Case of the Month

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A 54-year-old Caucasian female patient presented with gradual worsening of vision in both eyes over the past 2 1/2 years.

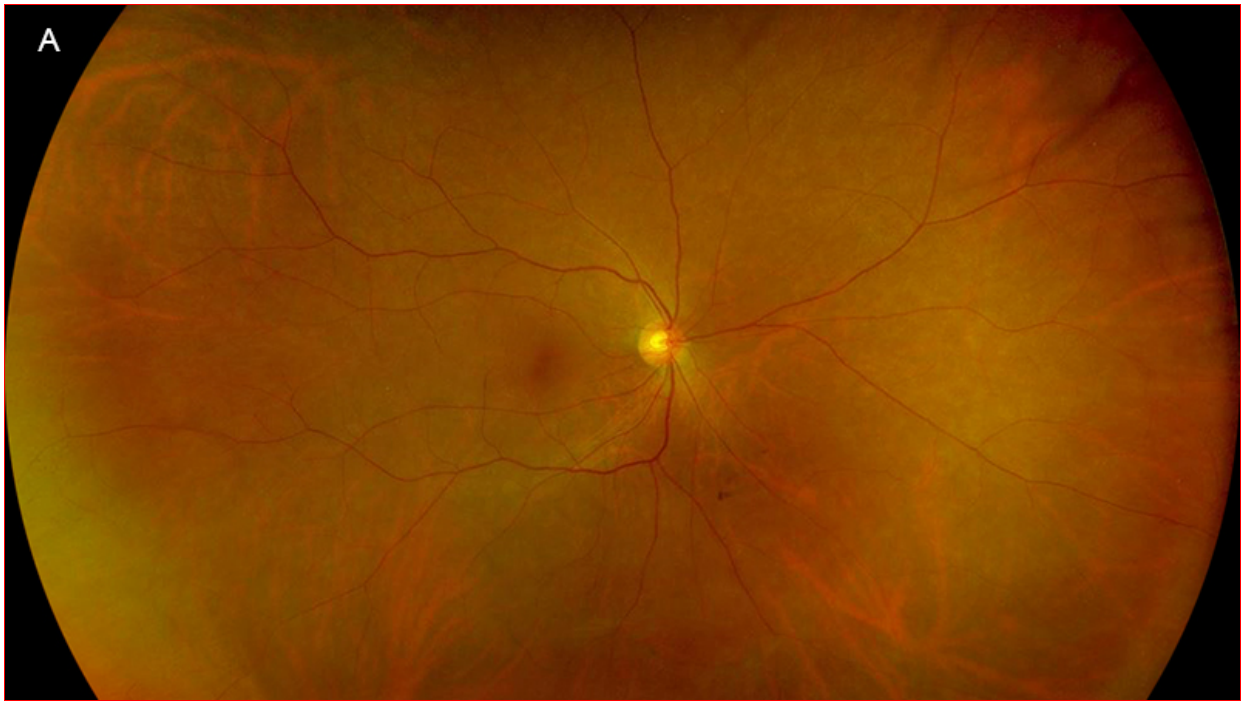


Figure 1A: Color photo of the right eye. Mild arteriolar narrowing is present.

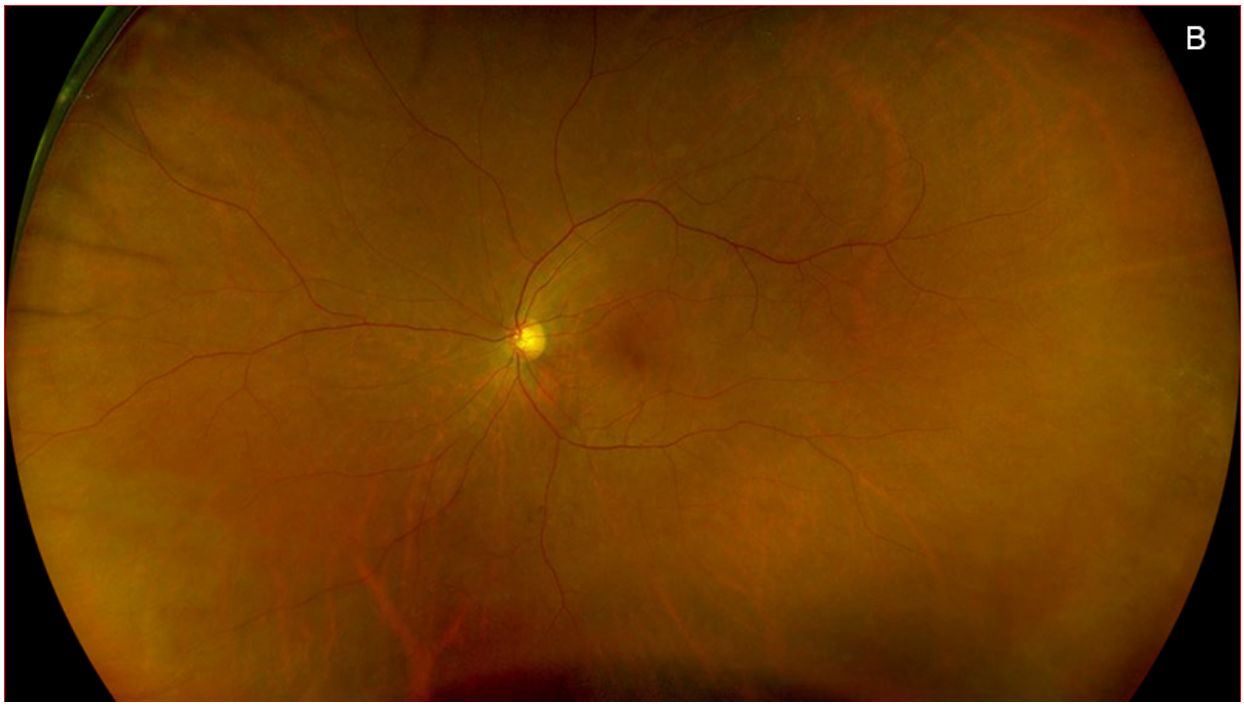


Figure 1B: Color photo of the right eye. Mild arteriolar narrowing is present.

