

**Subjects**

- 1) Chromosomes: mendelian genetics, enzyme anomalies, HLA 2**
- 2) Sex ratios of eye diseases 6**
- 3) Systemic syndromes with eye findings 7**
- 4) Ocular syndromes 19**
- 5) Phacomatoses 26**

## 1) Chromosome Defects

1p: Schnyder's crystalline corneal dystrophy  
**1p**: Stargardt's/fundus flavimaculatus  
 1q: juvenile OAG (AD)  
 2q: AR Alport's  
 2q: Oguchi's disease  
 3p: Von Hippel Lindau  
 3q: CSNB (rhodopsin)  
 3q: RP (AR & AD) (rhodopsin)  
 3q: Dominant (Kjer) optic atrophy  
 3q: blepharophimosis syndrome  
**4q25**: Axenfeld-Rieger's syndrome ("Rieg" gene)  
 4q25: Iridogoniodysgenesis syndrome  
 (Rieger's?)  
 a.k.a. iris hypoplasia (not aniridia)  
**5q31**: granular/lattice/Avellino/Reis B?  
 6p: RP-AD (RDS/peripherin)  
 6p11: retinal macular dystrophy ? (RDS/periph.)  
 6p11: retinitis punctata albescens (RDS/periph.)  
 6p25: iridogoniodysgenesis anomaly  
 6q14: North Carolina macular dystrophy  
 6q14: PBCRA - progr. bilat. chorioret. atrophy  
 9q: Tuberous scaposis  
 10q: gyrate atrophy  
**11p13**: aniridia (PAX6)  
**11p13**: Peter's anomaly (PAX6)  
 11q: oculocutaneous albinism (tyrosinase -)  
 11q: FEVR  
**11q**: Best's  
 11q: ataxia-telangiectasia (Louis Bar)  
 12: cornea plana  
 12q13: Stickler's  
 12q13: Wagner's  
**13q14**: RB  
 13: A.D. coloboma  
 14q: rod monochromacy  
 14q: oculopharyngeal dystrophy  
 15q: oculocutaneous albinism (tyrosinase +)  
 15q: Marfan's (fibrillin)  
 16q21: macular dystrophy of cornea  
**17q**: NF1  
 17p: central areolar choroidal dystrophy  
 17p: Leber's congenital amaurosis  
 20q11: posterior polymorphous dystrophy  
 21q: homocystinuria (cystathione  $\beta$  synthase)  
**22**: NF2  
 22q: Sorsby's dystrophy

## Autosomal Dominant Disorders

### A) Anterior segment

#### i) corneal dystrophies

- 1) post. embryotoxon
- 2) M-D-F
- 3) Reis Buckler's
- 4) gelatinous droplike dystrophy (amyloid)
- 5) Lattice/Granular
- 6) cornea farinata
- 7) Meesman's dystrophy
- 8) Schnyder's central crystalline dystrophy
- 9) CHSD
- 10) fleck dystrophy
- 11) posterior amorphous stromal dystrophy
- 12) Fuch's endoth. dystrophy
- 13) Posp. Polym. dystrophy - some
- 14) CHED - some (progressive)

#### ii) other

- 1) aniridia (2/3)
- 2) Axenfeld-Rieger's (75%)
- 3) sclerocornea
- 4) simple ectopia lentis
- 5) blepharophimosis syndrome
- 6) seborrheic keratosis
- 7) basal cell nevus syndrome
- 8) juvenile onset glaucoma - some
- 9) cong. cataracts - some
- 10) cornea plana - some
- 11) keratoglobus - some
- 12) microcornea - some
- 13) megalocornea - some

### B) Posterior segment

- 1) Most phacomatosis (NF, TS, VHL)
- 2) fleck dystrophy
- 3) RP - some - 22%
- 4) FEVR
- 5) Sorsby's
- 6) North Carolina dystrophy
- 7) Wagner's
- 8) Stickler's
- 9) familial drusen - common
- 10) disc drusen
- 11) central areolar choroidal dystrophy
- 12) Best's - incomplete penetrance - common
- 13) coloboma - some
- 14) Tritanopia/ tritanomalous

### C) Plastics

- 1) congenital fibrosis syndrome - some

- 2) euryblepharon - some
- 3) Apert's
- 4) Crouzon's
- 5) Treacher Collins (Kanski)
- 6) Goldenhar's (Kanski)

D) *Systemic*

- 1) osteogenesis imperfecta
- 2) Marfan's
- 3) Alport's - rare
- 4) Gardner's
- 5) myotonic dystrophy
- 6) Waardenburg's

**Autosomal Recessive Disorders**

A) *Anterior segment*

- 1) cornea plana
- 2) congenital glaucoma
- 3) macular dystrophy
- 4) sclerocornea
- 5) lipoid proteinosis
- 6) ectopia lentis et pupillae
- 7) post polymorphous dystrophy
- 8) CHED - some (non-progressive)
- 9) keratoglobus - some
- 10) cong. cataracts - some
- 11) megalocornea - rare

B) *Posterior segment*

- 1) RP - some - 10%
- 2) ocular albism - some
- 3) Goldman Favre
- 4) uveal coloboma - some
- 5) Bietti's crystalline dystrophy
- 6) Leber's congenital amaurosis
- 7) Oguchi's disease
- 8) oculocutaneous albinism
- 9) Stargardt's
- 10) rod monochromatism

