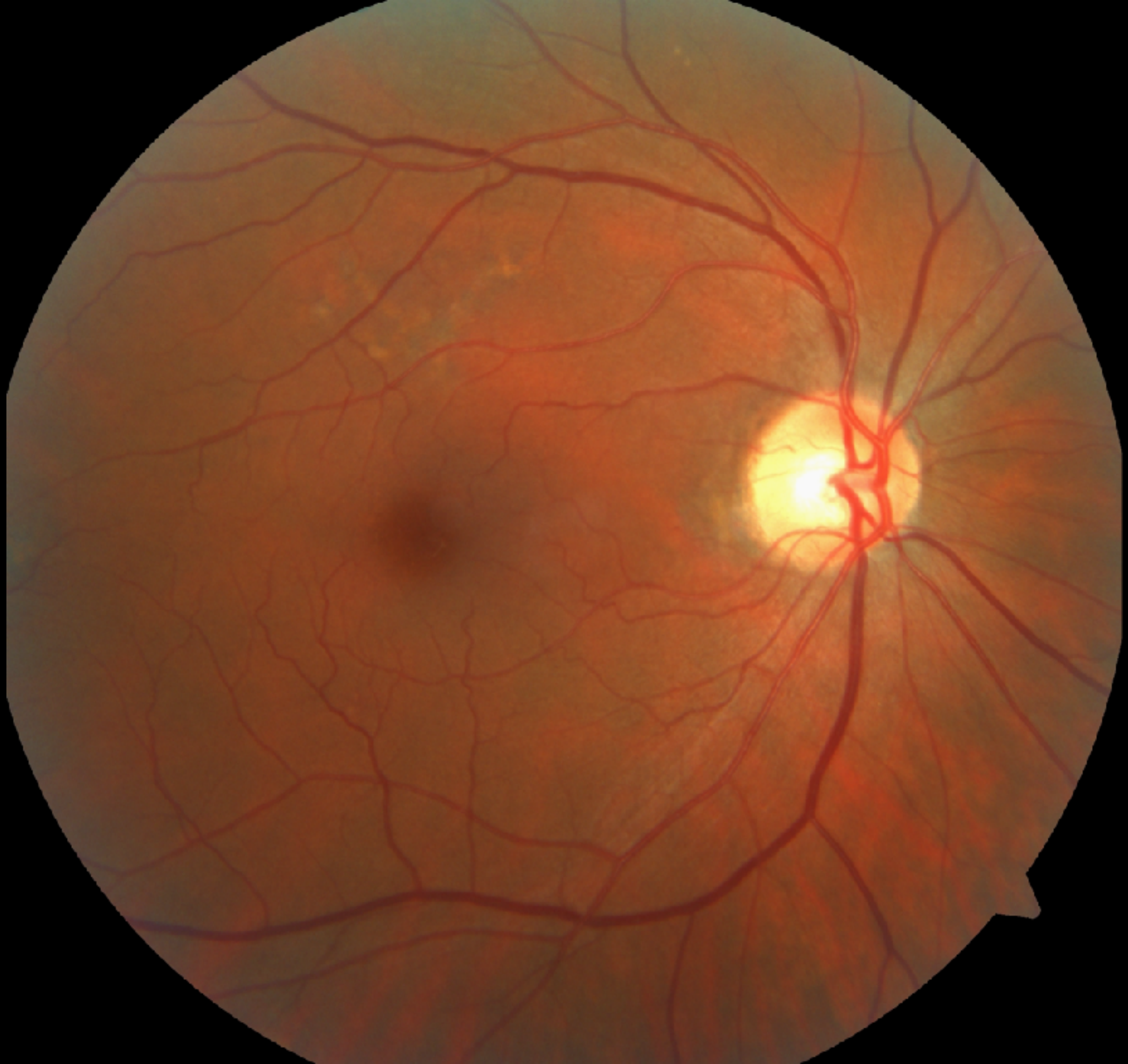
- Previous Medical History:
  - Type 2 Diabetes for 15 years. Use of Metformin
  - Bilateral hearing loss - neurosensory loss in audiometry exam

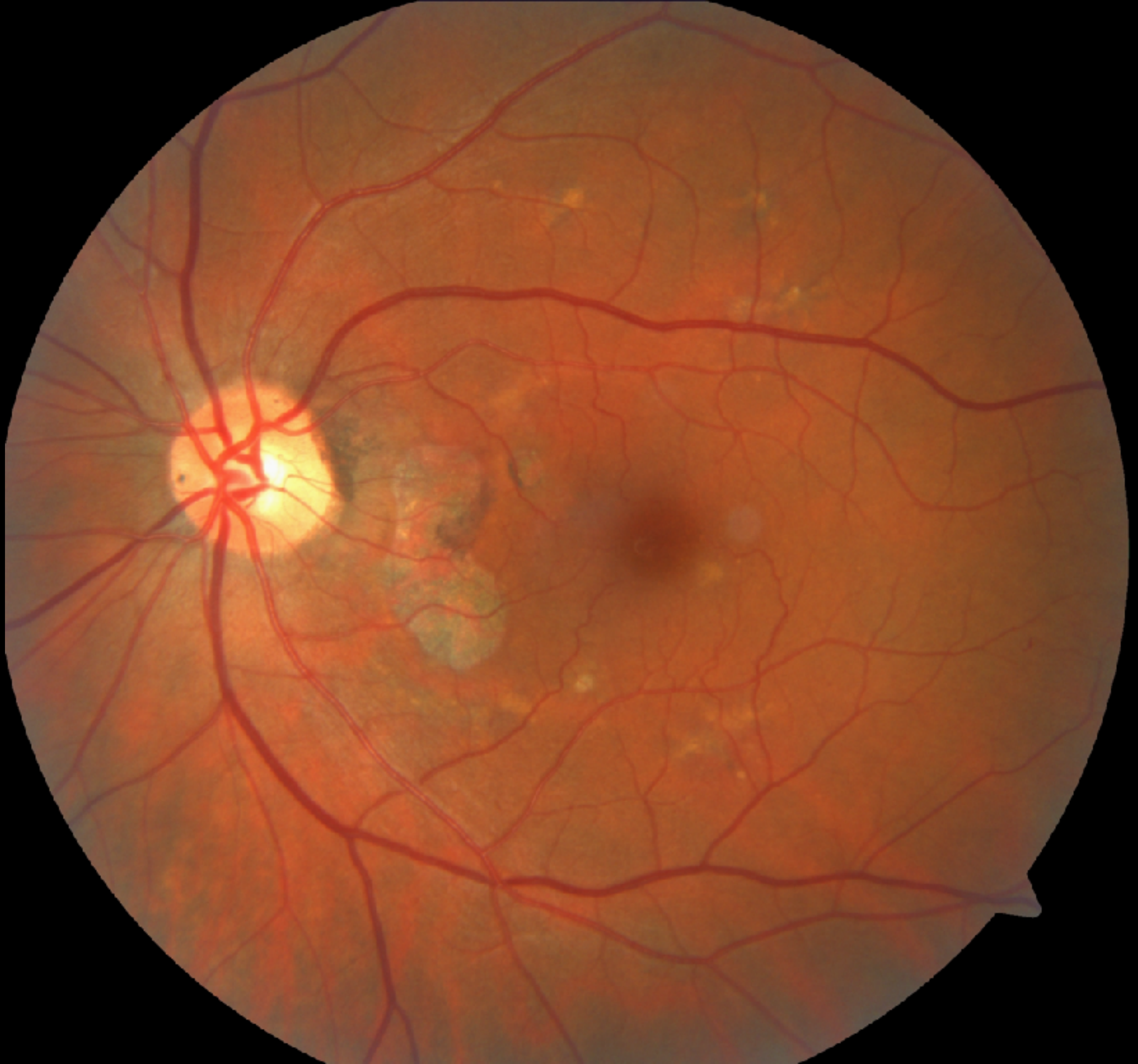
Medical History

- Family History
  - Mother and 2 female siblings with diabetes.
- Past Ocular History: unremarkable.

Ophthalmologic Exam

	OD	OS
Best Corrected Visual Acuity	20/20	20/20
Pupillary Reflex	D4+ / C4+	D4+ / C4+
Extra Ocular Motion	Full	Full
Slit Lamp Exam	Clear conjunctiva, clear cornea, clear lens, deep anterior chamber, no anterior chamber or vitreous cells	Clear conjunctiva, clear cornea, clear lens, deep anterior chamber, no anterior chamber or vitreous cells
Intra ocular pressure	14mmHg	12mmHg
Funduscopy	Photos	Photos





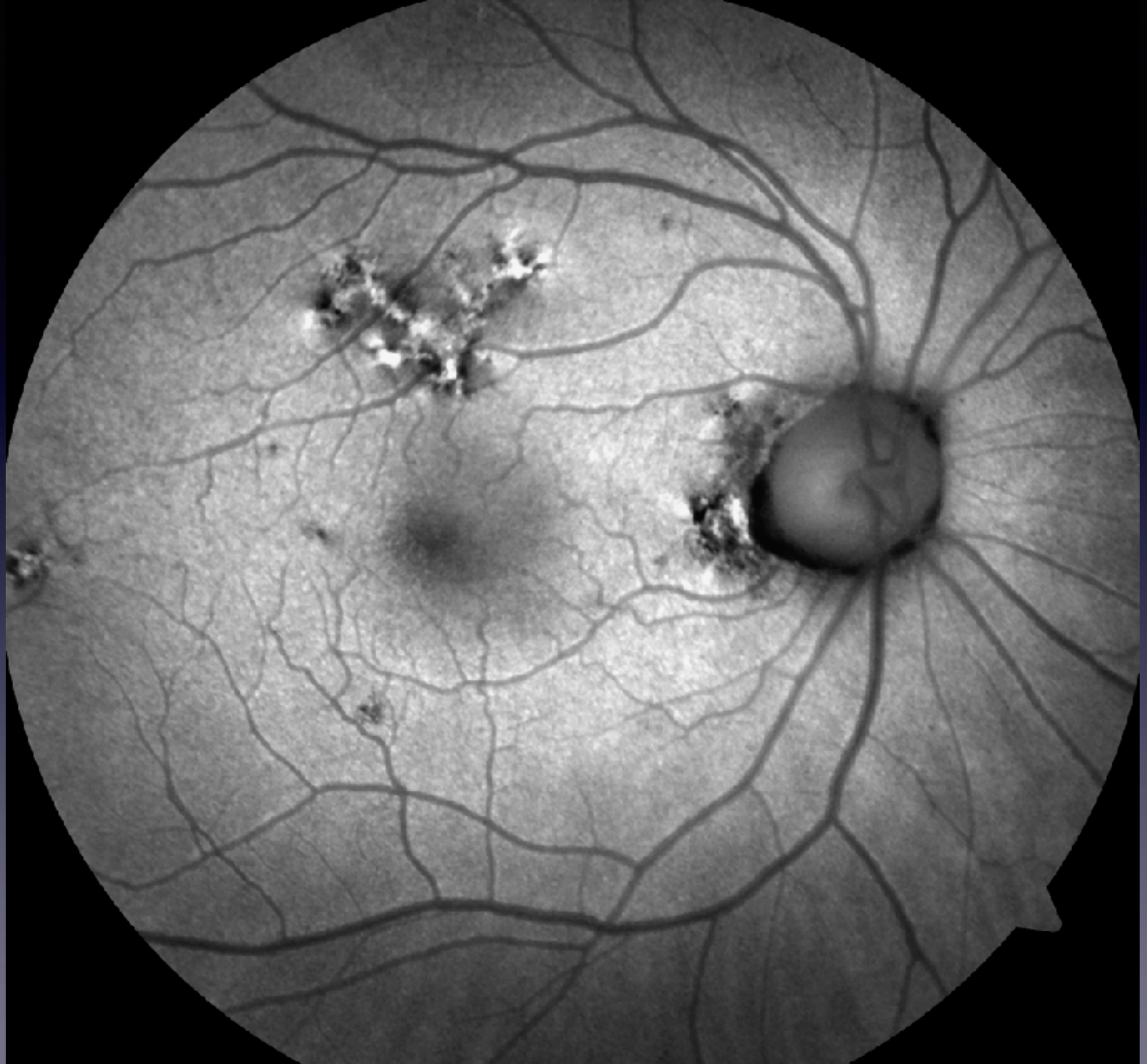
Diagnostic Hypothesis

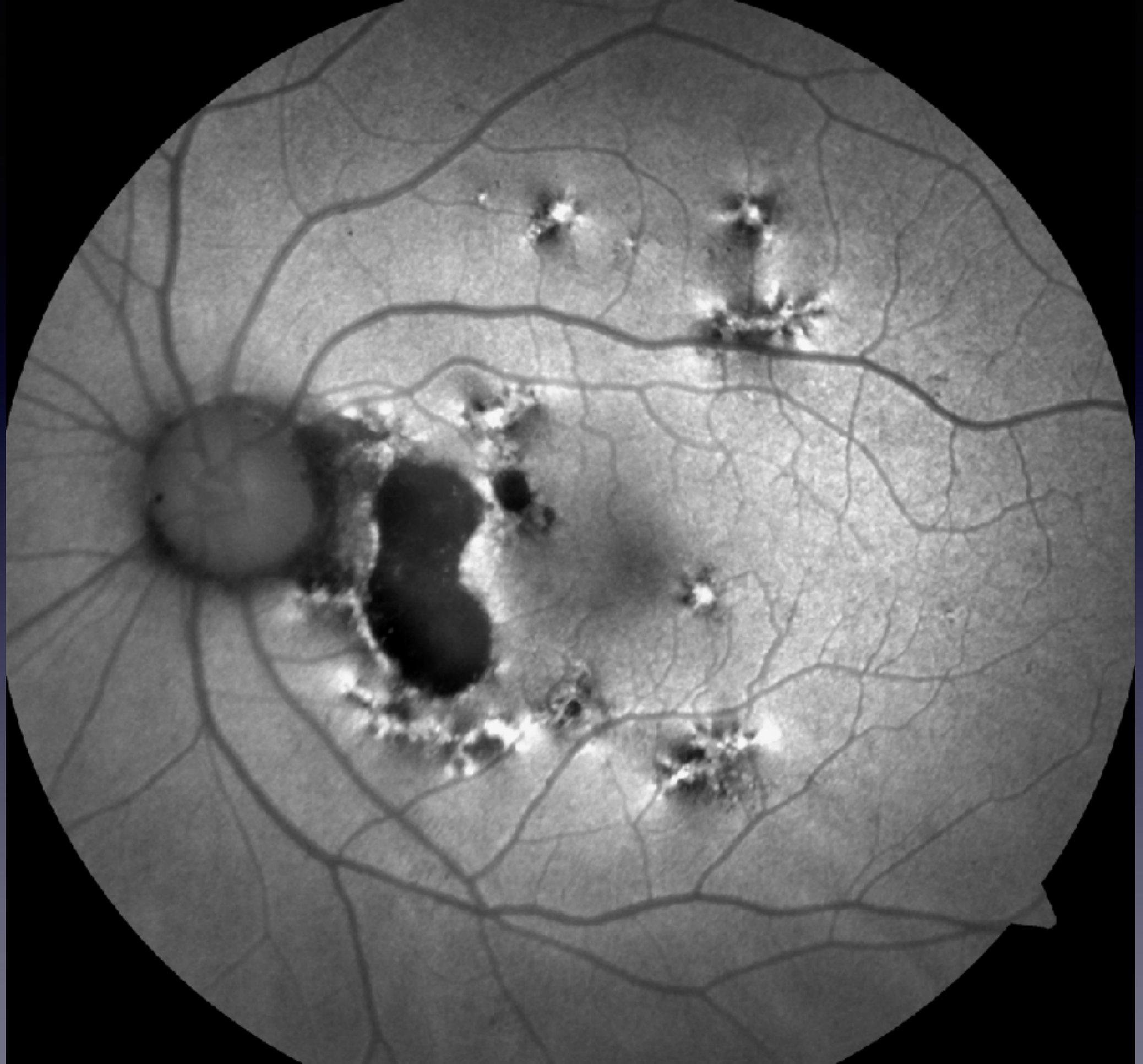
Diagnostic Hypothesis

- Pattern Dystrophies
- Macular Dystrophy - Sorsby
- Laser Marks
- Chorioretinitis Scar
    - Toxoplasmosis