Case History

The patient first noticed visual symptoms 2 ½ years earlier, experiencing central metamorphopsia and positive photopsias in the inferior field of her left eye. One month later, she developed symptoms in her right eye with "pixelation" of objects in her vision. Initial evaluation during her onset of symptoms was noted to be unremarkable except for bilateral posterior vitreous detachments (PVD). Her photopsias gradually worsened in both eyes over the following months as well as her ability to see complete images. She reported having 20/20 acuity in both eyes in 2013 and has since noted a progressive decline. She denied having a family history of related symptoms or ocular disease except for an older brother with a history of retinal detachment. Her past medical and ocular history are unremarkable, apart from mild myopia in both eyes. She takes Nasonex as needed for seasonal allergies. Her past medical history, surgical history, and social history is otherwise unremarkable. Her review of systems was negative. She was referred for our evaluation.

Her best corrected Snellen visual acuity measured 20/50 in the right eye (OD) and 20/100- in the left eye (OS). No afferent pupillary defect was noted. Her intraocular pressure was 15 mmHg in both eyes. Anterior segment examination was unremarkable in both eyes. The funduscopic examination of both eyes showed mild arteriole narrowing and faint yellow patches in the mid-periphery (Figure 1). Spectral domain optical coherence tomography (SD-OCT) showed loss of the photoreceptor inner and outer segments in the macula of both eyes (Figure 2). Fundus autofluorescence (FAF) of the posterior pole showed a mild increased autofluorescence in the posterior pole (Figure 3). Fluorescein angiography (FA) showed diffuse patchy hyperfluorescence, most prominent along the arcades and in the peripapillary regions of both eyes with late vascular leakage of the retinal veins (Figure 4).

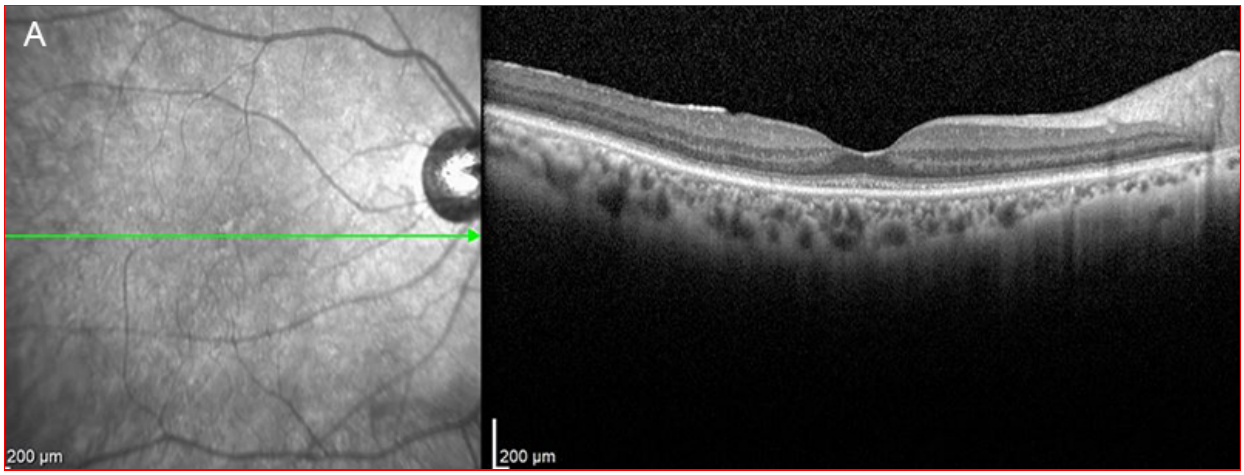


Figure 2A: Spectral Domain OCT of the right macula. Note the loss of photoreceptors in the juxtafoveal area.

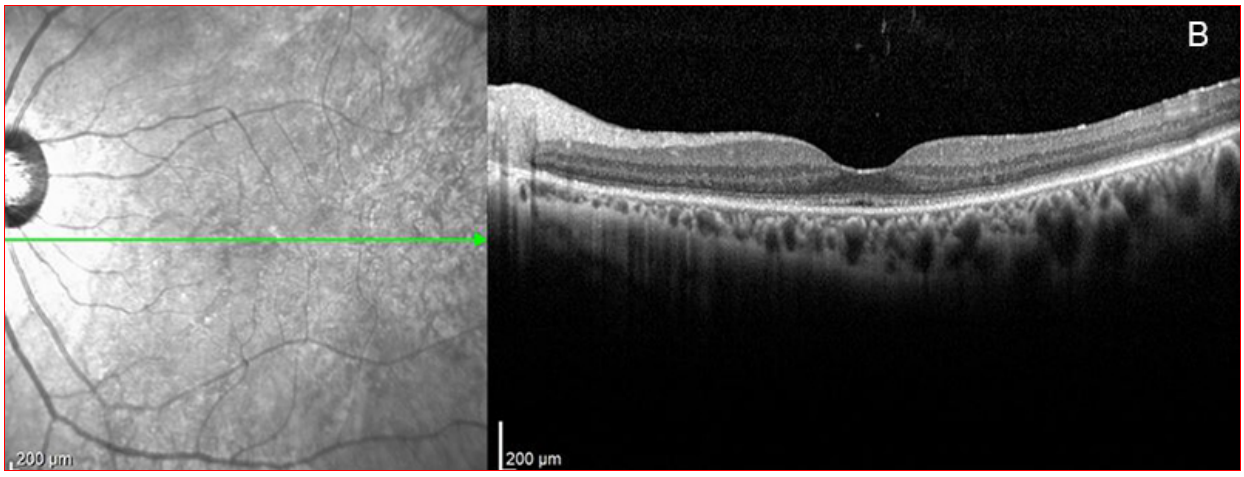


Figure 2B: Spectral Domain OCT of the left macula. A similar loss of photoreceptors in the juxtafoveal area, as seen in the right eye, is present.

