Autoimmune Retinopathy: A Systemic Approach

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INTRODUCTION

Autoimmune retinopathy (AIR) is a rare but visually devastating ocular condition. Patients often present with bilateral sub-acute decreased vision with a normal appearing fundus initially and vascular attenuation, optic nerve pallor, and macular ganglion cell loss later on in the clinical course. Autoimmune retinopathy may be caused by either non-paraneoplastic (nPAIR) or paraneoplastic (PAIR) etiology. A multidisciplinary approach is necessary for the diagnosis, treatment, and management of the patient. Visual prognosis is often poor and a referral to low vision rehabilitation is highly recommended as the patient may have difficulties with activities of daily living. This case report highlights the work-up that is pertinent to diagnosis as well as treatment and management of condition and patient's overall functional vision.

CLINICAL FINDINGS

A 53 y/o African-American male presented to clinic with a complaint of progressive bilateral vision loss over the course of six months.

	OD	OS
ANTERIOR SEGMENT		
Visual acuity	5ft/80 Feinbloom acuity	20/400 Snellen acuity
Color Vision (Ishihara)	3/14	4/14
Pupils, EOMs, CVF, SLE	NL	NL
POSTERIOR SEGMENT		
ONH	1-2+ temp pallor	2+ temp pallor, inf temp neuroretinal rim thinning
Macula, vitreous, periphery	NL	NL
Vessels	Arteriolar attenuation	Arteriolar attenuation, isolated resolving cotton wool spot present along the inferior temporal arcades

DIAGNOSIS AND DISCUSSION

There is no gold standard for diagnosis. One proposed criteria for diagnosing non-paraneoplastic autoimmune retinopathy is:

(1) no history of or current cause of decreased vision
(2) an abnormal but not specific pathologic pattern of ERG results +/- a visual field defect
(3) anti-glycolytic serum antibodies
(4) no intraocular inflammation

Furthermore, other testing that may aid in diagnosis are: OCT RNFL which may show optic nerve atrophy, OCT macula showing retinal thinning (such as with ganglion cells), fundus autofluorescence, unremarkable fluorescein angiography, and decreased color vision. Many of the systemic and ocular testing is done to rule out other causes of vision loss.