Management

- Fluorescein Angiography
- Optical Coherence Tomography
- Autofluorescence





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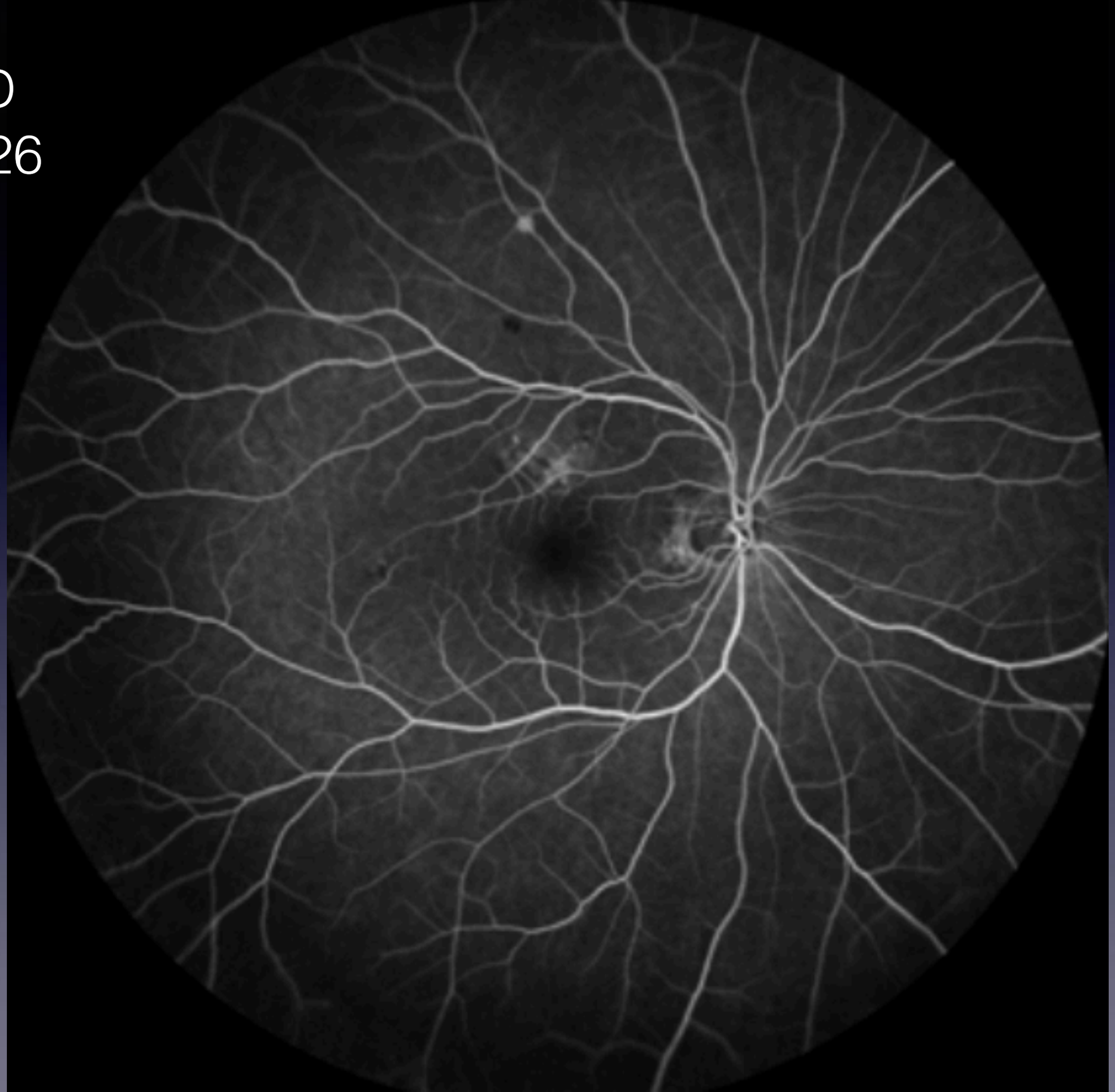

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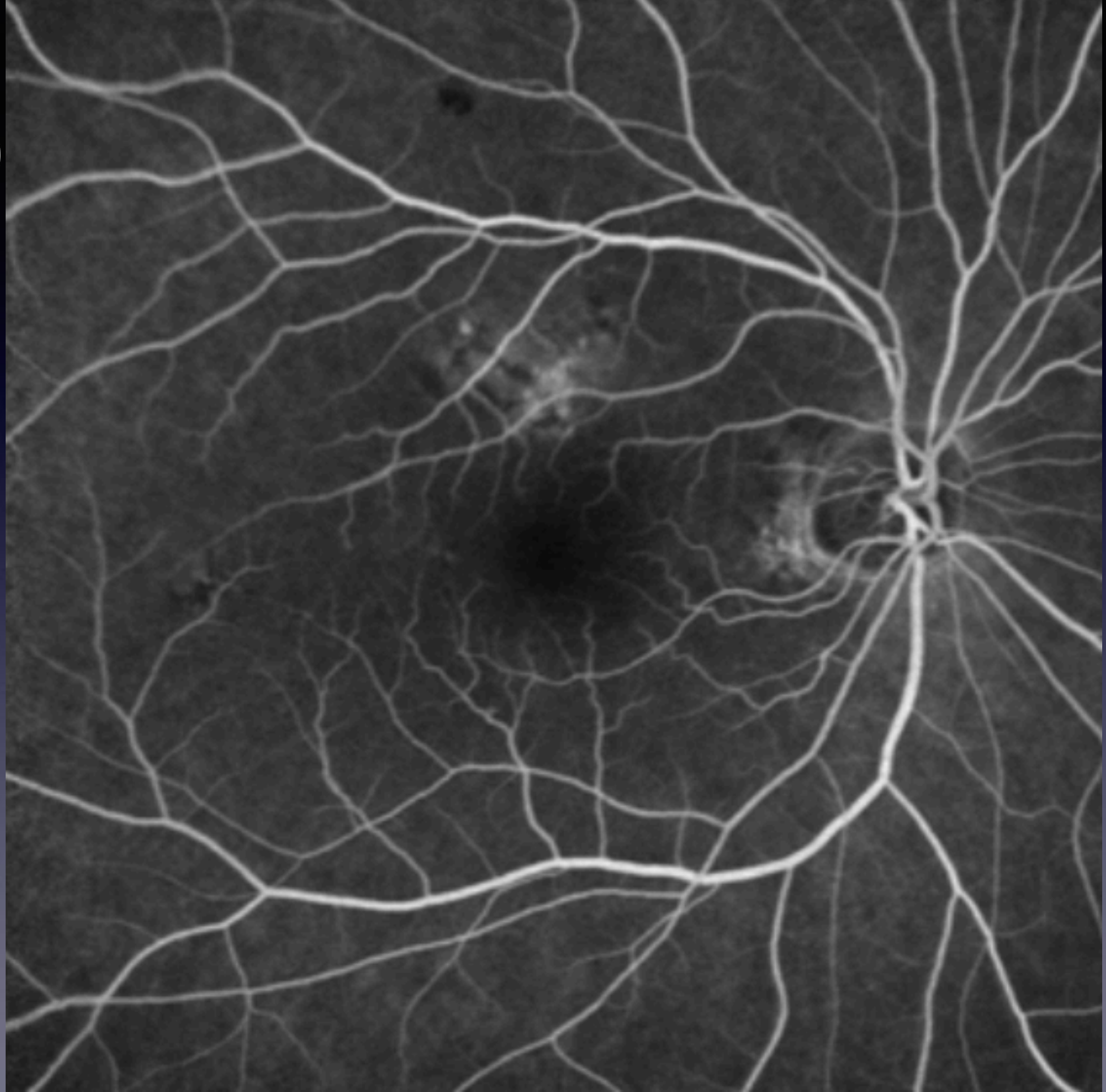

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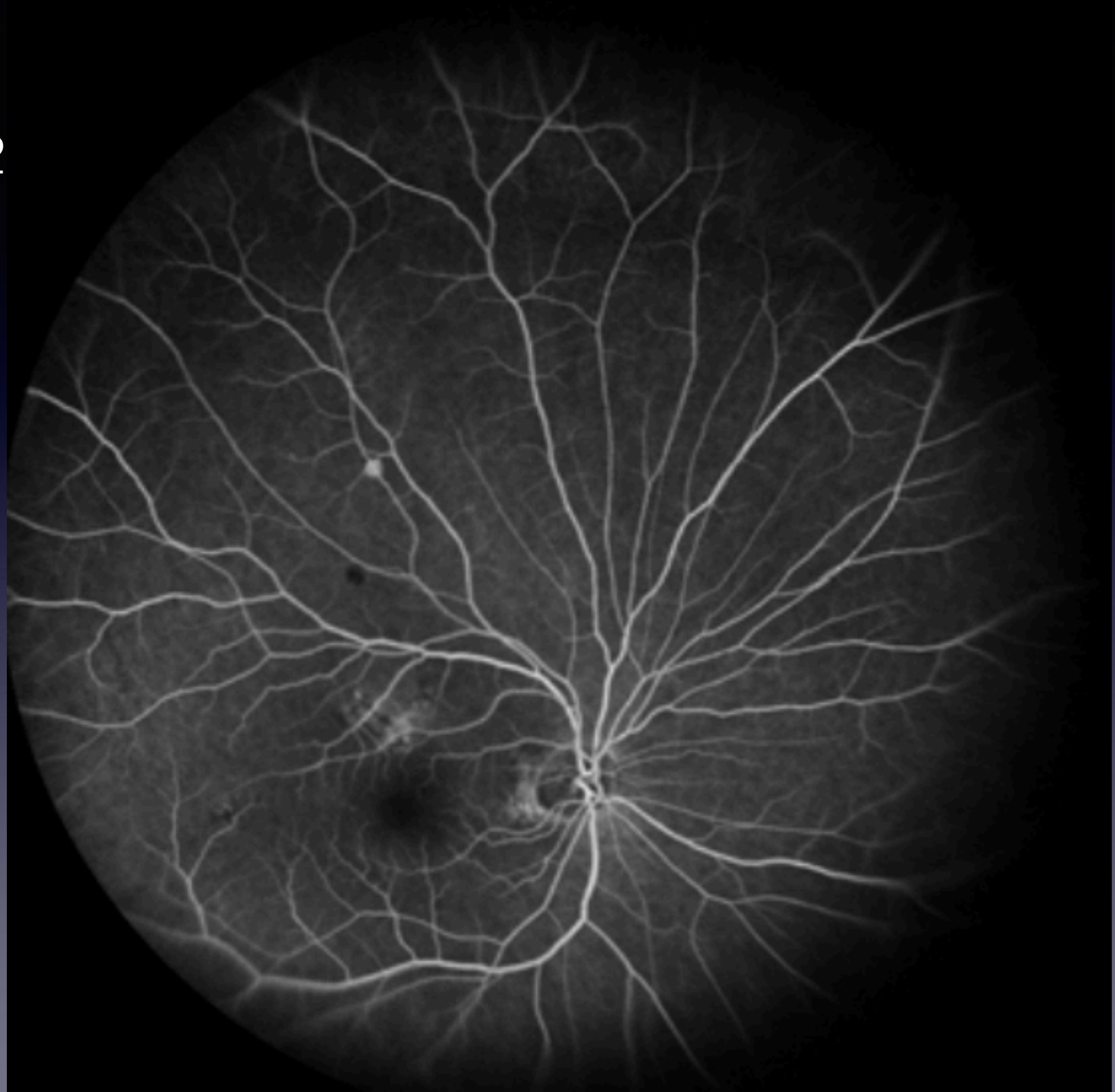

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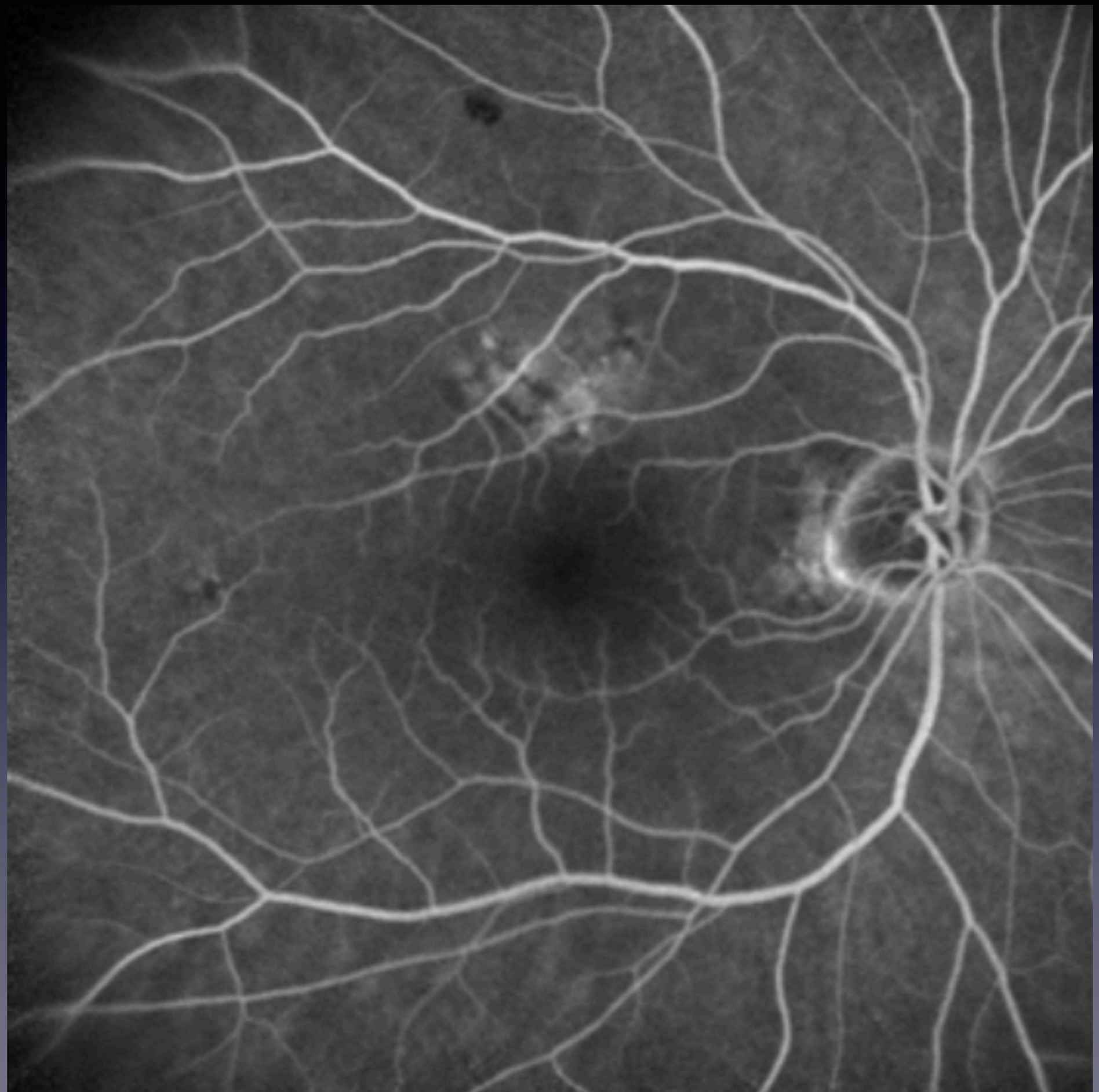

