A Case Report of Retinitis Pigmentosa Inversa: Review of Pathology, Treatment and Management

ABSTRACT

A rare form of retinitis pigmentosa in a Caucasian male causes a progressive loss of vision with subsequent glare and contrast impairment.

I. Case History
   a. Demographics: 74-year-old Caucasian male
   b. Chief complaint: Gradual contrast impairment, reduced peripheral and central vision, glare OS
   c. Goal: To improve contrast and glare OS
   e. Medical history: Hyperlipidemia, depression, diabetes
   f. Occupation: Retired, formerly served in military
   g. Hobbies: Very active, plays golf

II. Pertinent findings
   a. Clinical Findings (bolded items will be represented by images)
      i. Uncorrected vision: CF at 5 feet OD, light perception only OS
      ii. Anterior segment:
         1. Temporal TIDs of the iris OD, normal iris OS, corneas clear OU
         2. PC-IOL OD with no PCO, NO3 NC4 cataracts (per LOCS III) with 3+ dense brunescence and PSC OS
      iii. Posterior segment: Extensive chorioretinal atrophy in the posterior pole extending into the mid-periphery OU, mild pigmentary changes scattered throughout, vessel attenuation, waxy disc pallor OU
      iv. Past HVF 24-2, 10-2
      v. Past Fluorescein Angiography
      vi. Macula Star and Dense OCT: Diffuse chorioretinal atrophy OD 360 extending from macula into posterior pole; unable to discern foveal pit (no OS scans due to media opacity)
      vii. Fundus Autofluorescence: Diffuse area of non-fluorescence in posterior pole OD indicating PR cell loss and RPE atrophy (no OS scans due to media opacity)

III. Differential diagnosis for retinitis pigmentosa inversa
   a. Leber’s congenital amaurosis
b. Central gyrate atrophy
c. Central areolar choroidal sclerosis
d. Progressive cone-rod dystrophy
e. Syphilitic retinopathy
f. Thioridazine/chloroquine/hydroxychloroquine retinopathy

IV. Diagnosis and Discussion

a. Diagnosis: Retinitis pigmentosa inversa OU

b. Discussion: Retinitis pigmentosa (RP) encompasses a wide variety of hereditary disorders, both clinically and genetically, that involve progressive loss of the retinal pigmented epithelium and photoreceptors. It is one of the most common inherited retinal disorders, with a prevalence of 1 out of 4000. Retinitis pigmentosa inversa is an extremely rare variant of RP, with an unknown prevalence. Since it was first discovered in the 1800s, only a few dozen cases have been reported. Other variants of RP include sine pigmento (pigment changes are minimal or absent), sector RP (only one or two sectors of the fundus are affected), retinitis punctata albescens (scattered white dots located between posterior pole and equator), and unilateral RP (typical RP findings in one eye).

c. Modes of inheritance vary within RP subtypes and include: Sporadic, autosomal dominant (AD), autosomal recessive (AR), and X-linked forms (XL). It has been postulated that there is an AR pattern of retinitis pigmentosa inversa, however AD pattern and simplex forms have been reported. AD forms of RP are the most common and have the best prognosis with the rarest being XL and having the worst prognosis.

d. In classical RP, early signs involve arteriolar narrowing and attenuation. Very mild pigmentation can be noted. Later features show “bone-spicule” formation, and more definite pigmentary changes begin in the mid-periphery. “Waxy” optic disc pallor is noted at the advanced stage of the disease. Bilateral peripheral vision loss occurs with subsequent changes in color vision and night blindness.

e. Typical ocular findings that are associated with RP are optic nerve head drusen, vitreous particles and posterior subcapsular cataracts (common in all forms of RP). Cystoid macular edema, RPE atrophy, or ERM formation can also manifest over time.
Retinitis pigmentosa inversa (RPI), also known as pericentric retinitis pigmentosa, is an extremely rare form of RP with an unknown prevalence. Signs of RPI can vary depending on the stage of the disease, but are the same as typical RP. Pigmentary changes, RPE and choroidal atrophy occur in the posterior pole, along with vessel attenuation. The age of onset is not known; our patient was first detected with RP in the 1970s when he was in his 30s.

