

# **Retina II**

by

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**Part II Chapter 10 Retinal Degenerations Associated with Systemic Disease:**

**I. Disorders involving other organ systems:**

**A. Infantile-Onset to Early Childhood-Onset Syndromes**

1. Retinal dysfunction and low ERG
2. Differentiate from Leber congenital amaurosis
3. Neuronal ceroid lipofuscinoses (Batten disease)
4. Peroxisome disorders:
  - a. Refsum disease
  - b. Zellweger (cerebrohepatorenal) syndrome
  - c. Neonatal adrenoleukodystrophy
5. Leber's does not have seizures or deterioration in mental status

**B. Bardet-Biedl Complex of diseases**

1. Group of diseases with similar findings:
  - a. pigmentary retinopathy
  - b. obesity
  - c. polydactyly
  - d. hypogonadism
  - e. mental retardation
  - f. no bone spicules

**C. Hearing Loss and Pigmentary Retinopathy**

1. **Usher Syndrome**
  - a. Association of retinitis pigmentosa and congenital sensorineural hearing loss
  - b. 11 different genetic types
  - c. 10% of RP patients are profoundly deaf
  - d. Differentiate from Alport syndrome, Alström and Cockayne syndromes, dysplasia spondyloepiphysaria congenita, Hurler syndrome, and Refsum disease

**D. Neuromuscular Disorders**

1. Spinocerebellar degenerations: Friedreich's ataxia
2. Olivopontocerebellar atrophies
3. Charcot-Marie-Tooth disease
4. Myotonic dystrophy
5. Neuronal ceroid lipofuscinosis (Batten disease)
6. Progressive external ophthalmoplegia syndromes
7. Peroxisome disorders
8. Duchenne muscular dystrophy:

- a. No visual symptoms
- b. Characteristic ERG abnormality: normal A wave, reduced B wave

E. Renal Disorders

- 1. Familial juvenile nephronophthisis
  - a. autosomal recessive
  - b. juvenile-onset renal failure
  - c. Pigmentary retinal degeneration; may be sectorial
  - d. short stature
- 2. Bardet-Biedel: pyelonephritis and kidney damage
- 3. Alström disease
- 4. Alport disease
- 5. Type II membranoproliferative glomerulonephritis: drusenlike deposits

F. Gastrointestinal disease

- 1. Familial adenomatous polyposis (Gardner syndrome)
  - a. pigmented lesions similar to congenital hypertrophy of the RPE
  - b. smaller, more ovoid, more variegated in color, multiple and bilateral

G. Dermatologic diseases

- 1. Ichthyosis
  - a. Associated with Refsum disease and Sjögren-Larsson syndrome
- 2. Incontinentia pigmenti
  - a. Males do not survive
  - b. Females have dermatopathy
  - c. One third have pigmentary abnormalities in the retina
  - d. May develop retinal detachment
- 3. Pseudoxanthoma elasticum
  - a. angioid streaks
  - b. peau d'orange fundus

H. Paraneoplastic Retinopathy

- 1. Cancer-associated retinopathy (CAR)
  - a. Rapid visual field loss
  - b. Decreased ERG
  - c. Immunosuppressive therapy
- 2. Melanoma-associated retinopathy (MAR)
  - a. Visual loss and nightblindness
  - b. Abnormal ERG

**II. Metabolic Diseases:**

**A. Albinism:** a group of genetic disorders involving melanin pigmentation, characterized by congenital hypopigmentation of the hair, skin, and eyes.

- 1. General Ophthalmic Features:

- a. Hypopigmentation of fundus.
- b. Iris transillumination.
- c. High refractive errors, esp. high myopia.
- d. Strabismus: abnormal retinogeniculostriate projections (temporal nerve fibers decussate rather than project to ipsilateral geniculate body).
- e. Photophobia.

2. **True Albinism:** congenitally subnormal vision and nystagmus. **True albinism has foveal hypoplasia.**

3. **Oculocutaneous:** Eyes and skin effected.

- a. Decreased melanin in melanosome. Autosomal recessive. Tyrosinase positives have some pigment, whereas tyrosinase negatives have no pigment.
- b. Ocular findings include strabismus, subnormal acuity, abnormal VF with central scotomas, colobomas of iris, congenital pupillary membranes, partial aniridia, capsular cataract.
- c. Skin erythematous with increased frequency of cutaneous basal and squamous cell carcinoma.
- d. **Tyrosinase negatives:** milk-white hair; skin pink to red; eyes light grey to light blue, with no pigment flecks; prominent red reflex; severe photophobia and nystagmus; freckles very rare. Lowest vision. Negative hair bulb test.
- e. **Tyrosinase positives:** hair becomes yellow to brown with age; eyes blue to brown with flecks at iris border; fundi “blond”; photophobia and nystagmus present but less severe; freckles and pigmented nevi common. Vision 20/60- to 20/100. Hair bulb test positive.
- f. **Potentially Lethal Forms:**
  - i. **Chediak-Higashi Syndrome:** oculocutaneous, increased susceptibility to infection
  - ii. **Hermansky-Pudlak Syndrome:** oculocutaneous, platelet abnormality causes increased susceptibility to bruising and bleeding. More common in Puerto Rican population.
  - iii. Need hematology consultation.

g. Other types include Platinum, Yellow Mutant, Brown, Rufous.

