# AAO 2019 Course 630
# Cases with a Point
# Case 3

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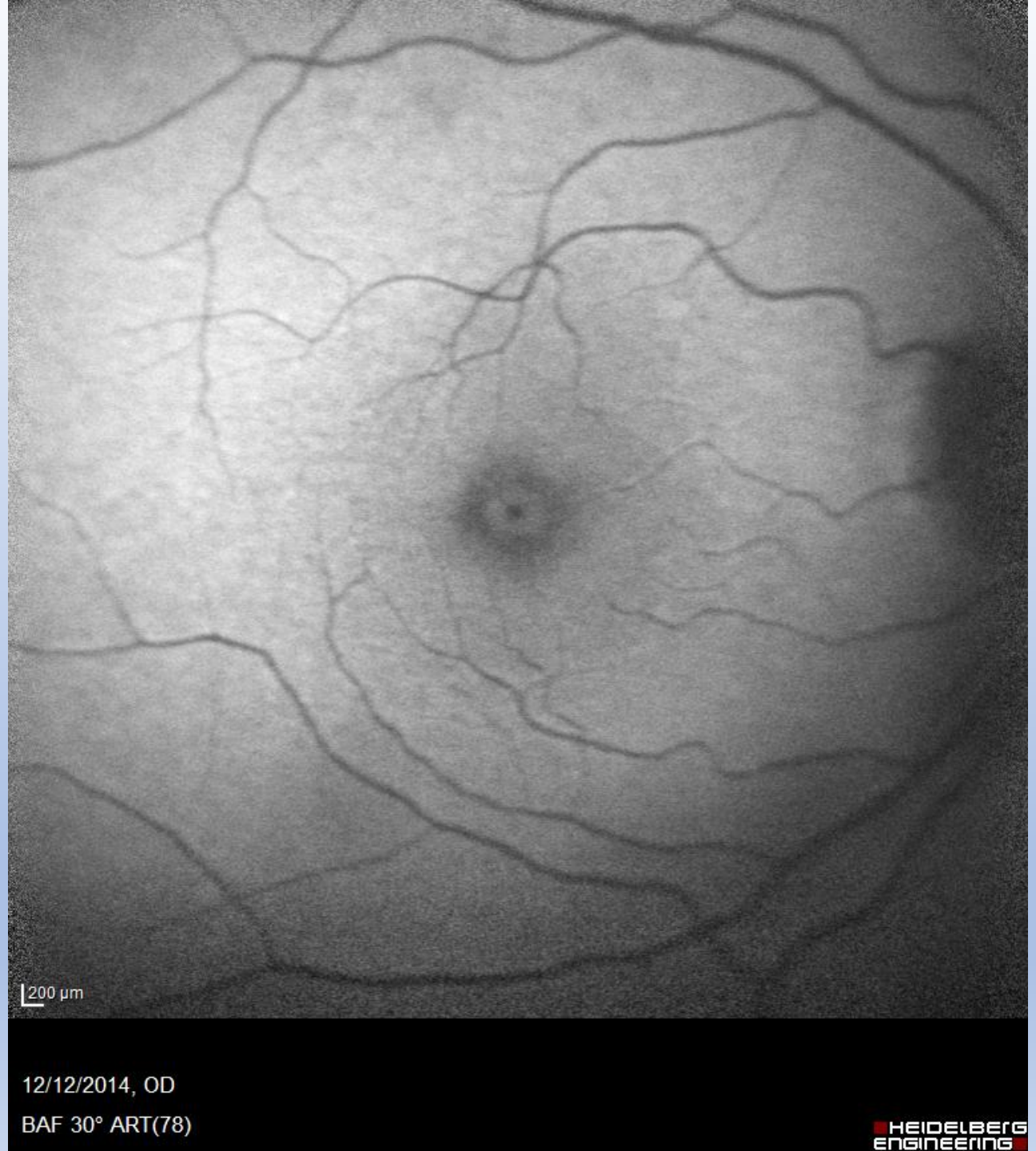
# History

- 74 yo female referred for macular changes found incidentally on recent exam
- Patient complains of blurred vision OS>OD
- PMH: Type 2 Diabetes, Hyperlipidemia, skin melanoma
- Meds: Glucovance, Metformin, Crestor  (no history of niacin use)
- Family Hx: Diabetes, no ocular pathology
- ROS unremarkable