FIGURE 1
Fundus photography

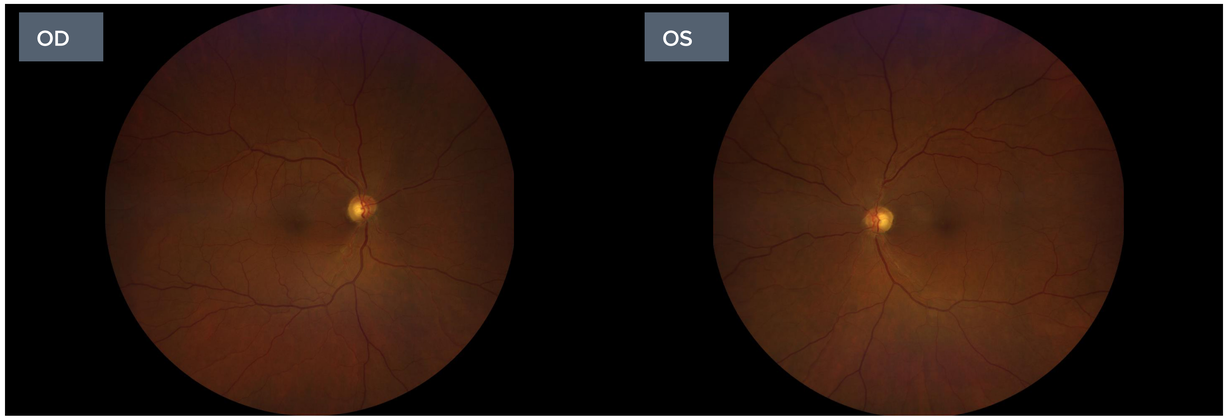

FIGURE 2
Ganglion cell analysis of the right and left eye respectively

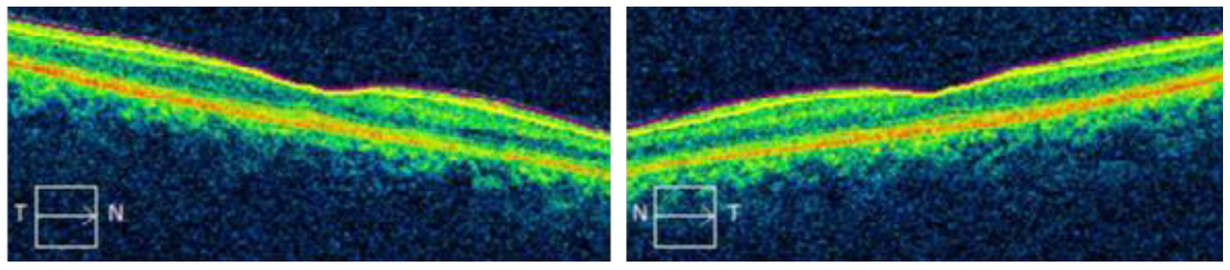

FIGURE 3
Lab testing for infectious, toxic, nutritional, and other categories

INFECTIOUS		**AUTOIMMUNE RETINOPATHY PANEL**	
Syphillis	Non-reactive	Carbonic anhydrase II	Negative
Quantiferon gold	Non-reactive	HSP27	Negative
Hepatitis B and C	NL	Aldolase	Negative
HIV	Non-reactive	α- Enolase	Positive
TOXIC		Arrestin	Positive
Lead, zinc, arsenic, copper	NL	Tubulin	Positive
NUTRITIONAL	NL	PKM2 (pyruvate kinase M2)	Positive
RBC folate	NL	GAPDH (glyceraldehyde- 3-phosphate dehydrogenase)	Negative
Vitamin B1, B6, and A	NL		
OTHER		CANCER-ASSOCIATED RETINOPATHY	
		PANEL	
Beta HCg	NL	Recoverin	Negative
ACE	NL	Carbonic anhydrase II	Negative
Alpha fetoprotein	NL		
Creatine	NL		

PATHOGENESIS

The overarching consensus is that auto-antibodies are made against glycolytic enzymes in the retina which degrade retinal cells and their function. The glycolytic enzymes targeted are: aldolase, α-enolase, pyruvate kinase M2 (PKM2), and glyceraldehyde- 3-phosphate dehydrogenase (GAPDH). Under normal conditions, these glycolytic enzymes are crucial to the function of retinal cells. Auto-antibodies are also made against tumor antigens or chronic inflammation (such as in systemic autoimmune conditions or bacterial infection) which then target the retinal antigens through molecular mimicry, thus producing an autoimmune reaction. The autoimmune reaction may cause apoptosis which clinically manifests as vision loss.

MANAGEMENT

The patient requires multidisciplinary care:
- retinal specialists
- rheumatology
- hematology/oncology
- neuro-opthalmology
- low vision

Repeat clinical testing such as OCT RNFL, OCT macula, color vision, are generally repeated every 3 months to monitor for any signs of improvement.

CONCLUSION

Each subtype of autoimmune retinopathy may be treated differently but generally long-term systemic immunosuppression is initiated.
- CAR: oral steriods, IVIG, immunosuppression, or antioxidants
- MAR : plasmapheresis, IVIG, or radiation
- nPAIR: local/systemic steroids and/or metabolites

Currently no gold standard for diagnosis, management, or treatment exists. A series of ocular and systemic testing is warranted for diagnosis. This includes an abnormal OCT macula and RNFL with normal appearing fundus and systemic work-up. Once diagnosis is made, long-term therapy of immunosuppressive agents is prescribed and the patient is followed every 3 months with poor visual prognosis. A low vision referral or vision rehabilitation consult may assist the patient in their activities of daily living and improve their quality of life.

BIBLIOGRAPHY

Available upon request

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