4. **Ocular:** effecting only eyes.

- a. Decreased number of melanosomes, but each melanosome may be normally pigmented. X-linked recessive.
- b. Carrier females - spotty iris transillumination, peripheral RPE mosaicism, macromelanosomes on skin biopsy.

5. **Albinoidism:** normal or minimally reduced vision, no nystagmus. Autosomal dominant.

6. Normal Pigmentation: pigmented cells, either melanocytes (neural crest @ 20 weeks gestation), or pigment epithelial cells (optic vesicle @ 5 weeks)

a. Melanosomes: organelles in which melanin is synthesized. Synthesis requires the enzyme tyrosinase. At least 7 separate genes can cause reduction in melanin production.

l-tyrosine → l-DOPA → dopaquinone → pheomelanin (red/yellow)

↓  
eumelanin (brown/black)

b. Early completion of melanogenesis in RPE is probably essential for normal foveal development.

7. Clinical Management:

- a. Refraction to minimize useful vision.
- b. Filters and sunglasses.
- c. Skin protection and regular examinations for malignant and precancerous lesions.
- d. Early diagnosis of lethal forms.
- e. Strabismus: decrease in nystagmus may follow repair.
- f. Prenatal diagnosis: normal fetus has pigmented scalp hair follicles @4 weeks.
- g. Genetic counseling.

## **B. Central Nervous System Metabolic Abnormalities**

Several metabolic diseases are known to affect the CNS and retina.

1. Neuronal ceroid lipofuscinosis (Batten disease)
  - a. autosomal recessive
  - b. accumulation of lipopigments within lysosomes of neurons, causing cell dysfunction and death.
  - c. progressive dementia, seizures, visual loss, pigmentary retinopathy (infantile and juvenile forms)
  - d. Findings: optic atrophy, macular pigmentary changes with mottling of the fundus periphery, low or absent ERG
  - e. Later onset cases may have bull's-eye-maculopathy
  - f. adult forms of NCL do not have ocular manifestations

2. Abetalipoproteinemia and vitamin A deficiency
  - a. autosomal recessive
  - b. apolipoprotein B is not synthesized. Leads to fat malabsorption and deficiencies of fat-soluble vitamins
  - c. therapy with vitamin A/E
  - d. most common cause of vitamin A deficiency retinopathy is in bariatric surgery patients or patients with Crohn disease following surgery
  - e. symptom of night blindness
  
3. Peroxisomal disorders and Refsum disease
  - a. Mostly autosomal recessive diseases
  - b. Dysfunction or absence of peroxisomes or peroxisomal enzymes
  - c. Zellweger syndrome:
    - i. severe infantile-onset retinal degeneration
    - ii. hypotonia, psychomotor retardation, seizures, renal cysts, hepatic interstitial fibrosis, death in infancy
  - d. Neonatal adrenoleukodystrophy
    - i. present in infancy
    - ii. survive until age 7-10
  - e. Infantile Refsum disease
    - i. elevation of serum phytanic acid
  - f. Refsum disease
    - i. may not be a peroxisomal disorder
    - ii. may be diagnosed in adulthood
    - iii. pigmentary retinopathy, reduced ERG, cerebellar ataxia, polyneuropathy, anosmia, hearing loss, cardiomyopathy
    - iv. night blindness
    - v. elevated plasma levels of phytanic acid or reduced phytanic acid oxidase activity
  
4. Mucopolysaccharidoses
  - a. excessive quantities of incompletely metabolized acid mucopolysaccharides/complex lipids stored in lysosomes
  - b. autosomal recessive except for type II (Hunter) – x-linked
  - c. Retinal dystrophy caused by storage of heparan sulfate only:
    - i. MPS IH (Hurler syndrome) and MPS IS (Scheie syndrome): coarse facies, mental retardation, corneal clouding, retinal degeneration (may be subtle, abn. ERG)
    - ii. MPS II (Hunter syndrome): pigmentary retinopathy, no corneal clouding
    - iii. MPS III (Sanfilippo syndrome): severe pigmentary retinopathy, milder somatic stigmata