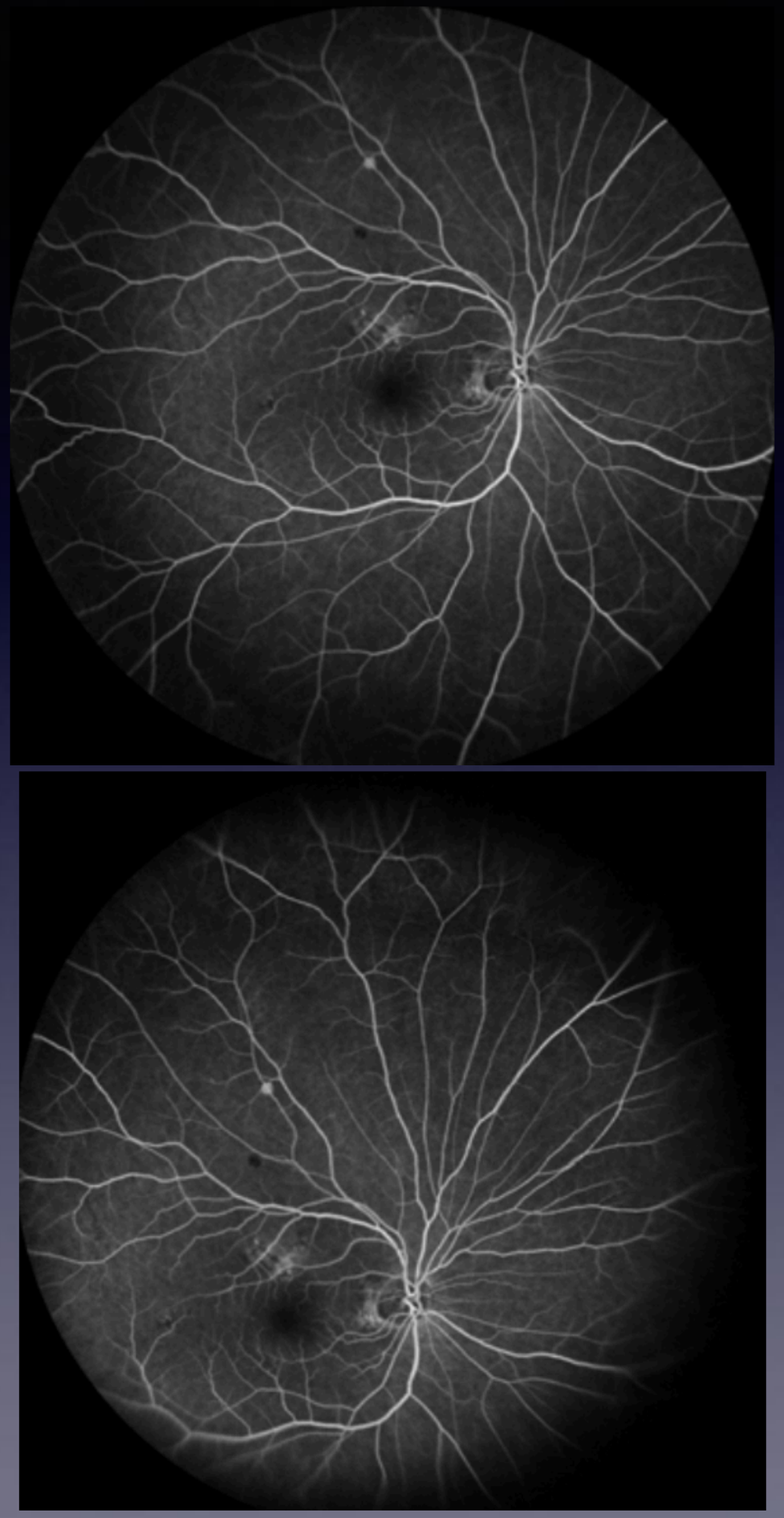

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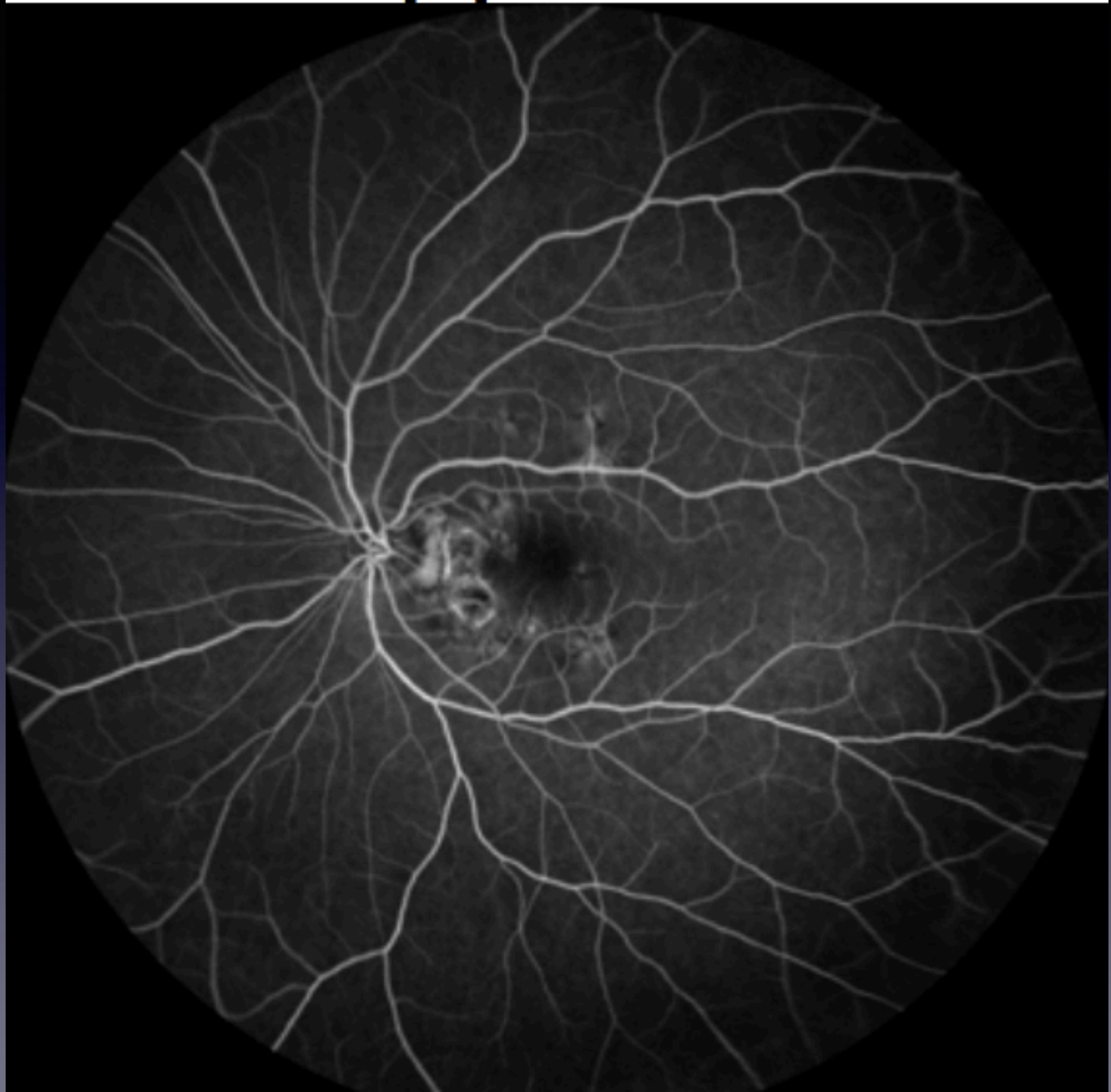

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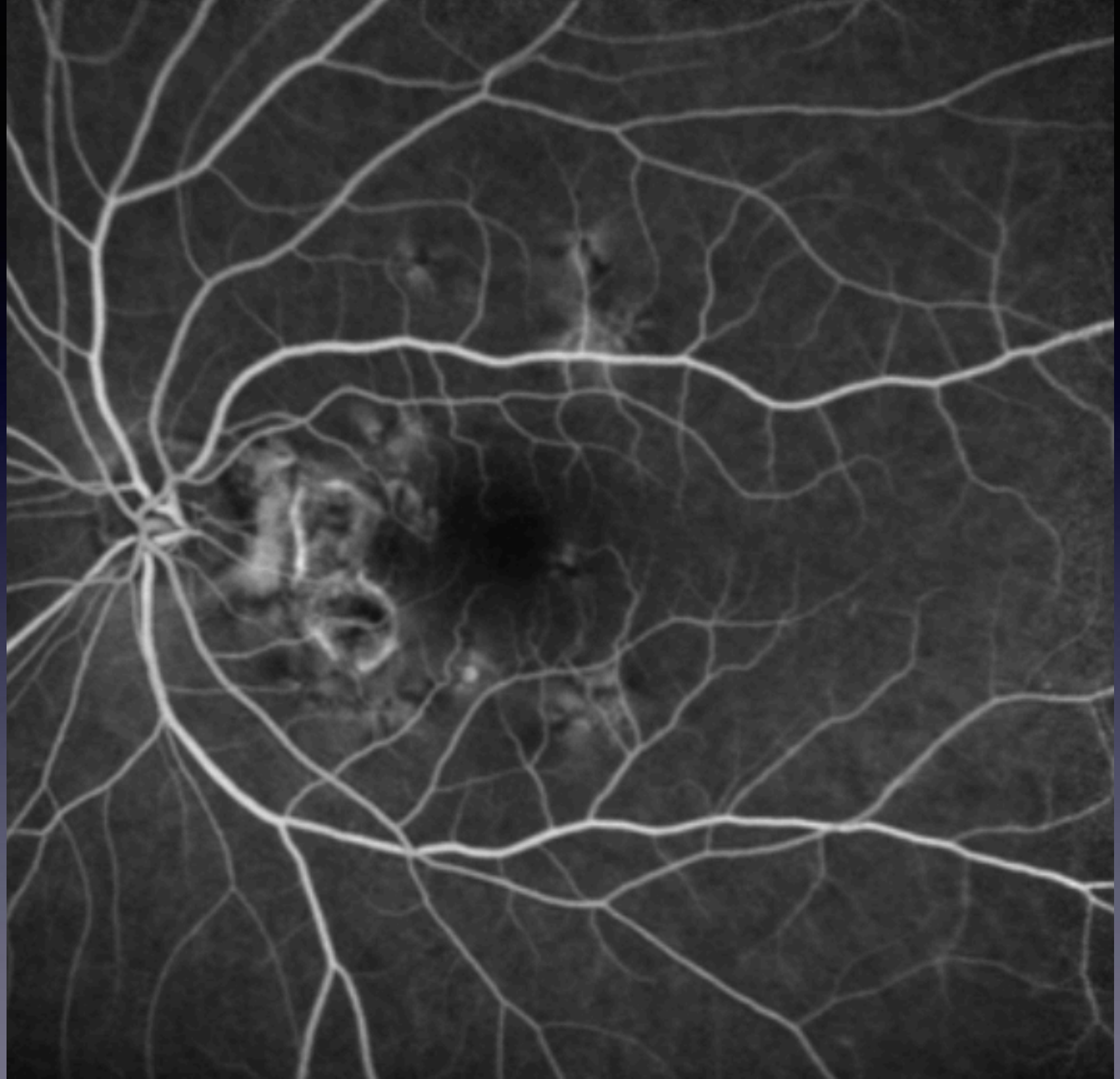

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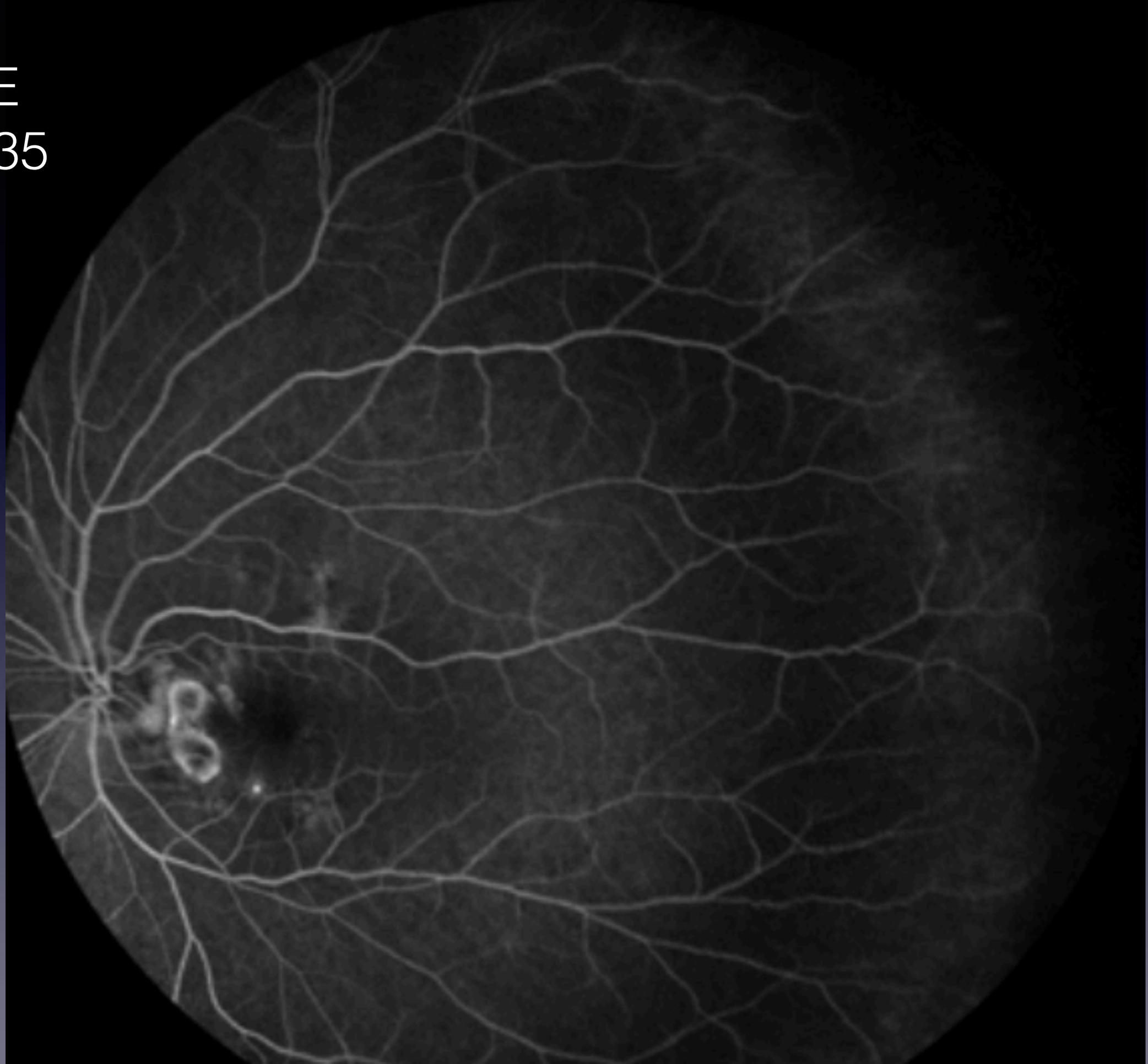

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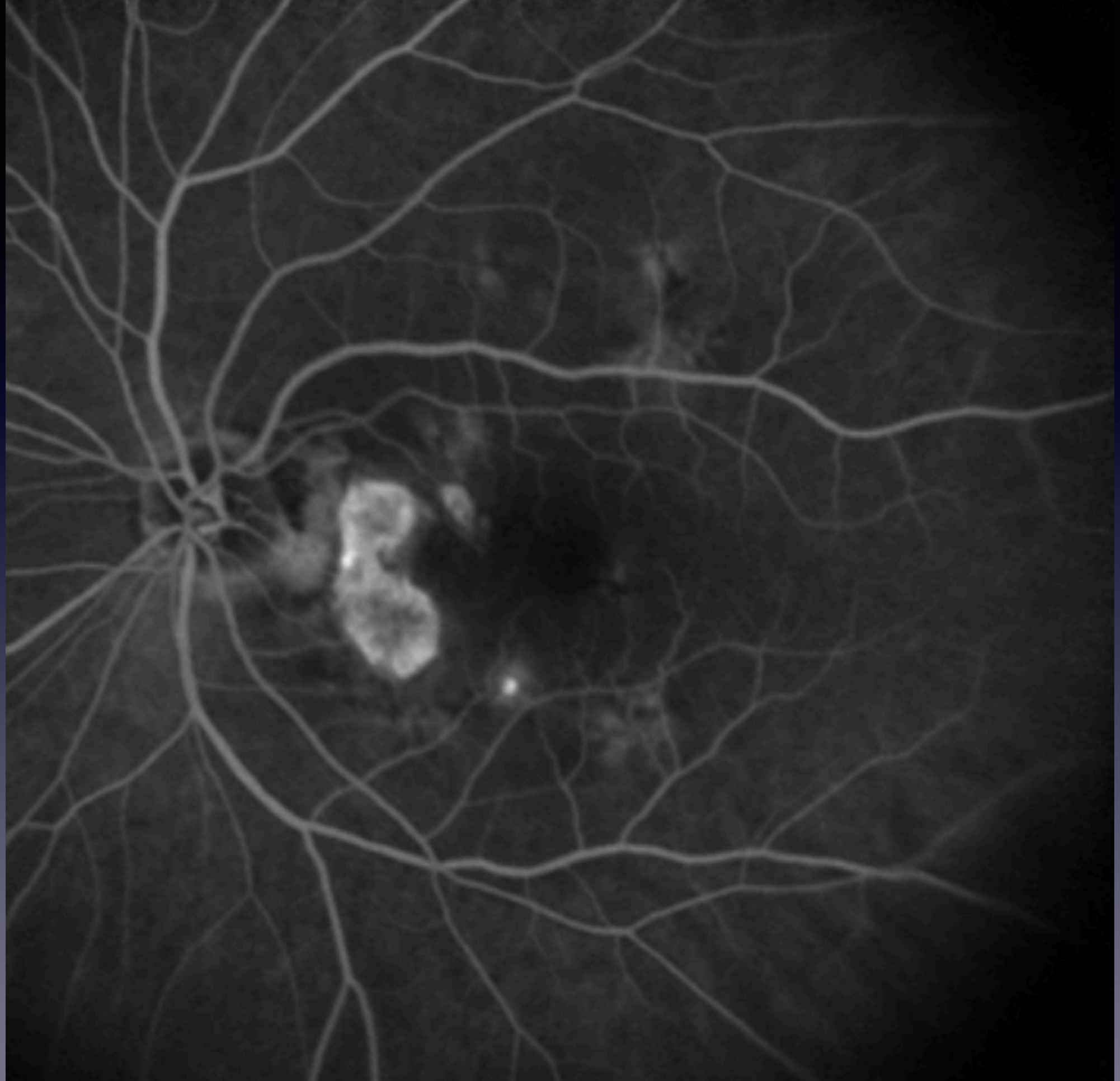

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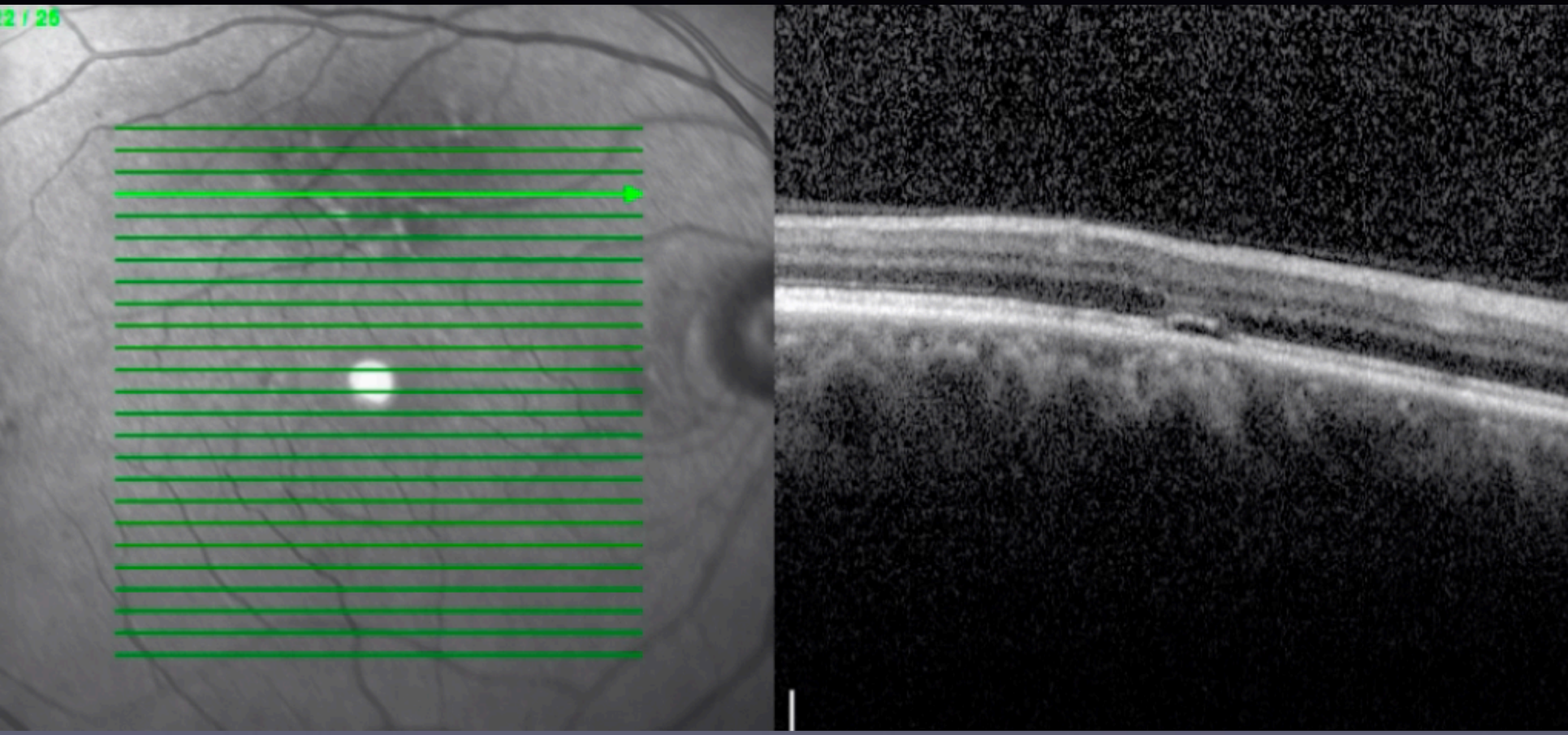

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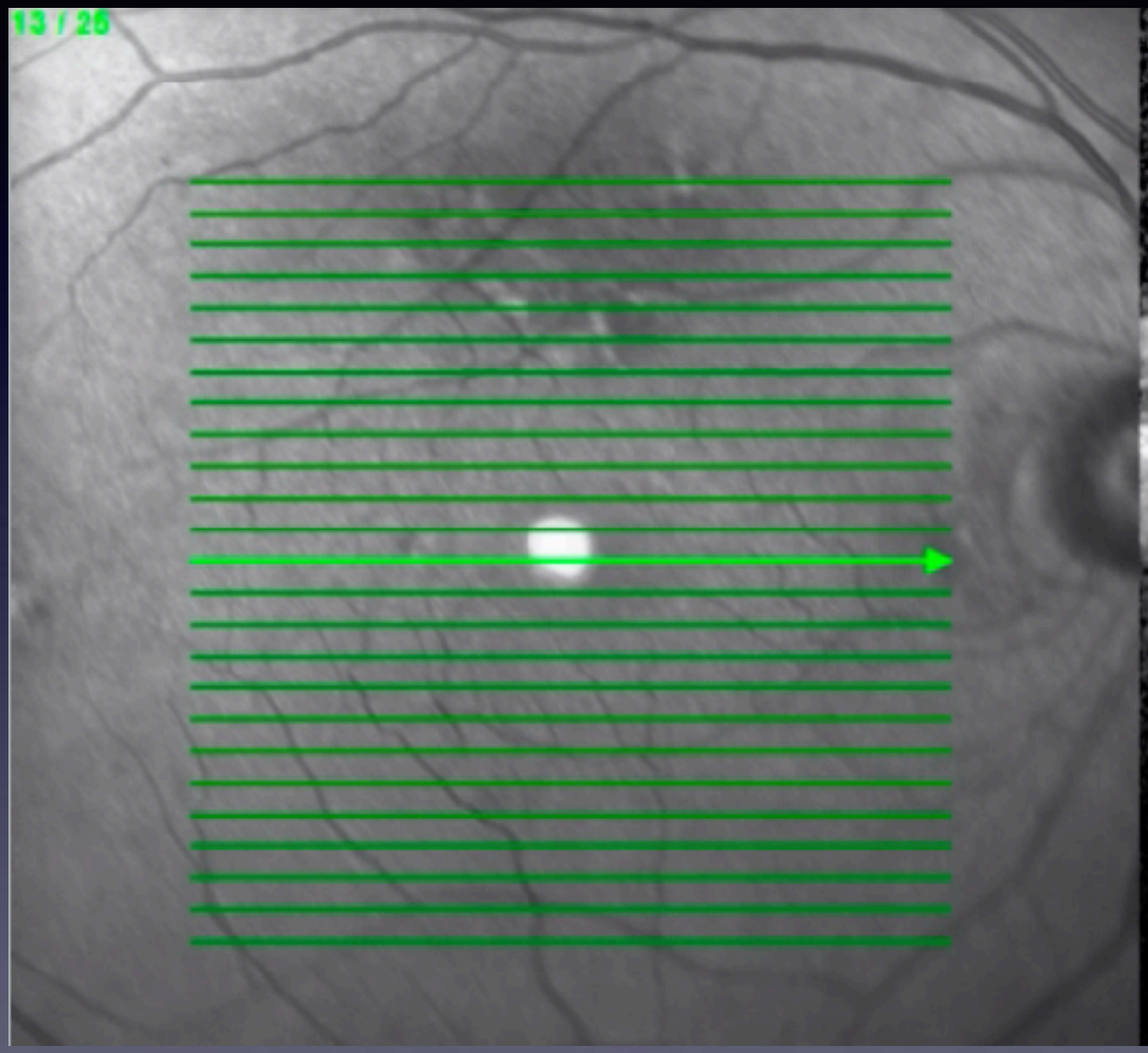
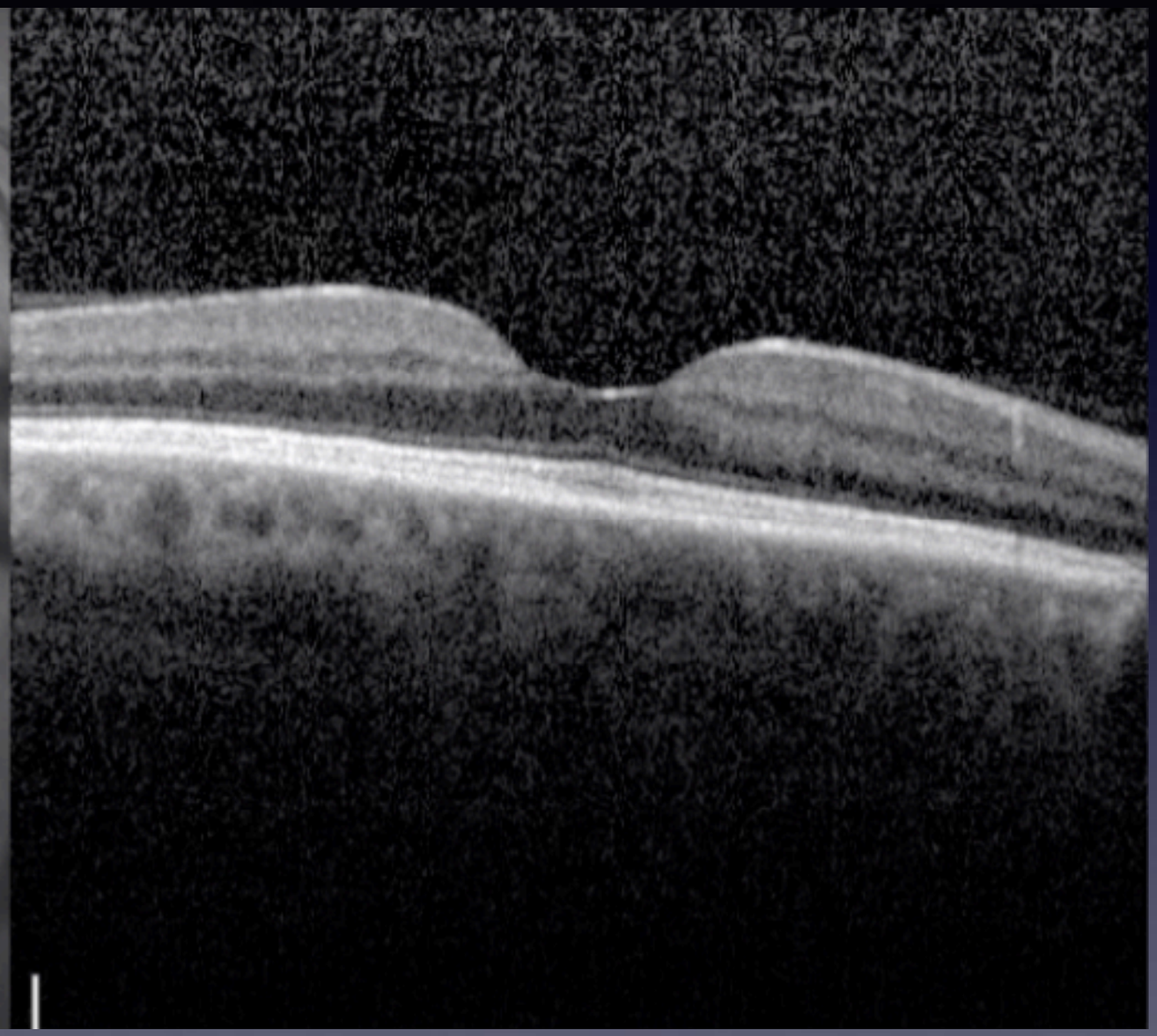
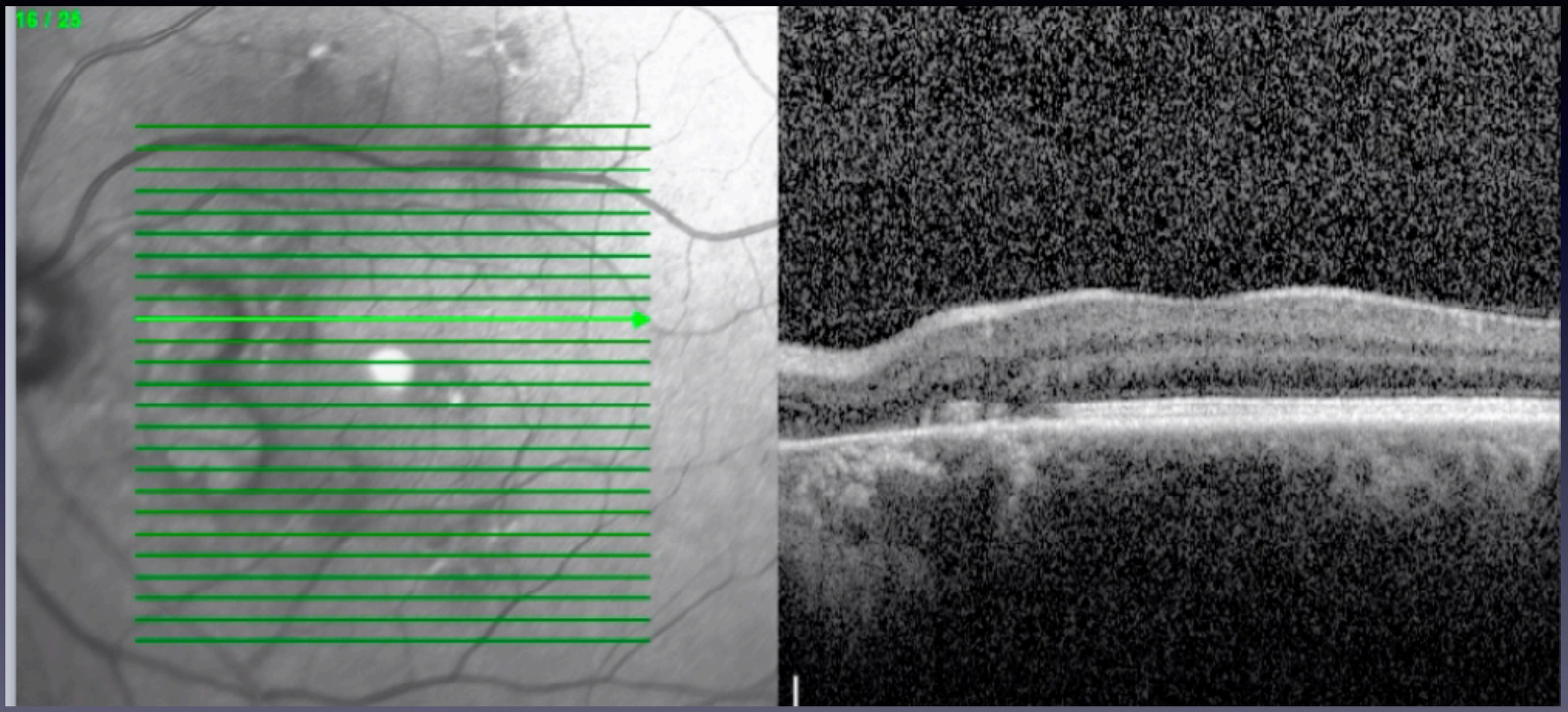




