MULTIFOCAL PATTERN DYSTROPHY: A CASE REVIEW AND THE MOST COMMON DIFFERENTIAL DIAGNOSIS

ABSTRACT: A 65 year old male with previously diagnosed bilateral macular retinal pigmented epithelial atrophy that has been stable for many years, has shown progression on examination. Imaging confirmed multifocal pattern dystrophy.

CASE HISTORY:

- Chief Complaint: A 65 year old African American male with a history of previously diagnosed bilateral macular retinal pigmented epithelial atrophy stable for many years presents for a dilated fundus examination.
- Patient demographics:
  - 65 year old African American male
- Ocular History
  - Ocular hypertension OU: treated with topical medication
  - Confirmed optic nerve head drusen OD
  - Cataracts OU
- Medical History:
  - Hypertension
  - Hyperlipidemia
  - Diabetes Mellitus type 2
  - Resected prostate cancer
- Medications:
  - Alprostadil 20MCG/cartridge
  - Aspirin 81mg
  - Brimonidine Tartrate 0.2%
  - Cholecalciferol 400unit
  - Divalproex 500mg
  - Fluoxetine HCl 20mg
  - Hydromellose 0.4% solution
  - Insulin, 100UN/ML novo flexpen
  - Losartan 10mg/HCTZ 25mg
  - Metformin HCl 500mg
  - Metoprolol Tartrate 25mg
  - Mometasone Furoate 220mcg
  - Sildenafil citrate 100mg
  - Simvastatin 20mg

PERTINENT FINDINGS:

- BCVA: 20/25 OD/OS (stable for many years)
- Extraocular motilities: full OU
• Pupils: PERRL/APD-
• Anterior segment unremarkable except NS grade 1 OU
• Goldmann Tonometry: OD: 12mm Hg OS: 12mm Hg at 10:24am
• Dilated fundus examination:
  o C/D: 0.2 pink/distinct rims OD/OS
  o Vasculature: normal caliber OU
  o Macula: dense pigment centrally surrounded by an area of yellow lesions in a radiating pattern OU
  o Periphery: Within the arcades, there are scattered yellow pigmented lesions in both OD and OS. Otherwise, retina appears flat and intact with no holes or tears 360 OU.
• Amsler Grid:
  o OD: mild metamorphopsia superior temporally and inferiorly
  o OS: (-) metamorphopsia (-) scotoma
• A review of the patient’s previous fundus photo from 2008 reveals an increase in number of lesions present in the posterior pole on this exam compared to previously.
• Optical coherence tomography: multiple retinal pigment epithelium detachments and drusen in the perifoveal region OU
• Fundus autofluorescence: hyperfluorescence of yellow lesions OU
• Fluorescein angiography: the pigmented areas showed early hypofluorescence, surrounded by hyperfluorescence, with some degree of late staining OU. No leakage was noted.
• Humphrey Visual field 24-2 Sita Standard: reliable and essentially full OD/OS

DIFFERENTIAL DIAGNOSIS:
• Multifocal pattern dystrophy
• Age related macular degeneration
• Stargardt’s disease
• Adult vitelliform macular dystrophy

DIAGNOSIS AND DISCUSSION:
• Multifocal pattern dystrophy is an autosomal dominantly inherited dystrophy of the retinal pigment epithelium that is characterized by lipofuscin deposits arranged in various patterns along with pigmentation in the macular region as well as some scattered throughout the posterior pole.
• These patients often maintain good acuity until later in life.
• In advanced cases, patient can develop CNV and/or geographic atrophy.
• A case of unilateral choroidal neovascularization has previously been documented in a patient with butterfly like pattern dystrophy.
• Mutations in the PRPH2 gene are the most common cause of the different pattern dystrophies.

TREATMENT:
• Patient’s fluorescein angiography showed no leakage indicating that there is no underlying neovascularization process occurring.
• At this time, no treatment is indicated.
• Patient is scheduled for follow-up in 3 months, and has been given a copy of Amsler Grid to monitor for visual changes, and strongly advised to return sooner if any changes in vision occur.

CONCLUSION:

• This case delineates the differential diagnosis of pattern dystrophy and demonstrates the importance of proper imaging in the establishment of a diagnosis in a patient with macular lesions.

REFERENCES:


