CLINICAL VIGNETTE

A Case of Autoimmune Retinopathy

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Case Report

A 56-year-old woman with long standing discoid lupus presented with 6 months of progressive vision loss and night blindness. Her discoid lupus was diagnosed after biopsy 10 years prior and had been managed with as needed topical steroid. She had no history of other lupus related symptoms including hair loss, oral ulcers, joint pain, pleurisy or nephritis. She had never required systemic treatment. She had no other significant past medical or surgical history. Her medications included triamcinolone ointment as needed, multivitamin and probiotics. She did not smoke or drink alcohol and had no family history of autoimmune disease.

Her initial Ophthalmology exam was normal, however, electroretinography testing showed moderate to severely reduced rod responses and moderate reduced cone responses in both eyes, the right eye worse than the left. Lab testing indicated a positive ANA of titer 1:80 homogenous with subsequent negative antibodies for double stranded DNA, smith, RNP, cardiolipin, Ro and La. Anti-retinal antibodies though were positive. To rule out a paraneoplastic process, malignancy testing and was normal, including PAP smear, mammogram, colonoscopy and CT scans of the chest, abdomen and pelvis.

A diagnosis of non-paraneoplastic autoimmune retinopathy was made. She was started on prednisone 1mg/kg daily as well as mycophenolate 1000mg twice daily as a steroid sparing agent. Her prednisone was tapered off over 3 months and she was maintained on mycophenolate alone. Her vision stabilized on this regimen.

Discussion

Autoimmune retinopathy encompasses a spectrum of rare autoimmune diseases that primarily affect retinal photoreceptor function.[1] It is characterized by progressive unexplained vision loss, visual field deficits, photoreceptor dysfunction and the presence of circulating anti-retinal antibodies.[2] Autoimmune retinopathy can be divided in 2 groups: paraneoplastic and non-paraneoplastic. The paraneoplastic group is further subdivided into cancer-associated retinopathy and melanoma-associated retinopathy.[3] Therefore, all patients with suspected autoimmune retinopathy without a past medical history of cancer need to be thoroughly investigated for an occult malignancy before a diagnosis of non-paraneoplastic autoimmune retinopathy can be made.[1]

Although it is believed to be rare, the prevalence of autoimmune retinopathy is currently unknown. Onset is typically in the fifth to sixth decade of life with a female predominance. A history of autoimmune disease is common. Symptoms present as painless subacute vision loss, scotoma, photopsia, nyctalopia and dyschromatopsia. The disease is usually bilateral but can be asymmetric.[3] On clinical examination, the fundus is typically normal, especially initially, but abnormalities can be detected later on in the disease. Usually there are minimal or no signs of intraocular inflammation.[4] Electrophysiologic tests are a valuable diagnostic aid in autoimmune retinopathy, allowing objective measurement of the electrical activity of the eye. Specifically, the electroretinogram provides information about the rod and cone systems,[1] the main target in autoimmune retinopathy.

The presence of circulating anti-retinal antibodies is considered important in the diagnosis of autoimmune retinopathy. However, autoantibodies can be seen in both healthy and diseased patients. Furthermore, antiretinal antibodies have been described in a variety of systemic autoimmune diseases such as Behcets, inflammatory bowel disease, systemic lupus, and multiple sclerosis as well as degenerative ocular diseases such as age-related macular degeneration and both infectious and non-infectious uveitis.[4] Therefore, the clinical context of the lab result is crucial to an accurate diagnosis and the presence of the antibody alone is not sufficient enough to make the diagnosis.

Utility of anti-retinal antibodies is also limited by absence of standardization for the detection and measurement of anti-retinal antibodies. Multiple laboratory techniques can be used to detect antibodies including western blot, immune-histochemistry and ELISA. Many variables in each technique limit reliability across laboratories as well as varying clinical ranges of anti-retinal antibodies which may also create conflicting diagnoses.[2]

The diagnosis of non-paraneoplastic autoimmune retinopathy was made in this patient due to her clinical presentation of subacute vision loss, night blindness, abnormal electro-retinogram and presence of anti-retinal antibodies in the setting of a negative malignancy work-up.

There are no established treatment protocol for autoimmune retinopathy. There is general agreement to treat underlying malignancies if present.[3] For non-paraneoplastic autoimmune

retinopathy, treatment strategies have focused on immunomodulatory therapy[4] to modulate the immune system and reduce the autoimmune attack on the retina before irreversible damage occurs.[1] Various approaches including, corticosteroids, intravenous immunoglobulin, plasmapheresis, mycophenolate mofetil, azathioprine, cyclosporine and rituximab have shown varying improvement or stability of the ocular disease. However, there are also reports of patients that remain stable in the absence of treatment, as well as progressive disease despite immunomodulatory therapy.[3] Thus it is difficult to assess the most appropriate long term treatment strategy. This patient responded to mycophenolate therapy with stabilization of her retinopathy.

In conclusion, this case highlights clinical aspects of autoimmune retinopathy and the potential challenges faced with diagnosis and treatment. Malignancy should be ruled out as paraneoplastic forms exist. It is important to be aware of this condition so that it can be recognized more readily and treated prior to the occurrence of retinal damage and irreversible vision loss.

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