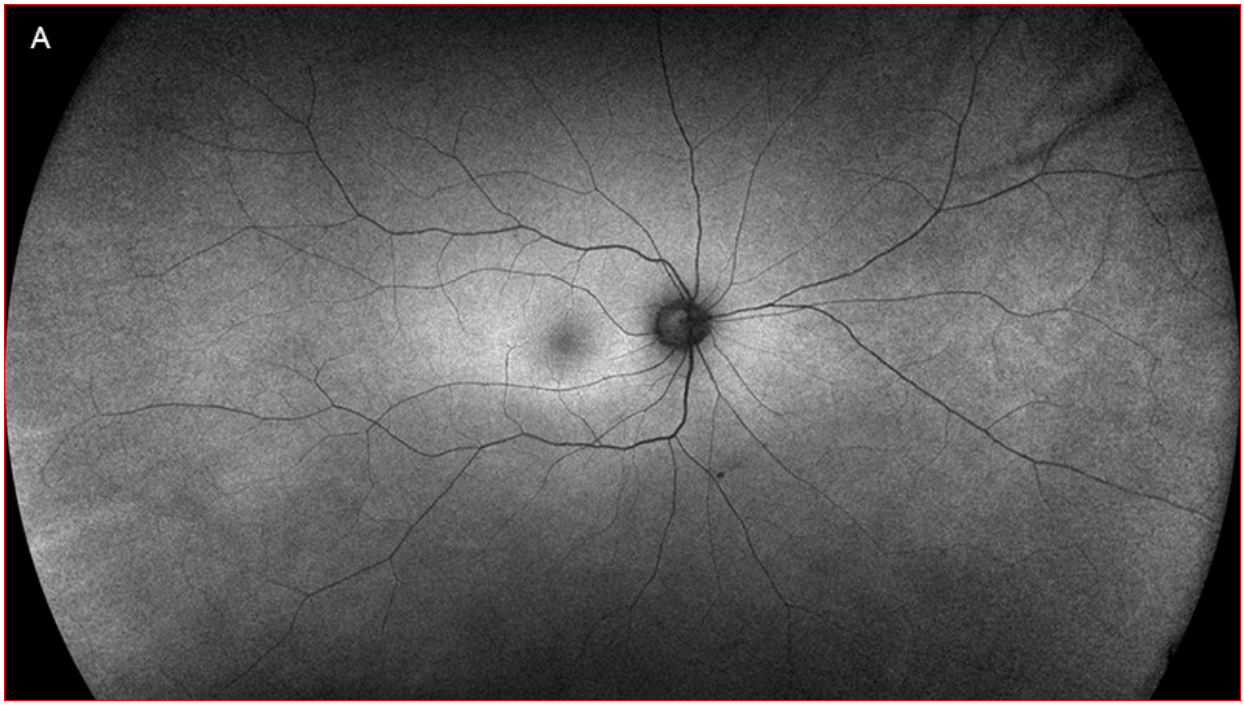


Figure 3A: Wide-field fundus autofluorescence of the right eye showed a mild increased autofluorescence in the posterior pole.

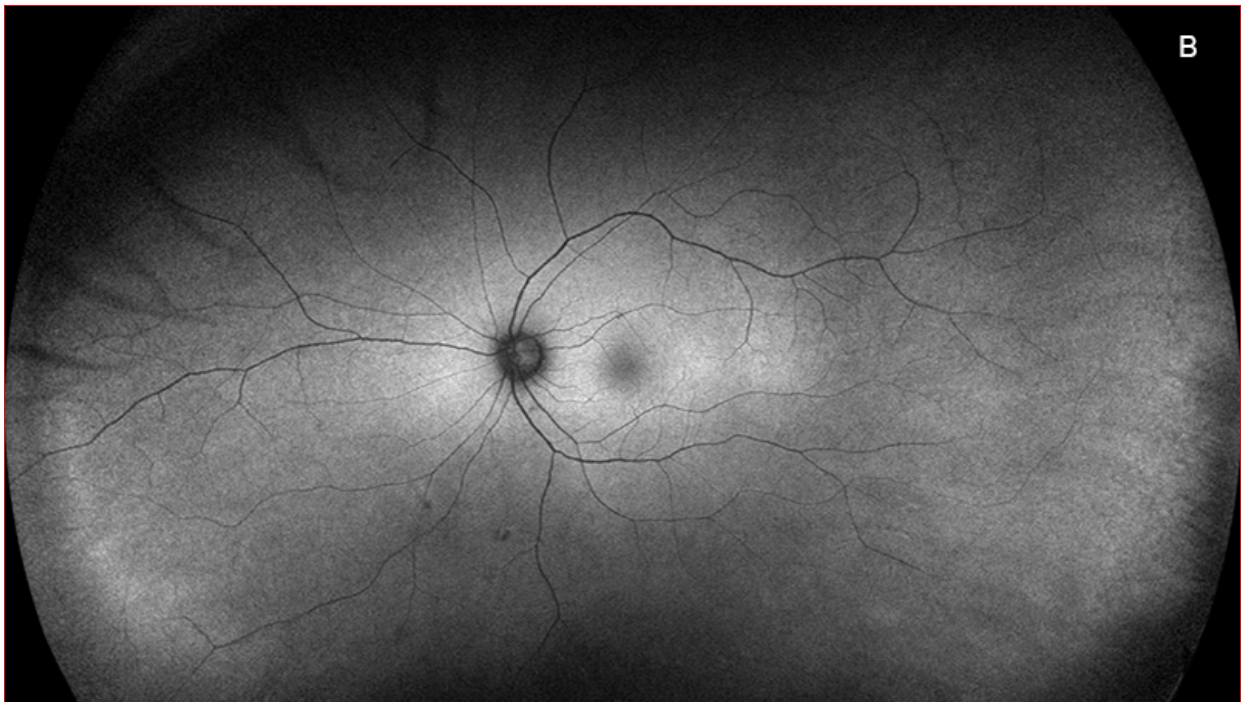


Figure 3B: Wide-field fundus autofluorescence of the right eye showed a mild increased autofluorescence in the posterior pole.

