Retinal Manifestations of Systemic Disease – Part 2
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The Retina and Systemic diseases
- Retinitis/Vasculitis
- Vitreous cells
- Serous detachments
- Choroidal lesions
- Pigmentary retinopathies
- Choroidal folds/Choroidal masses
- Retinal Vascular abnormalities

Pigmentary retinopathies with systemic associations

40 yo with unilateral VA loss. Va = 20/400

Psudoretinitis pigmentosa
- Syphillis
- Other inflammatory disease – Lyme, AZOOR, DUSN
- Phentothiazine toxicity
- Congenital rubella
- Autoimmune / Cancer associated retinopathy
- Vitamin A deficiency
- Resolved exudative detachment – VKH, toxemia of pregnancy
- Retained foreign body after penetrating trauma
Diffuse Unilateral Subacute Neuroretinitis (DUSN)

- Unilateral progressive pigmentary retinopathy, vascular attenuation, optic atrophy, vitritis
- Baylisascaris procyonis (raccoon parasite), Ancylostoma caninum (dog hookworm)
- Fecal-oral route

Rubella

- Can be unilateral or bilateral after congenital or acquired infection. “Salt and Pepper” appearance. Vision and ERG usually normal. Vessels usually normal. Cataract may be present.

Measles

- Typically bilateral, post-infectious. “Salt and Pepper” appearance. ERG/VF diminished, but may recover.

Vitamin A Deficiency

- Night blindness, severe dry eye. Common in children in developing world. Can occur after bariatric surgery, with malabsorption, or liver disease. ERG/VF changes. Pigmentary retinopathy. Treatment: Vitamin A

Acute Zonal Occult Outer Retinopathy (AZOOR)

- Late stage
Acute Zonal Occult Outer Retinopathy (AZOOR)

Typically young females in mid 30’s. Present with photopsias, scotomas, VF loss with ERG changes, minimal fundus changes with delayed development of zones of atrophic RPE. Start unilateral, but often become bilateral. Poor prognosis.

Autoimmune Retinopathy – high degree of suspicion

HVF / multifocal ERG

Gardner’s syndrome

Systemic Diseases associated with Retinitis Pigmentosa

Consider in young patients with RP
- Refsum’s disease – recessive, associated with increased phytanic acid levels.
- Hereditary abetalipoproteinemia – recessive, RP with fat intolerance, deficiency of vitamins A, D, E, and K
- Kearns-Sayre syndrome - recessive, salt and pepper appearance, normal arterioles, CPEO, ptosis, heart block by age 15
Kearns–Sayre syndrome

Angioid Streaks
- Pseudoxanthoma Elasticum -60-90%
- Ehlers–Danlos syndrome
- Paget's disease
- Sickle cell disease
- Idiopathic

Pseudoxanthoma Elasticum
- Peau d'orange
- Main concerns: Cardiovascular disease from abnormal elastic tissue in blood vessel walls and GI bleeds

Angioid Streaks
- Hypercalcemia
- Marfan syndrome
- Hemochromatosis
- Acromegaly
- Sturge-Weber
- Myopia

Choroidal Folds
- Hypotony
- Choroidal Inflammation
- Posterior Scleritis
- Hyperopia
- Hyperthyroidism
- Hyperopia
- Postoperative
- Retinal detachment repair
- CNV/Disciform scar
- Choroidal mass
- Orbital mass
- Idiopathic
Posterior Scleritis

50% have associated systemic disease – collagen vascular disease, infectious disease (including Lyme, TB, parasitic).

Posterior Scleritis

Present with pain, tenderness, vision loss. Choroidal folds/serous RD. Papilledema may be present. Posterior scleral thickening on B scan. Angle closure can occur from choroidal thickening.

Hypotony Maculopathy associated with Carotid insufficiency

Hypotony Maculopathy may develop rebound glaucoma after reperfusion surgery.

