Visual Electrodiagnostic Testing in a Case of Late Onset Tapetoretinal Degeneration: An Atypical Variant of Retinitis Pigmentosa

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I. Case History:

a. Patient Demographics: 65 year old Caucasian male

b. Chief Complaint:
   1. The patient was referred to the Visual Neurophysiology Service (VNS) Clinic at Rosenberg School of Optometry to evaluate progressive visual field loss in both eyes (OU).
   2. The patient reported being bothered by bumping into things when he was walking. He noted his left eye appeared to be worse than his right (OD), and that the symptoms seemed to be gradually worsening over the past two years. He also mentioned that the symptoms were worse at night or in dim lighting.

c. Ocular History:
   1. Possible Retinitis Pigmentosa OU (2014)
      i. The patient had been to two separate retina specialists to evaluate the diagnosis. At one specialist he had been diagnosed with retinitis pigmentosa and at the second specialist he had been diagnosed with cobblestone degeneration.
   2. Cobblestone Degeneration OU (2014)
   3. Longstanding Vitreous Floaters OU (Unknown Date of Diagnosis)
   4. Possible Color Vision Deficiency OU (Unknown Date of Diagnosis)
   5. Cataracts OU (Unknown Date of Diagnosis)
   6. High Myopia OU

d. Medical History:
   1. Type 2 Diabetes Mellitus (6-7 years)
      i. The patient had a stable HbA1c and fasting blood glucose level.
   2. Heart Disease with Cardiac Bypass (2002)
   3. Hypertension (1999)
   4. Hypercholesterolemia (20 years)

e. Medications:
   1. Metformin (Unknown dosage)
   2. Crestor (Unknown dosage)
   3. Lisinopril (Unknown dosage)
f. Other Salient Information:
1. The patient had no family history of retinitis pigmentosa or other tapetoretinal
degenerations; however his father was color deficient.
2. The patient had no history of ocular or medical trauma.

II. Pertinent Findings:

a. Clinical Findings:
1. The patient’s corrected (cc) entering visual acuity at distance (DVA) during his first visit was:
   i. OD: 20/50 -1 (Pinhole to 20/50+1)
   ii. OS: 20/50+1 (Pinhole to 20/40+2)
2. The patient showed improvement in vision with the following prescription:
   i. OD: -9.50-0.50x075 ADD: +2.50 DVA: 20/40+2
   ii. OS: -8.75-0.50x045 ADD: +2.50 DVA: 20/50+2
3. The patient’s entering DVA during his second visit with contact lenses was:
   i. OD: 20/30 -1
   ii. OS: 20/30-2
4. All of the entrance testing that was performed at both visits (pupil, extraocular
   muscle testing, confrontations, and cover test) was unremarkable.
5. Perimetry:
   i. At the patient’s first visit a Full Field 120 Point Screening Test was performed with the following results:
      a. OD: Reliable results with only 14/120 points seen (restricted to <20 degrees)
      b. OS: Decreased reliability with only 15/120 points seen (restricted to <20 degrees)
   ii. At the patient’s second visit a Full Field 120 Point Screening Test was repeated with the following results:
      a. OD: Decreased reliability but comparable results with only 11/120 points seen (restricted to <20 degrees)
      b. OS: Reliable and repeatable results with only 19/120 points seen (restricted to <20 degrees)
6. Color Vision Testing:
   i. Color vision testing was performed via the Rabin Cone Contrast Test which showed decreased red, green and blue cone scores attributable to retinal changes OD and OS.
      a. These findings exemplify the fact that both rods and cones are affected in this condition.
7. Corneal Topography:
   i. Corneal Topography was performed due tapetoretinal degenerations sometimes being associated with keratoconus. The results were as follows:
a. OD: 47.47/46.70
b. OS: 47.47/46.67
c. Findings: The values were within normal limits indicating a combined axial and refractive myopia that was the result of both axial length and corneal changes. No keratoconus was seen.