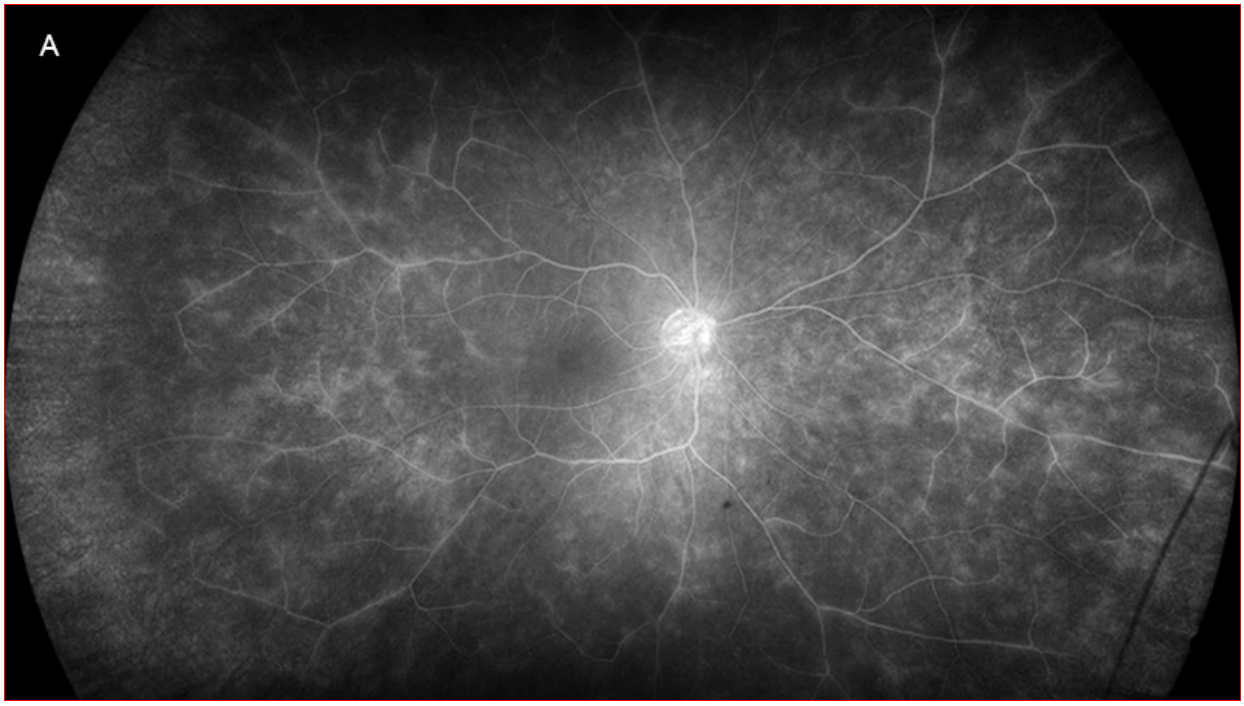


Figure 4A: Wide-field fluorescein angiography of the right eye is most notable for widespread venular fluorescein leakage

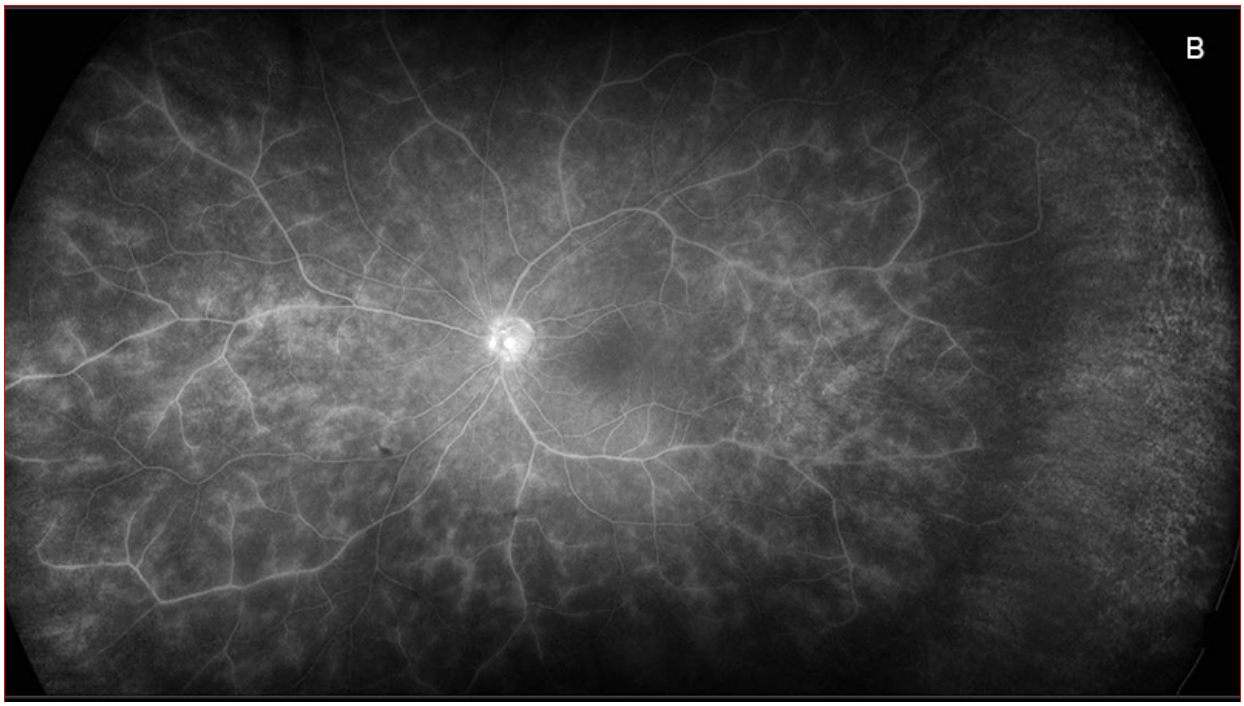


Figure 4B: Wide-field fluorescein angiography of the right eye is most notable for widespread venular fluorescein leakage

What is Your Diagnosis?

Differential Diagnosis

- Autoimmune Retinopathy (AIR)
- Cone-Rod Dystrophy
- Retinitis pigmentosa
- Acute zonal occult outer retinopathy (AZOOR)
- Inflammatory/infectious retinopathy
- Toxic retinopathy

Additional History, Testing and Patient Course

Prior to her retinal evaluation, the patient underwent testing with multifocal electroretinography (mfERG) which showed diminished central amplitudes in both eyes (Figure 5). After her initial visit with our clinic, further workup was performed, including a Goldman visual field which demonstrated a relative, central scotoma in both eyes (Figure 6). Full-field ERG showed generalized rod and cone dysfunction (Figure 7). Her electrooculogram (EOG) had a significant subnormal Arden ratio in both eyes: 1.1 and 1.3 (normal ≥ 1.80) (Figure 8).