Other Choroidal lesions and Choroidal masses
55 yo male with photopsias and distortion for 2 months

Choroidal mass lesion

Choroidal Mass - OCT

Choroidal Mass (EDI)

Choroidal Mass (Ultrasound)

Ocular Melanoma – worrisome findings on exam/testing
- Thickness - > 2 mm
- Fluid/ Fluorescein leakage
- Symptoms - Photopsias
- Orange Pigment
- Margins near the optic disc (within 3 mm)
- Acoustic hollowness (Low internal reflectivity)
- Absence of Halo/ Absence of Drusen
Uveal Melanoma

Ocular Melanosis

Congenital, may be associated with glaucoma and increased risk of melanoma

Astrocytic Hamartoma in Tuberous Sclerosis

Classic calcific “Mulberry lesion.” CNS astrocytoma, seizures, mental retardation, adenoma sebaceum, ash leaf spot, subungual fibroma, café au lait spots. Usually bilateral in TS.

Astrocytic Hamartoma

Non calcified appearance

Astrocytic Hamartoma

Can be isolated and idiopathic. Less common, but can be associated with Neurofibromatosis.

Astrocytic Hamarotoma

Isolated lesions possible, but need to rule out systemic TS and NF.
Retinal capillary hemangioblastoma in Von Hippel-Lindau Syndrome

Often earliest manifestation of VHL. Mean age is 25. 42% unilateral. Most that have multilocular bilateral, have VHL. Can also be isolated. Genetic testing is best way to screen for the condition.

Von Hippel Lindau disease

Feeder arteriole and draining venule. FA show rapid filling and leakage. VA loss from exudation, traction, VH, NVG. Small ones can be observed. Laser/Cryo/Radiation/surgical excision. +/- steroids/anti-VEGF. Prognosis depends on tumor size.

Wyburn-Mason Syndrome

Non-hereditary. Congenital Unilateral, can be bilateral. Syndrome involves retina and midbrain – seizures, hemiparesis. Vision can be normal to severe loss due to intraretinal heme and NVG. No clear Rx.

Wyburn-Mason Syndrome

Cavernous Hemangioma

Benign vascular hamartoma. Most are sporadic, they can be autosomal dominant and associated with brain and skin hemangiomas. Grape-like clusters with stagnant blood flow cause layering on FA. No leakage. Usually no Rx and asymptomatic, unless traction or VH develops in which case cryo/laser may help. May involve optic nerve or macula.

Systemic Histiocystosis

20/20 OD 20/400 OS
Sclerochoroidal Calcification

Most cases are idiopathic – should test patients for abnormal calcium-phosphorous metabolism. Check parathyroid hormone, vitamin D and calcium levels. Chronic Kidney disease can be associated.

B-Scan OD

Choroidal Osteoma

A benign osteochondroma where bone replaces the choroid. More commonly unilateral females in late teens or early twenties. Oval/round, peripapillary with well defined borders. Slow growing with CNVM in 30-40%.

BDUMP

A rare paraneoplastic condition with diffuse uveal melanocytic infiltration associated with systemic malignancy (ovarian, uterine, lung most common). Often associated with rapidly progressive cataracts. Cutaneous and mucosal involvement has been observed.

Cirumscribed Choroidal Hemangioma

Red-orange, round to oval shape in the posterior ⅓ of the fundus. Usually 1-3 mm thick and 3-7 mm in diameter. Can get fibrous changes on surface. Often get a serous detachment and cystic degeneration. PDT is quite effective. Radiation in severe cases. Can also combine anti-VEGF with PDT.

Choroidal Hemangioma

FA – very early hyperfluorescence of large choroidal vessels with late staining of entire lesion.
Choroidal Hemangioma (EDI)

Most are within 2DD of the nerve. Cystoid degeneration and SRF on OCT. B scan shows high internal reflectivity.

Circumscribed Choroidal Hemangioma

Exudation and serous RD cause vision loss. Diffuse choroidal hemangioma occurs in Stuge-Weber syndrome in which case glaucoma can result.