# Exam

- Va OD sc 20/63 PH 20/25
- Va OS sc 20/40 PH 20/25
- Normal color vision and fields
- Anterior segment: 2+ NS OU
- Posterior segment: ON pink with sharp margins 0.1 C/D OU
- Macula unremarkable OD, Macular striae OS
- Periphery attached OU without retinoschisis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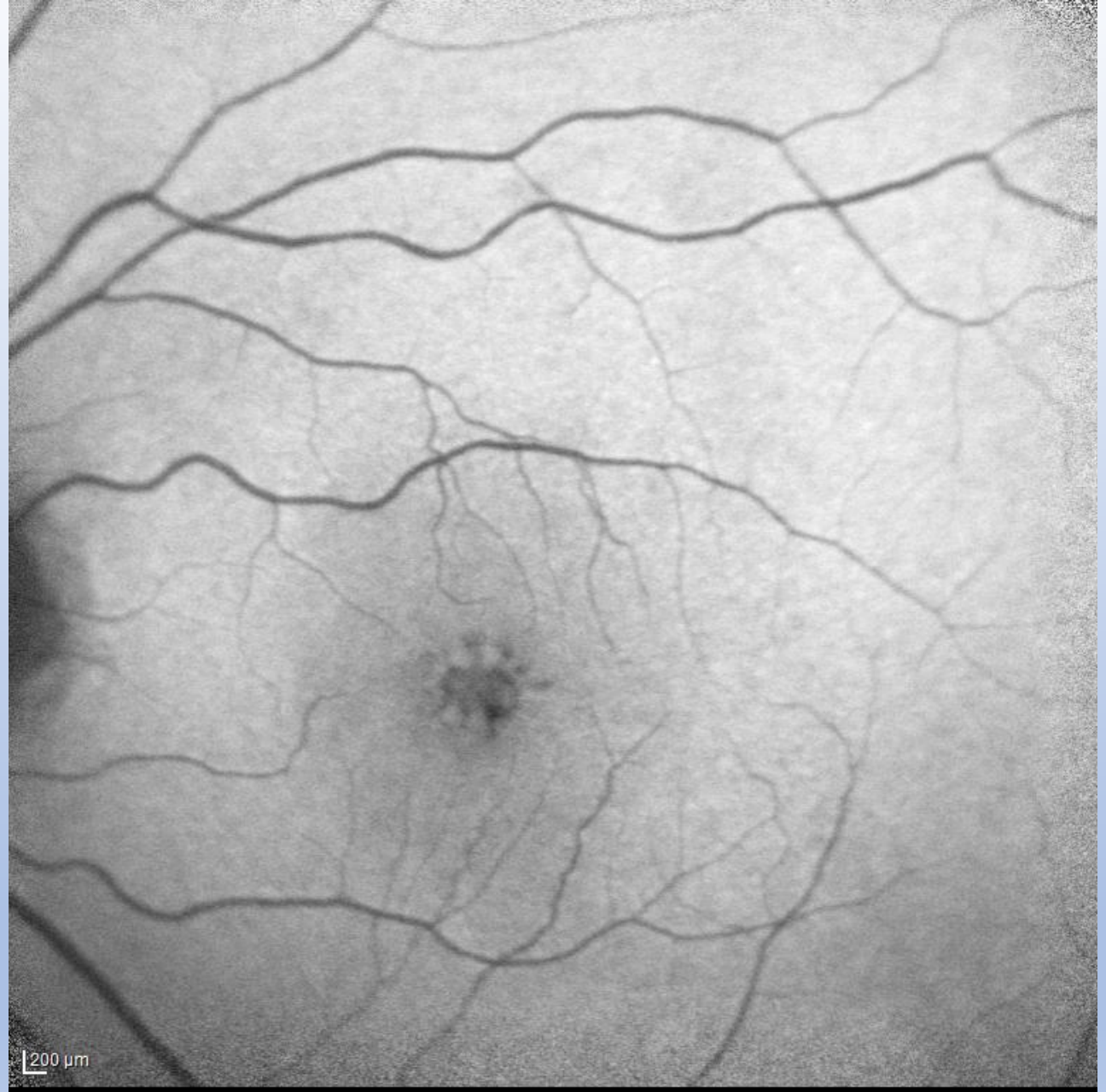
12/12/2014, OD
BAF 30° ART(78)
HEIDELBERG ENGINEERING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