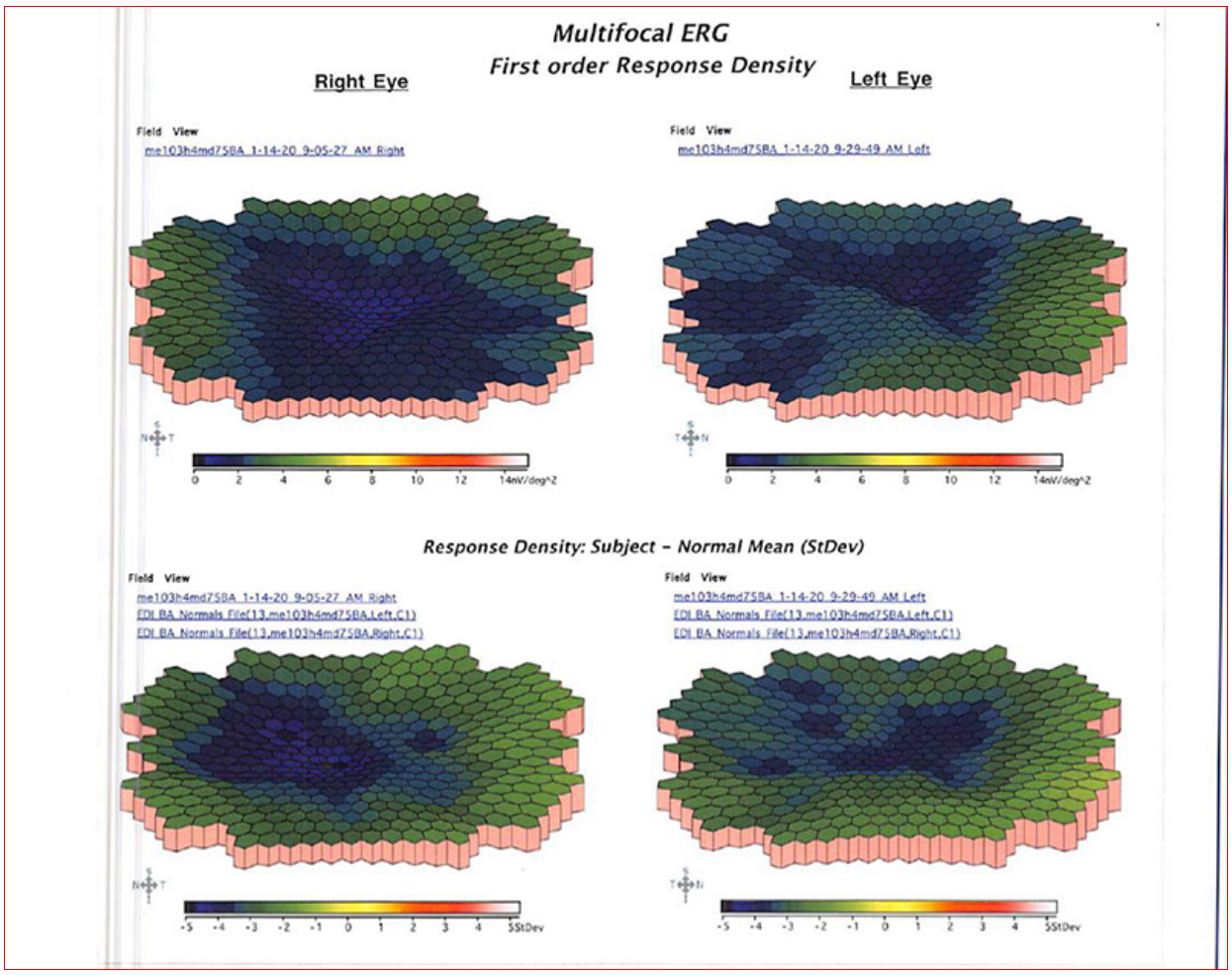


Figure 5: Multifocal ERG showing reduced central amplitudes

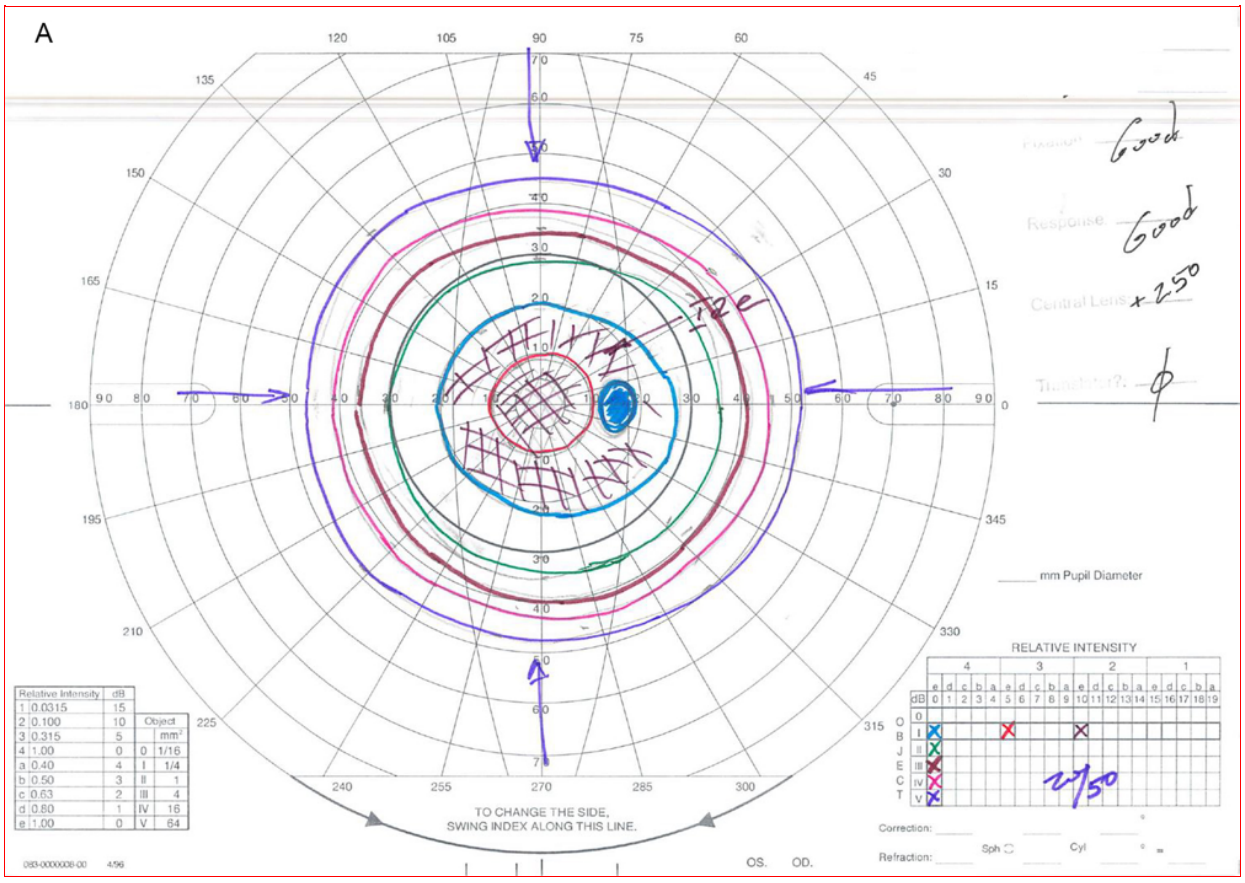


Figure 6A: Goldmann visual field of the right eye showing a relative central scotoma.