C) *Plastics*

- 1) congenital fibrosis syndrome - some

D) *Systemic*

- 1) Wilson's disease
- 2) ochronosis - alkaptonuria
- 3) Cockayne syndrome
- 4) xeroderma pigmentosum
- 5) ataxia-telangiectasia

- 6) Weill Marchesani (1 parent usually short - carrier)

- 7) cystinosis
- 8) tyrosinemia
- 9) homocysteinuria
- 10) Alport - rare
- 11) oculocutaneous albinism
- 12) abetalipoproteinemia

**X-linked disorders**

- underlined were on exam

A) *Anterior segment*

- 1) megalocornea - most
- 2) cong. cataracts - some

B) *Posterior Segment*

- 1) choroideremia (geranylgeranyl transferase)
- 2) congenital stationary night blindness - some
- 3) blue cone monochromatism (Xq28)
- 4) Norrie's disease
- 5) red or green color deficiencies
- 6) juvenile retinoschisis
- 7) ocular albinism ("Nettleship Falls")
- 7) Aicardi's
- 8) primary retinal dysplasia - some (same gene as Norrie's)
- 9) FEVR - some (same gene as Norrie's)
- 10) cone dystrophy - some

C) *Systemic*

- 1) RP (some) - worst
- 2) ocular albinism (Nettleship-Falls type)
- 3) Lowe's (oculocerebral)
- 4) Fabry's
- 5) incontinentia pigmenti (woman only; males lethal)
- 6) M.L.S. (microphthalmia with linear skin defects)
- 7) Alport - some; XL dominant
- 8) Nance Horan
- 9) Hunter's

**Sporadic disorders**

A) *Anterior Segment*

- 1) ICE syndrome
- 2) Peter's
- 3) cong. cataracts - 60%
- 4) post. lenticonus
- 5) aniridia - 1/3
- 6) sclerocornea (50%)

- 7) Axenfeld-Rieger's (some)
- 8) Fuch's endoth. dystrophy - some

#### B) *Posterior Segment*

- 1) RP
- 2) Wyburn Mason (racemose angiomatosis)

#### C) *Plastics*

- 1) congenital fibrosis syndrome - some
- 2) euryblepharon - some

#### D) *Posterior Segment*

- 1) Sturge Weber
- 2) Prader Willi

#### **Mitochondrial inherited**

- 1) MERRF (myoclonus, epilepsy, ragged red fibers)
- 2) MELAS (mitochondrial encephalopathy, lactic acidosis, )
- 3) Leber's optic neuropathy (11778)
- 4) Kearns syndrome / CPEO
- 5) Leigh's

#### **Multiple chromosome Defects**

- 1) Bardet-Biedl (AR RP): 11q, 16q, 3q, 15q:
- 2) Waardenburg: 2q, 3p, 11p13 (PAX3)
- 3) congenital cataracts: 1p, 1q, 2q, 16q, 17p, 17q, 22q
- 4) congenital glaucoma: 2p, 1p,
- 5) Stickler's: 6p, 12q
- 6) ARRP: 1q, 3q, 4p, 6p, 11q
- 7) ADRP: 3q
- 8) sporadic RP: 6p, 7p, 7q, 8q, 17p, 1p, 17q, 19q
- 9) Usher's: 1q, 3q, 10q, 11p, 11q, 14q, 21q
- 10) cone rod dystrophy: 18q, 19q
- 11) congenital fibrosis of the EOM's: 12, 7p

#### **Enzyme deficiencies or gene anomalies**

- 1) choroideremia: geranylgeranyl transferase
- 2) gyrate atrophy: ornithine aminotransferase
- 3) galactosemia: gal-1-P uridyl transferase, (classic), galactokinase, galactose epimerase
- 4) RP: rhodopsin, peripherin, PDE, ROM1
- 5) retinitis punctata albescens, RP: peripherin
- 6) Marfan's, ectopia lentis: fibrillin
- 7) Norrie's, XL FEVR, XL retinal dysplasia: norrin
- 8) Von Hippel: novel mb protein
- 9) oculocutaneous albinism (tyr-): tyrosinase
- 10) Oguchi's: arrestin, PDE

- 11) CSNB - some: rhodopsin
- 12) NF1: neurofibromin
- 13) NF2: Schwannomin
- 14) Ochronosis (alkaptonuria): homogentisic acid oxidase (homogentisic acid is interm. in breakdown of tyrosine)
- 15) Wagner's, Stickler's: collagen type II
- 16) Fabry's: alpha galactosidase
- 17) Sorsby dystrophy: metalloproteinase inhibitor
- 18) homocystinuria: cystathione  $\beta$  synthase

#### **Cone locations**

- 1) Red cone pigment: Xq28,
- 2) Green cone pigment: Xq28
- 3) Blue cone pigment: 7q31

