and he desired to have further testing and evaluation for macular degeneration. The patient denied visual loss or metamorphopsia. His medical history was unremarkable, and he denied having a family history of ocular disease.

The best corrected visual acuity was 20/30+1 in the right eye and 20/20 in the left eye. There was no afferent pupillary defect. Confrontations were full to finger counting in each eye. Color vision was tested with Ishihara plates and was normal for each eye. The anterior segment exam was unremarkable for both eyes. Intraocular pressure measured by Goldmann applanation tonometry was 15mmHg in the right eye and 16mmHg for the left eye.

The dilated ophthalmoscopic examination revealed a one-half disc diameter yellow disciform macular lesion in the right eye (Figures 1 and 2) with macular retinal pigment epithelial clumping in the left eye (Figures 3 and 4).

**Abstract**
Adult-onset vitelliform macular dystrophy is a bilateral dystrophy characterized by round subretinal yellow macular lesions. The dystrophy manifests between the third and fifth decades of life and has a variable genetic inheritance. A case of adult-onset vitelliform macular dystrophy is presented with the use of electrophysiological testing, fluorescein angiography, fundus autofluorescence and optical coherence tomography.

Key Words: Vitelliform macular dystrophy, electroretinogram

**Introduction**
Adult-onset vitelliform macular dystrophy (AVMD) manifests between the third and fifth decades of life and it occurs in approximately 1 in 8,000 individuals\(^1\). The dystrophy is characterized by bilateral round subretinal yellow macular lesions that can be mildly elevated and are one third to one disc diameter in size\(^2,3\). A significant amount of lipofuscin in the macular retinal pigment epithelium with loss of the retinal pigment epithelium and photoreceptor cell layers have been found in studies of AVMD patients\(^4\). Patients with AVMD may have normal or subnormal electro-oculograms and they may present with mild metamorphopsia, mild decrease in vision or they may be asymptomatic\(^2,4\). The long-term visual prognosis varies among individuals. For most adults, the clinical course is benign and progresses slowly, but for some individuals, especially older patients, central vision can be reduced due to macular atrophy and the development of a choroidal neovascular membrane\(^5\). Other possible complications of AVMD include full-thickness macular holes and retinal detachments\(^6\).

**Case Report**
A 37-year-old white male presented for a new comprehensive eye examination. The patient stated that he was diagnosed with “macular degeneration” in both eyes two months prior...
The patient did not return for five months. Although he denied visual loss or metamorphopsia, his best corrected visual acuity decreased to 20/40-, not improving with pinhole or refraction in the right eye and his visual acuity remained stable at 20/20 in the left eye. The dilated fundus exam findings appeared stable in both eyes, however, the macular OCT revealed mild progression of the retinal pigment epithelial detachments in both eyes.

An electro-oculogram (EOG) and a multifocal electroretinogram (ERG) were ordered due to suspicion for vitelliform macular dystrophy from the yellow disciform macular lesion in the right eye. The EOG was normal for both eyes, however, the multifocal ERG revealed decreased retinal function in the right eye with suspicious findings in the left eye. Latency was increased and amplitudes were diminished within the central 5-7 degrees for the right eye. The left eye was suspicious for underlying pathology due to an attenuated response within 5 degrees of the center. There were no signs of changes in amplitude or implicit times in the central 5-7 degrees for the left eye (Figures 11 and 12).

Macular optical coherence tomography analysis revealed paracentral thickening in the left eye greater than the right eye and a retinal pigment epithelial detachment in the right eye greater than the left eye (Figures 5 and 6). Fundus autofluorescence analysis revealed a central dark lesion bordered by hyperautofluorescence in the right eye and a central dark lesion with adjacent hyperautofluorescence in the left eye (Figures 7 and 8). Fluorescein angiography revealed central hypofluorescence right eye and isolated areas of hyperfluorescence within the macular region in the left eye greater than the right eye (Figures 9 and 10). HVF 10-2 testing revealed a relative central scotoma right eye greater than the left eye. The patient reported mild metamorphopsia right eye on Amsler grid evaluation and he denied having a scotoma or metamorphopsia left eye.