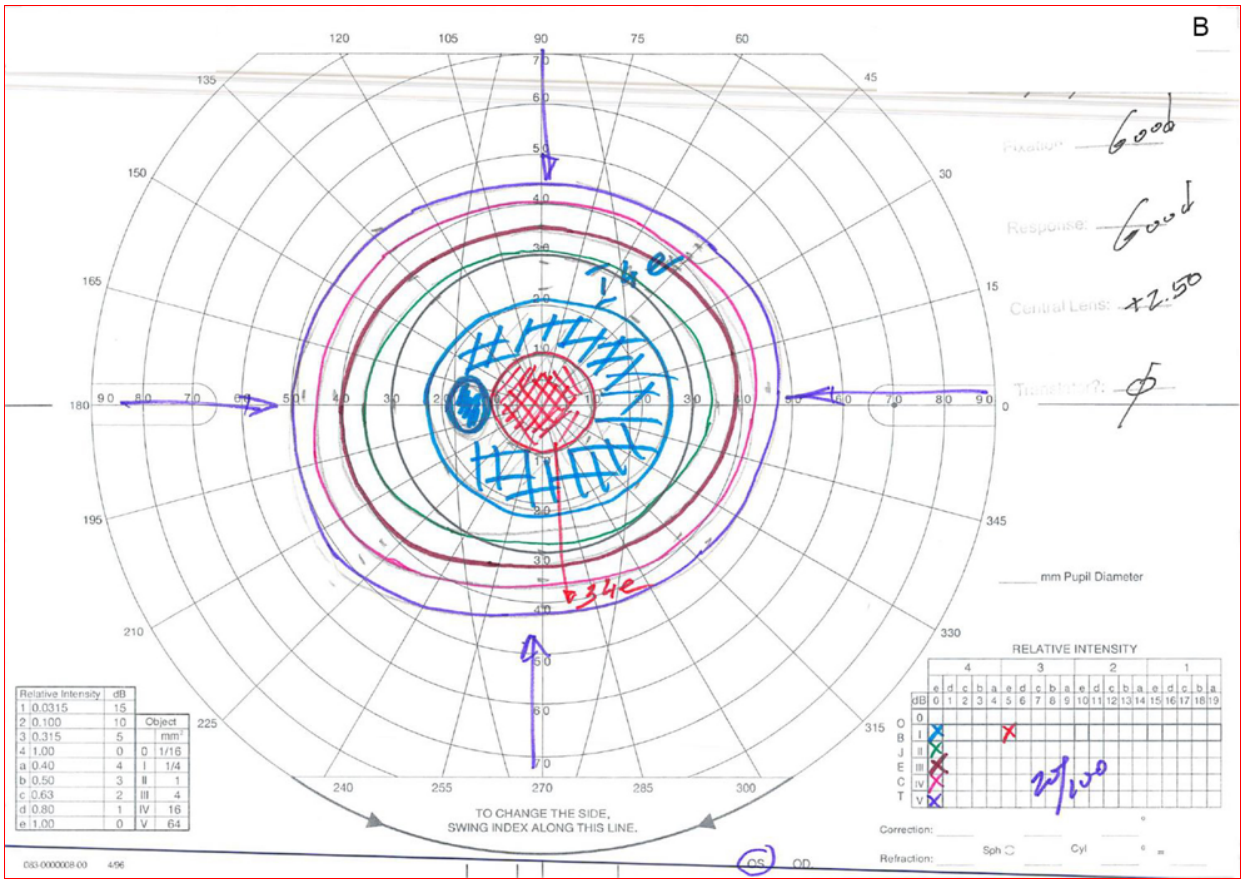
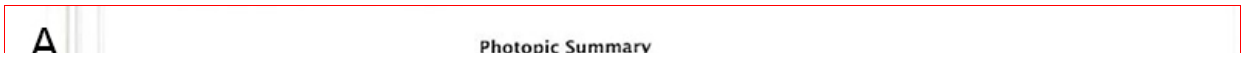


Figure 6B: Goldmann visual field of the left eye showing a relative central scotoma.