#### **Treatable systemic diseases with retinopathy**

- 1) gyrate atrophy  
Dx: increased ornithine  
Tx:: diet
- 2) cobalamin C defects  
Dx: homocystine in urine?  
Tx: hydroxycobalamin
- 3) abetalipoproteinemia  
Dx: low serum lipids  
Tx: Rx: Vit A and E
- 4) cystinosis  
Tx:: cysteamine
- 5) mucopolysaccharidosis  
Tx: bone marrow transplant
- 6) Refsum's disease  
Dx: increased phytanic acid  
Tx: plasmaphoresis
- 7) osteopetrosis  
Tx: B.M.T.
- 8) Alagille syndrome  
Tx: Vit A and E
- 9) syphilis  
Tx: Penicillin
- 10) Vit A deficiency  
Tx: Vit A
- 11) cancer
- 12) oxalosis  
Tx: pyridoxine (B6)
- 13) Cogan's syndrome  
Tx: Immunosuppressants
- 14) hypercalcuria  
Tx: treat kidney stones
- 15) homocystinuria  
Tx: pyridoxine, diet, ASA

**HLA associations**

**A29:** birdshot retinochoroidopathy

**B5:** Behcet's (Bw51 subset)

**B7:** POHS

**B8:** ant. uveitis in blacks, primary Sjogren's syndrome

**B27:** ant. uveitis with IBD, AS, psoriasis, Reiter's

**Bw54:** Posner-Schlossman, VKH

**DR2:** MS, POHS

**DR3:** Thygeson's, Sjogren's

**DR4:** OCP, VKH

**DR5:** pauciarticular JRA

## 2) Sex Ratios of eye diseases

### Disorders more common in women

#### A) *Autoimmune conditions*

- 1) SLE
- 2) Graves' (4:1)
- 3) Hashimoto's
- 4) RA
- 5) DM type I
- 6) polymyositis
- 7) primary Sjogren's syndrome (9:1)
- 8) SLK
- 9) optic neuritis
- 10) scleritis
- 11) erythema multiforme (SJ)
- 12) pemphigoid (2:1)
- 13) sarcoidosis (2:1)
- 14) OCP (2:1)

#### B) *Retinal conditions*

- 1) macular hole (3:1)
- 2) epiretinal membranes
- 3) ARMD
- 4) acute macular neuroretinopathy
- 5) MEWDS (4:1)

#### C) *Anterior segment*

- 1) dry eyes
- 2) ICE syndromes
- 3) post. keratoconus
- 4) blepharitis
- 5) M-D-F
- 6) blepharochalasis
- 7) sebaceous cell carcinoma (2:1)
- 8) Salzmann's nodular degeneration
- 9) PACG
- 10) xanthelasma
- 11) chronic iridocyclitis (of young girls)

#### D) *Neuro*

- 1) morning glory disc (2:1)
- 2) idiopathic intracranial hypertension
- 3) meningioma (3:1)
- 4) CVA
- 5) essential blepharospasm
- 6) Duane's (2:1)

#### E) *Orbit/plastics*

- 1) capillary hemangioma (2:1)
- 2) NLD obstruction (2:1)
- 3) fibrous dysplasia
- 4) cavernous hemangioma

### Disorders more common in men

#### A) *anterior segment*

- 1) Mooren's ulcer
- 2) spheroidal degeneration
- 3) anterior lenticonus
- 4) pigment dispersion
- 5) vernal
- 6) Reiter's (90%)

#### B) *Retina*

- 1) Coat's (retinal telangiectasia)
- 2) Eales' (primary idiopathic retinal vasculitis)
- 3) idiopathic juxtafoveal telangiectasia (type 1)
- 4) Leber's (90%)

#### C) *Neuro*

- 1) medullated nerve fibers
- 2) Behr's optic atrophy
- 3) glioblastoma (optic nerve)
- 4) oculomotor apraxia
- 5) CPEO

#### D) *Other*

- 1) traumatic eye disease (all)
- 2) pleomorphic adenoma (benign mixed)
- 3) primary systemic amyloidosis

### Disorders more common in blacks

- 1) sarcoid
- 2) limbal vernal
- 3) COAG
- 4) racial melanosis
- 5) tyrosinase positive oculocutaneous albinism
- 6) nevus of Ota
- 7) SCA

### Disorders more common in Asians

- 1) vernal
- 2) VKH
- 3) Behcet's
- 4) racial melanosis
- 5) Nevus of Ota

### Diseases more common in whites

- 1) ARMD
- 2) BCC, SCC, melanoma of skin
- 3) uveal melanoma in nevus of ota

### 3) Syndromes with Eye Findings

#### Abetalipoproteinemia

(Bassen-Kornzweig Syndrome)

- AR

- 1) abetalipoproteinemia
- 2) malabsorption of fat
- 3) acanthocytosis (crenated RBC's)
- 4) ataxia, neuropathy
- 5) retinitis pigmentosa
- 6) cardiac myopathy
- 7) angioid streaks (rare)
- 8) retinopathy can be arrested by vitamin E supplements

#### Aicardi's

- 1) XL dominant (females only)
- 2) MR
- 3) agenesis of corpus callosum
- 4) large round depigmented chorioretinal lesions
- 5) optic head colobomas