Choroidal Hemangioma

Would treat with PDT – reduced fluence.

Retinoblastoma

B cell lymphoma

Mass infiltration

Yellow spots

Metastasis (Breast)

Amelanotic, shallow, round-oval, usually posterior to the equator, may be multifocal
Metastasis

Usually bright, hyperfluorescent late, hyperechoic with high internal reflectivity.

Metastatic Breast Cancer

Lung Metastasis
Macroaneurysm with old subretinal hemorrhage

Will fluctuate with valsalva maneuver

Varix

Will fluctuate with valsalva maneuver

Cotton wool spot

50 yo with new blurry spot and no known medical problems

Cotton Wool spots

- Hypertension – usually DBP >110
- Diabetes – seen in 44% of cases of DR
- Retinal vein occlusion
- Inflammatory – GCA, Wegener’s granulomatosis, Polyarteritis nodosa, Systemic Lupus, Scleroderma
- Infectious – CMV, HIV retinopathy, Lyme, Toxoplasmosis, Mucormycosis, Leptospirosis
- Coagulopathies – Sickle cell disease, omocysteinemia, Lupus anticoagulant syndrome, Proteins C, S, antithrombin III deficiencies
- Emboli – Carotid and Cardiac disease
- Miscellaneous – Migraine, severe anemia, Leukemia/lymphoproliferative disorders, Interferon therapy, Radiation, Purtscher’s retinopathy, Papilledema
Hypertensive Crisis

Acute Leukocytic Leukemia

Multiple Myeloma

Purtscher’s Retinopathy

Radiation Retinopathy

Other Systemic Retinal Vascular Disease

Classic is after crush injury, long bone fractures. Associated with pancreatitis, amniotic fluid embolism, collagen vascular disease, TTP.
Ocular ischemic syndrome
Symptoms of vision loss, transient vision loss, pain. May be asymptomatic. Delayed choroidal filling and A/V transit. Unilateral cataract, iritis, low IOP initially, but may go into NVG. Asymmetric Diabetic Retinopathy.

Retinal Arterial Macroaneurysm
Elderly females with HTN. Treat those with leakage/exudation. Laser, but anti-VEGF seems to help regress as well. Those that bleed often fibrose.

Emboli
Recommend Carotid imaging and Transesophageal echo. Anticoagulation.

Susac’s Syndrome
Multiple mid-arteriolar occlusions, tinnitus, mental status changes. MRI. Immunosuppression.

Susac’s syndrome
Mid arteriolar obstruction

Lipemia Retinalis
Hyperglycemia with levels > 1000 mg/dL. Genetic hyperlipoproteinemia. Starts peripherally, usually affects posterior vessels at levels of 3500-5000.
Peripheral Retinal Neovascularization

- Diabetes
- Vein occlusion (branch)
- Sickle cell retinopathy (SC)
- Sarcoidosis
- Drug abuse embolization/ t alc retinopathy
- Chronic uveitis/pars planitis
- Vasculitis
- Leukemia/anemia
- Eales’ disease

PDR

PDR after West Nile Virus

Diabetic Retinopathy rapidly progresses after ocular inflammatory disease

Talc Retinopathy

SS Disease: Macular infarct

16 year old HM vision after Sickle crisis lead to NVG within 1 month
SC disease
Black sunburst lesions, salmon patches, peripheral ischemia, neovascularization. Hgb electrophoresis.

Retinal Vasculitis

Familial Exudative Vitreoretinopathy (FEVR)
OCT may show temporal thinning

Conclusions
- Many retinal - systemic associations
- Keep in mind patterns of retinal abnormalities that should raise clinical suspicion
- May be bilateral or unilateral
- Often no overt systemic symptoms at the time of ocular presentation. However, many times the symptoms are there if specifically looked for.
- The ophthalmologist can have a life-saving role

Thanks very much for attending!