The impression after the initial examination was unspecified macular degeneration in the right eye greater than the left eye with differential diagnoses of vitelliform macular dystrophy, chronic idiopathic central serous retinopathy, pattern dystrophy and Stargardt disease. The patient was dispensed a home Amsler grid and was educated to return in three months or sooner if he noticed visual loss or metamorphopsia.
The patient was diagnosed with adult-onset vitelliform macular dystrophy right eye greater than the left eye due to the retinal appearance, age of onset, the electrophysiologic results and the analysis of the macular OCT, fundus autofluorescence and fluorescein angiography. The patient was educated and counseled about the inheritance pattern and nature of adult-onset vitelliform macular dystrophy. He was advised to monitor his vision with an Amsler grid and return immediately if he noticed metamorphopsia or loss of vision. The patient was also advised to return to the retinal specialist in six weeks for a follow-up examination due to the mild progression of the pigment epithelial detachments in both eyes. The patient did not return for his scheduled appointment.

Discussion
Adult-onset vitelliform macular dystrophy is thought to be an inherited disorder, however, the precise function of genetics is not completely established. Some studies propose an autosomal dominant inheritance with variable penetrance, yet, AVMD can occur without evidence of a familial inheritance pattern. Other investigators believe that AVMD may be degenerative with a genetic predisposition. In addition, mutations in the genes encoding for VMD2 and RDS/peripherin have been found in a small population of patients who are diagnosed with AVMD. Other studies found five missense mutations associated with AVMD including A1α243Val and Thr6Pro and a minority of patients with AVMD have been reported to have mutations in the PRPH2, Best1, IMPG1 or IMPG2 genes.

Electroretinogram (ERG)
The ERG is an electrical response of the retina that is evoked by a flash of light or a bright pattern. The photic stimulation elicits a negative a-wave that is derived from the rods and cones and a positive b-wave that is derived from the midretina. A full-field ERG is performed in order to determine the summed retinal response. Multifocal ERGs can isolate focal retinal dysfunction in the central 40 degrees of visual field. The multifocal ERG of adult-onset vitelliform macular dystrophy, amplitudes rather than implicit times are affected. The amplitudes were diminished within the central 5-7 degrees for the right eye in this case study.

Electrooculogram (EOG)
The EOG measures the standing potential between the retina and electropositive cornea during scotopic and photic adapted states. The voltage lowers in the dark, known as the dark trough, and the potential rises in the light, known as the light peak. The electrooculographic potentials are derived from the retinal pigment epithelium interacting with the midretina. In order for the potential to increase in the light, both the retinal pigment epithelium and midretina must be functioning normally. For adult-onset vitelliform macular dystrophy, the EOG can be normal or mildly subnormal. The EOG was normal in this case study.

Fluorescein Angiography
In cases of adult onset-vitelliform macular dystrophy, hypofluorescence in often seen in the location of the vitelliform lesion on fluorescein angiography. A ring of hyperfluorescence that surrounds the vitelliform lesion is also seen on fluorescein angiography in cases of AVMD. In this case study, fluorescein angiography revealed central hypofluorescence right eye and isolated areas of hyperfluorescence within the macular region in the left eye greater than the right eye.
**Fundus Autofluorescence**

Fundus autofluorescence is a non-invasive retinal imaging technique used to evaluate irregularities of the retinal pigment epithelium, photoreceptors and macular pigment. Fundus autofluorescence allows visualization of fluorophores from excessive accumulation of lipofuscin in retinal pigment epithelial cells to aid in evaluation of macular dystrophies and hereditary retinal diseases. Vitelliform lesions on fundus autofluorescence appear as circumscribed hyperautofluorescence in the macula and as a dark lesion surrounded by hyperautofluorescence in the vitelliruptive stage. In this case study, fundus autofluorescence analysis revealed a central dark lesion bordered by condensations of hyperautofluorescence in the right eye and a central dark lesion with adjacent hyperautofluorescence in the left eye.