5. Other lysosomal metabolism disorders
  - a. Tay-Sachs disease (GM2 gangliosidosis type I)
    - i. deficiency of subunit A of hexosaminidase A
    - ii. glycolipid accumulation in brain and retina
    - iii. mental retardation and blindness
    - iv. death by age 2-5 years
    - v. cherry red spot caused by whitening of ganglion cells
  - b. cherry red spot can also be seen in Sandhoff disease (GM2 gangliosidosis type II) and generalized gangliosidosis (GM1 gangliosidosis type I)
  - c. Gaucher disease
    - i. glucosylceramide in liver, spleen, lymph nodes, skin and bone marrow
    - ii. may have cherry red spot
    - iii. whitish subretinal lesions in midperiphery
  - d. Niemann-Pick disease: absence of sphingomyelinase isoenzymes
    - i. Type B: mildest. Macular halo is diagnostic.
    - ii. Type A: cherry red spot in 50%.
  - e. Cherry red spot also found in:
    - i. mucopolipidosis I
    - ii. cherry-red spot – myoclonus syndrome
    - iii. Goldberg-Cotlier syndrome (GM1 gangliosidosis type IV) (also diffuse retinal degeneration)
  - f. Fabry disease
    - i. x-linked recessive
    - ii. mutations in the alpha-galactosidase A gene
    - iii. accumulation of ceramide trihexoside in kidneys, skin, gastrointestinal tract, CNS, heart, and RE system
    - iv. corneal verticillata, tortuous conjunctival and retinal vessels, lens changes
6. Amino Acid Disorders
  - a. Cystinosis: defect in transport out of lysosomes
    - i. accumulation of intralysosomal cystine
    - ii. all autosomal recessive
    - iii. 3 types, all develop corneal crystals
    - iii. nephropathic type: develops retinopathy; patchy depigmentation of RPE with pigment clumps; good vision
    - iv. late-onset type
    - vi. benign type
7. Mitochondrial Disorders
  - a. Chronic progressive external ophthalmoplegia
    - i. mitochondrial myopathy with "ragged-red" fibers on muscle biopsy

- ii. atypical RP
- iii. With cardiomyopathy, known as Kearns-Sayre syndrome
- iv. variable pigmentary retinopathy
- v. variable visual function and ERG
- b. NARP syndrome (neurogenic muscle weakness, ataxia, RP)
- c. MELAS (mitochondrial encephalomyopathy, lactic acidosis, stroke)

### III. Systemic Drug Toxicity:

**A. Chloroquine and hydroxychloroquine:** used in collagen-vascular diseases and malaria prophylaxis.

1. Toxicity related to duration of treatment, total dose, and patient age.
2. Blurred vision is major symptom. May have decreased color vision or dark-adaptation.
3. “Bull’s Eye maculopathy most common. Loss of foveal reflex with increased pigment in the macula.
4. **Chloroquine (Aralen):** Used in malarial prophylaxis. Ocular toxicity rare if total dose is less than 300 g. Daily dose of less than 250 mg. recommended.
5. **Hydroxychloroquine (Plaquenil)** more commonly used. Used to treat rheumatoid arthritis. Lower incidence of toxicity. 5-7 mg/kg/day is safe.
6. **No single test reliable in indicating early, reversible toxicity.**
  - a. Damage may progress despite stopping drug.
  - b. Baseline examination should include ophthalmoscopy, fundus photography, visual acuity, color vision, and Humphrey-type VF.
  - c. Follow-up examinations q. 6-12 mos.
  - d. Ophthalmoscopic changes may precede symptoms.
  - e. FA shows typically more than seen ophthalmoscopically.
  - f. EOG and ERG may be abnormal early.
7. **“Bull’s Eye” Maculopathy: Diff. Dx.:**
  - a. Chloroquine Retinopathy
  - b. Acquired Cone Dystrophy
  - c. Benign Annular Foveal Dystrophy
  - d. Stargardt’s Fundus Flavimaculatus
  - e. Senile Macular Degeneration

### B. Phenothiazines:

1. Bind to melanin granules. Concentrated in uvea and RPE.
2. **Chlorpromazine (Thorazine):** abnormal pigmentation of lids, conjunctiva, cornea, lens capsule and lens. Retinopathy rare, but reported.
3. **Thioridazine (Mellaril):** severe retinopathy.
  - a. Can develop rapidly.
  - b. Rare at doses of 800 mg/day or less.
  - c. Blurred vision presenting symptom.

- d. Pigment stippling in post. pole early, becomes widespread but patchy.
- e. Atrophy of RPE and choriocapillaris. Areas of hypo and hyperpigmentation.
- f. Vision may deteriorate despite stopping drug.