#### AIDS

##### new CDC HIV definition

- 1) HIV+

##### and any of the following:

- 1) CD<200
- 2) TB
- 3) cervical CA
- 4) recurrent pneumonia

##### Eye findings

- 1) *lids*
  - a) Kaposi's
  - b) HSV & HZV
  - c) HPV
  - d) molluscum
- 2) *conjunctiva*
  - a) Kaposi's
  - b) CMV, HSV, HZV
- 3) *cornea*
  - a) HSV, HZV, Candida
  - b) microsporidia
- 4) *iris*
  - a) toxo iritis
- 5) *retina*
  - l) *Infectious*
    - a) CMV
    - b) ARN: HSV, HZV
    - c) toxo
    - d) candida
    - e) syphilis

ll) *non-infectious*

- a) CWS
- b) hemorrhages
- c) IRMA
- d) ischemic maculopathy
- 6) *choroid*
  - a) PCP
  - b) MAI
  - c) Candida
  - d) Toxo
- 7) *vitreous*
  - a) bacterial endophthalmitis
- 8) *optic nerve*
  - a) CMV
  - b) syphilis
  - c) optic atrophy (CNS cryptococcus)
- 9) *orbit*
  - a) Kaposi's
  - b) Burkitt's
- 10) *neuro*
  - a) cranial nerve palsies

#### Albinism - Oculocutaneous

A) *Tyrosinase negative*

- 1) AR
- 2) path: no melanin pigment seen at all
- 3) no pigment in skin or eye (oculo-cutaneous)
- 4) iris transillumination
- 5) acuity < 20/100
- 6) nystagmus
- 7) strabismus
- 8) photophobia
- 9) foveal hypoplasia (no foveal pit)
- 10) increased or decreased crossing? at chiasm
- 11) high refractive errors

B) *Tyrosinase positive*

- 1) AR
- 2) path: normal # of pigment granules, decreased melanin within the granules; see macromelanosomes in Chediak-Higashi type
- 3) associated with various syndromes (Chediak-Higashi, Hermanski Pudlak)
- 4) ocular findings similar to tyrosinase negative but not as severe
- 5) skin findings more variable
- 6) more common in blacks

#### Albinism - Ocular ("Nettleship Falls")

- 1) X-linked

- 2) path: macromelanosomes (classic) - large pigment granules
- 3) poor acuity
- 4) nystagmus
- 5) photophobia
- 6) foveal hypoplasia
- 7) iris: "cartwheel transillumination"

### Carriers of X-linked albinism

- 1) pigment changes in iris
- 2) fundus pigment changes
- 3) macromelanosomes on path
- 4) ? light hair

### Albinoidism

- 1) normal acuity (or slight decrease)
- 2) normal fovea

### Alport Syndrome

- x-linked dominant (80%); AD and AR described

- 1) hereditary nephritis
- 2) deafness
- 3) platelet anomalies

Ocular

- 1) anterior lenticonus +/- ant. polar cataract
- 2) post. lenticonus (less common) +/- PSCC
- 3) spherophakia
- 4) retinal stippling

### Ankylosing Spondylitis

- 1) iritis
- 2) sacroiliitis
- 3) lung apex fibrosis
- 4) aortitis (5%)

### Apert's syndrome

A) *Ocular*

- 1) strabismus
- 2) optic atrophy
- 3) exposure
- 4) anti-mongoloid slant

B) *Systemic*

- 1) craniosynostosis
- 2) syndactyly of hands ("mitten hands")
- 3) raised intracranial pressure
- 4) congenital heart disease
- 5) MR in 1/3
- 6) lung
- 7) kidney

### Behcet's diagnosis

**Complete:** - 4 main Sx.

**Incomplete**

a) 3 main Sx.

or b) recurrent eye disease + 1 main

**Suspect:** 2 major (not eyes)

**Possible:** 1 major

main signs:.

- 1) aphthous ulcers
- 2) genital ulcers
- 3) eye lesions
- 4) skin lesions

A) *Anterior Segment*

- 1) anterior uveitis
- 2) fine KP's (upper cornea)
- 3) can have mutton fat KP's
- 4) hypopyon

B) *Posterior segment*

- 1) retinal vasculitis
- 2) retinal hemorrhages
- 3) macular edema
- 4) retinal necrosis
- 5) AION
- 6) vitritis

B) *Skin lesions*

- 1) erythema nodosa
  - 2) acne
  - 3) hypersensitivity
- C) *Other features*
- 1) arthropathy
  - 2) CNS involvement (meningismus, CVA's, palsies, confusion): 25%
  - 3) GI lesions
  - 4) occlusive b.v. disease
  - 5) aneurysm formation
  - 6) epididymitis
  - 7) thrombophlebitis

### Behr's optic atrophy

- 1) M > F
- 2) AR
- 3) increased DTR
- 4) ataxia
- 5) MR
- 6) hypotonia
- 7) external ophthalmoplegia
- 8) visual acuity ?

### CHARGE

- 1) coloboma

- 2) heart disease
- 3) choanal atresia
- 4) mental retardation
- 5) genital hypoplasia
- 6) ear anomalies (deafness)

### **Cockayne syndrome**

- AR

#### A) *Ocular*

- 1) band keratopathy
- 2) lens: cataracts
- 3) iris hypoplasia → poor dilation (characteristic)
- 4) pigmentary retinopathy
- 5) strabismus
- 6) nystagmus
- 7) optic atrophy
- 8) enophthalmos
- 9) hyperopia

#### B) *Systemic*

- 1) bird like facies
- 2) cachexia
- 3) growth retardation
- 4) CNS: MR, seizures
- 5) deafness

### **Congenital Rubella**

#### A) *systemic*

- 1) congenital heart disease
- 2) myocarditis
- 3) pneumonitis
- 4) hepatosplenomegaly
- 5) jaundice
- 6) thrombocytopenic purpura
- 7) microcephaly
- 8) MR
- 9) deafness
- 10) encephalitis