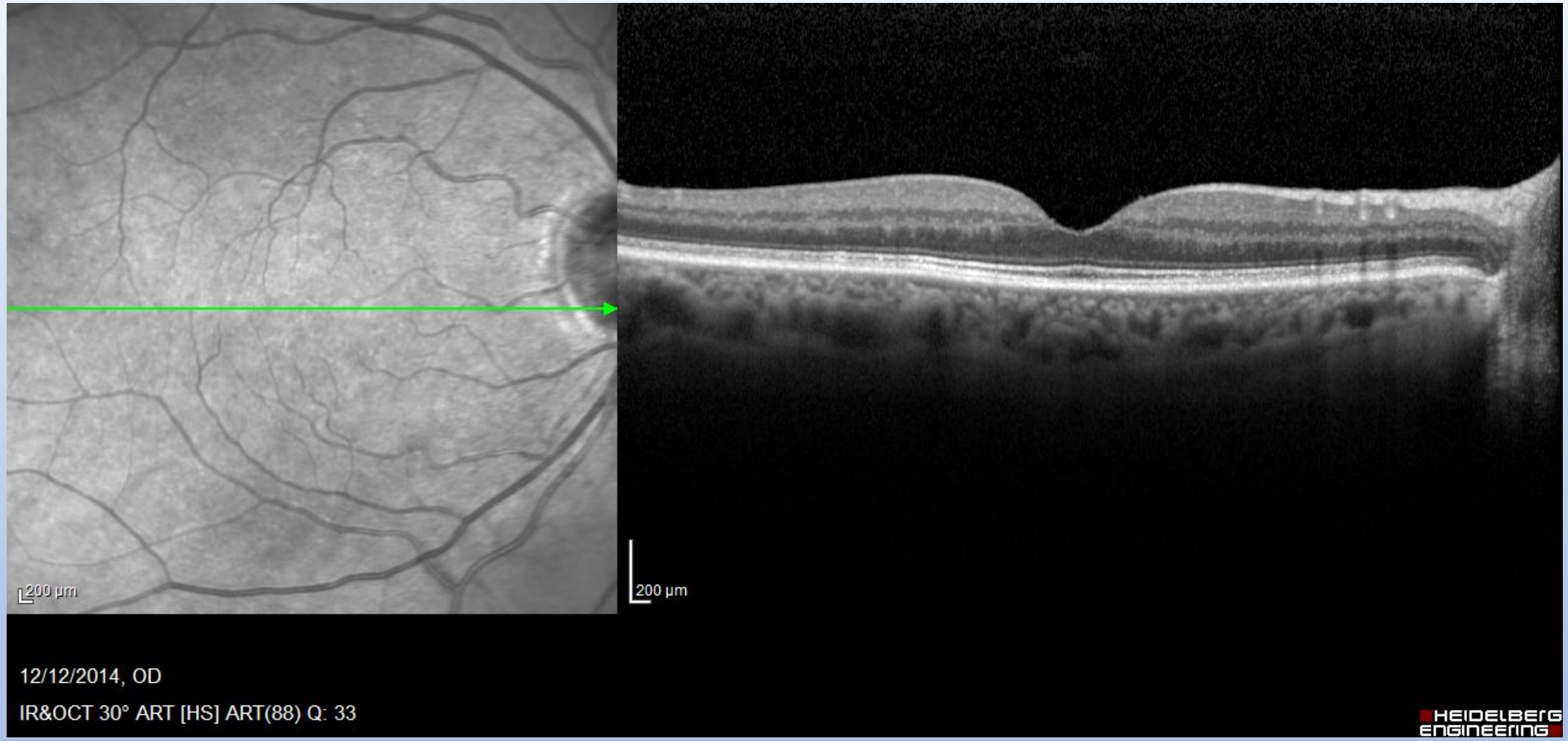
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200 µm
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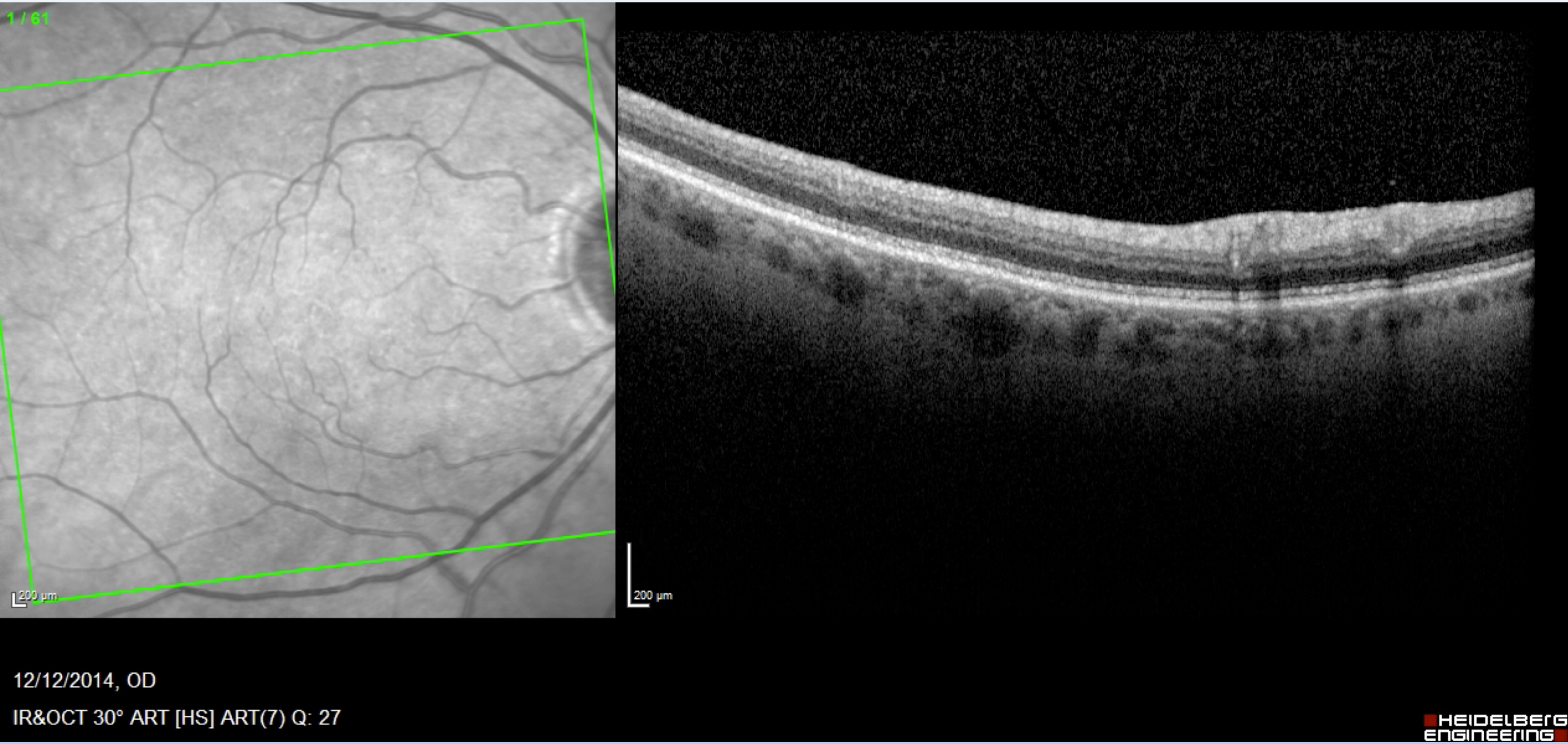
1 / 61
200 µm
200 µm