OCT - Olho Esquerdo



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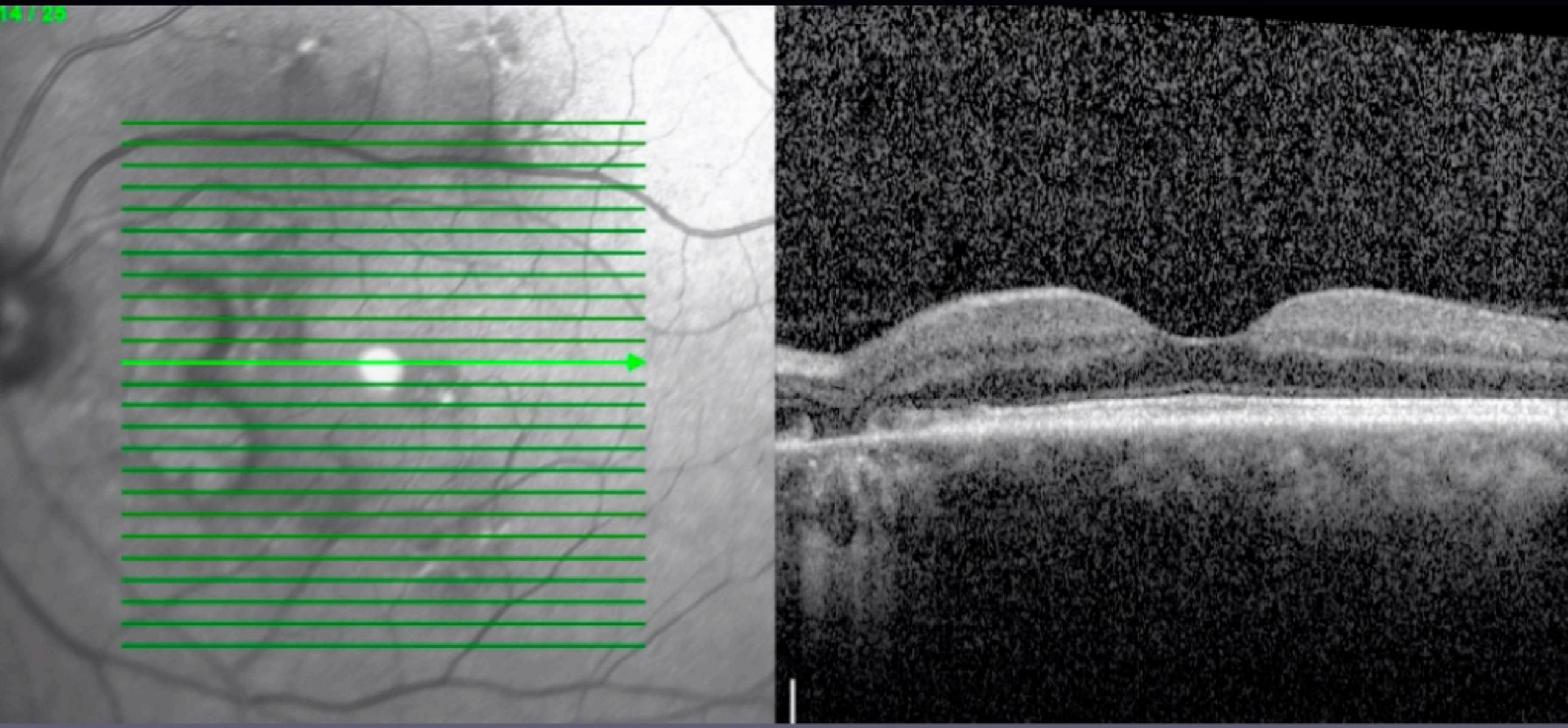

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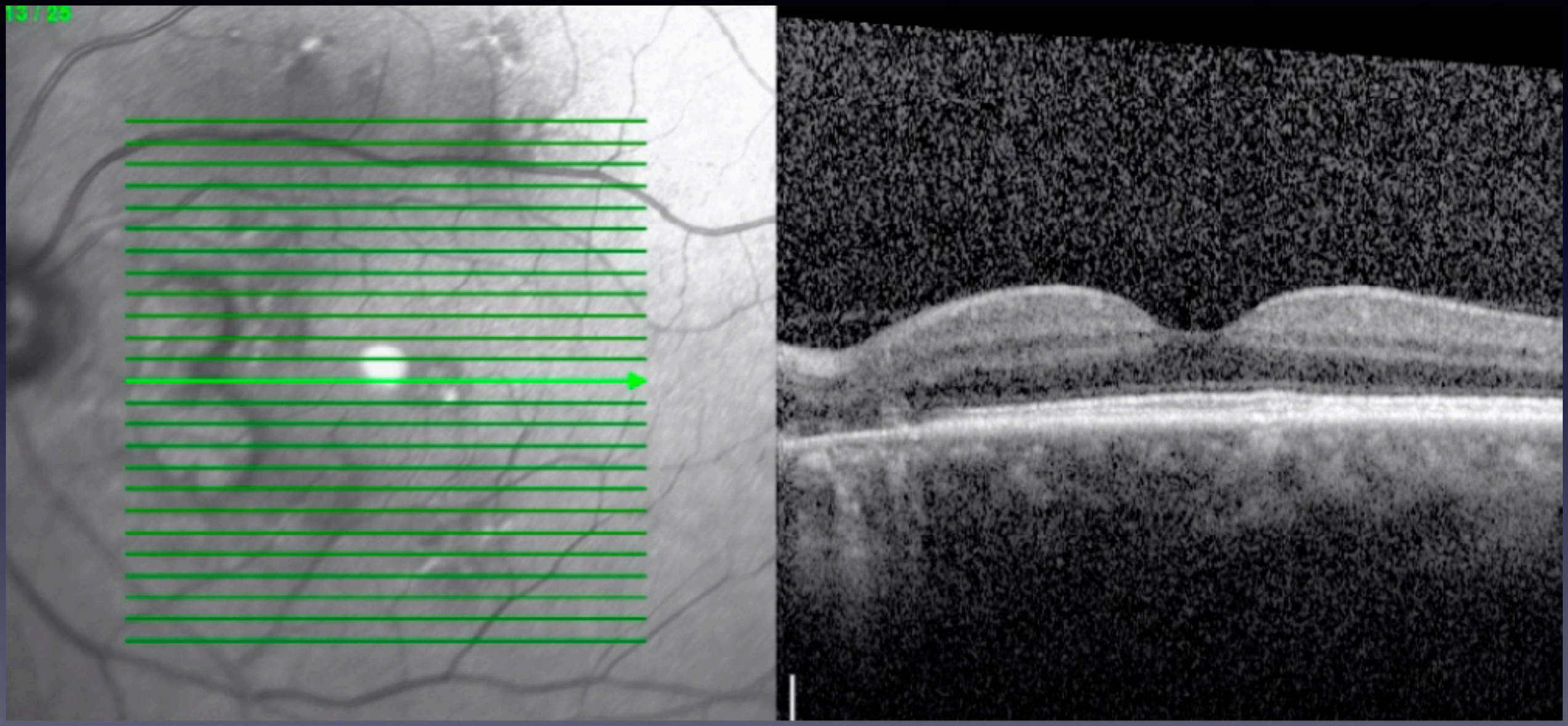
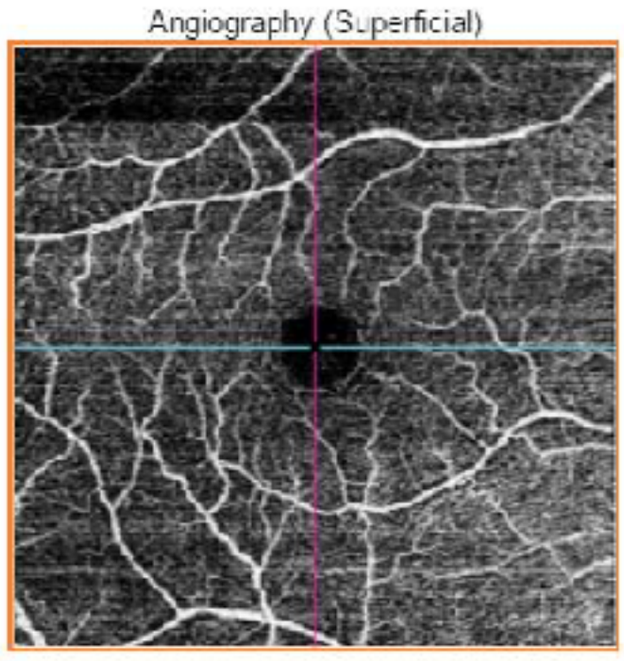
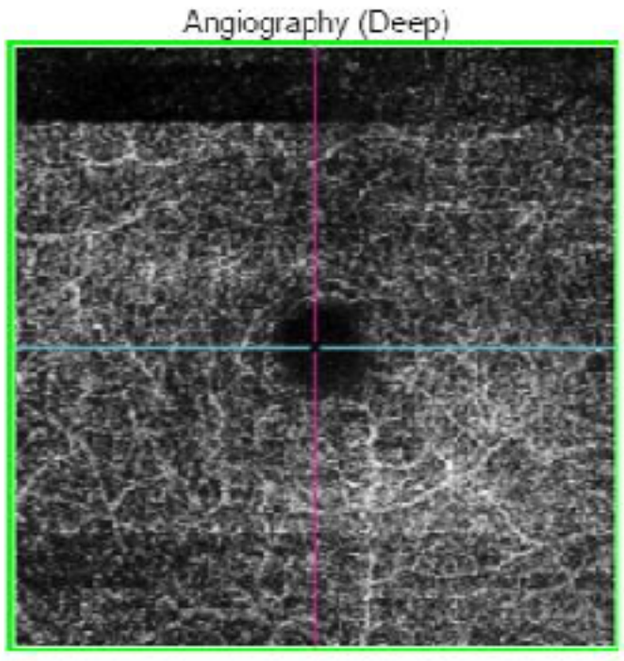
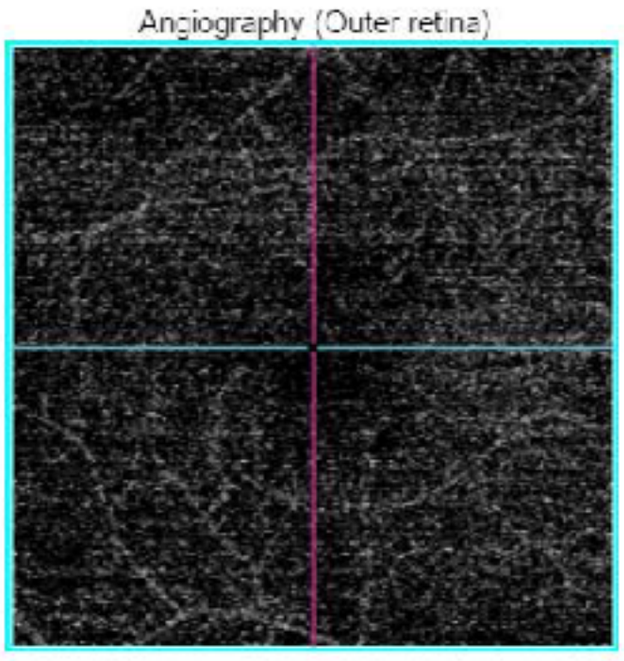
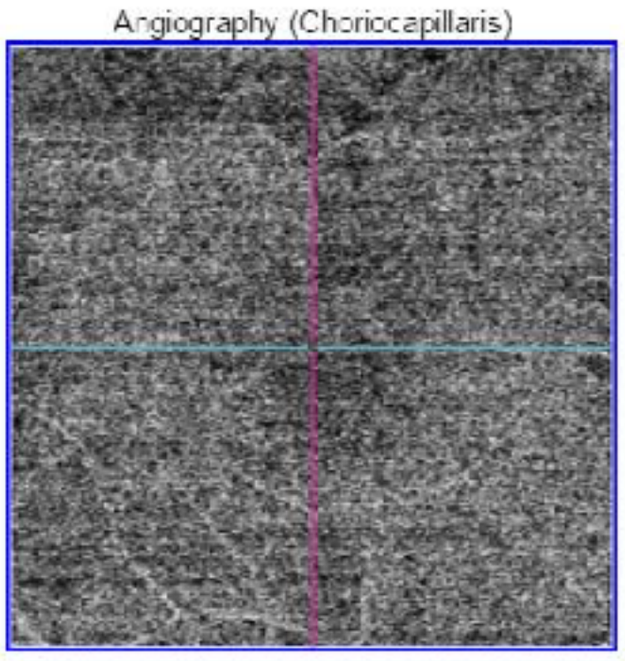