#### B) *Ocular*

- 1) vision good (20/20-20/60)
- 2) normal ERG and EOG, dark adaptation
- 3) corneal clouding
- 4) cataracts (20%)
- 5) angle dysgenesis
- 6) iris dilator hypoplasia
- 7) microphthalmia
- 8) pigment retinopathy
- 9) chorioretinitis
- 10) congenital glaucoma
- 11) nystagmus

### **Congenital Syphilis**

#### A) *Systemic*

- 1) notched incisors
- 2) frontal bossing
- 3) saddle nose
- 4) saber shins
- 5) CN 8 deafness
- 6) rhagades (circumoral lines)
- 7) MR

#### B) *Ocular*

- 1) deep IK as child or adolescent
- 2) active IK may have salmon patch appearance
- 3) KP's during active IK
- 4) bilat in 80% (acquired unilateral in 60%)

### **Cogan's syndrome**

- young adult

- URTI 1-2 weeks prior to symptoms

#### A) *Systemic*

- 1) verigo
- 2) tinnitus
- 3) hearing loss
- 4) systemic vasculitis (PAN)

#### B) *Ocular*

- 1) patchy IK

treatment: topical and systemic steroids

### **Crouzon's disease**

- AD

#### A) *Ocular*

- 1) proptosis - exposure keratopathy
- 2) hypertelorism
- 3) exophthalmos
- 4) strabismus (V pattern XT most common)
- 5) papilledema
- 6) optic atrophy

#### B) *Systemic*

- 1) craniosynostosis
- 2) hypoplastic mandible
- 3) respiration abnormal

#### C) *Emergencies*

- 1) globe dislocation: lift lid over globe to treat
- 2) exposure: tarsorrhaphy needed
- 3) papilledema from increased ICP and ON compression?

### **Cystinosis**

- AR

- high intracellular cystine
- plasma levels normal

- treatment: oral and topical cysteamine

A) *Ocular*

- 1) corneal deposits
- 2) prominent photophobia
- 3) conjunctiva deposits
- 4) iris crystal deposits
- 5) retinopathy

B) *Systemic*

- 1) kidney: causes rickets, renal failure
- 2) bone marrow deposits
- 3) WBC's
- 4) three forms: infantile, adolescent, adult

**De Morsier's**

clinical triad: 1-3

- 1) short stature
- 2) ON hypoplasia
- 3) nystagmus
- 4) agenesis of corpus callosum
- 5) panhypopit (hyperglyc, DI, hypothyroid)
- 6) absence of septum pellucidum

**Duane's**

- 1) F>M 2:1
- 2) OS>OD 3:2
- 3) bilateral 10%
- 4) epibulbar dermoids
- 5) pre-auricular appendages
- 6) deafness
- 7) webbed neck

**Fabry's disease (and other sphingolipidoses)**

- X-linked
- alpha galactosidase deficiency
- Tay Sachs is also a sphingolipidosis

A) *Ocular signs*

- 1) cornea vorticillata
- 2) spokelike cataracts
- 3) aneurysmal dilation of conj. veins
- 4) retina: vessel tortuosity
- 5) papilledema
- 6) optic atrophy

B) *Systemic*

- 1) skin changes: angiokeratoma
- 2) myocardial ischemia
- 3) cardiopulmonary hypertension
- 4) renal failure

**Fetal Alcohol Syndrome - eye findings**

- 1) blepharophimosis
- 2) ptosis
- 3) telecanthus
- 4) anterior segment dysgenesis
- 5) retinal vascular tortuosity
- 6) esotropia
- 7) ON hypoplasia

**Floppy eyelid**

A) *Ocular*

- 1) lax upper tarsus
- 2) easily everted
- 3) papillary conjunctivitis
- 4) mucous D/C
- 5) keratoconus
- 6) pannus
- 7) punctate keratitis

B) *Systemic*

- 1) Obese

**Gardner's syndrome (familial polyposis)**

- 1) AD
- 2) chromosome 5
- 3) osteomas
- 4) fibromas
- 5) dental abnormalities
- 6) CHRPE - bear tracks

**Goldenhar's**

triad: 1-3

- unilateral (80%)
- sometimes AD? (Kanski)

A) *Ocular*

- 1) upper lid colobomas
- 2) antimongoloid slant
- 3) limbal dermoids
- 4) decreased corneal sensation
- 5) Duane's

B) *Systemic*

- 1) vertebral deformities
- 2) pre-tragus fistulas
- 3) preauricular appendage
- 4) deafness
- 5) large mouth
- 6) mandibular hypoplasia

**Histoplasmosis**

- 1) fever
- 2) chills
- 3) cough

- 4) chest discomfort
- 5) headache
- 6) arthralgias and myalgias
- 7) erythema nodosum
- 8) erythema multiforme may accompany
- 9) Healed pulmonary lesions usually resolve completely
- 10) calcified granulomas may later be seen in lung parenchyma, hilar and mediastinal lymph nodes, and spleen

### Homocystinuria

Rx: pyridoxine (B6), diet, ASA

test: sodium nitroprusside

- AR

#### A) Systemic

- 1) MR
- 2) thromboemboli
- 3) marfanoid habitus
- 4) arteriosclerosis
- \*\* Beware of general anesthesia