### C. Other Agents:

1. **Tamoxifen:** treatment of breast cancer
  - a. Retinopathy rare at normal dosage.
  - b. Crystalline retinopathy in very high doses.
  - c. Can have macular edema
  - d. Vision can be reduced.
2. **Canthaxanthine:** carotenoid, sun-tanning.
  - a. Asymptomatic
  - b. Crystalline maculopathy
  - c. Resolves when drug stopped.
3. **Methoxyfluorane (Penthrane):** general anesthetic
  - a. Broken down to oxalic acid + calcium, then to calcium oxalate salt.
  - b. Oxalate crystals accumulate in inner retina and RPE.
  - c. Also can be seen with ingestion of ethylene glycol
4. **Desferrioxamine**
  - a. used IV to treat transfusional hemosiderosis
  - b. rapid bilateral visual loss
  - c. nyctalopia
  - d. ring scotoma and widespread mottled pigmentary changes
  - e. reduced ERG
  - f. vision returns slowly
5. **Isotretinoin (Accutane):** acne treatment
  - a. Assoc. with poor night vision and decreased dark-adaptation
  - b. felt to be reversible
6. **Rifabutin**
  - a. used in HIV patients as prophylaxis against *Mycobacterium avium*
  - b. Severe uveitis with hypopyon
7. **Cardiac glycosides (Digitalis):**
  - a. Blurred vision
  - b. Defective color vision
  - c. Xanthopsia (“yellow vision”)
  - d. Pericentral scotomata
8. **Sildenafil (Viagra)**
  - a. Transient blue tinting of vision
  - b. Temporary abnormal ERG
  - c. May occur in 50% of patients taking more than 100 mg.
  - d. No permanent effects have been reported.

## Chapter 11. Peripheral Retinal Abnormalities:

**I. Retinal Breaks: full-thickness defect in neurosensory retina.**

- A. Horseshoe tear: flap of retina pulled anteriorly secondary to PVD.
- B. Operculated hole: piece of retina pulled completely free.
- C. Atrophic retinal hole
- D. Macular hole
- E. Dialysis: break along the ora serrata.
- F. Giant break: extending more than 90 degrees circumferentially
- G. Traumatic Retinal Breaks:
  - 1. Associated with closed or penetrating injuries
  - 2. **Blunt trauma:**
    - a. Direct contusion (coup) causes contusion necrosis
    - b. Contrecoup injuries caused by compression of globe and “shock-waves”
    - c. Stretch injuries to ocular coats: choroidal ruptures most common, with tearing of RPE and choriocapillaris
    - d. Stretch injuries to vitreous cause tears along anterior or posterior vitreous base.
    - e. Dialysis: circumferential linear break at the vitreous base
    - f. **Avulsion of vitreous base is pathognomonic of trauma**
    - g. Can also cause horseshoe tears or holes
    - h. **Traumatic Retinal Detachment:**
      - i. In young patients, typically not acute because vitreous detachment not present.
      - ii. If chronic, may show demarcation lines, thin retina, intraretinal cysts, subretinal precipitates, normal or elevated intraocular pressure

**II. Posterior Vitreous Detachment:**

- A. Syneresis of central vitreous
- B. Separation of posterior hyaloid from the retina
- C. Normally, firm attachment of vitreous at vitreous base, optic disc, macula, major vessels, lattice degeneration, scars.
- D. Factors which increase prevalence:
  - 1. axial length
  - 2. patient age
  - 3. aphakia
  - 4. inflammation
  - 5. trauma
- E. Symptoms may include flashes and/or floaters. Commonly asymptomatic.
- F. **If symptomatic, 15% have retinal tear.**
- G. If associated with vitreous hemorrhage, 70% have retinal tear.

### III. Lesions Predisposing to Retinal Detachment:

- A. **Lattice Degeneration:** full-thickness vitreoretinal abnormality
  - a. 6-10% of population (familial)
  - b. Bilateral in 30-50%
  - c. Histology: discontinuity of the internal limiting membrane of the retina, overlying pocket of liquefied vitreous, condensation and adherence of vitreous at margin of lesion, atrophy of inner retina.
  - d. Underlying cause of 20-30% of all retinal detachments
  - e. Will cause detachment in only about 1% of patients with lattice
  - f. Typical ophthalmoscopic findings: white lines of Vogt, vascular sheathing, RPE hyperplasia, atrophic holes, tears along margin (both can cause detachment)
  
- B. **Vitreoretinal Tufts:** small retinal elevations caused by focal vitreoretinal traction
  - 1. noncystic
  - 2. cystic
  - 3. zonular-traction tufts.
  - 4. cystic and zonular-traction tufts may predispose to retinal tear/hole
  
- C. **Meridional Folds:** folds of redundant retina at ora. Occasional tear at posterior margin.
  
- D. **Enclosed ora bays:** islands of pars plana epithelium posterior to ora surrounded by normal peripheral retina
  
- E. **Peripheral Retinal Excavations:** may be atypical lattice

### IV. Lesions Not Predisposing to Retinal Detachment:

- A. **Paving-stone Degeneration:** discrete areas of ischemic atrophy of outer retina, RPE, and choriocapillaris. Usually inferior.
  
