Title:

Vitreoretinal Interface Abnormalities in Retinitis Pigmentosa, The Forgotten Vision Threatening Complications

Abstract:
50 yo Hispanic male presents with Retinitis Pigmentosa (RP). In addition to common signs of RP, vitreoretinal interface abnormalities including epiretinal membrane (ERM) formation are found on clinical examination and on optical coherence tomography studies.

I. Case History

Patient demographics:
50 yo Hispanic Male

Chief complaint:
Worsening distance and near vision during the day and poor vision at night

Ocular History:
Retinitis Pigmentosa diagnosed at young age
Cataract extraction with PCIOL OU

Medical History: unremarkable

Medications: None

Family Ocular History:
2 younger brothers have similar vision problems, but pt reports his condition is most advanced

II. Pertinent findings

Clinical:
UCVA: 20/100 OS 20/80
Pupils: PERRL(-) APD
EOMs: Full OD/OS
CVF: Constricted 360 OD/OS
Manifest: +1.25-1.50x040 VA 20/80-2
    +0.25-1.75x115 VA 20/80-2
Anterior Segment:
    Cornea: clear with mild endothelial pigment deposition OD/OS
    Lens: PCIOL OD/OS clear centrally
Posterior Segment:
    C/D: OD 0.4/0.4 OS 0.45/0.45
    Disc: waxy pallor greatest superiorly OD/OS
    Macula: ERM with radial striae OD/OS
    Vessels: arteriolar attenuation OD/OS
    Periphery: heavy mid peripheral bone spicule RPE hyperplasia to arcades; retina flat and intact 360 OD/OS
    Vitreous: mid peripheral vitreous condensation with strands 360 OD/OS

Imaging:
    OCT: ERM OU with mild cystic edema
    Optos: color fundus photos showing vitreoretinal interface abnormalities

Physical: N/A
Laboratory studies: N/A
Radiology studies: N/A

III. Differential diagnosis

Retinitis Pigmentosa
X-Linked Juvenile Retinoschisis
Wagner Hereditary Vitreoretinal Degeneration
Goldman Favre Vitreotapetoretinal Degeneration

IV. Diagnosis and discussion

-- RP is group of a hereditary retinal disorders characterized by progressive degeneration of photoreceptors and retinal pigment epithelium, leading to constriction of visual field, nyctalopia, and loss of visual acuity
-- common findings are waxy optic disc pallor, arteriolar narrowing, pigment clumping/”bone spicule” deposits and atrophy of the RPE

-- RP typically affects peripheral vision first sparing central vision until in the later stage
-- central vision threatening complications include optic nerve pallor, cystoid macular edema, and posterior subcapsular cataracts
-- however, lesser known changes in the vitreous and vitreo-macular
interface (5) can also affect central vision

-- vitreoretinal interface is affected in 94% of RP patients (5)
-- an OCT study has also shown that ERMs were found in 64.3% of eyes with RP (3)

-- electron microscopic studies show RP vitreous consists of: pigment epithelial cells, uveal melanocytes, retinal astrocytes, and macrophage-like cells (1)
-- additionally, tightly packed filaments were found together with normal collagen fibers and loose pigment granules (1)
-- in control groups, macrophages and loose pigment were only occasionally found (1)
-- the above reason and the progressive loss of hyaluronic acid (4) are what have been proposed as the reason responsible for these vitreoretinal interface abnormalities

-- this vitreous degeneration process is divided into 4 stages (1-- being the mildest form – colorless, dust like particles / 2 – complete separation of posterior hyaloid from vitreous / 3 – posterior hyaloid has moved anteriorly creating a matrix of interconnecting fibers and large opacities / 4 – vitreous gel has collapsed with a great volume reduction) (4)
-- vitreous abnormalities can be found as early as 1st decade, no sex preference, and affects both hereditary and sporadic types (4)
-- moreover, it is bilateral, symmetrical and progresses rapidly (4)
-- the progression of this vitreous degeneration parallels the retinal malfunction (4)

-- another OCT study found that all eyes presenting with micropseudocysts or cystoid macular edema also presented with ILM thickening or epiretinal membrane showing importance of involvement of vitreoretinal interface abnormalities and suggesting a multifactorial pathogenesis of CME in RP (5)

V. Treatment, management

-- vitrectomy and ILM removal have been shown to effectively reduce macular edema with improvement of visual acuity, but must consider that dystrophic retinas do not tolerate surgical trauma well and there are other mechanisms besides vitreous tractions that are causing these macular complications ie. failed RPE pumping mechanism (3)
-- therefore, in this patient an ERM peel was not considered

VI. Conclusion

-- vitreoretinal interface abnormalities including ERMs have been shown to be highly associated with RP and should be considered as potential reason for
decrease in vision in these patients
-- however, there are multiple mechanisms at play including RPE atrophy/dysfunction and surgical outcomes may not always be favorable in these patients
-- the OCT is a very powerful tool in following these patients, assessing macular complications and deciding the appropriate management/treatment

References:


