A Walk on the Sunny Side

The Differential Diagnosis of Vitelliform Lesions

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| D | Dystrophy Degeneration |
| I | Infectious Inflammatory Immunologic Iatrogenic Idiopathic |
| T | Tumor Trauma Toxicity |
| H | Hereditary Hematologic |
| C | Congenital |
| O | Other |

Dystrophies?
**Best Vitelliform Macular Dystrophy**

- Autosomal dominant disorder with variable penetrance and expressivity
- Yellowish material attributed to lipofuscin in the RPE and/or subretinal space
- Mutation in the VMD2 gene coding for bestrophin
  - Calcium sensitive chloride channel protein located on the basolateral membrane of RPE cells
- Stages: Pre-vitelliform, vitelliform, scrambled egg, pseudohypopyon, atrophic
- Abnormal EOG universally present

**Adult-onset Vitelliform Macular Dystrophy**

**Terminology**
- Adult-onset foveomacular dystrophy
- Adult-onset foveal pigment epithelial dystrophy
- Pattern dystrophy of the RPE
- Adult Best disease
- Group of disorders which may be inherited AD
- Incomplete penetrance, variable expression
Adult-onset Vitelliform Macular Dystrophy

Mutation in the RDS gene (peripherin 2, PRPH2)
Third to fifth decade
Yellow subretinal deposits exhibiting fundus hyperautofluorescence
Exudative lesion may appear similar to Best vitelliform dystrophy
Fluorescein Angiography
   Vitelliform detachment
   Block early, stain late

Degenerations?
**Cuticular Drusen**

- **Appear as numerous, small, yellow semi-translucent macular RPE lesions**
- **Present between 40-60 years of age**
- **Histology**
  - Nodular excrescences of a thickened inner aspect of Bruch’s membrane, under the RPE

**Fluorescein angiography**
- Milky-way pattern of multiple hyperfluorescent dots against a dark background

**Autofluorescence**
- Innumerable hypoautofluorescent dots
- May develop large vitelliform macular detachment

**Reticular pseudodrusen**

- **Interlacing pattern of yellowish material not well seen on angiography**
- **First appear in superior temporal macula, progress to other quadrants**
- **OCT shows “saw tooth” patterns on anterior surface of RPE, in the subretinal space**
- **Amorphous debris is seen between the IS-OS junction of the photoreceptors and the RPE appears as a vitelliform detachment**

**Vitelliform macular detachment**

- The result of numerous abnormalities
  - Best vitelliform macular dystrophy
  - Adult onset vitelliform dystrophy
  - Cuticular drusen
  - Reticular pseudodrusen

- **When an acquired vitelliform-like detachment occurs in the macula it is referred to as a vitelliform macular detachment**

- **Material composed largely of degenerated photoreceptor outer segments found between the RPE and the IS/OS junction**
Vitelliform macular detachment

Angiography
Blocks early, stains late
The dark hypofluorescence on angiography indicates lipofuscin

Autofluorescence
Intense hyperautofluorescence which helps differentiate the lesion from CNV
The hyperautofluorescence also suggests lipofuscin

A vitelliform detachment is avascular without CNV
Vitelliform detachments may result in RPE atrophy
Vitelliform detachment with choroidal neovascularization

Infections?
Syphilitic retinitis

Uveitis
  Granulomatous: 50%
Vitritis
Retinitis
Vasculitis
Papillitis
Chorioretinitis

Blastomycosis

Chronic granulomatous fungal infection common in Mississippi river valley
Pulmonary and cutaneous manifestation
A yellow granulomatous lesion with overlying detachment is typical presentation

Cysticercosis

Larval stage of pork tapeworm
  Taenia solium
Ingestion of ova
  Contaminated food → ingestion of infected feces → the larva penetrate intestinal wall, travel by lymphatics and vascular system
Cysticercosis can be seen in vitreous body or subretinal space

Acute Idiopathic Maculopathy

Rare disorder affecting healthy young adults
Usually present with sudden central visual loss in one eye
May follow viral prodrome, presumed to be coxsackievirus
Neurosensory detachment overy RPE lesion, intraretinal hemorrhage, mild disc swelling
Spontaneous recovery over weeks to near normal vision, bulls-eye RPE change
Inflammations?