12/12/2014, OD
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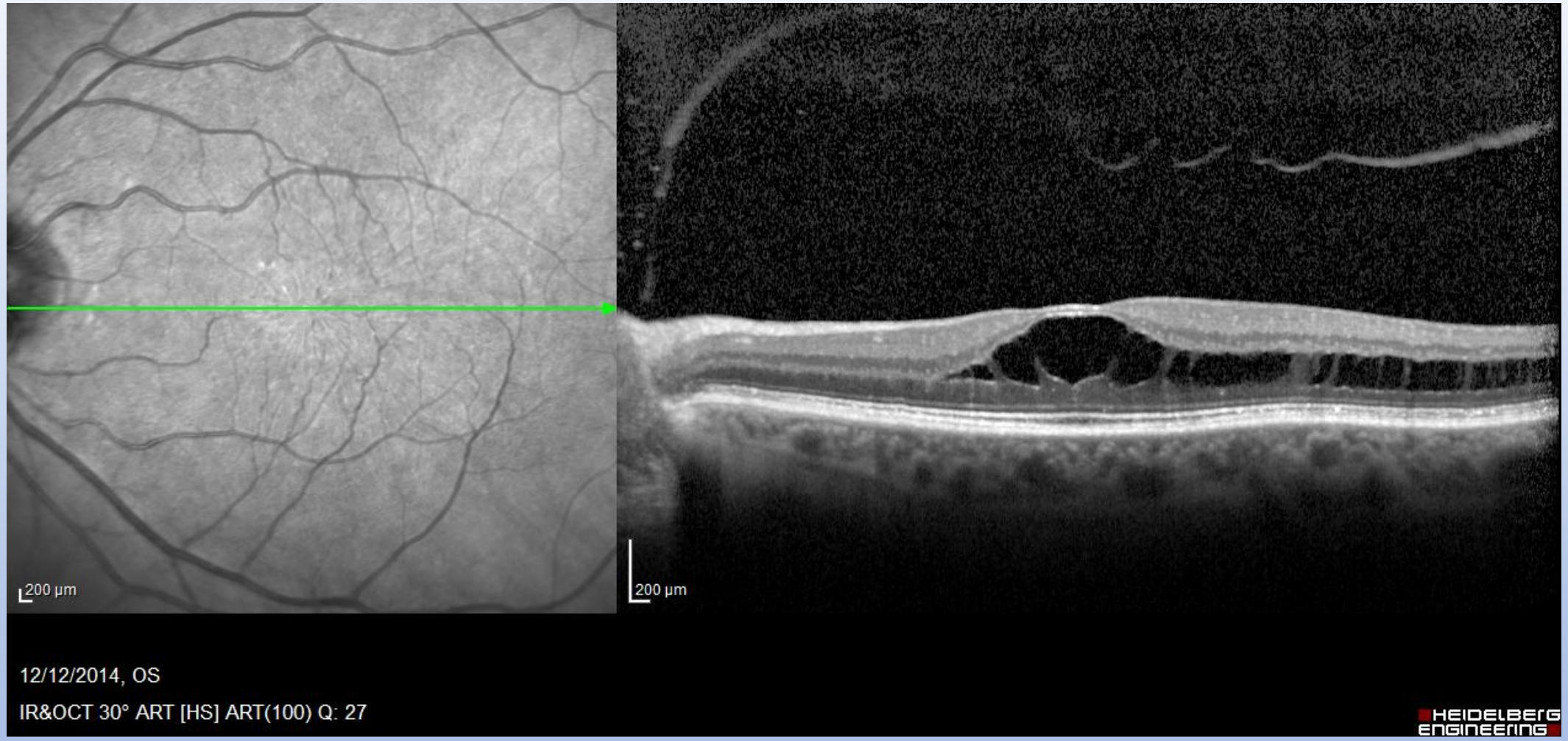
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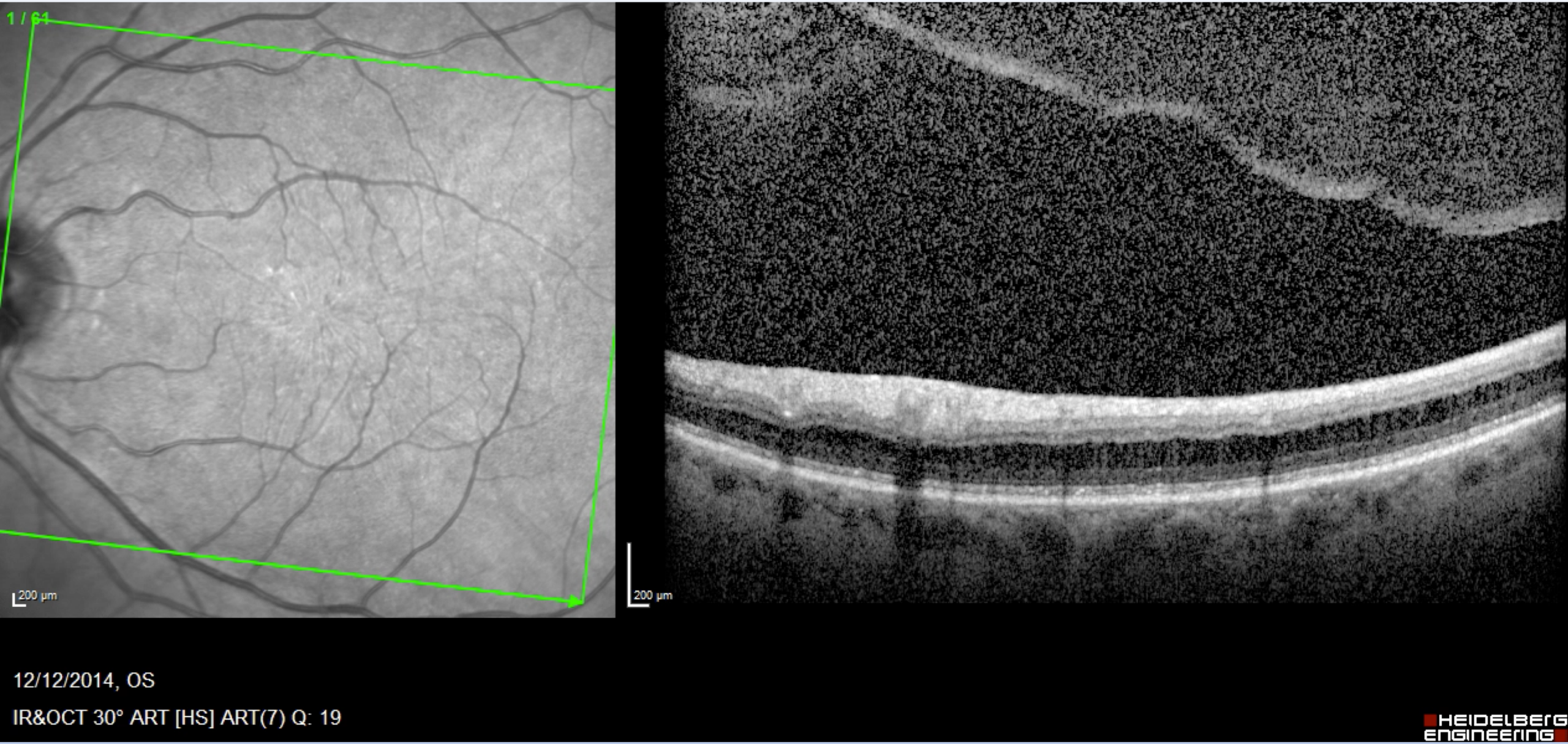
