IT'S NOT ALWAYS WHAT IT SEEMS

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Case-report

- 67 years old female
- Sent to retina department due to a progressive decrease in visual acuity.
- Previously diagnosed with wet AMD, treated with intravitreal injections of bevacizumab.

Fundus Flavimaculatus:

- Macular distrophy variant due to ABCA4 gene mutation.
- Fundus flavimaculatus and Stargardt Disease are the same pathology with different time-presentation.
- Progressive decrease in visual acuity
- Macula: non-specific mottling, oval "snail-slime", "beaten-bronze" appearance and geographic atrophy in end-stage disease
- Bilateral yellow-white lesions: Flecks.
- Genotype-fenotype correlation is well-described.

June 2013

Visual Acuity:
RE 3/10 LE 2/10

Biomicroscopy
Cortical and Nuclear Cataract OU

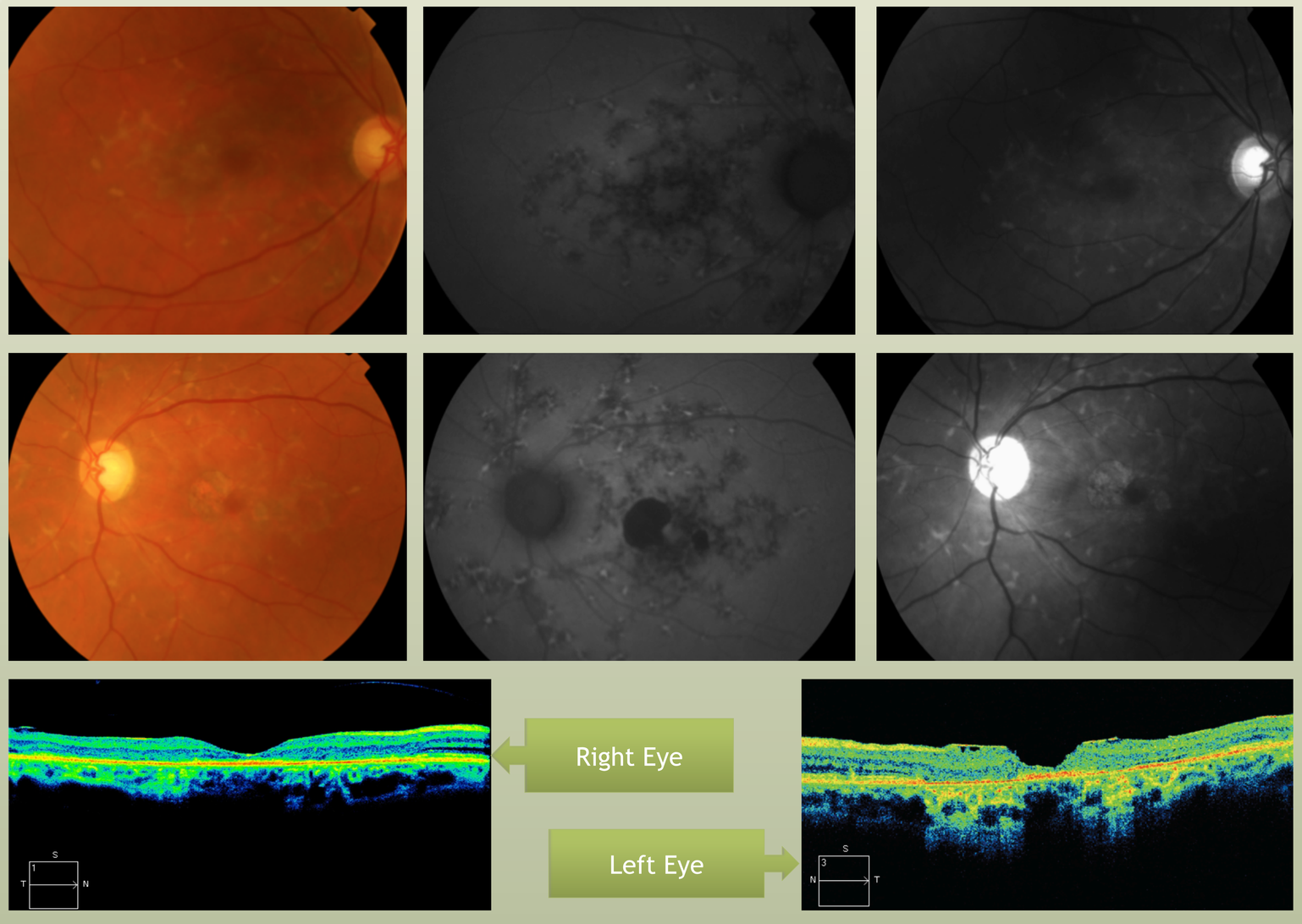
Intraocular Pressure by Goldmann Aplannation:
RE 19 mmHg LE 16 mmHg

Fundoscopy:
Yellow and flecked-like lesions at posterior pole OU
LE Geographic Atrophy and Macular Pucker
Glaucomatous Optic Disc OU

ERG: OU subnormal photopic
EOG: LE subnormal

Fluorescein Angiography was not performed due to allergy suspicious.