- B. **RPE Hyperplasia:** occurs after trauma, inflammation, tumor, or degeneration
  
- C. **RPE Hypertrophy:** degenerative or congenital. RPE looks dark or black. Flat.
  - 1. **Congenital hypertrophy of RPE (“bear tracks”)**, usually in groups
    - b. With aging as degenerative change in periphery.
    - c. Histology: larger than normal cells with large spherical melanin granules

### V. Prophylactic Treatment of Retinal Breaks:

- A. 6% of all eyes have breaks, but incidence of RD in population is only 0.07%

lifetime.

- B. Symptomatic flap tears have high risk of RD, therefore usually treated.
- C. Acute operculated holes less dangerous because vitreous traction relieved.
- D. Atrophic holes have low risk.
- E. Asymptomatic flap tears: usually low risk. May treat with lattice, myopia, subclinical detachment, aphakia with detachment in fellow eye
- F. Lattice Degeneration: may treat in presence of high myopia, retinal detachment in fellow eye, flap tears, aphakia.
- G. Fellow eye with RD: Phakic 10% risk  
Aphakic 20-36% risk

H. Subclinical detachment: subretinal fluid extends more than 1 disc diameter from the break but not more than 2 dd posterior to the equator.

## VI. Retinal Detachment

### A. Rhegmatogenous Retinal Detachment:

1. Liquid vitreous passes through break in the retina into subretinal space.
2. Break found in 97% of cases.
3. **Shafer's sign:** pigmented cells in anterior vitreous.
4. PVR most common cause of failure.
5. Surgical management:
  - a. Find all breaks
  - b. Close all breaks
  - c. Create a scar around each break
  - d. Scleral buckling:
    - i. Permanent buckling
    - ii. Temporary buckling (Lincoff "balloon")
  - e. Pneumatic retinopexy
    - i. Breaks in superior half of fundus
    - ii. Gas tamponade
  - f. PPV discussed later
6. Anatomic reattachment rate overall is 90%.
7. Post-operative vision: dependent on whether macula detached and how long
  - a. Sparing macula: 87% have 20/50 or better
  - b. Macula detached: 37% have 20/50 or better
    - i. If less than 1 week, 75% have 20/70 or better
    - ii. 1-8 weeks detached, 50% have 20/70 or better
  - c. Complications effecting final vision:
    - i. macular edema
    - ii. macular pucker

### B. Tractional Retinal Detachment:

1. Caused by vitreous membranes pulling on retina.
2. Usually membranes can be seen on examination.

3. Retina has smooth surface, concave, and is immobile.
4. Rarely extends to ora serrata.
5. May resolve with vitrectomy.
6. If rhegmatogenous component present, may require vitrectomy + buckle.

**C. Exudative Retinal Detachment:**

1. Treatment is usually directed at underlying disorder.
2. Usual causes are inflammatory or neoplastic.
3. Shifting fluid highly suggestive.
4. Smooth appearance of detached retina.

**VII. Differential Diagnosis of Retinal Detachment:**

**A. Retinoschisis**

1. Causes absolute scotoma
2. Smooth dome-shaped surface
3. Reaction to photocoagulation
4. Bilateral in 50-80%
5. Most common inferotemporally

**B. Typical peripheral cystoid degeneration**

1. Seen in most adults
2. Cystoid cavities in outer plexiform layer
3. Benign; may progress to typical degenerative retinoschisis

**C. Reticular peripheral cystoid degeneration**

1. Linear pattern following vessels
2. Cystoid spaces in nerve fiber layer.
3. Less common than TPCD.

**D. Degenerative retinoschisis**

1. Typical and reticular forms, difficult to differentiate clinically.
2. Reticular form: split occurs in nerve fiber layer
  - a. Associated with posterior extension and RD
  - b. Inner layer may be markedly elevated
  - c. 23% have outer layer breaks (rolled edges)
3. Typical form: split occurs in outer plexiform layer.
  - a. White dots: remnants of Muller cells bridging cavity

**E. Differentiation from Retinal Detachment:**

1. Causes absolute scotoma
2. Not associated with "tobacco dust" or vitreous hemorrhage
3. Smooth surface
4. No demarcation lines, cysts, or RPE atrophy

- F. Association with Retinal Detachment
  - 1. Schisis associated with 3% of retinal detachments
  - 2. Holes present in outer layer only:
    - a. associated with slowly developing detachments
    - b. Usually does not require treatment
  - 3. Holes in both inner and outer layers
    - a. Collapse of cavity may occur, causing RD
    - b. Usually require surgery
    - c. Difficult to repair

## Chapter 12. Diseases of the Vitreous

### I. Normal Anatomy

- A. Vitreous consists of collagen and hyaluronic acid
- B. Vitreous Base: from 1.5-2.0 anteriorly and 1.0-3.0 posteriorly to the ora serrata
- C. Vitreous Cortex:
  - 1. Anterior Vitreous Cortex: anterior to vitreous base
  - 2. Posterior Vitreous Cortex: posterior to vitreous base
    - a. Adherent to basal lamina of the internal limiting membrane of the retina

### II. Posterior Vitreous Detachment

#### See Notes from Chapter 11

- A. Vitreomacular Traction Syndromes:
  - 1. Persistent attachment of vitreous to macula causes distortion and elevation of the macula
  - 2. Attachment to fovea can cause macular hole
  - 3. Remnants of vitreous on inner retina can cause ERM

### III. Developmental Abnormalities

#### A. Tunica Vasculosa Lentis

- 1. Mittendorf's dot
- 2. Bergmeister's Papilla
- 3. Entire hyaloid artery may persist

#### B. Prepapillary Vascular Loops

- 1. Not remnants of hyaloid artery
- 2. Normal retinal vessels which grew into Bergmeister's papilla
- 3. Supply part of retina
- 4. May be arterial or venous
- 5. Associated with BRAO, amaurosis fugax, vitreous heme

#### C. Persistent Hyperplastic Primary Vitreous (PHPV)

- 1. Results from failure of primary vitreous to regress.
  - a. New terminology: Persistent fetal vasculature (PFV)

2. Found in normal infants
3. Unilateral in 90%
4. Anterior PHPV:
  - a. Persistent hyaloid artery
  - b. White vascular membrane behind lens
  - c. microphthalmos
  - d. leukocoria
  - e. glaucoma
  - f. Extremely poor prognosis
5. Posterior PHPV:
  - a. Occurs with anterior PHPV or isolated
  - b. Usually normal anterior chamber
  - c. May have microphthalmos
  - d. Retinal (falciform fold) extending from the disc
  - e. Differentiate from ROP, toxocariasis, and FEVR

#### IV. Hereditary Hyaloideoretinopathies with Optically Empty Vitreous

##### A. Ocular only

1. Wagner's (not associated with RD): autosomal dominant
2. Jansen's (high incidence of RD): autosomal dominant
3. Lattice Degeneration
4. Goldmann-Favre
5. Familial Exudative Vitreoretinopathy

##### B. Associated with Systemic Abnormalities

1. Stickler's (Marfanoid): **most common**
2. Weill-Marchesani (stiff joints)
3. Spondyloepiphyseal dysplasia, variant and congenital forms
4. Kniest Syndrome
5. Vitreoretinal Degeneration in Facial Clefting Syndrome

##### C. Wagner's and Jansen's Syndromes

1. Autosomal dominant
2. Myopia, strabismus, cataract, vitreous degeneration, chorioretinal changes
3. Abnormal ERG late

##### D. Hereditary Progressive Arthroophthalmopathy of Stickler

1. Autosomal dominant
2. Myopia, open angle glaucoma, cataract, perivascular pigmentation, lattice
3. **RD common**, posterior breaks, poor prognosis
4. Optically empty vitreous
5. Flattening of mid-face and nasal bridge, cleft palate, Pierre-Robin anomaly (micrognathia, cleft palate, glossoptosis)

6. Hearing loss
7. Marfanoid habitus with joint hyperextensibility (stiffness and soreness in childhood), arthritis, skeletal dysplasia, scoliosis, arachnodactyly, hip joint deformity

#### E. Other Syndromes with Dwarfing

1. Spondyloepiphyseal Dysplasia
  - a. Congenita form: manifest in 1st year of life; short limbs, cleft palate, club feet
  - b. Childhood form: dwarfism with short trunk
  - c. Tarda form: dwarfism with short trunk, hip and back abnormalities
  - d. Associated with myopia and RD
2. Kniest Disease
  - a. Form of dwarfism
  - b. abnormalities of vertebral bodies, can cause paralysis
  - c. Facial abnormalities with cleft palate
  - d. Deafness
  - e. Myopia and RD
3. Vitreoretinal Degeneration in Facial Clefting Syndrome
  - a. Probably autosomal dominant
  - b. Hallmarks are family history of RD, facial clefting, vitreoretinal degeneration
  - c. Skeletal abnormalities
  - d. Hearing loss
  - e. Facial abnormalities and cleft palate
  - f. Optically empty vitreous with condensations and epiretinal membranes, perivascular retinal pigmentation, lattice, retinal breaks and RD

#### V. Familial Exudative Vitreoretinopathy (FEVR)

- A. Autosomal dominant; full-term infants
- B. Temporal retina does not vascularize
- C. Tractional RD's with temporal dragging of disc vessels
- D. Retinal exudation, especially in babies and adolescents
- E. Rhegmatogenous RD's may occur
- F. Usually bilateral

#### VI. Asteroid Hyalosis

- A. Oval, white bodies adherent to framework of vitreous gel.  
**Calcium phospholipids**
- B. Most frequently seen in patients over 50; overall incidence 1 in 200.
- C. Unilateral in 75% of cases.
- D. Associated with diabetes in 30%; also associated with HTN.
- E. Rarely causes significant visual symptoms.

F. Fluorescein angiography images well.

**VII. Cholesterolosis**

- A. Also known as **Hemophthalmos, Synchrony Scintillans.**
- B. Highly refractile golden crystals freely floating in vitreous gel and anterior chamber. **Cholesterol crystals.**
- C. Related to injury or inflammation.
- D. Often settle inferiorly.

**VIII. Amyloidosis**

- A. Associated with dominantly inherited form
- B. Bilateral vitreous opacities, “glass wool”
- C. Amyloid also can be deposited in retinal vessels, choroid, TM
- D. Associated with hemorrhages, exudates, CW spots, peripheral retinal neov.
- E. Systemic findings: polyneuropathy, CNS abnormalities. Deposits in heart, skin, GI tract

**IX. Spontaneous Vitreous Hemorrhage**

- A. About half caused by diabetic retinopathy
- B. Retinal Break (12-17%)
- C. PVD (7-12%)
- D. Rhegmatogenous RD (7-10%)
- E. Neovascularization with BRVO or CRVO (3-10%)
- F. Trauma
- G. Congenital retinoschisis and pars planitis
- H. Re-examine until cause is found!

**X. Pigment Granules**

- A. **Shafer’s Sign:** pigmented cells in anterior vitreous in absence of trauma, uveitis, or RP
- B. Highly suggestive of retinal break

**XI. Vitreous Abnormalities Secondary to Cataract Surgery**

- A. Vitreous loss associated with incarceration in wound.
  - 1. Poor wound closure
  - 2. Increased risk of endophthalmitis
  - 3. epithelial ingrowth
  - 4. Hypotony
  - 5. Chronic inflammation
  - 6. Cystoid macular edema
  - 7. Irvine-Gass Syndrome (with disc edema)
  - 8. Retinal Detachment

## **I. Evaluation of the Patient Following Ocular Trauma**

- A. Detailed history of injury
  1. How and when
  2. Estimate of force involved
  3. Work- related
  4. Systemic injuries
  5. Last meal
  6. Prior ocular history
  7. Possibility of FB
- B. Careful complete examination
  1. Avoid putting pressure on globe
  2. Afferent pupillary defect
  3. IOP
  4. Indirect ophthalmoscopy if possible
  5. Ultrasonography
  6. CT scanning

## **II. Blunt Trauma (no break in tissue)**

- A. Object causing injury does not penetrate eye**
- B. Blunt trauma may cause rupture of the eyewall**
- C. Anterior and posterior segment injuries may coexist
- D. Look for angle recession, hyphema, vitreous hemorrhage, retinal tears, RD, lens injury, commotio retinae, choroidal rupture, macular hole, avulsed optic nerve, scleral rupture, orbital fracture
- E. Vitreous hemorrhage**
  1. From injury to ciliary body, retina, or choroid
  2. Retinal tears or scleral rupture
  3. Ultrasound if posterior segment cannot be visualized.
  4. Bed rest may allow clearing.
- D. Commotio Retinae**
  1. Contrecoup injury to outer retinal layers by shock waves
  2. Retinal whitening
  3. In posterior pole, called Berlin's edema
  4. Cherry red spot in macula because of lack of photoreceptors in fovea
  5. Clears within weeks
  6. Visual recovery [variable; poor if increased foveal thickness found on OCT]
  7. No treatment
- E. Choroidal Rupture**
  1. Stretch injury secondary to compression of globe in A-P axis
  2. Tear in Bruch's membrane, RPE, and choriocapillaris
  3. Usually temporal to and concentric to disc
  4. Overlying retina usually intact.
  5. Associated with subretinal hemorrhage
  6. Can develop subretinal neovascularization

**F. Traumatic macular hole**

1. Caused by contusion necrosis, vitreous traction, Berlin's edema, or combination
2. May also occur after subretinal hemorrhage
3. Lightning and electrical injury can also cause macular hole

**G. Retinitis Sclopetaria**

1. Produced by high-speed missile injuries to the orbit
2. Choroidal and retinal injury
3. Subretinal and retinal hemorrhages early
4. Extensive pigment alteration and scar formation late
5. If macula is involved, visual prognosis is poor
6. Reported after paintball injuries

**H. Scleral Rupture**

1. Most common locations are at limbus or under muscle insertions
2. Conjunctival chemosis and hemorrhage, decreased ductions, deep anterior chamber, vitreous hemorrhage. IOP can be low.