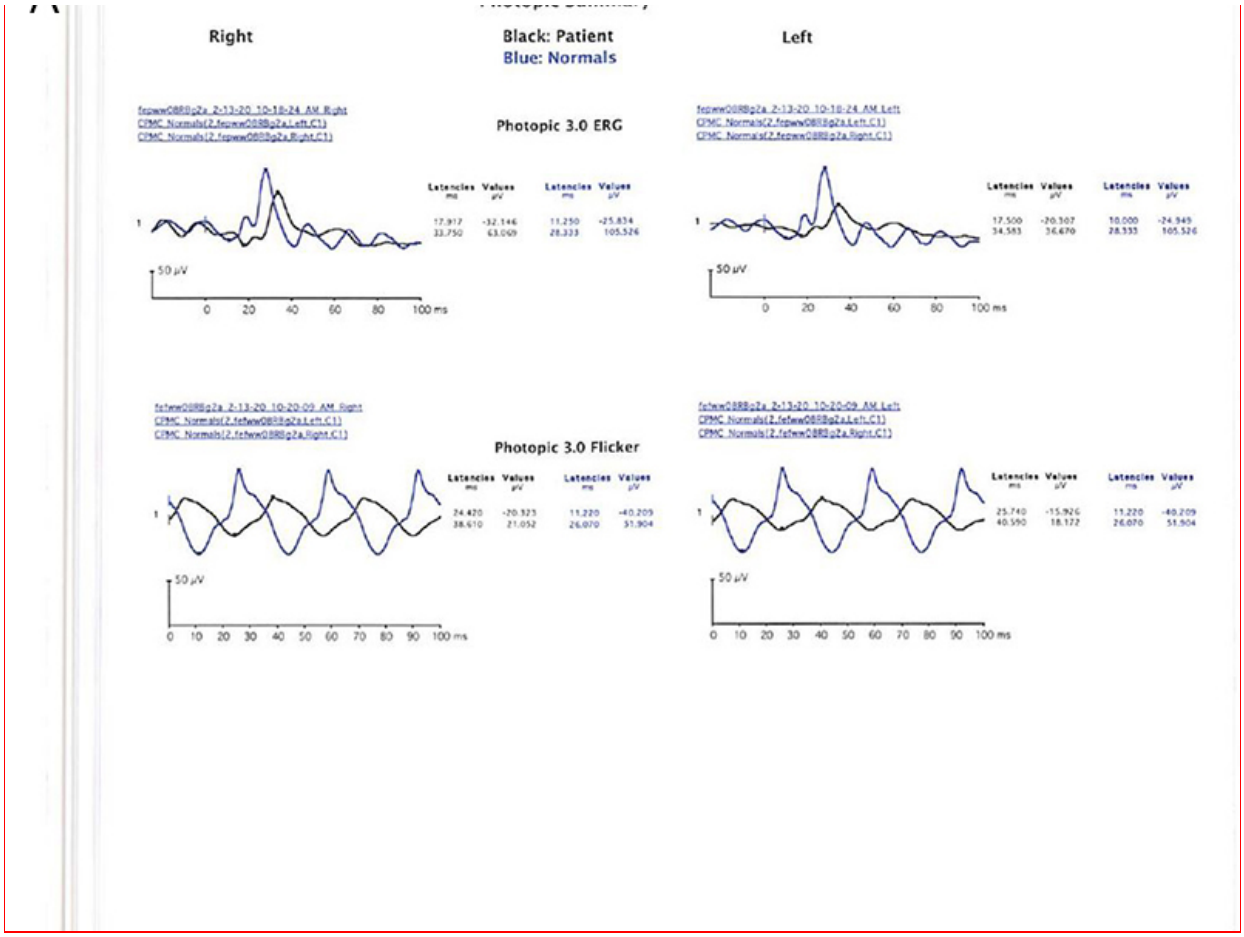


Figure 7A: Photopic ERG of the right and left eye showing generalized rod and cone dysfunction in both eyes

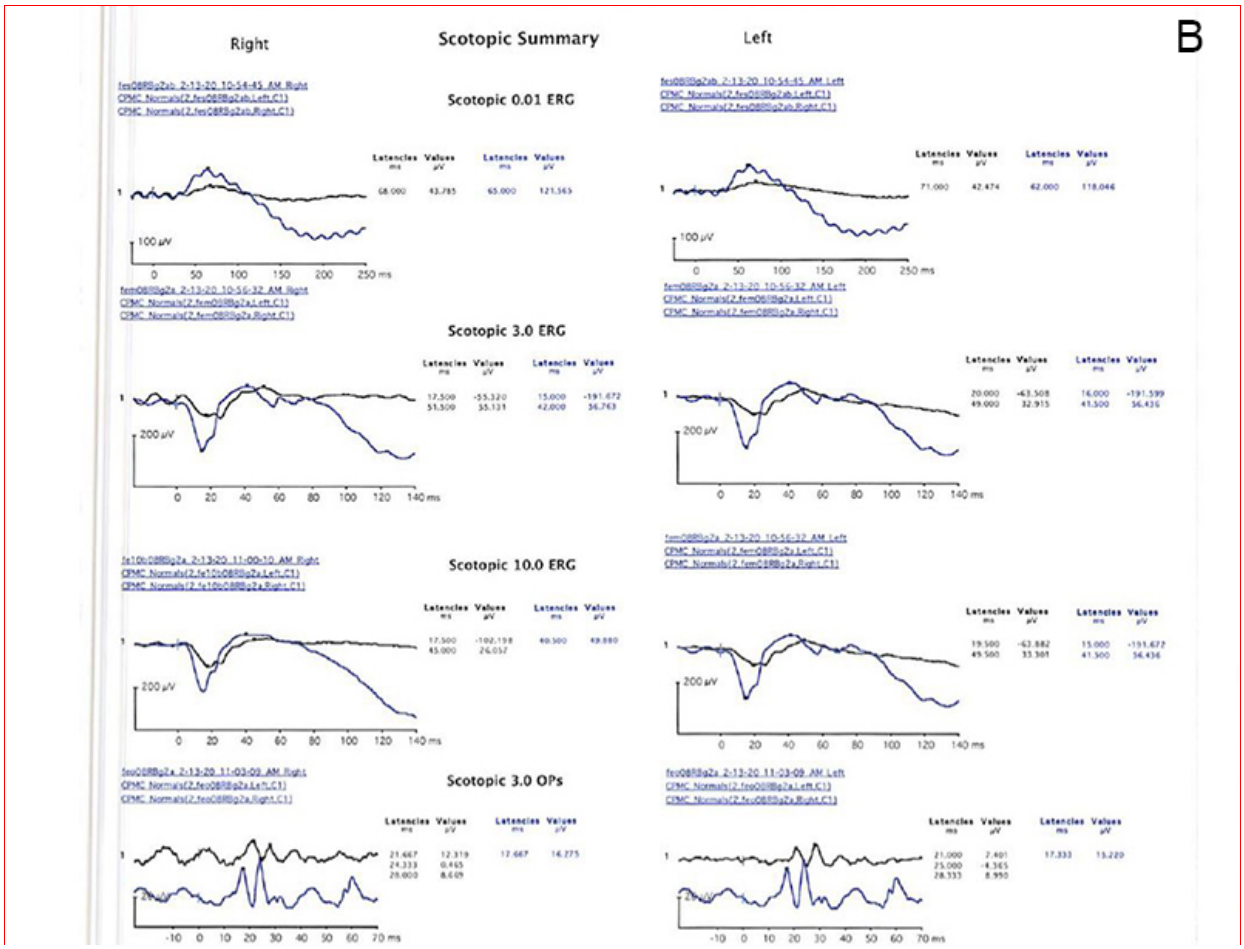


Figure 7B: Scotopic ERG of the right and left eye showing generalized rod and cone dysfunction in both eyes