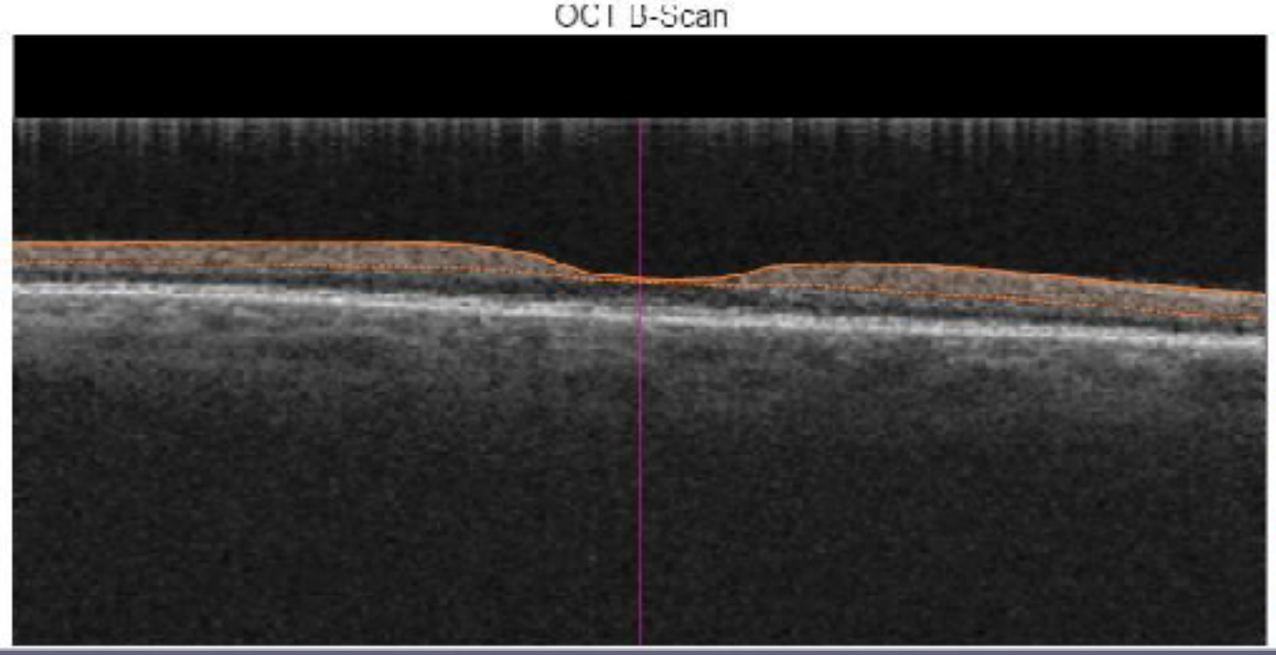
OCT angiography - Right Eye

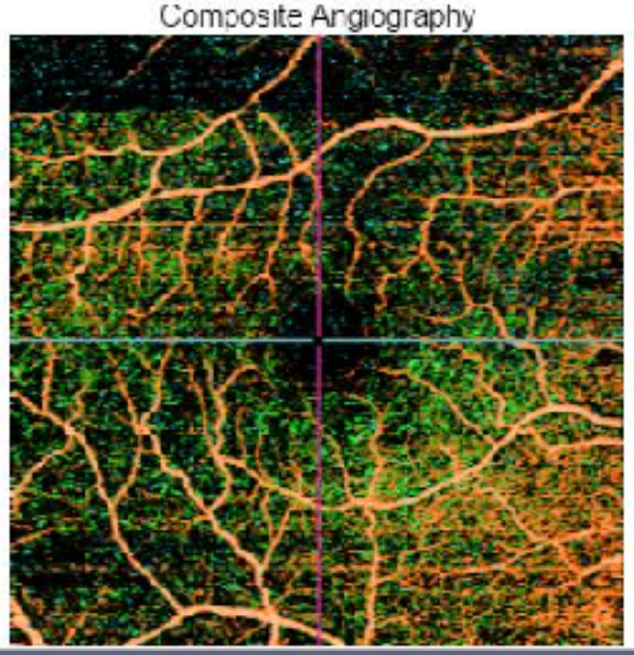


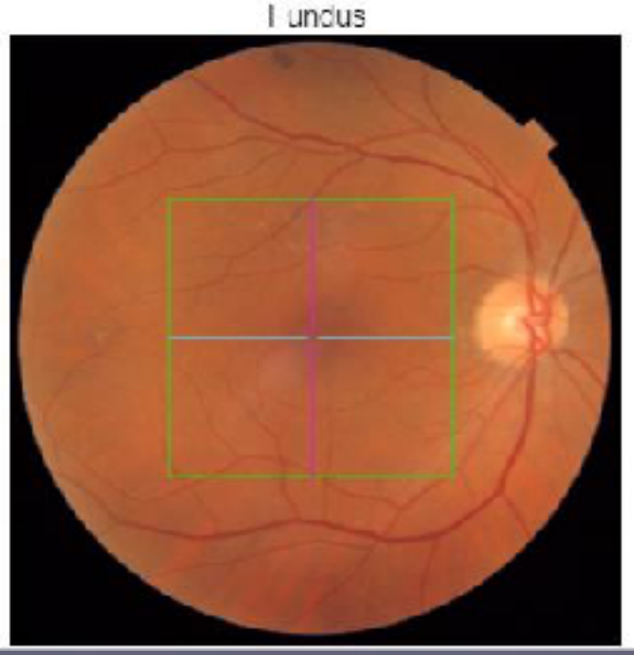




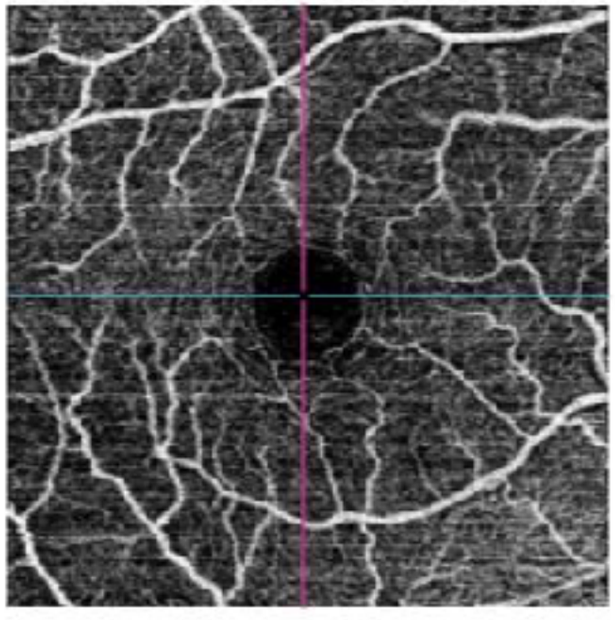


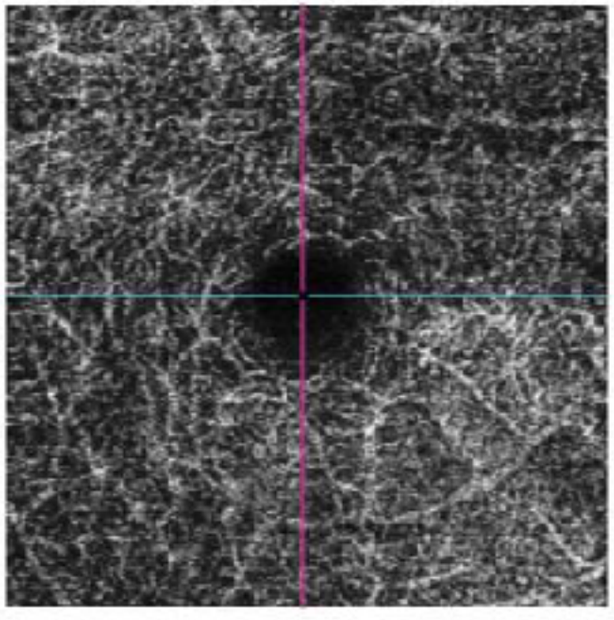


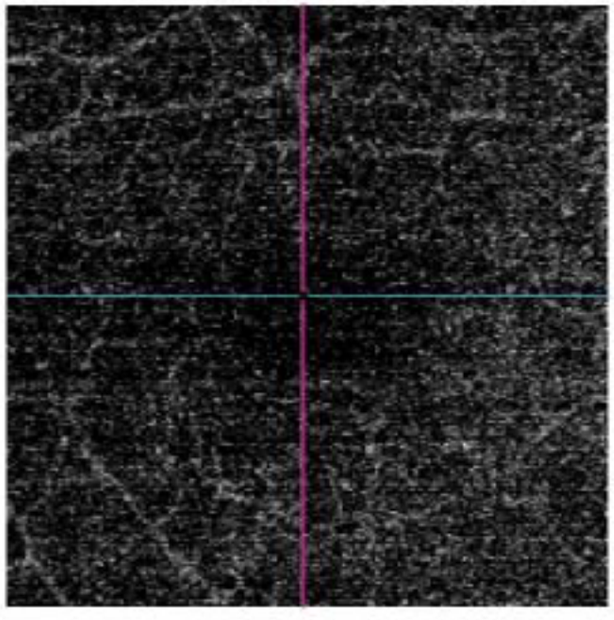


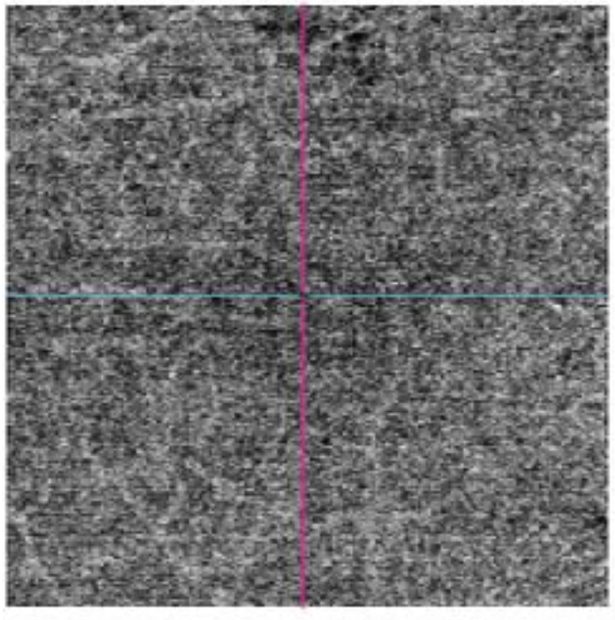


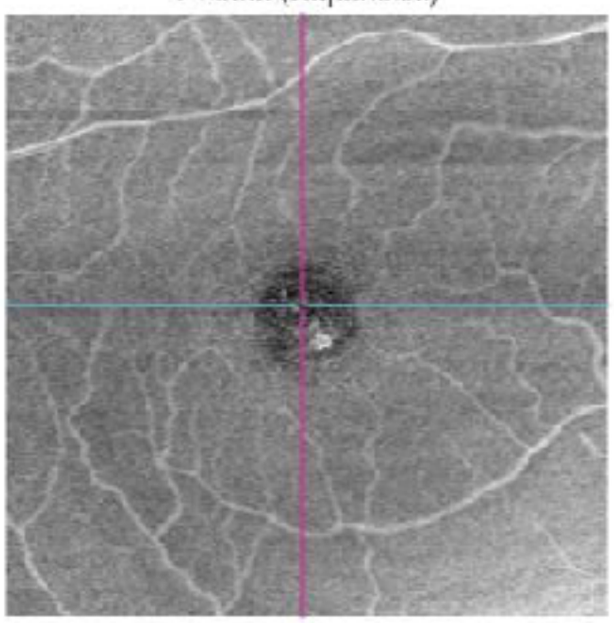
OCT angiography - Right Eye

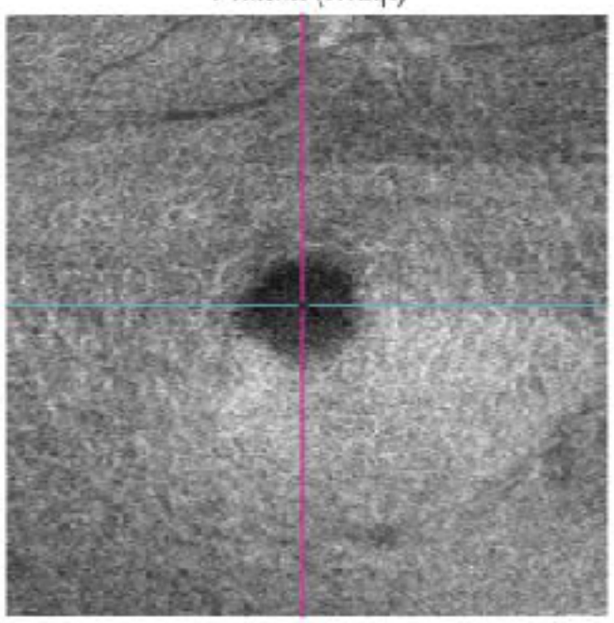


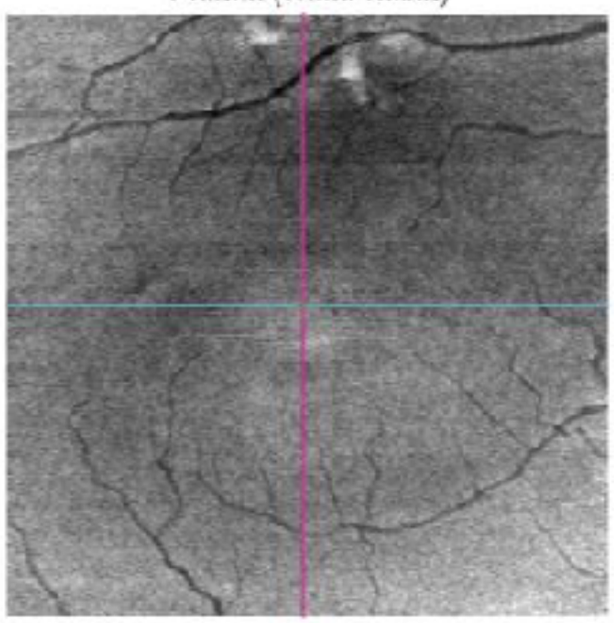


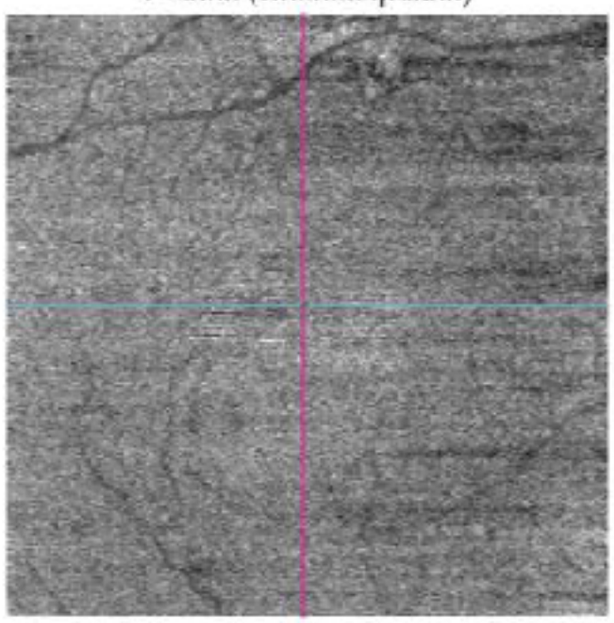








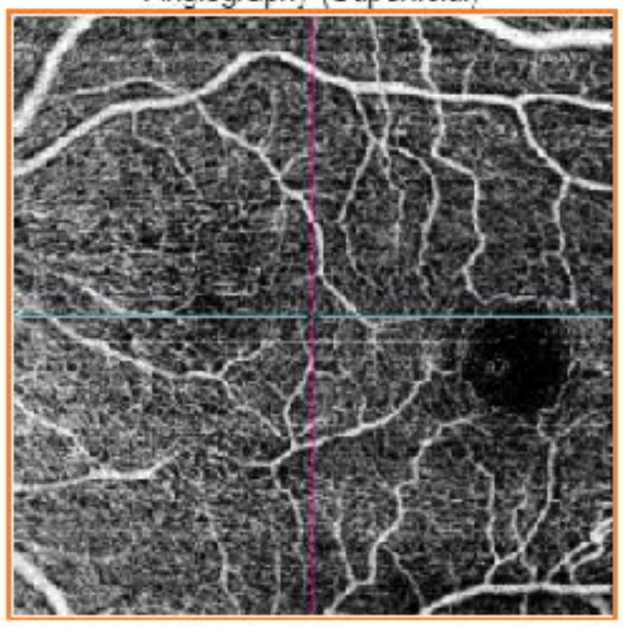




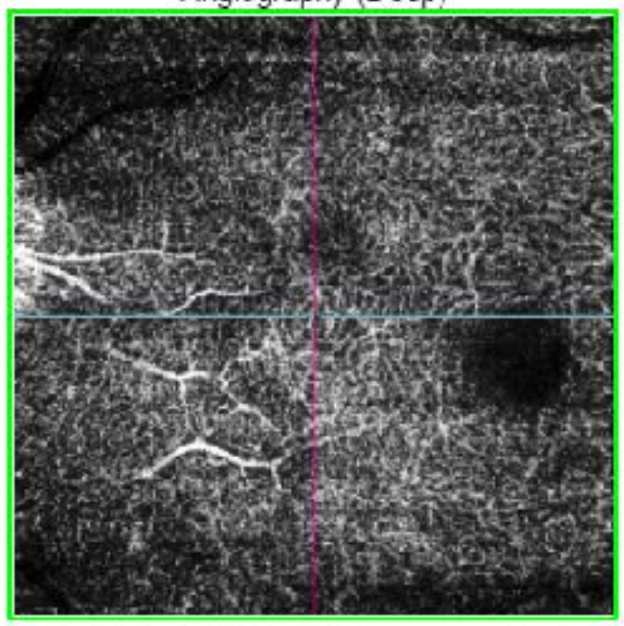
OCT angiography - Left Eye

Capture Date: 8/29/2017

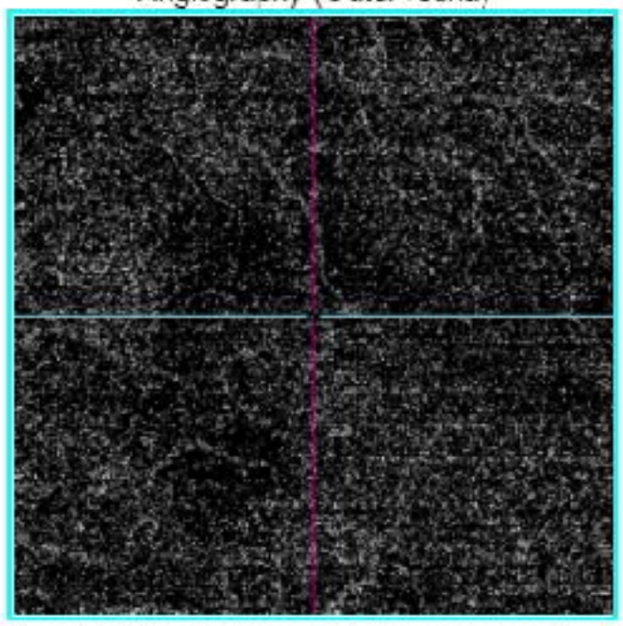
Angiography (Superficial)



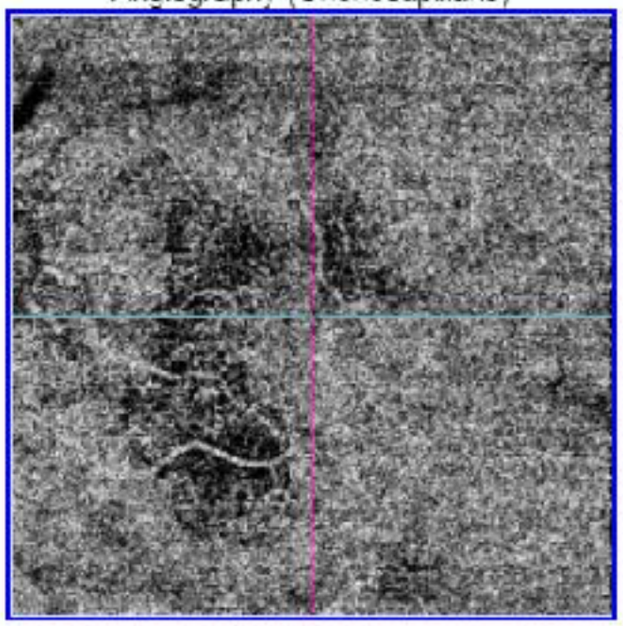
Angiography (Deep)



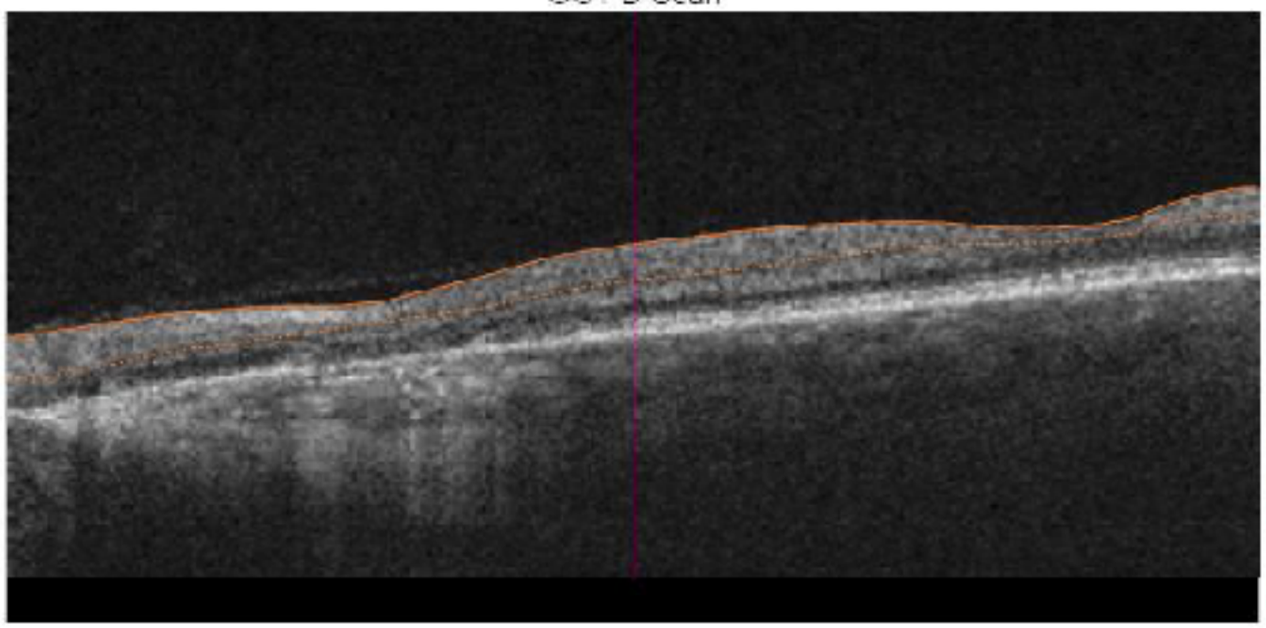
Angiography (Outer retina)



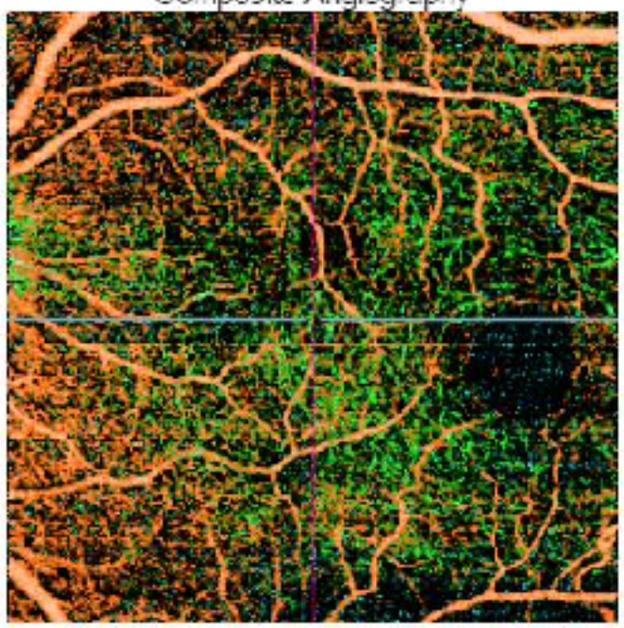
Angiography (Choriocapillaris)