**III. Lacerating and Penetrating Injuries (entrance wound only)**

**A. Cutting or tearing of eyewall by sharp object**

**B. Penetrating injury is caused by a laceration at a single site on the globe.**

C.. Careful wound closure of total extent of wound

D. Very posterior wounds may be left open

E. Complications include traction RD, cyclitic membrane, phthisis bulbi.

F. Optimal timing of vitrectomy is unknown.

1. Defer until 10-14 days to reduce risk of bleeding, allow cornea to clear, and allow spontaneous PVD to occur

**IV. Perforating Injuries (entrance and exit wounds)**

A. Close anterior wounds. Small posterior wounds may be left unrepaired.

B. Vitrectomy useful in vitreous hemorrhage. Usually delay 7 days to allow posterior wounds to close by proliferation.

C. Remove posterior hyaloid during vitrectomy.

**V. Intraocular Foreign Bodies**

A. Rule out after any trauma

B. X-rays and CT scanning may be helpful.

C. MRI contraindicated.

D. Surgical techniques:

1. PPV: free FB from blood or tissue
2. Pars plana magnet extraction if small, non-encapsulated, easily seen, non-embedded.
3. Intraocular magnet extraction following PPV

4. Intraocular forceps if non-magnetic or large
- E. Retained foreign bodies
  1. Inert sterile FB's well-tolerated  
(stone, sand, glass, porcelain, plastic, cilia)
  2. Minimal inflammation: zinc, aluminum
  3. Pure copper highly toxic (**chalcosis**)
    - a. causes severe inflammation.
    - b. affinity for limiting membranes (Descemet's).
    - c. signs:
      - i. green discoloration of iris
      - ii. "sunflower cataract"
      - iii. opacities in aqueous, vitreous, and in retina.
  4. **Iron (siderosis):**
    - a. deposited in epithelial tissues of iris, ciliary body, lens, retina and RPE.
    - b. Ferric ions cause production of oxidants causing Haber-Weiss reaction (transition metal ions catalyze generation of hydroxyl radicals, leading to lipid peroxidation, sulphydryl oxidation and depolymerization)
    - c. damage primarily to photoreceptors and RPE.
    - d. symptoms:
      - i. nyctalopia
      - ii. constriction of VF
      - iii. blindness.
    - e. ocular signs:
      - i. corneal staining
      - ii. iris heterochromia
      - iii. pupillary mydriasis
      - iv. deposits on lens, cataract
      - v. retinal pigmentation
      - vi. optic atrophy
      - vii. secondary open angle glaucoma.
    - f. ERG eventually extinguished

## VI. Posttraumatic Endophthalmitis

- A. Occurs in 2-7% of penetrating injuries
- B. Higher incidence with FB's and in rural settings (remember gardening!)
- C. *Bacillus cereus* in 25% of cases
- D. Signs and symptoms:
  1. Pain
  2. Decreased vision
  3. Inflammation with fibrin, hypopyon, vitreous inflammation
  4. Corneal opacification
- E. Culture and sensitivity of anterior chamber and vitreous
- F. Antibiotics:

1. subconjunctival
2. intravenous
3. intravitreal
4. topical
5. prophylactic use if no sign of endophthalmitis is controversial

### **VII. Sympathetic Ophthalmia**

- A. Avoid primary enucleation if globe can be repaired
- B. Enucleation within 2 weeks if no potential for recovery
- C. Incidence 1 in 500 cases of penetrating injury
- D. Onset usually within 3 months to 1 year, but can be many years later
- E. Symptoms in fellow eye:
  1. loss of vision
  2. loss of accommodation
  3. photophobia
  4. pain
- F. Signs in fellow eye:
  1. panuveitis
  2. multifocal infiltrates in RPE (Dalen-Fuchs nodules) or choroid
  3. exudative detachment
  4. optic nerve swelling
  5. uveal thickening on B-scan

### **VIII. Shaken Baby Syndrome/Child Abuse**

- A. Infants less than 1 year of age
- B. Systemic signs:
  1. bradycardia, apnea, hypothermia
  2. lethargy, irritability, seizures, hypotonia
  3. failure to thrive
  4. bulging fontanelles
  5. bruises, fractures
  6. subdural/subarachnoid hemorrhage
- C. Ocular signs:
  1. Retinal hemorrhages and CW spots
  2. Retinal folds
  3. hemorrhagic schisis cavities
  4. resolve rapidly
  5. resemble Terson syndrome, Purtscher's, or CRVO

### **IX. Avulsion of the Optic Disc**

- A. Can occur from extreme dislocation of globe
- B. Penetrating orbital injury
- C. Sudden increase in IOP

**Be sure to review Chapter 14 "Adverse Effects of Electromagnetic Energy on the Retina",**

**Chapter 15 "Laser Therapy for Posterior Segment Diseases", and Chapter 16 "Vitreoretinal Surgery".**

**If you can get a copy of it, I also highly recommend the AAO Monography "Retinal Detachment Principles and Practice" by Hilton, McLean, and Brinton, 1995 which is unfortunately out of print.**

**Revised 12/27/10**