#### B) Ocular

- 1) lens: sublux
- 2) retina: pigm. changes

### Hyperparathyroidism

- 1) band keratopathy
- 2) kidney stones
- 3) ulcers

### Hypoparathyroidism

- 1) blepharospasm
- 2) punctate keratitis
- 3) polychromatic cataract (50%)
- 4) pannus
- 5) papilledema (increased ICP)
- 6) extraosseous calcification

### Idiopathic Intracranial HTN

#### A) Systemic

- 1) CSF pressure > 250 cm H<sub>2</sub>O
- 2) normal CSF chemistry
- 3) unilateral or bilateral CN 6 palsies
- 4) 80% women
- 5) teens - 50s
- 6) obese
- 7) headaches
- 8) normal to small ventricles on CT

#### B) VF defect

- 1) blind spot enlargement

- 2) NFB defects
- 3) generalized depression

### JXG

#### A) Ocular

- 1) lesions develop in first 2 years of life
- 2) lesions resolve in 3-6 years
- 3) ocular lesions: tens to involve iris or c.b.
- 4) retina, choroid, conj, sclera, cornea have all had documented lesions
- 5) glaucoma
- 6) treatment of complications only (topical steroids, pressure drops, excision)

#### B) Systemic

- 1) kidney and liver nodules
- 2) lesions of face, neck, upper body

### Juvenile retinoschisis

#### A) Ocular

- 1) peripheral schisis 50%
- 2) foveal schisis 100%
- 3) VH
- 4) macular dragging
- 5) strabismus
- 6) nystagmus
- 7) carriers may show some changes
- 8) ERG: decreased b wave
- 9) NFL schisis
- 10) vitreous veils
- 11) tritan defect??

### JRA

#### A) Ocular

- 1) band keratopathy
- 2) cataracts
- 3) post. synechia
- 4) glaucoma
- 5) uveitis
- 6) phthisis

### Kawasaki Syndrome (infantile PAN)

- systemic steroids contraindicated
- use cycloplegics or mild topical steroid for iritis (synechia have not been reported)

#### A) Systemic

- 1) fever
- 2) skin rash
- 3) mucous membrane congestion
- 4) lymphadenopathy
- 5) systemic vasculitis

- 6) coronary artery aneurysms  
 B) *Ocular*  
 1) conjunctivitis  
 2) iritis (mild)

### **Kearns-Sayre**

- A) *Ocular*  
 1) Pigmentary retinopathy  
 2) CPEO  
 B) *Systemic*  
 1) ataxia  
 2) nerve deafness  
 3) heart block  
 4) elevated CSF protein

### **Lateral Medullary syndrome (Wallenberg)**

- 80% are post inf. cerebellar artery (PICA)  
 A) *Ipsilateral*  
 1) Horner's  
 2) CN 5, 9, 10, 11  
 3) voice affected (palate); hiccups  
 4) cerebellar ataxia  
 B) *Contralateral*  
 1) no pain or temp sensation  
 2) nystagmus  
 C) *Ocular*  
 1) skew?

### **Ligneous Conjunctivitis**

- Involves  
 1) conj (upper > lower)  
 2) buccal mucosa  
 3) nasopharynx  
 4) vagina, cervix  
 5) middle ear

### **Leber's optic atrophy**

- bilateral; mito-linked  
 A) *Ocular*  
 1) disc swelling  
 2) vision loss to 20/200  
 3) papillary telangiectasia  
 4) swelling of NFL  
 5) +/- hemorrhage or exudate  
 B) *Systemic*  
 1) deafness  
 2) migraine  
 3) vertigo  
 4) dementia

### **Lowe's syndrome (oculocerebral syndrome)**

- 1) x-linked recessive (males only)  
 2) female carrier: cortical lens opacities  
 A) *Ocular*  
 1) cornea: keloids  
 2) lens: cataracts; small, disc shaped lens  
 3) glaucoma  
 B) *Systemic*  
 1) renal rickets  
 2) amino aciduria  
 3) CNS: MR, seizures  
 4) MSK: growth retarded

### **Lyme disease**

- caused by *Borrelia Burgdorferi*  
 A) *Ocular*  
 1) conjunctivitis  
 2) stromal keratitis  
 3) iritis  
 4) pars planitis  
 5) posterior uveitis  
 6) papilledema  
 7) cranial nerve palsies  
 B) *Systemic*  
 1) skin rash  
 2) meningitis  
 3) cardiac disease

### **Stages of Lyme Disease**

- 1) *first month*  
 - skin: erythema chronicum migrans  
 - eye: follicular conjunctivitis  
 - constitutional symptoms  
 2) *1-4 months after*  
 - neurologic: meningitis  
 - MSK: arthritis  
 - cardiac: myocarditis  
 - ocular: uveitis, keratitis  
 3) *> 5 months*  
 - skin: atrophic skin changes  
 - ocular: keratitis  
 - neuro: chronic meningitis  
 - lungs: ARDS

### **Malignant Hyperthermia**

- 1) AD or spontaneous  
 2) 1 per 10000 (higher in kids)  
 3) triggered by:  
 i) inhalation anesthetics: halothane, enflurane, isoflurane  
 iii) muscle relaxant: succinylcholine

- 4) first sign:  
 i) tachycardia (most common)  
 ii) elevated end tidal CO<sub>2</sub>  
 iii) masseter spasm