There are two types of retinitis pigmentosa inversa: Pericentral and central form. In the pericentral form, chorioretinal atrophy and pigment migration concentrate around the macula, leaving central vision unaffected. This type tends to have a favorable prognosis with minimal progression after three to five years. On the contrary, the central form of RPI is characterized by bilateral retinal changes central to the posterior pole, affecting the macula. Pigmentary clumps and bone-spicules form, primarily along the horizontal axis. Our patient exhibits the signs and symptoms of the central form.

V. Evaluation and Prognosis, Treatment and Management

a. Evaluation

i. Typically objective data, in conjunction with a dilated fundus exam, and patient symptoms establish a diagnosis of RPI
   1. A complete review of medical history and medications should be done to rule out hydrochloroquine, chloroquine, or thioridazine toxicity.
   2. Laboratory testing for syphilis (including RPR and fluorescent treponemal antibody absorption) should be done to rule syphilitic retinopathy.
   3. Visual fields should be performed. An incomplete or absolute ring scotoma would be indicative of typical RP. In our case, a visual field showing a bilateral central scotoma would be expected.
   4. Color vision testing with Farnsworth-Munsell or HRR can be helpful, but abnormalities are not usually detected at late stages of disease. Most patients with RP tend to exhibit tritanopia; however deuteranopia and protanopia can be present.
   5. Electroretinography can be helpful in situations where the fundus shows minimal changes. Expected findings are severely reduced or absent scotopic ERG response, and an unaffected normal photopic ERG.
ii. Prognosis
   1. With RP inversa, expected visual acuities can range from 20/400 to light perception only in both eyes. Patients are usually legally blind by the 40s and 50s.4

iii. Treatment and Management
   1. Annual dilation, perimetry and serial FAF should be performed to detect any status change. Findings such as cystoid macular edema or a worsening cataract that have a course of treatment, could improve the patient’s overall quality of vision.
   2. ERG testing can show reduced or absence of scotopic amplitudes preceding reduction of photopic amplitudes. Performing ERG can also be helpful to identify female carriers of X-linked RP for genetic counseling as it has shown to be abnormal in over 90% of female carriers.6,9
   3. Genetic testing and laboratory testing should be pursued to confirm or rule out any atypical forms of RP with possible systemic associations.6
   4. Mega doses of Vitamin A can be prescribed for patients 18 and older, although its role is controversial. This recommendation is for typical forms of RP and has not been studied in atypical forms. It may slow the progression of RPE atrophy, shown by reduction of ERG amplitudes.9
   5. Vitamin E should be avoided for all RP patients as a daily dose of 400IU led to faster progression, according to one study.
   6. Systemic acetazolamide (Diamox 500mg IV or PO) for CME is also controversial due to the lack of research. It may be considered if CME persists after several weeks and visual acuity has not improved.9
   7. Cataract surgery can be indicated, as in our patient, with complaints of glare, contrast reduction, depending on retinal function.

VI. Conclusion
   a. Retinitis pigmentosa inversa is a very rare disease that slowly destroys the RPE layer and photoreceptors of the macula and posterior pole, leading to a progressive loss of central bilateral vision. Patients affected by this condition usually retain their peripheral vision until very late in the disease. Our patient had extreme peripheral vision loss with little field remaining.
b. The lack of research and studies due to the rarity of the disease presents a clinical challenge. Although there is no direct treatment, offering support through low vision devices, filters, night vision aids, field expanders, and orientation and mobility treatment can greatly improve a patient’s quality of life.

c. Our patient was extremely independent, and did not demonstrate a need for a cane. He was previously examined in the low vision clinic and was issued several low vision devices. At his most recent appointment, he did not express the need for any more devices, but simply desired an improvement to his visual problems. Since he had already undergone successful cataract surgery OD, a cataract consultation was scheduled for the OS.

d. In conclusion, appropriate steps should be taken to educate the patient and families involved of the visual expectations, and to offer any feasible treatment, surgical or palliative care that could enhance remaining vision.

VII. References