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200 µm
200 µm
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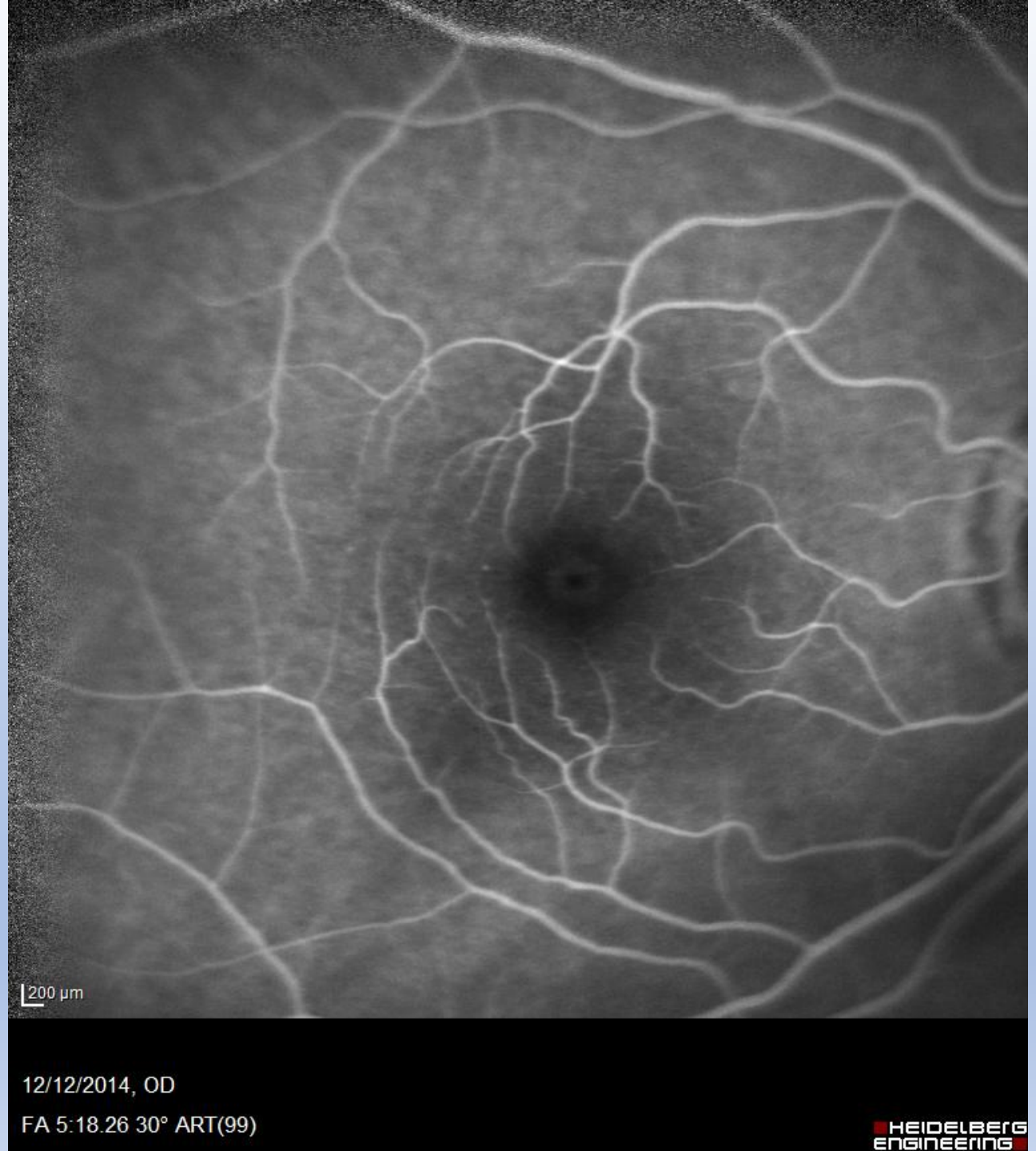
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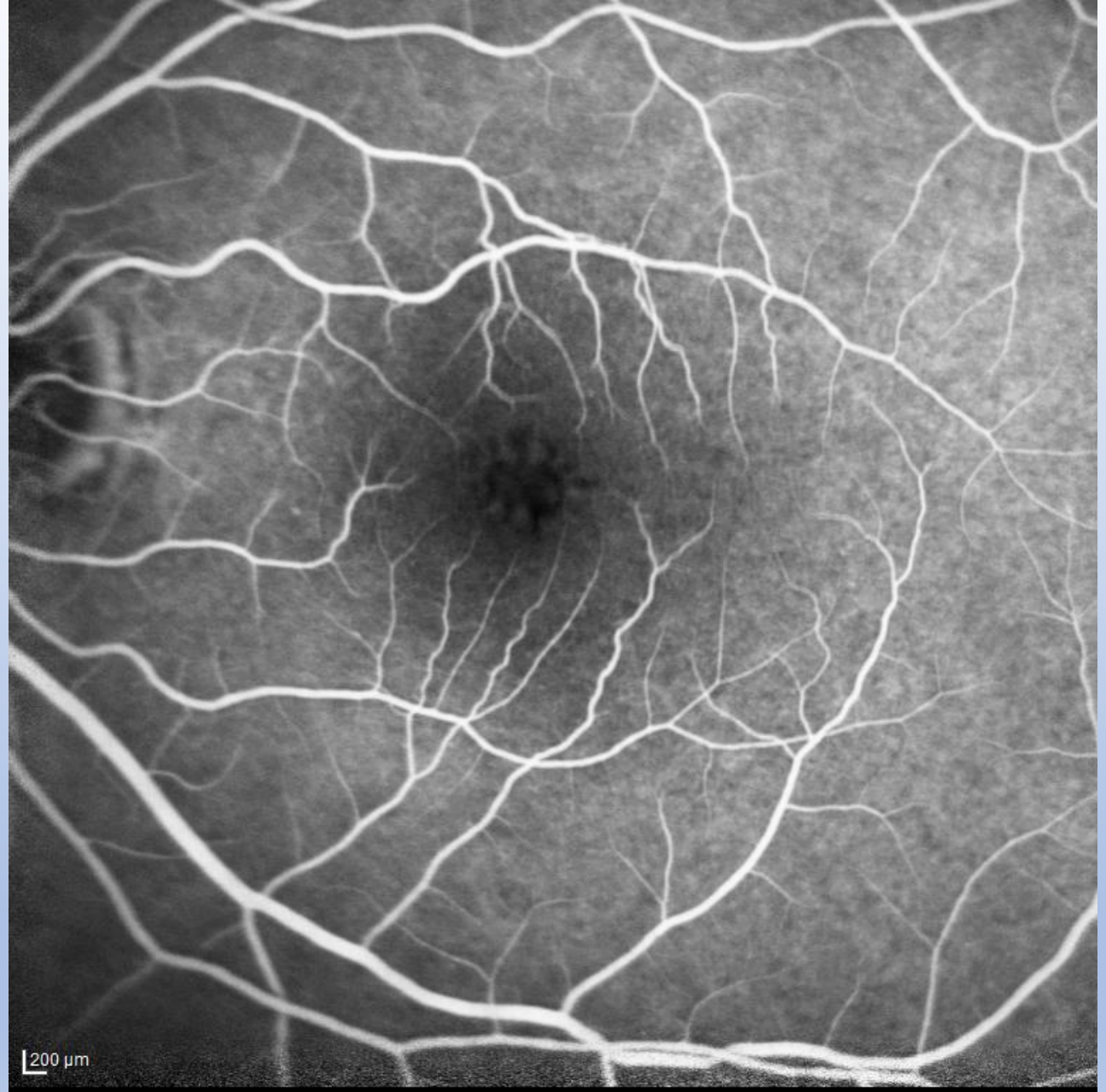