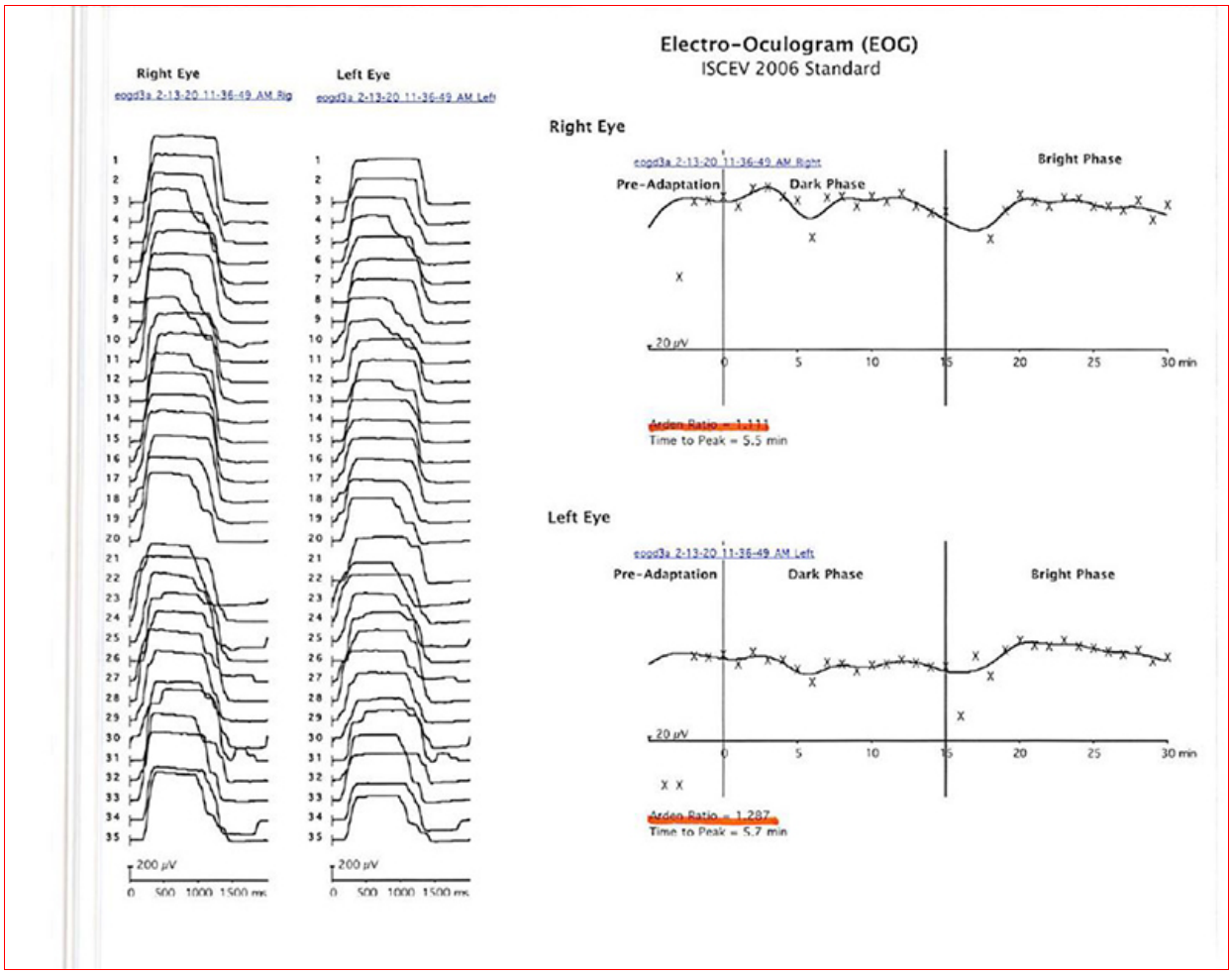


Figure 8: Electro-oculogram of the right and left eye showing a subnormal arden ratio.

Discussion

Autoimmune retinopathy (AIR) is commonly divided into two categories: paraneoplastic (PAIR) and nonparaneoplastic (nPAIR). PAIR is associated with the presence of antiretinal antibodies that occurring in the presence of cancer. Cancer-associated retinopathy (CAR) and melanoma-associated retinopathy (MAR) are more specific subtypes of the PAIR category. In contrast, antiretinal antibodies from nPAIR are thought to arise from the presence of systemic inflammatory or infectious disease.

Diagnosing AIR is challenging, as the differential is rather extensive. Commonly presenting symptoms of AIR can be found in Table 1.[1,2] Fox et al used a modified Delphi process to establish essential diagnostic criteria, supportive diagnostic criteria, and core diagnostic testing recommended for AIR which can be seen in Table 2.[3] Patients should generally be monitored every 3 months with repeat ancillary testing to assess for disease progression.[3]

The treatment of AIR involves the use of steroids (local or systemic), conventional immunosuppressives (such as antimetabolites or T-cell inhibitors), biologics (such as monoclonal antibodies), or intravenous immunoglobulin (IVIG), with steroids and conventional immunosuppressives recommended as first and second line treatments.[3] Most recent studies have focused on the use of immunomodulating and biological agents. Combination therapy or monotherapy with rituximab has shown some limited success with the use in these patients.[4-6] In a case series of 16 patients with AIR, Davoudi et al showed seventy-seven percent of eyes had stable or improved VA 6 months after rituximab initiation.[4] Immunosuppressive therapy, however, does not prove to be beneficial once widespread retinal degeneration has occurred.[7] In general, the prognosis for patients with AIR is not well-defined, but the clinical course is generally progressive, with a small percentage of patients showing clinical improvement.

Table 1: Common symptoms of AIR[1,2]

1. Photopsia

2. Night blindness
3. Scotomata
4. Dyschromotopsia
5. Central vision loss

Table 2: Essential Criteria for AIR[3]

1. No apparent cause for visual function abnormality (e.g. malignancy, inflammation, infection, surgery, drug toxicity, trauma, hereditary retinal degeneration)
2. ERG abnormality with or without visual field abnormality
3. Presence of serum antiretinal antibodies
4. Absence of fundus lesion and retinal degeneration or dystrophy that may explain visual function loss
5. Absence of overt intraocular inflammation

Supportive Diagnostic Criteria for AIR[3]

1. Personal or family history of systemic autoimmune disease
2. Presence of photopsias, scotoma, nyctaloopia or photoaversion, dyschromatopsia
3. Rapidity of onset of vision change (acute, 0-3 months or subacute, 3-6 months).

Core Diagnostic Tests[3]

1. Evaluation for malignancy
2. Serum antireitnal antibody testing
3. Electroretinogram (ERG)
4. Fluorescein angiography (FA)
5. Fundus autofluorescence (FAF)
6. Optical coherence tomography (OCT)

Take Home Points

- AIR is divided into paraneoplastic autoimmune retinopathy and nonparaneoplastic autoimmune retinopathy.
- AIR patients may present with photopsia, night blindness, scotomata, dyschromotopsia and/or central vision loss.
- The diagnosis of AIR is typically a diagnosis of exclusion to exclude a broad differential diagnostic list of disorders.
- Diagnostic evaluation of AIR includes ERG, FA, FAF, OCT, antiretinal antibody testing and a careful search for malignancy.
- Management of AIR involves steroid and conventional immunosuppresive therapy as 1st and 2nd line treatment options.

References

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