I. Case History

- 29 year old Caucasian Female
- Chief complaint: Mild increase in floaters OU. (-) Flashes of light, (-) curtain/veil covering vision, worse when looking at a bright background.
- Ocular Hx: Soft contact lens wearer otherwise unremarkable. (-) family hx of ocular disease
- Medical Hx: 24 weeks pregnant, history of migraines with auras before pregnancy.
- Medications: Pre-natal vitamins, Adapalene, Sumatriptan, Folic Acid

II. Pertinent findings

- **Ocular findings:**
  - VA cc OD: 20/20+ OS: 20/20+
  - SLE: OD: congenital pinpoint cataract. All other slit lamp findings unremarkable OU
  - IOP: 18/18
  - DFE: OD: Mild perifoveal pigment mottling, OS: Perifoveal pigment mottling, hypopigmented yellow lesion in the fovea OS>OD
- **Physical:** Pregnant, otherwise unremarkable
- **Fundus Photos:** OU: Perifoveal mottling with yellow lesion at the fovea OS>OD
- **FAF:** Normal OU
- **OCT:** OD: Mild perifoveal mottling OS: Focal drusenoid sub-RPE lesion at fovea with perifoveal mottling

III. Differential diagnosis

- Primary/Leading Dx: Adult-Onset Vitelliform Dystrophy
- Ddx:
  - Reticular dystrophy
  - Butterfly dystrophy
  - Fundus Pulverlentus
  - Multifocal pattern dystrophy simulating fundus flavimaculatus
  - Best’s Disease
  - Age-Related Macular Degeneration

IV. Diagnosis and discussion

- Adult-Onset foveomacular Vitelliform Dystrophy (AFVD) is one of the many forms of pattern dystrophies.
- AFVD is typically characterized by bilateral subretinal yellow lesions found at the macula
- Fundus Autoflourescence: the “egg yolk-like deposit” appears hyperautoflourescent but this is not always the case as such with our patient
- Optical Coherence Topgraphy: the vitelliform lesion appears as a dome shaped hyper-reflective elevation and has been hypothesized to involve the layers above the RPE, specifically between the RPE and IS/OS interface (which differentiates this from the appearance of drusen).
The exact pathophysiology of AFVD is unknown, but it has been theorized that alterations in the RPE and photoreceptors cause the accumulation of lipofuscin-like material at the fovea.

Patients with AFVD have been found to have mutations in genes BEST 1, IMPG1, IMGPG2, PRPH2, HTRA 1.

Onset of AFVD is variable but usually occurs after 40 years of age.

Patients are typically asymptomatic until the 5th decade of life.

Symptoms that patients may experience are a decrease in visual acuity, metamorphopsia, and a gradual decline in central vision.

Severe vision loss from the development of CNVM or chorioretinal atrophy can occur in up to 50% of patients after the age of 70.

V. Treatment, management

There is no current validated treatment or preventative therapy for AFVD.

Gene therapy may be an option in the near future.

Bevacizumab has been used for patients who develop CNVM with guarded visual outcome due to the progression of the lesions.

Monitoring and offering low vision options in the future may help to prepare patients to adapt to visual changes.

References:

VI. Conclusion

Our patient was without subjective or objective visual decrease, yet was found to have an obvious retinal change consistent with a pattern dystrophy, in this case Adult Foveomacular Vitelliform Dystrophy. We cannot assume that our young patients with no chief complaint are entirely exempt from intriguing ocular conditions.

Early diagnosis can allow clinicians to educate patients on prognosis of this condition and future management.

Because pattern dystrophies are a genetic disorder, detection of this condition in a patient can encourage the evaluation of other family members as well.