12/12/2014, OS

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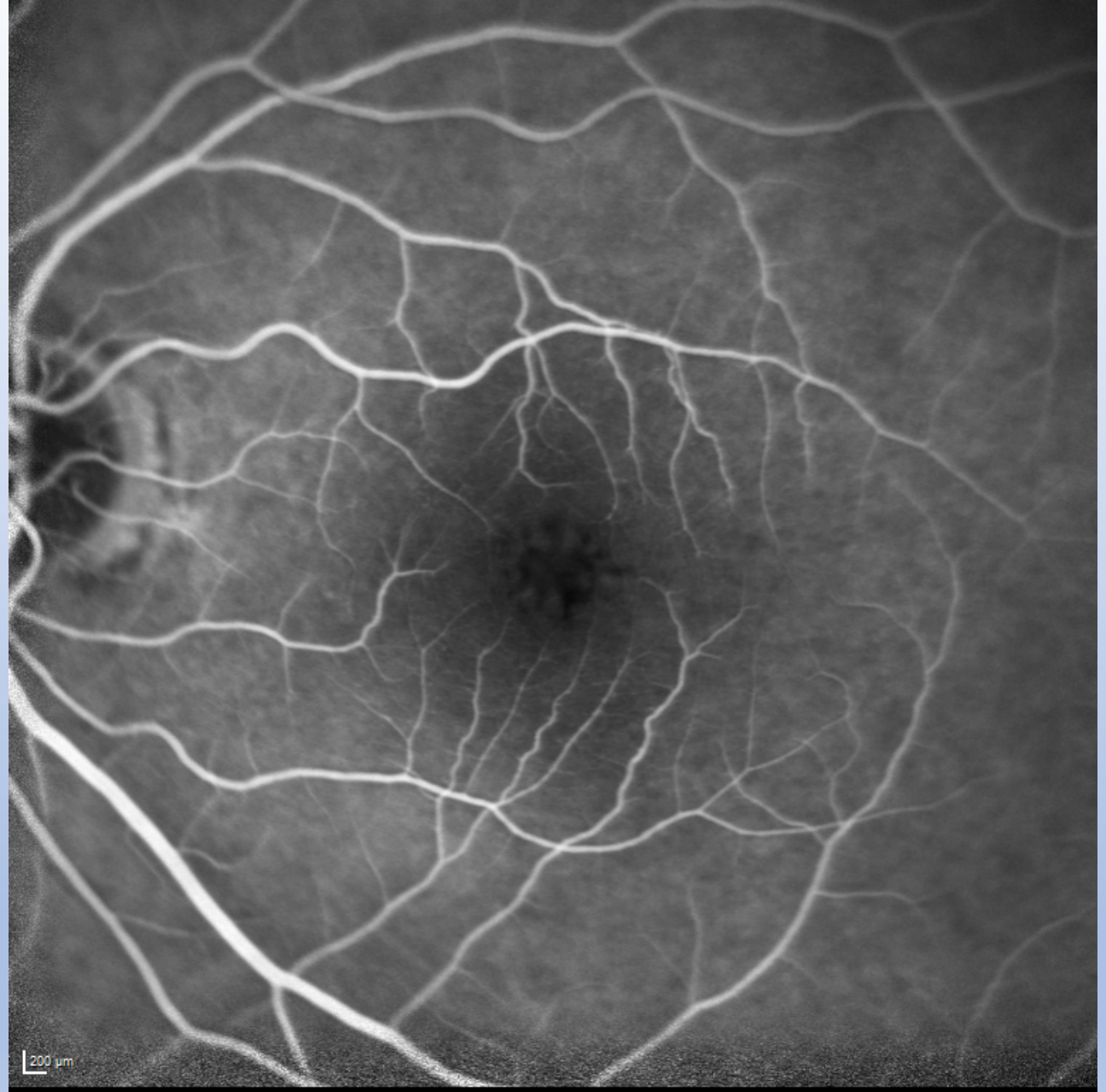


12/12/2014, OS

FA 4:56.21 30° ART(100)

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# Diagnosis?

# Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis

Michael D. Ober, MD,[1] K. Bailey Freund, MD,[2] Manthan Shah, MD,[3] Shareef Ahmed, MD,[3]
Tamer H. Mahmoud, MD, PhD,[4] Thomas M. Aaberg, Jr., MD,[5] David N. Zacks, MD,[6] Hua Gao, MD,[3]
Krishna Mukkamala, MD,[2] Uday Desai, MD,[3] Kirk H. Packo, MD,[7] Lawrence A. Yannuzzi, MD[2]

Idiopathic foveomacular retinoschisis

No leak on fluorescein angiography

Female predominance (as opposed to Congenital X-linked Retinoschisis)

Usually Unilateral

Sometimes associated with peripheral schisis

Visual prognosis is good

# Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis (SNIFR): A Diagnosis of Exclusion

- Hereditary conditions (congenital X-linked retinoschisis, *CRB1*-associated maculopathy, familial foveal retinoschisis, Enhanced S-cone syndrome )
- Optic disc disease (congenital pits, glaucoma)
- Vitreo-retinal interface disease (ERM-induced tractional schisis, VMT)
- Myopic traction maculopathy
- Drug-related FA-negative macular edema (niacin, taxane derivatives)
- Pachychoroid Peripapillary Syndrome

# Points of the Case

- SNIFR is a diagnosis of exclusion.
- SNIFR will likely be separated into more definitive diagnoses over time (e.g. pachychoroid peripapillary syndrome).
- Our case may have been due to past undocumented vitreomacular traction (VMT) with spontaneous separation.
- Consider *RS1* (retinoschisin) genetic testing for congenital X-linked retinoschisis, especially in bilateral cases.