8. Optical Coherence Tomography (OCT):
   i. Optic Nerve OCT:
      a. Unreliable due to lens changes (media obscuration)
   ii. Macula OCT:
      a. Unreliable due to lens changes (media obscuration)

9. Electrodiagnostic Testing:
   i. The patient had dark adaptometry and electroretinography (ERG) performed. Dark adaptometry was performed prior to ERG testing and the results were as follows:
      a. Dark adaptation was found to be elevated 1.50 log units above normal
   ii. ISVEC standard full field flash ERGs were performed under scotopic, maximum amplitude (rod and cone) and photopic conditions and the results were:
      a. ERG’s were found to be non-recordable under most testing conditions and only small amplitudes could be observed.

b. Physical Findings:
   1. Lids and Lashes: clear OU
   2. Conjunctiva: white and quiet OU
   3. Cornea: trace endothelial pigment OU
   4. Anterior Chamber: deep and quiet OU
   5. Iris: flat and intact with no transillumination defects or neovascularization OU
   6. Lens: 2+ Nuclear Sclerosis (NS), +2 Posterior Subcapsular Cataract (PSC) superior central, and +1 Anterior Cortical Cataract (ACC) superiorly OU
   7. Angles: Open, Grade 3 OU
   8. Intraocular Pressure (IOP): 15mmHg OD /16mmHg OS
   9. Optic Disc: 0.25 Horizontal x 0.25 Vertical with parapapillary atrophy temporally OU
   10. Macula: normal color and contour OU
   11. Vessels: normal size and contour with no nicking or attenuation OU
   12. Periphery: Retinal Pigment Epithelium (RPE) clumping 360 degrees OU with Pavingstone Degeneration inferiorly OS
   13. Vitreous: clear with no cells OU

c. Laboratory Studies: Not Available

d. Radiology Studies: Not Available

e. Others: Not Available
III. Differential Diagnosis
a. Tapetoretinal Degeneration – Retinitis Pigmentosa (Primary)
b. Glaucoma (Secondary)
d. Pigmented Paravenous Retinochoroidal Atrophy (PPRCA) (Secondary)
e. Vitamin A Deficiency with Nyctalopia (Secondary)

IV. Diagnosis and Discussion:

a. Elaborate on the condition:
   1. The term “tapetoretinal degeneration” encompasses diseases that cause degeneration of the tapetum (RPE). One of the most common types of peripheral tapetoretinal degeneration is retinitis pigmentosa.
   2. Tapetoretinal degeneration is considered one of the most common retinal diseases occurring in 1 out of every 3,500-5,000 people.
   3. Common clinical signs and findings of patients include nyctalopia, loss of peripheral vision, and loss of central vision. Traditional retinitis pigmentosa, specifically, produces arterial attenuation, bone-spicules, lenticular changes, and waxy pallor of the optic disc.
   4. Retinitis pigmentosa is thought to stem from mutations in rhodopsin, peripherin, and retinol acetyltransferase genes which results in a lack of transfer of rhodopsin into the photoreceptor outer segments. This failure to transfer rhodopsin leads to apoptosis and overall destruction of rods. The majority of other tapetoretinal degenerations derive from a similar mechanism.

b. Expound on unique features:
   1. The unique feature of this case is the patient’s age. Patients with this condition are usually diagnosed at a young age, and this patient was 65. The patient may have had decreasing visual fields for years, but may have just noticed them within the last two years due to the increasing encroachment on his central vision.
   2. Another unique feature of this case is the use of Electrodiagnostic testing in characterizing the disorder. ERG is considered the gold standard for diagnosis of this condition.

VI. Treatment and Management:

a. Treatment and Response to Treatment:
   1. There is no cure for retinitis pigmentosa and like tapetoretinal degenerations. The majority of treatment is palliative care.
   2. There are four major areas of research in possible future treatment options:
      i. Retinal gene therapy to correct gene abnormality.
      ii. Use of pharmaceutical or nutritional supplements to prevent destruction of photoreceptors.
      iii. Use of electronic retinal implants to take the place of photoreceptors.
      iv. Use of gene manipulation/transplantation to regenerate photoreceptors.
   3. A low vision evaluation was recommended to our patient for aid in activities of daily living. Patients should also be given genetic counseling due to the often hereditary nature of the disease process.
   4. Vitamin A supplements are also thought to slow down further visual field loss.
V. Conclusion:

a. Tapetoretinal degenerations include a wide range of genotypes and phenotypes producing a diverse number of signs and symptoms.

b. The best method of diagnosis includes a thorough record of the patient’s symptoms and familial history combined with extensive physical testing to eliminate differentials and manage the proper diagnosis. Visual electrodiagnosis, dark adaptometry and appropriate visual field testing remain mainstays for diagnosis and monitoring.

c. Although visual prognosis is generally poor low vision care is essential to improve peripheral, central and night vision.