Retinography, Autofluorescence Angiography and OCT were performed.



- AMD diagnosis was questioned.
- No family history of Stargardt Disease or Ophthalmic Pathology was verified.
- ABCA4 gene (Stargardt) genetic test based on flexible and cost-effective Arrayed Primer EXtension (APEX) genotyping was performed.

Blood genetic test revealed an **Heterozygous Single Mutation.**

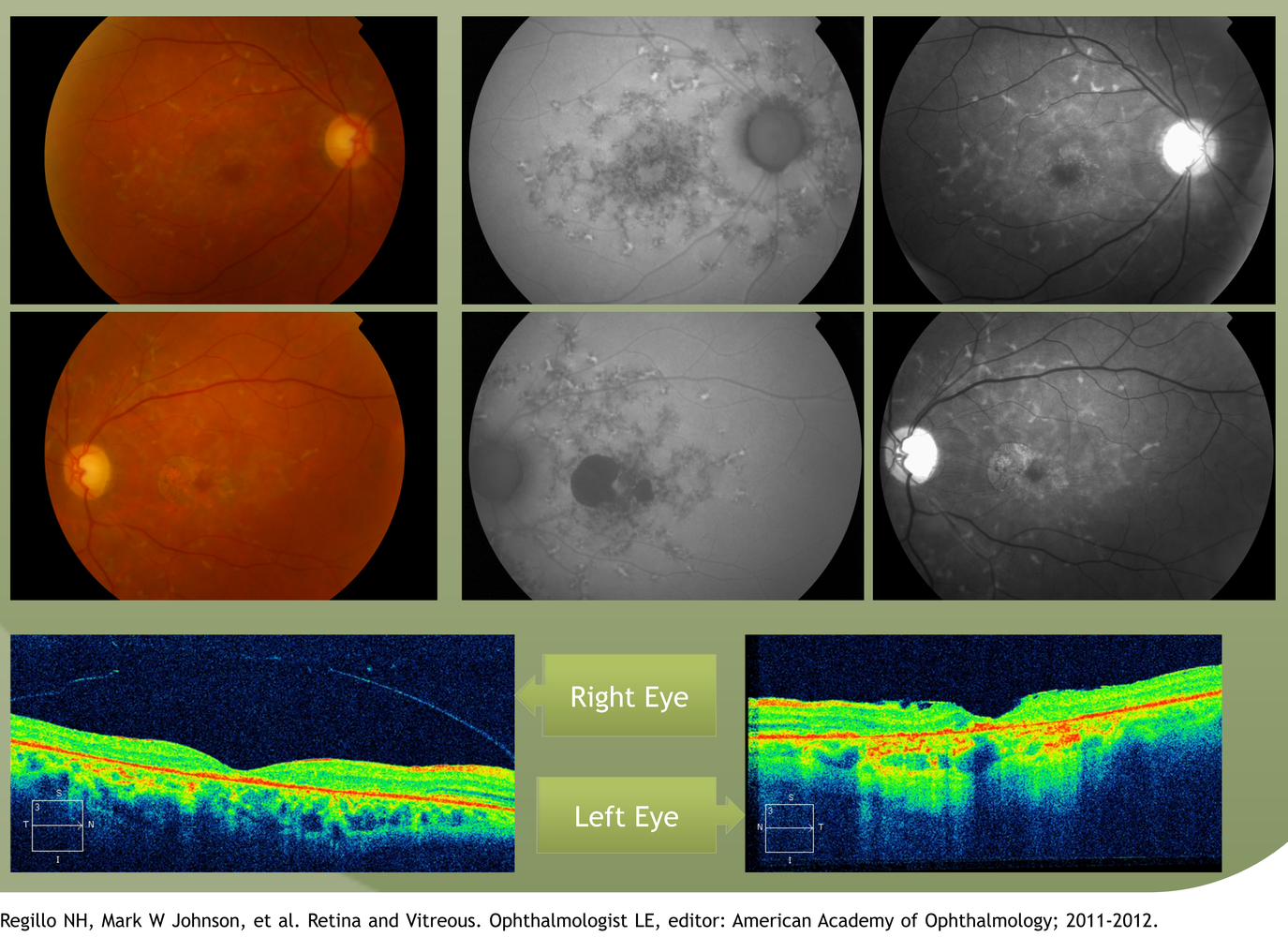
June 2014

- Late-Onset of Stargardt Disease diagnosis was made.
- No more relatives afected.
- Started Brimonidine drops bid

- Visual Acuity
RE 1/10 LE <1/10

- Intraocular Pressure
(w/ brimonidine)
RE 12 mmHg LE 11 mmHg

- Fundoscopy:
Yellow and flecked-like lesions at posterior pole OU
LE Geographic Atrophy and Macular Pucker
Glaucomatous Optic Disc OU



References: Kanski JJ BB. Clinical Ophthalmology - A Systematic Approach: Elsevier Saunders; 2011. Carl Regillo NH, Mark W Johnson, et al. Retina and Vitreous. Ophthalmologist LE, editor: American Academy of Ophthalmology; 2011-2012. Yanoff M, Ducker JS. Ophthalmology: Elsevier; 2009.