Sarcoid

Chronic idiopathic multi-system inflammatory disorder characterized by non-caseating granulomas
Posterior segment ocular findings include vitritis, focal or diffuse vasculitis, vascular occlusion, choroidal or optic nerve head granulomas

Idiopathic Helioid Choroidopathy

Idiopathic uveal scleral granuloma
Idiopathic solitary choroiditis
Yellow mass with overlying retinal vascular changes and detachment
Responds to steroids
May be associated with CNV
Late phase has bright yellow helioid appearance

Immunologic?
Melanoma Associated Retinopathy

Multiple serous detachments of retina
Multiple pale to yellow-orange, round or ovoid circular lesions
Autofluorescence shows hypofluorescence at first then hyperfluorescence as the lesions mature
EOG is normal
Slow and incomplete resolution of the vitelliform lesions occur
Possibly a paraneoplastic syndrome for cutaneous and ocular melanoma

Acute exudative polymorphous vitelliform maculopathy

Iatrogenic?

Retrobulbar Injection Optic Nerve Sheath Injection of Anesthetic
Idiopathic?

Central Serous Retinopathy

Idiopathic disorder involving leaks through RPE into subretinal space
Often small pigment epithelial detachments
Usually males 30 to 50, unilaterally
Natural course, spontaneous resolution after 3-4 months
Occasionally, chronic, recurrent, persistent detachment with RPE loss
Fibrin deposition, gutters, bullous detachment
Tumors?

Lymphoma
Masquerades as chronic posterior uveitis
Complaints of floaters common
Multiple fundus lesions
Solitary, yellow sub RPE tumors
Extends into choroid, retina
Reticular pattern of flecks

Trauma?

Solar Retinopathy
Photochemical reactions from light dependent on duration, intensity and spectral content
Lesions are usually bilateral and asymmetric with more severe disease in dominant eye
Vitelliform appearance only seen in acute stage
**Choroidal Rupture**

Blunt trauma causes uveal and RPE breaks
Usually concentric to optic nerve
Bleeding common, secondary choroidal neovascularization
Dehemoglobinization of blood causes yellow, white lesion
Valsalva, RAM, Tersons

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**Deferoxamine**

Patients with transfusion dependent anemias may develop hemosiderosis
Deferoxamine is an iron chelator
Ocular findings
- Cataract
- Optic neuropathy
- RPE toxicity
  - Vitelliform appearance
  - Macular RPE clumping

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**Toxicity**

- Presentation
  - Decreased vision
  - Nyctalopia
  - Visual field loss

- Angiography
  - Early blocked fluorescence and late hyperfluorescence
  - Late hyperfluorescence is a sign of ongoing toxicity
  - These changes precede RPE mottling

- Vision improves with cessation
Hematologic?

**Thrombotic Thrombocytopenic Purpura**

Dessemiated intravascular coagulopathy (DIC) is an excessive formation of fibrin clots within small vessels

TTP is related to DIC

Basic lesion is a subendothelial thrombus in the wake of an acute infection

Causes occlusive disease in the choriocapillaris and adjacent vessels by fibrin-platelet clots

Results in bilateral serous detachment

Congenital?

**Torpedo Maculopathy**

Zonal area of congenital amelanotic retinal pigment epithelium

These lesions have a halo of fundus hyperautofluorescence surrounding the lesion, which is hypoautofluorescent
Differential Diagnosis
Vitelliform Lesions

Dystrophy
- Best vitelliform dystrophy
- Adult onset vitelliform dystrophy

Degenerations
- Pseudovitelliform detachment
- Cuticular drusen
- Reticular drusen
- Sclerochoroidal calcification
- Pseudoxanthoma elasticum

Infectious
- Syphilis
- Nocardia
- Fungal
- Cysticercosis
- Toxocara canis
- Coxiella burnetii (AIM)

Inflammatory
- Sarcoid
- APMPPE
- Relentless placoid (Ampiginous)
- Heliod choroidopathy

Immunologic
- Paraneoplastic
- MAR
- Acute exudative polymorphous vitelliform
- Laryngitis
- Optic nerve injection
- Idiopathic
- Central Serous Retinopathy
- Tumor
- Lymphoma
- Leukemia
- Metastatic carcinoma
- Astrocytic hamartoma
- Retinoblastoma
- Benign reactive lymphoid hyperplasia
- Combined retina-RPE hamartoma
- Toxicity
- Deferinaxine
- Solar retinopathy
- Trauma
- Choroidal rupture
- Valsalva
- Hematologic
- Thrombotic thrombocytopenic purpura
- Congenital
- Torpedo maculopathy

Diagnosis?

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