### Marfan's syndrome (1:20000)

- defect: fibrillin
- Propanolol treatment for heart
- IV ABC's before surgery
- A) *Ocular*
  - 1) cornea: flat cornea; megalocornea
  - 2) lens: ectopia, spherophakia, earlier age of cataract
  - 3) iris: poor dilation, iridodonesis, atrophy of dilator muscle
  - 4) EOM: strabismus
  - 5) angle: COAG
  - 6) retina: RD's
- B) *Systemic*
  - 1) aortic root dilatation and aortic aneurysms
  - 2) upper: lower segment increased
  - 3) MV prolapse
  - 4) arachnodactily
  - 5) tall

### MEN IIB

- 1) enlarged corneal nerves
- 2) dry eyes
- 3) typical facies
- 4) Marfanoid habitus
- 5) submucosal neuromas
- 6) pheochromocytomas
- 7) medullary thyroid cancer

### MEWDS - described in 1984 (Jampol)

- 1) unilateral
- 2) females 4:1
- 3) outer retina, inner RPE
- 4) blurred vision, depressed fields
- 5) mild papillitis
- 6) may follow flu
- 7) good outcome (no scars left)

### Moebius

- nerves: CN 5-10,12 (typically 6 and 7)
- 1) absence of pectoralis (Poland anomaly)
  - 2) polydactily (extra digits)
  - 3) syndactily
  - 4) tongue defect
  - 5) hypoplastic limbs

- 6) Pierre Robin sequence

### Mucopolidoses

- defect in lysosomes

  - 1) cherry red spot
  - 2) corneal clouding

### Mucopolysaccharidosis (AAO cornea p.238)

- defect in carbohydrate metabolism
- Ocular findings:
  - 1) corneal clouding (except Hunter's and S-F)
  - 2) retinopathy
  - 3) optic atrophy
  - 4) papilledema
  - 5) Hurler's, Scheie's, Hunter's, Morquio, Sanfilippo

### Myotonic dystrophy (Steinert's)

- AD; 1 per 30000
- A) *Ocular*
  - 1) ptosis
  - 2) blepharitis
  - 3) christmas tree cataract
  - 4) pigmentary retinopathy
  - 5) pattern dystrophy
  - 6) light near dissociation
  - 7) sluggish (tonic), miotic pupils
  - 8) exodeviations
  - 9) decreased ERG
  - 10) ocular hypotony
- B) *Systemic*
  - 1) baldness
  - 2) testicular atrophy
  - 3) myotonia - don't release grip
  - 4) MR
  - 5) abnormal cardiac conduction
  - 6) skull abnormalities

### Multiple myeloma

- A) *Anterior segment*
  - 1) follicles
  - 2) cornea crystals
  - 3) copper in Descemet's
  - 4) iridocyclitis
- B) *Posterior segment*
  - 1) pars plana cysts → anterior lens displacement
  - 2) stasis retinopathy
  - 3) papilledema
  - 4) NV??

**Norrie's disease:**

- x-linked
- bilateral
- A) *Ocular*
  - 1) cornea opacifies
  - 2) cataract develops
  - 3) total retinal detachments
  - 4) retinal dysplasia
  - 5) phthisis by age 10
- B) *Systemic*
  - 4) deafness
  - 5) MR

**Ochronosis (1:250000)**

- A) *Ocular*
  - 1) scleral pigment (over muscles)
  - 2) peripheral corneal pigment
- B) *Systemic*
  - 1) brown urine
  - 2) skin pigment (axilla, genitals, over cartilage (nose, hands, ribs))
  - 3) joint disease (depos.)
  - 4) kidney disease (depos.)

**Ocular Cicatricial Pemphigoid**

- A) *Organs*
  - 1) mouth
  - 2) oropharynx
  - 3) genitals
  - 4) anus
  - 5) skin
  - 6) lids
- B) *Eyes*
  - 1) dry eyes
  - 2) loss of goblets
  - 3) shortened fornices
  - 4) symblephera
  - 5) rarely, EOM restriction
  - 6) entropion
  - 7) trichiasis

**Olivipontocerebellar atrophy**

- hereditary and sporadic forms
- 1) pigmentary retinopathy
- 2) CPEO
- 3) optic atrophy
- 4) ataxia
- 5) slurred speech
- 6) young adults

**Osteogenesis Imperfecta (1:20000)***Eye findings*

- 1) blue sclera
- 2) keratoconus
- 3) post. embryotoxon
- 4) megalocornea
- 5) Saturn ring - whitening of paralimbal sclera
- 6) papilledema/optic atrophy - from bone compression

**Paget's Disease of Bone**

- aka osteitis deformans
- heavily calcified bones
- exuberant osteoclastic reaction with a secondary osteoblastic response
- an elevated serum alkaline phosphatase

*A) Ocular*

- 1) angioid streaks
- 2) blue sclera

*B) Systemic*

- 1) pelvis
- 2) skull
- 3) femur
- 4) humerus

**Paraneoplastic Retinopathy***A) Ocular*

- 1) constricted VF
- 2) decreased night vision
- 3) photopsia
- 4) color blindness

*B) associated cancers*

- 1) oat cell
- 2) Hodgkin's

**Parinaud's dorsal midbrain***A) Findings*

- 1) light-near dissociation
- 2) lid retraction
- 3) convergence retraction nystagmus
- 4) decreased upgaze, later decreased downgaze
- 5) papilledema
- 6) skew deviation
- 7) impaired convergence?