# Thank You

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Case 3
Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis (SNIFR)

Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis (SNIFR) was described in 2014 by Ober and associates[1] as a new category of macular schisis. It is a diagnosis made by excluding known hereditary and acquired causes. SNIFR is usually a monocular macular schisis detected by optical coherence tomography (OCT) in adult females, though cases of bilaterality and male sex also occur. In some cases, flat or bullous peripheral retinoschisis may be present in one or both eyes. Our case (case3) fits the description of SNIFR, though further work-up may be needed to rule out other know causes of macular schisis.

Macular schisis is commonly recognized with congenital X-linked retinoschisis (CXLR). It presents with bilateral macular schisis in young males often in association with peripheral schisis. However, female cases have been reported with homozygous mutations, Turner syndrome, and carrier states.[1] The diagnosis of CXLR is confirmed with genetic testing of the *RS1* gene.[2] Not all cases in the original description of SNIFR underwent genetic testing.[1] Our case of schisis was female and reported no family history of decreased vision or known CXLR. The diagnosis of CXLR is unlikely and genetic testing was not done, but *RS1* gene testing would have been reasonable.

CRB1-associated maculopathy may present with macular schisis in association with rod-cone degeneration. Both the rod-cone degeneration and CRB1 testing are helpful diagnostically.[3.] Our case lacked symptoms and signs of rod-cone dystrophy. No genetic testing was done.

Enhanced S-cone syndrome (formerly Goldmann-Favre syndrome) may present with foveal schisis, but patients usually report a history of long-standing nyctalopia and show peripheral pigmentary degeneration. Electroretinographic (ERG) testing shows a pathognomonic pattern. The rod response is extinguished, yet an increased response is elicited with cone testing using an orange background (suppressing red and green cones) suggestive of functioning S-cones (short wavelength/blue cones).[4.] ERG testing was not performed on our case due to the lack of symptoms and signs of enhanced S-cone syndrome.

Macular schisis-like changes may occur with optic disc abnormalities including congenital and acquired pits, as well as glaucomatous optic neuropathy. The optic disc findings differentiate these conditions from SNIFR.[1] No optic disc abnormalities were seen in our case.

Epiretinal membranes (ERM) and vitreomacular traction may cause macular schisis. These conditions are easily recognized on OCT and differentiate themselves from SNIFR. However, spontaneous resolution of VMT may leave residual schisis-like macular changes making diagnosis difficult.[5.] Although there was no epiretinal membrane on OCT in our case, we

documented vitreomacular detachment. We cannot completely rule out spontaneous separation of vitreomacular traction with residual schisis in our case. Furthermore, OCT scans should have been performed temporal to the macula in our case to rule out peripheral traction with posterior extension.

Myopic traction maculoschisis is readily distinguished from SNIFR with the history of high myopic and findings of the concave staphylomatous appearance of the macula on OCT. There were no myopic changes in our case.

Niacin and taxane-derived medications (used to treat prostate cancer) may cause cystic maculopathy resembling SNIFR.[1] Fluorescein angiography shows no leakage. Our patent was not on either of these medications.

Familial foveal retinoschisis has been reported in females. However, no family history was elicited in the original description of SNIFR[1] or in our case. However, we were unable to examine any family members to definitively rule out familial foveal retinoschisis.

Subsequent to the description of SNIFR, pachychoroid peripapillary syndrome (PPS) was described by Sarraf and associates.[6] PPS presents with outer retinal edema resembling schisis that emanates from a defect in the blood-retinal barrier at the level of the retinal pigment epithelium in the peripapillary area. It has also been described as posterior uveal effusion[7] and pachychoroid macular effusion.[8] The diagnosis is suggested by the findings of a thick choroid with the typical distribution of edema/schisis at the temporal disc border extending to the macula.

Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis (SNIFR) is a diagnosis of exclusion and likely represents a collection of conditions likely to be further separated with improved understanding. Upon revisiting the original description[1] of SNIFR, I am impressed that some of the cases may not be "idiopathic." For example, case 6 (and possibly case 8) might be reclassified today as pachychoroid peripapillary syndrome. Case 10 demonstrated eccentric vitreoretinal tractional schisis temporal to the macula, which may have explained the presence of macular schisis due to posterior extension.

Points of the case:

1.) SNIFR is a diagnosis of exclusion.
2.) Cases of SNIFR will likely be separated into more definitive diagnoses over time.
3.) Consider RS1 (retinoschisin) genetic testing to rule out congenital X-linked retinoschisis.

References


1.) Ober et al. Stellate Nonhereditary Idiopathic Foveomacular Retinoschisis. Ophthalmol 2014;121:1406-1413.
2.) Tantri et al. X-linked retinoschisis: a clinical and molecular genetic review. Surv Ophthalmol 2004;49:214-230.
3.) Mucciolo et al. Long-term follow-up of a CRB1-associated maculopathy. Ophthalmic Genet 2018;39:522-525.
4.) Tsang et al. Enahanced S-cone syndrome (Goldmann-Favre syndrome). Adv Exp Med Biol 2018;1085:153-156.
5.) Errara et al. A study of the natural history of vitreomacular syndrome. Ophthalmol 2018;125:701-707.
6.) Sarraf et al. Pachychoroid Peripapillary Syndrome. Retina 2018;38:1652-1667.
7.) Pautler SE, Browning DJ. Isolated posterior uveal effusion: expanding the spectrum of the uveal effusion syndrome. Clin Ophthalmol 2014;9:43-49.
8.) Pautler et al. Clinical findings of the posterior uveal effusion syndrome. AAO Poster Presentation P0476, Chicago. October 2015.