Mystery Retina 2017
Interactive Discussion of Challenging Cases

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Case BD
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A 48-year old female experienced acute complete visual loss OD while having lid filler injections in the glabellar region OU, along with botulinum injections around the temporal margins of her eyelids. She was NLP OD, and there was no visual recovery.

Diagnosis

(Retrograde) Ophthalmic artery occlusion/central retinal artery occlusion OD secondary to cosmetic lid filler injection

References


Case 2
William F Mieler, MD, Felix Chau, MD, Chicago, IL

This 33 year old Hispanic female was initially evaluated in 12/2004, when she experienced painless progressive loss of vision to 3/200 OD, and 1/200 OS. Systemically, the patient was in excellent health, and a systemic evaluation for underlying disease was negative. On clinical examination, optic disc swelling was noted along with retinal arteriolar aneurysms, and macular exudation. The VA gradually improved to 20/80 OD, while the left eye remained compromised at 1/200 OS. Intravitreal triamcinolone was administered OS in 03/2005 and 08/2005, along with peripheral scatter photocoagulation OU. In 2009, an inferotemporal BRAO developed. VA was 20/100 OD, and 1/200 OS. Cataract surgery was performed OU in 10/2009 and 11/2009. VA improved to 20/30 OD, and 20/200 OS. Finally, the patient developed an epiretinal membrane OD for which she underwent vitrectomy removal. Final VA was 20/40 OD, and 20/200 OS. With time, the retinal arteriolar aneurysms resolved, along with the macular exudates.

Diagnosis

Idiopathic retinal vasculitis, aneurysms, and neuroretinitis (IRVAN) syndrome OU, BRAO OS
References


Case 3
William F Mieler, MD, Chicago, IL

A 74 year old male was referred for evaluation of a pigmented choroidal lesion with an overlying white “fibrous cap”. The lesion was noted on a routine ocular examination, and the patient was visually asymptomatic. Color photos will be shown, along with a FA, and SD-OCT. The lesion has not changed in six months

Diagnosis

Turbid pigment epithelial detachment (PED) overlying a choroidal nevus versus presumed solitary circumscribed retinal astrocytic proliferation (SCRAP)

References


Case 4
Jennifer I Lim, MD, William F Mieler, MD, Chicago, IL

A 59-year-old male noted a two-week history of visual blurring OD, and three days of blurring OS. VA was 20/40 OD, and 20/25 OS. Systemically in good health. Color photographs, FA, FAF, red free, and SD-OCT images will be presented. Choroidal neovascularization eventually developed OS and was treated with bevacizumab.

Diagnosis

Persistent placoid maculopathy OU with CNV OS
References


Case 1

Carol L Shields, MD, Philadelphia, PA

A 2-month old girl was noted to have a white pupil at 6 weeks of age. She was referred for retinoblastoma management. On examination, the right eye showed posterior synechia temporally and funduscopically there was a fibrotic retinal detachment with neovascularization, confirmed on IVFA. Ultrasound revealed total retinal detachment with dense tissue and no calcification. The OS showed no visible abnormalities. There was no retinoblastoma.

Skin evaluation disclosed hyperpigmented streaking (lines of Blaschko), with occasional blister ulceration over the entire body, consistent with incontinentia pigmenti. The mother showed no cutaneous or dental abnormalities. Given the advanced stage of scarring in the right eye, observation was advised and warning regarding enucleation for pain was discussed.

Incontinentia pigmenti affects the skin, brain, teeth, and eyes. In the skin, there are 4 stages with erythema, vesicles, linear hyperpigmentation (lines of Blaschko), and pale scarring. The brain features include ischemic changes that can lead to seizures, developmental delay, mental retardation, stroke, coma, and death. The dental features include adontia, microdontia, and pegged teeth. The eye findings include retinal ischemia with traction detachment, retinal pigment epithelial hyper- or hypo pigmented spots, and foveal hypoplasia.

Diagnosis

Total ischemic retinal detachment in a child with incontinentia pigmenti

References


Case 2  
Carol L Shields, MD, Philadelphia, PA

A 92 year-old woman with blurred vision, initially treated as vitritis, was later found to have biopsy-proven bilateral posterior vitreoretinal lymphoma (PVRL). She had severe vitreous tumor infiltration in the OD and mild infiltration OS with sub-RPE tumor OS. Visual acuity was 20/30 OD and 20/40 OS.

Given her age and difficulty in keeping appointments, she wanted a “quick fix” for this problem. Knowing that methotrexate injections might take approximately 25 injections, she preferred fewer trips. We decided to treat with intravitreal Melphalan, as we were comfortable with its use for retinoblastoma. She was treated with low dose intravitreal melphalan (10 ug/0.1 ml) OU. Following initial injection, complete clearance of vitreous tumor OU was noted within 3 weeks. Over 16 month follow up, the OD required 5 injections off and on for minor seed recurrence and OS remained stable without recurrence and without need for further injections. There were no toxicities OU, and final visual acuity was 20/50 OU.

Diagnosis

Vitreoretinal lymphoma treated with intravitreal melphalan

The right eye had advanced PVRL (left photo) treated with intravitreal melphalan and showed rapid response in 3 weeks (right photo) with complete clearing of cells.

References


Case 3
Carol L Shields, MD, Philadelphia, PA

A 67 year-old university vice president was urgently referred for mild loss of vision OS from an intraocular mass. On examination, the OD was normal. The affected OS demonstrated mild conjunctival injection with shallow anterior chamber angle. The left fundus revealed shallow retinal detachment and underlying brown elevated mass. Ultrasound showed uveal effusion with a single area of a solid appearing mass. Red-free wide-angle imaging confirmed choroidal folds for 360 degrees, suggestive of effusion. To be certain that there was no intraocular tumor, MRI scan was obtained and showed 360 degrees effusion with no solid enhancing mass.

Options were offered including oral, pericocular, or intravitreal corticosteroids vs scleral windows. Oral prednisone was given (80 mg/day with slow taper over 6 weeks) and the effusion completely settled. We have found oral or pericocular steroids to be successful for management of uveal effusion and the need for scleral windows is rare.

Diagnosis

Uveal effusion syndrome responsive to oral corticosteroids

References


A 10 year-old girl had blurred vision OD for 1 year, with cause unknown. Visual acuity was 20/100 OD and 20/20 OS. The left eye was normal.

Evaluation of the right eye showed normal fundus confirmed on fundus photography and fluorescein angiography. Autofluorescence demonstrated a slightly mottled fovea and red-free showed the mottling more prominently. OCT held the answer as the fovea showed a multicavitary “bee hive” appearance of a cavitory tumor, located primarily in the nerve fiber layer and consistent with cavitory retinal astrocytic hamartoma. There was no history of tuberous sclerosis complex (TSC). Observation was advised.

Retinal astrocytic hamartoma develops in the nerve fiber layer in all cases and demonstrates “moth-eaten” spaces on OCT in 91% of cases. The spaces represent either calcification or cavitation. Veronese et al were the first to recognize the cavitory pattern on OCT. Evaluation for TSC is suggested and observation of the retinal tumor is advised.

**Diagnosis**

Cavitary retinal astrocytoma

**References**


Case 1
Jerry A Shields, MD, Philadelphia, PA

At age 9 years, a circumscribed choroidal mass lesion compatible with uveal melanoma was diagnosed, in a patient with a past history of a cervical Burkitts lymphoma. A FNAB of the choroidal mass at the time of plaque application revealed Burkitts lymphoma and not melanoma. This well circumscribed choroidal mass was believed by oncologists to be a non-Burkitts lymphoma that occurred secondary to the immunosuppression used to treat the cervical Burkitts lymphoma several years earlier. This is a well described, though relatively infrequent occurrence.

Diagnosis
Intraocular non-Burkitts lymphoma
Case 2
Jerry A Shields, MD, Philadelphia, PA

An 11 year-old girl presented with a ciliary body mass that occupied one third of the globe nasally. How was she treated and what was the eventual outcome?

She and her parents refused enucleation and she was therefore managed by partial lamellar sclerouvectomy (PLSU). The outcome was excellent and she has had almost 6/6 vision after 20 years with no cataract or other complications. Although this case was reported previously, it is now presented with long term follow up.

Diagnosis
Intraocular leiomyoma, s/p partial lamellar sclerouvectomy (PLSU)

Selected References
Intraocular leiomyomas. Observations on seven cases. Annual Byron Demorest Lecture, Davis, CA Arch Ophthalmol 112;54.521-8D


Case 3
Jerry A Shields, MD, Philadelphia, PA

A middle aged woman was seen for a dark ciliary body mass and she wanted no treatment. About 2 years later the lesion had enlarged and now covered more than one-half of the pupil and was causing vision loss. What is your diagnosis and how should she be managed?

This patient had a mid-zonal iris pigment epithelial cyst, which can sometimes resemble a ciliary body melanoma. Such cysts are be usually diagnosed with slit lamp biomicroscopy, and UBM and they usually remain stable. This case is unique in that it showed progressive enlargement and occluded the pupil. After successful aspiration the cyst collapsed and has not recurred after 3 years. Patients with such cysts are often referred to rule iris or ciliary body melanoma.

Diagnosis

References


Case 4
Jerry A Shields, MD, Philadelphia, PA

A 4-year-old child presents with a total retinal detachment and ultrasound shows a dome shaped mass with intense echoes compatible with calcium. Should treatment be modified because of presence of the presumed calcification. Are these clinical and ultrasound findings pathognomonic of retinoblastoma or are there other conditions can have similar findings?

Although these ultrasound findings are highly suggestive, and almost pathognomonic, of retinoblastoma, this child had tuberous sclerosis with astrocytic hamartomas with large calcospheres that occur in the brain tuberous sclerosis. When they occur in the retina in children with tuberous sclerosis the echoes may be identical to the echoes seen with retinoblastoma.

Diagnosis

Retinal astrocytoma in tuberous sclerosis complex (TSC)

References


Case 1  
David Sarraf, MD, Los Angeles, CA

A 71-year old male presented with sudden vision loss in the right eye. Retinal examination illustrated macular hemorrhage with a radial pattern in the right eye and retinal crystals in the temporal foveal region of the left eye. Spectral domain OCT displayed hyper-reflective lesions in the outer plexiform layer of Henle in the right eye and retinal cavitation in the left eye. A diagnosis of macular telangiectasia Type 2 was made complicated by macular hemorrhage in the deep retinal capillary plexus OD.

Diagnosis

Macular telangiectasis type 2 with intraretinal hemorrhage

Reference

A 72-year old female with Sjogren syndrome presented for hydroxychloroquine screening evaluation. Retinal examination was normal in each eye but spectral domain OCT illustrated subtle thinning and depression of the perifoveal ellipsoid zone in each eye. Formal 10-2 Humphrey visual field was normal but the patient was high risk for toxicity with a total cumulative dosage of 1952 grams. The patient had been overdosed at 400 mg/day (maximum safe dosage at 5.0 mg/kg/day). Subsequent multifocal ERG and microperimetry confirmed macular toxicity. SD OCT can detect early hydroxychloroquine maculopathy in association with normal 10-2 HVF.

**Diagnosis**

Early hydroxychloroquine maculopathy

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**References**


Case 3
David Sarraf, MD, Los Angeles, CA

A 44-year old male presented with chronic bilateral vision loss and a history of chronic central serous chorioretinopathy status post multiple laser and anti-VEGF procedures in each eye. Spectral domain OCT illustrated bilateral macular detachment and pachychoroid syndrome. Fundus autofluorescence demonstrated mixed autofluorescent lesions in the central macula OU. EOG confirmed the diagnosis of Best disease with absent light rise OU.

Diagnosis

Vitelliform dystrophy

Reference

A 68-year-old male presented with a two-week history of decreased vision in both eyes and photopsia in his right eye. His ocular history included neovascular AMD in his right eye. His medical history included multiple myeloma. Visual acuity was 20/100 in his right eye and 20/40 in his left eye. Dilated funduscopic examination showed multiple atrophic chorioretinal lesions. The lesions were in a linear distribution in several areas. Corresponding outer retinal thinning and RPE loss with central aggregates were noted on OCT. Fluorescein angiography showed peripheral areas of capillary non-perfusion with window defects consistent with chorioretinal scarring.

The patient had recently been hospitalized for confusion following a fall at home. His altered mental status was mistakenly thought to be delirium associated with a urinary tract infection. A post-admission review of his medical records, combined with the new retinal findings, led to a suspicion that West Nile virus had contributed to his delirium manifesting as an encephalopathy. This was confirmed on serology.

While West Nile virus usually presents as a mild febrile illness, it can also manifest as an encephalopathy.

**Diagnosis**

West Nile Viral Chorioretinopathy
References


Case 2  
K Bailey Freund, MD, New York, NY

73-year-old male with no significant past ocular history presented with a recent change in vision of his left eye. Visual acuity was 20/30 in his right eye and 20/50 in his left eye. Multimodal imaging showed subretinal fluid associated with type 1 (sub-RPE) neovascularization in his left eye. Increased choroidal thickness with dilated Haller layer vessels was present in both eyes. One year later, a new finding was noted in his left eye. This new finding remained unchanged over 4 additional years of follow-up.

Diagnosis

Late-onset acquired melanocytic nevus in a patient with pachychoroid neovasculopathy

References


Case 3
K Bailey Freund, MD, New York, NY

A 58-year-old male was referred for evaluation of bilateral pigmentary changes and a mild reduction in visual acuity of his left eye. There was a history of diabetes mellitus type 2. Clinical examination and multimodal imaging showed findings of pachychoroid disease in both eyes. *En face* OCT projections of the choroid demonstrated sharply delimited hyporeflective areas around large pachyvessels in areas where ICGA showed hyperpermiability. Cross-sectional OCT of these lesions showed posterior hypertransmission. We speculate that caverns are lipid-rich. Friedman lipid globules, with similar sizes and tissue locations in AMD and healthy subjects, are candidates for histologic correlates of caverns.

**Diagnosis**

Choroidal caverns (AKA Friedman lipid globules)

**References**


Case 1
Lee M Jampol, MD, Chicago, IL

- Clinical History
- 67 year old patient with decreased vision OS x 4 years, OD x 3 years
- PDT OD x 2
- Bevacizumab x 1
- Dorzolamide
- VA 20/70 OD, 20/25 OS

**Diagnosis**
Severe central serous chorioretinopathy (CSCR) without subretinal fluid

**References**
None
Case 2
Lee M Jampol, MD, Chicago, IL

- 60 year old patient with five week history of decreased VA OD
- Subretinal fluid OD > OS
  - Bevacizumab OD of no help
  - Systemic prednisone-possible response
- Examination
  - Giraffe skin appearance OD
  - Mild pigmentary changes OS
  - White debris under macula OD

**Diagnosis**
Bilateral Diffuse Uveal Melanocytic Proliferation (lymphoma)
References


Case 3  
Lee M Jampol, MD, Chicago, IL

- 40 year old female  
- Blurry VA over months to years, with strabismus surgery OD in the past, and subsequent amblyopia OD  
- Vision 20/100 OD, and 20/40 OS  
- No nystagmus, no apparent foveas, iris transillumination  
- Fair skin, dark blonde hair

Diagnosis

Questionable albinism

References


Case 4
Lee M Jampol, MD, Chicago, IL

- 34 year old African American female
- Vision loss OD, inflammation OS
- 2010 diagnoses with possible white dot syndrome
- Treated with corticosteroids and methotrexate
- 2016-Inflammation OD, treated once again with corticosteroids
- In 2001, the son had TB encephalitis
- Patient had INH x 6 months
- Quantiferon gold positive

Diagnosis

Tubercular Multifocal Serpiginoid Choroiditis

References

Patel SS, Saraiya NV, Tessler HH, Goldstein DA. Mycobacterial Ocular Inflammation: Delay in Diagnosis and Other Factors Impacting Morbidity. JAMA Ophthalmol 2013;131:752-758


Case 5
Lee M Jampol, MD, Chicago, IL

- 44 year old male patient with history of renal failure and hearing loss
- White dots on retina
- Thinning of temporal retina versus thickening nasal.

**Diagnosis**

Alport syndrome

References