**Optical Coherence Tomography (OCT)**

OCT is a non-invasive diagnostic device that provides information of the morphological characteristics of adult-onset vitelliform macular dystrophy. In cases of AVMD, the vitelliform lesion is located in the retinal pigment epithelium or between the retinal pigment epithelium and photoreceptor layer. In this case study, the macular optical coherence tomography analysis revealed paracentral thickening in the left eye greater than the right eye and a retinal pigment epithelial detachment in the right eye greater than the left eye.

**Differential Diagnoses**

Differential diagnoses for this case study of adult-onset vitelliform macular dystrophy include chronic idiopathic central serous retinopathy, pattern dystrophy and Stargardt disease. Chronic idiopathic central serous retinopathy typically affects males in their third to fifth decades of life. It is characterized by an idiopathic detachment of the neurosensory retina in the macula due to retinal pigment epithelial dysfunction. Optical coherence tomography confirms the presence of serous neurosensory detachments and pigment epithelial detachments. Fluorescein angiography demonstrates focal leakage at the level of the RPE and fundus autofluorescence reveals increased autofluorescence at the leakage site. The electro-oculogram is typically normal, while the entire posterior pole is depressed in multifocal ERGs of patients with central serous retinopathy.

Pattern dystrophy is a bilateral macular dystrophy with an autosomal dominant inherited disorder that often presents in the fourth to fifth decades of life. Multiple fundus presentations including butterfly-shaped pigment dystrophy and reticular dystrophy have been documented and the patterns are dependent on the pigment deposition at the level of the retinal pigment epithelium. Fundus autofluorescence often reveals a speckled hyperautofluorescent pattern due to pigment deposits in the RPE and optical coherence tomography results depict hyper-reflectivity at the level of the RPE in patients with pattern dystrophy. Fluorescein angiography reveals early hyperfluorescence that delineates hypofluorescent lesions and electroretinograms are generally normal or mildly subnormal in pattern dystrophy.

Stargardt disease is an inherited bilateral macular dystrophy that typically presents in childhood or adolescence. Stargardt disease is characterized by diffuse deposits of lipofuscin that appear as retinal flecks in the retinal pigment epithelium. The retinal flecks are often concentrated around the macula with some flecks in the midperipheral retina. New flecks appear hyperautofluorescent on fundus autofluorescence while older flecks become hypautofluorescent and some flecks may be surrounded by a ring of decreased autofluorescence. Optical coherence tomography analyzes the amount of photoreceptor layer loss and RPE atrophy by displaying hyper-reflectivity at the base of the foveal outer nuclear layer in Stargardt disease. Fluorescein angiography may reveal a dark choroid and electroretinogram patterns are abnormal in patients with Stargardt disease.

**Treatment and Management**

There is currently no cure available to prevent the progression of adult-onset vitelliform macular dystrophy. For most cases of AVMD, the clinical course is benign and progresses slowly, but for some patients, choroidal neovascularization can develop and threaten central vision. Treatment with anti-vascular endothelial growth factor therapy, including ranibizumab, is beneficial if a choroidal neovascularization is present. Low vision consultation and rehabilitation is warranted for advanced cases of AVMD with significantly impaired vision. Furthermore, dilated comprehensive eye examinations should be performed every 6 to 12 months in order to monitor for possible complications of AVMD, including choroidal neovascularization, full-thickness macular holes and retinal detachments. Family members of individuals with AVMD should also have dilated comprehensive eye examinations in order to detect early signs of this rare condition.

**Conclusion**

This case demonstrates the role of clinical observation, electrophysiology studies, fluorescein angiography, fundus autofluorescence and ocular coherence tomography in diagnosing adult-onset vitelliform macular dystrophy. The information derived from these diagnostic modalities is essential to detect abnormalities in retinal function and to exclude other differentials with irregularities in the retinal pigment epithelium. It is important for clinicians to educate the patient about the natural course of this macular dystrophy and to counsel the patient and family members of possible visual and genetic outcomes. Although the prognosis for adult-onset vitelliform macular dystrophy is generally good, patients should continue to self-monitor their vision, as sight-threatening complications may develop.
References