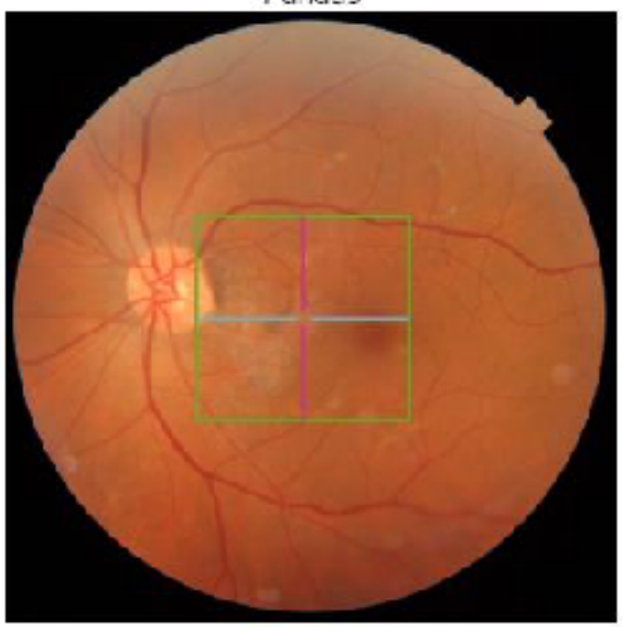
OCT B-Scan



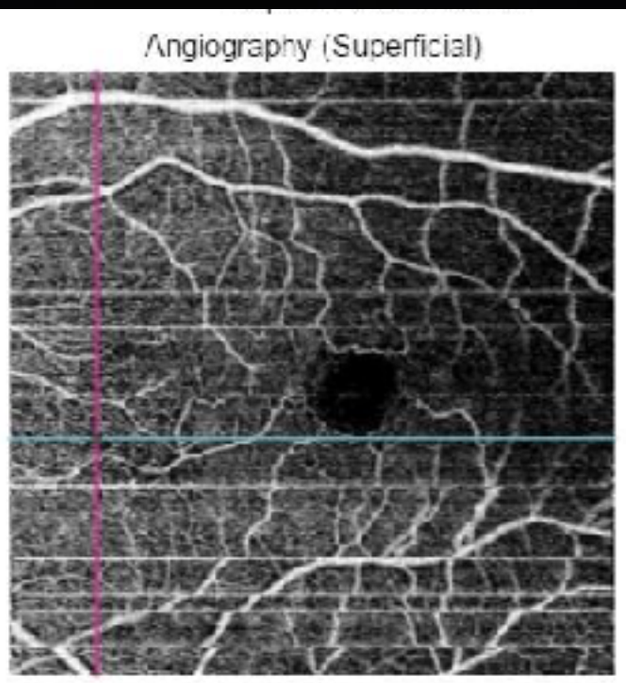
Composite Angiography

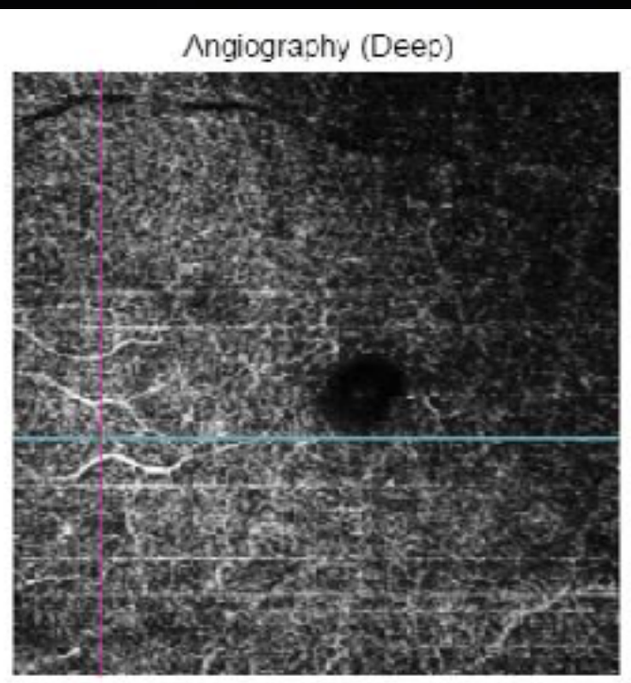


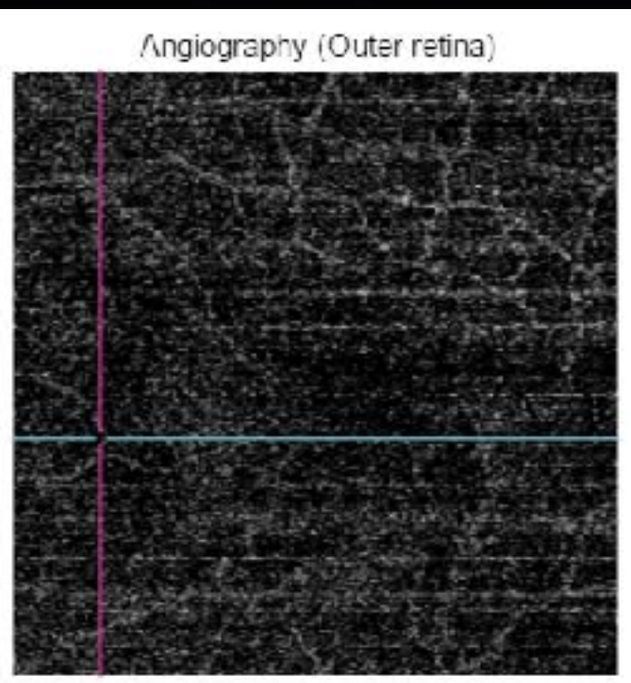
Fundus

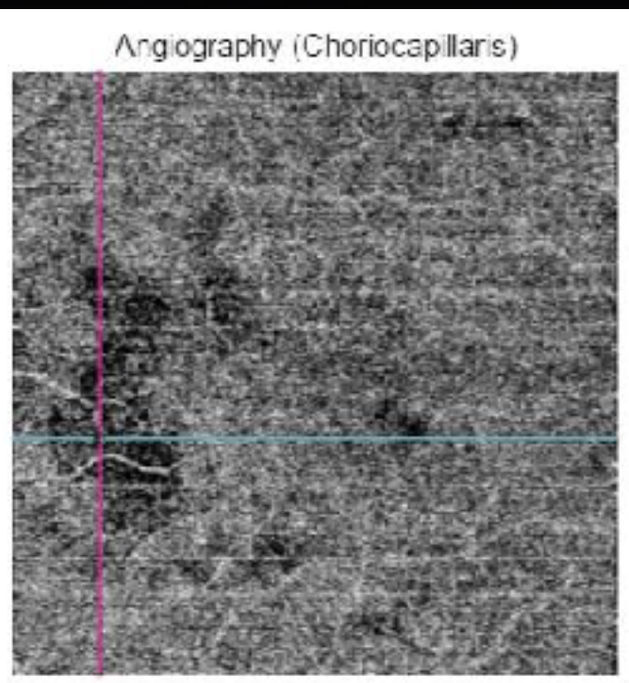


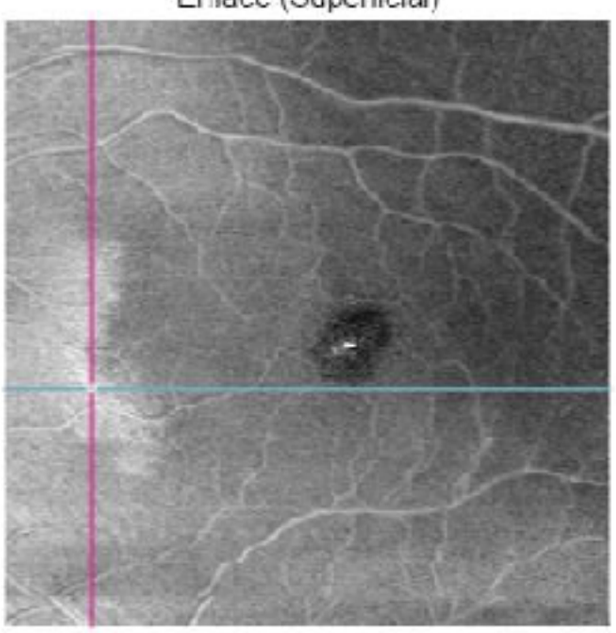
OCT angiography - Left Eye

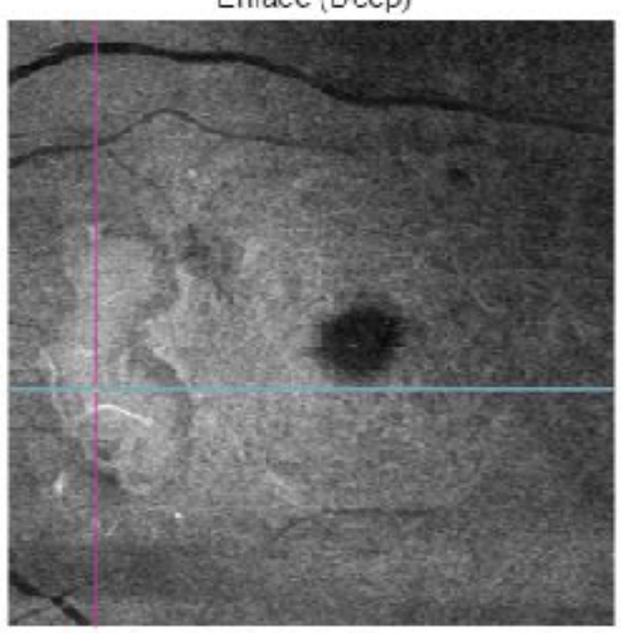


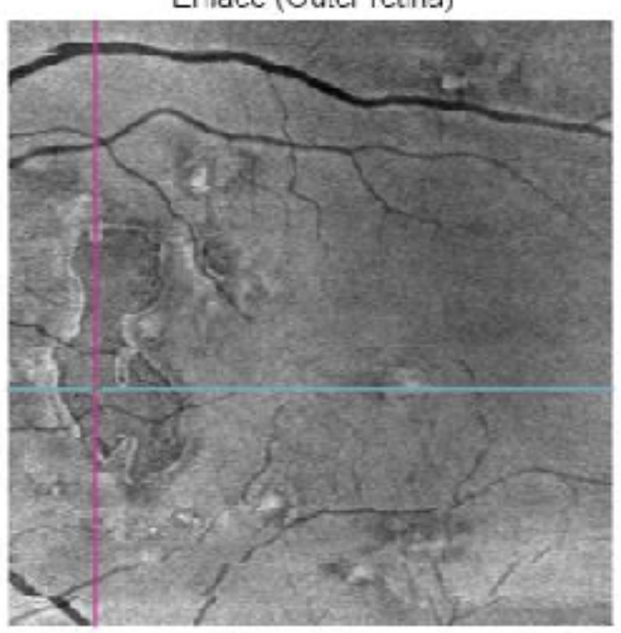


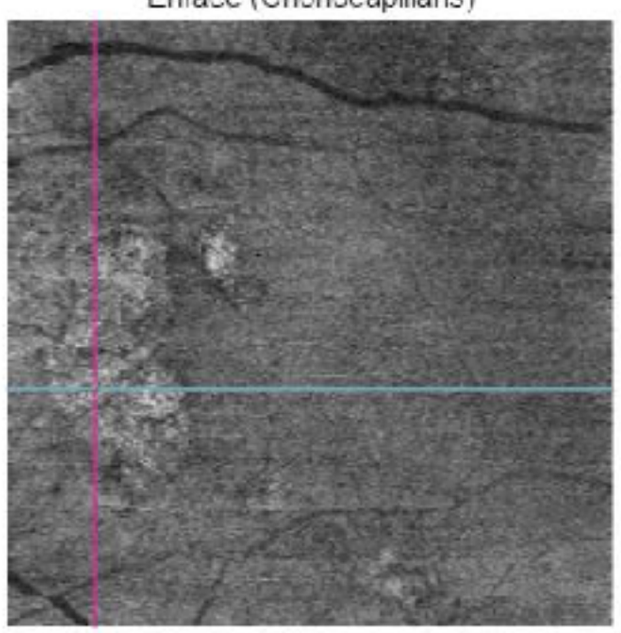












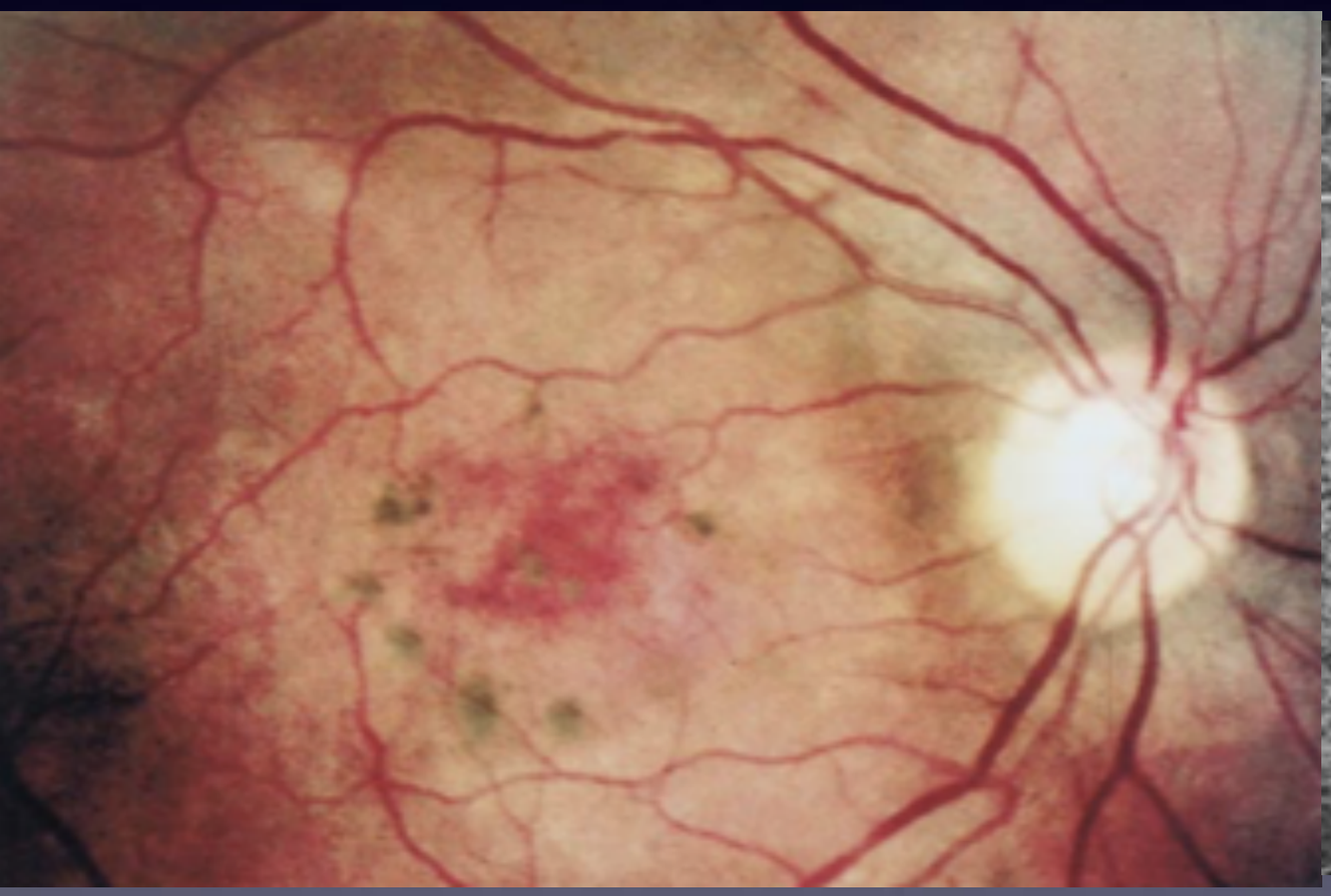
Diagnostic Hypothesis

- Maternally Inherited Diabetes and Deafness (MIDD)
  - Genetic test:
    - Mithocondrial diabetes confirmed
      - Substitution of Guanine for Adenine at position 3243 of leucine transfer RNA in the mitochondrial DNA.
        - salt and pepper retinopathy
        - abnormal pigmentation at posterior pole
        - pigmentary retinopathy
        - macular pattern distrophy

Ophthalmology. 1999 Sep;106(9):1821-7.

Prevalence of macular pattern dystrophy in maternally inherited diabetes and deafness. GEDIAM Group.

Massin P[1], Virally-Monod M, Vialettes B, Paques M, Gin H, Porokhov B, Caillat-Zucman S, Froguel P, Paquis-Fluckinger V, Gaudric A, Guillausseau PJ.



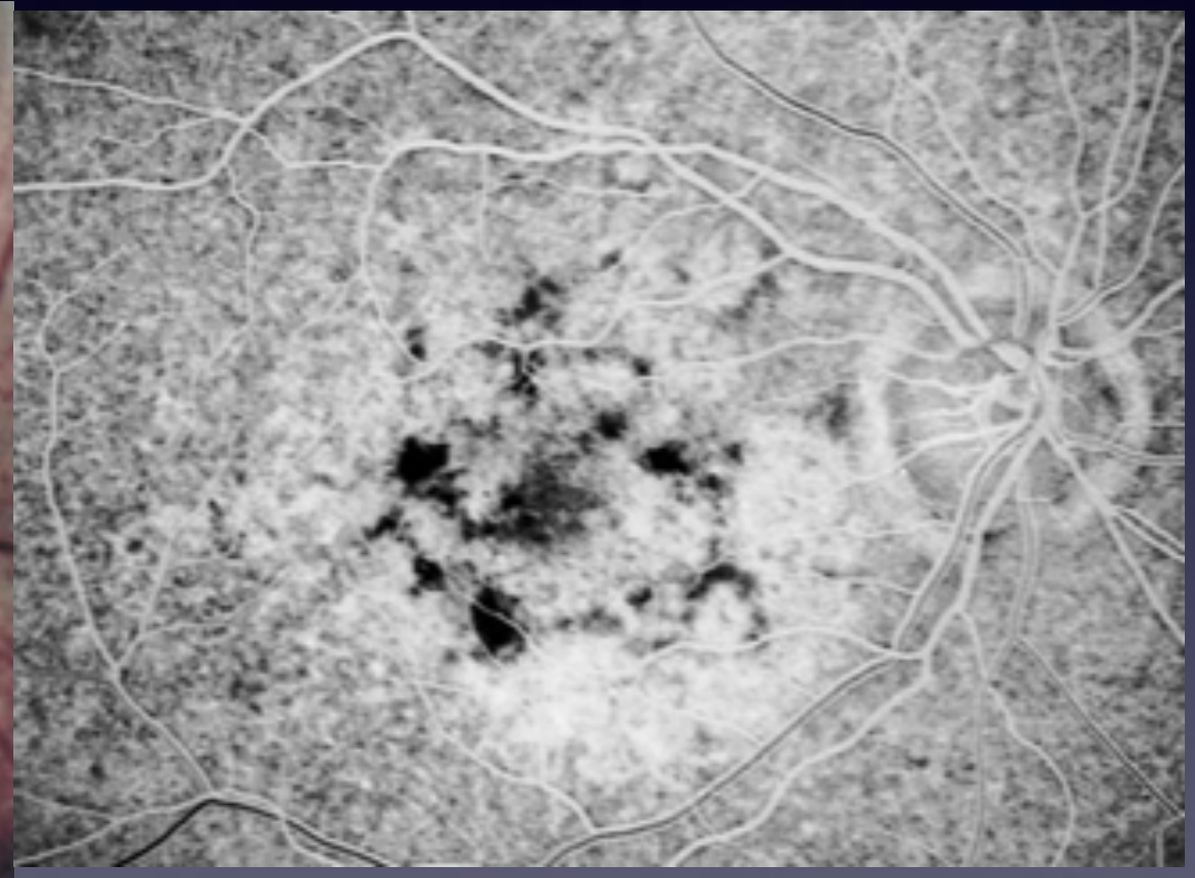
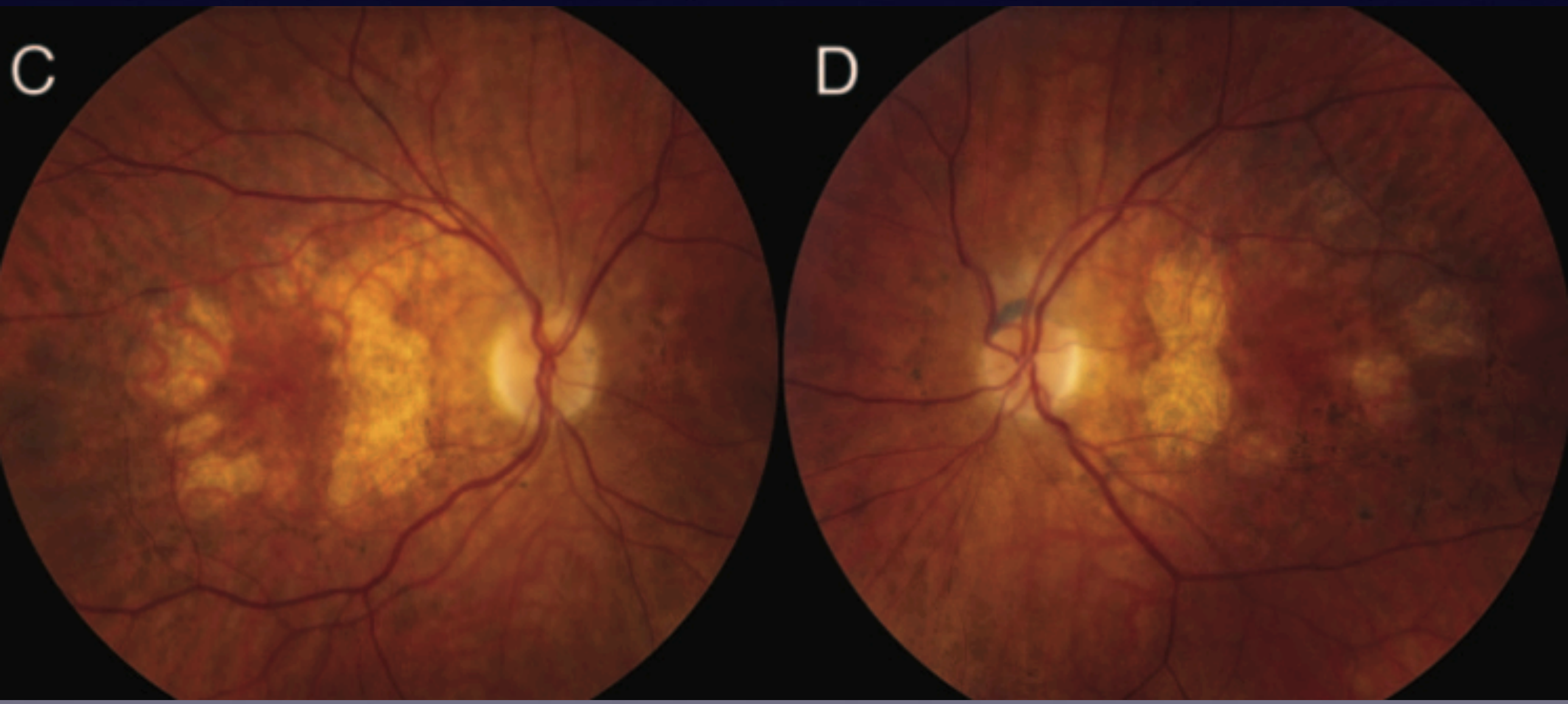


Figure 1. Macular pattern dystrophy. **A,** color fundus photograph shows pigmented deposits surrounding the macula. **B,** fluorescein angiography shows mild atrophy of the retinal pigment epithelium at the posterior pole and anular and radiate hypofluorescence corresponding to the pigmented deposits.

Neurology. 2012 Aug 7;79(6):e54-6. doi: 10.1212/WNL.0b013e31826356ad.

Pearls & oy-sters: maternally inherited diabetes and deafness presenting with ptosis and macular pattern dystrophy.

Ogun O[1], Sheldon C, Barton JJ.



Thank You!

Ophthalmology. 1999 Sep;106(9):1821-7.

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⊕ Author information

Abstract

OBJECTIVE: To evaluate the prevalence of macular pattern dystrophy (MPD) in maternally inherited diabetes and deafness (MIDD), a new subtype of diabetes mellitus that cosegregates with a mutation of mitochondrial DNA (i.e., the substitution of guanine for adenine at position 3243 of leucine transfer RNA) and to report the clinical characteristics of MPD.

DESIGN: Prospective cohort study.

PARTICIPANTS: Forty-six patients from 29 families with an adenine-to-guanine mutation of mitochondrial DNA were recruited from a French collaborative multicenter study. Thirty-five patients had MIDD, 8 were asymptomatic children of MIDD patients, and 3 had MELAS syndrome (mitochondrial myopathy, encephalopathy, lactic acidosis, and strokelike episodes). The 33 MIDD patients with diabetes were matched for diabetes duration and gender with 33 patients with "common" type-2 diabetes to compare the prevalence of diabetic retinopathy (DR) in both series.

METHODS: All patients had a full ophthalmologic examination and fundus photographs.

MAIN OUTCOME MEASURES: The presence and severity of MPD and DR were assessed in each patient.

RESULTS: Thirty MIDD patients (85.7%) of 35 exhibited bilateral MPD characterized by linear pigmentation surrounding the macula and optic disc. In 24 of these 30 patients, visual acuity was 20/25 or more in both eyes. The prevalence of DR was 6% in MIDD patients with diabetes versus 15% for patients with common type-2 diabetes (a difference that was not significant, P = 0.23). The fundus of each of the eight asymptomatic children was normal. MPD was present in one of the three cases of MELAS.

CONCLUSION: The prevalence of MPD in MIDD is high. Its detection may be helpful for the diagnosis of this new subtype of diabetes, for which specific treatments may be proposed.