*B) Causes*

- 1) pinealoma
- 2) hydrocephalus
- 3) vascular

- 4) mets
- 5) astrocytoma

### **Polyarteritis Nodosa**

#### A) *Ocular*

- 1) orbit vasculitis
- 2) scleritis
- 3) retinal vasculitis

#### B) *Systemic*

### **Prader Willi syndrome (1:20000)**

- deletion of 15 q11

#### A) *Ocular*

- 1) strabismus (common)
- 2) increased crossing of nerve fibers
- 3) nystagnus
- 4) glaucoma
- 5) ectropion uvea

#### B) *Systemic*

- 1) hypotonia
- 2) obesity
- 3) hypogonadism
- 4) short stature
- 5) small hands and feet

**PXE-** damage to elastic tissues (eg. blood vessels)

#### A) *Ocular*

- 1) angioid streaks
- 2) peau d'orange: pathognomonic
- 3) "punched out" retinal lesions
- 4) o.n. drusen?

#### B) *Systemic*

- 1) plucked chicken skin (neck, axilla, genital)
- 2) GI bleeding (b.v.)
- 3) CNS bleeds
- 4) peripheral vascular disease

### **Reiter's syndrome**

- occurs after G- dysentery or chlamydia urethritis

#### A) *Ocular*

- 1) conjunctivitis: 50%
- 2) iritis: 10%
- 3) keratitis
- 4) corneal infiltrates
- 5) corneal pannus

#### B) *Systemic*

- 1) urethritis
- 2) polyarthritis
- 3) fever

- 4) scaling skin eruption: keratoderma blenorrhagicum
- 5) balanitis
- 6) aphthous ulcers
- 7) lymphadenopathy
- 8) pneumonitis
- 9) pericarditis

### **Refsum's Disease**

- phytanic acid storage disease

#### A) *Ocular*

- 1) atypical RP \*\*
- 2) cataracts (rare)
- 3) pupil abnormalities

#### B) *Systemic*

- 1) polyneuritis
- 2) dry skin
- 3) ataxia
- 4) deafness
- 5) anosmia

### **Rubinstein-Taybi syndrome (1:30000)**

- sporadic

#### A) *Ocular*

- 1) antimongoloid slant
- 2) NLD obstruction
- 3) cataract
- 4) strabismus
- 5) congenital glaucoma

#### B) *Systemic*

- 1) CNS: MR
- 2) broad thumbs
- 3) beaked, broad nose
- 4) ear anomalies
- 5) cong. heart disease
- 6) lung anomalies
- 7) MSK anomalies
- 8) GU anomalies

### **Sarcoidosis**

- Mikulicz's syndrome (dry mouth and eyes)

- Heeford's syndrome

#### A) *Ocular*

- 1) lids: granulomas
- 2) sclera: scleritis, episcleritis
- 3) iris: acute and chronic iritis
- 4) choroid: choroiditis with D-F nodules, SRNV
- 5) retina: - retinal vasculitis; seafans (NV)
- 6) disc: NVD, disc edema

**no** EOM or orbital c.t. involvement

**B) Systemic**

- 1) hilar lymphadenopathy
- 2) erythema nodosum
- 3) hepatosplenomegaly
- 4) arthritis
- 5) lymphadenopathy
- 6) cardiac abnormalities
- 7) myositis
- 8) meningitis with CN2 and CN 7 involved

**Sickle cell findings****A) Ocular Findings**

- 1) comma shaped conj. vessels
- 2) higher IOP with hyphema
- 3) salmon patch
- 4) iridescent spots
- 5) black sunburst
- 6) sea fans, NV(SC, SThal)
- 7) autoinfarcts
- 8) vitreous hemorrhage
- 9) CRAO (SS)
- 10) ON damage with moderately elevated IOP

**Sjogren's**

- lacrimal gland infiltration with tear deficiency and either xerostomia or a CTD
- young to middle age women

**A) CTD associated**

- 1) RA
- 2) SLE
- 3) PAN
- 4) others

**B) Systemic findings**

- 1) swollen lacrimal gland
  - 2) swollen parotid gland
  - 3) exocrine dysfunction
  - 4) atrophy of gastric mucosa
  - 5) atrophy of vaginal mucosa
  - 6) kidney infiltration
  - 7) muscle infiltration
  - 8) lung infiltration
  - 9) Raynaud's
  - 10) arthralgias
  - 11) myalgias
  - 12) fever
- C) Serology**
- 1) ANA
  - 2) anti SS-A or SS-B

**Sticklers (1:20000)**

(eye findings only: Wagner's)

- AD

**A) Ocular**

- 1) lens sublux. (rare), early cataracts
- 2) retina: lattice, RD, sheathing, pigment around vessels
- 3) myopia
- 4) glaucoma (5%)
- 5) optically empty vitreous

**B) Systemic**

- 1) Pierre Robin anomaly
- 2) joint abnormalities
- 3) MV prolapse
- 4) deafness
- 5) cleft palate
- 6) abnormal facies

**Temporal arteritis findings****A) Ocular**

- 1) AION
- 2) CRAO
- 3) CN 3, 4, 6 palsy
- 4) anterior segment ischemia
- 5) hyperemia of the conjunctiva
- 6) iris rubeosis
- 7) cataract
- 8) considerable disc cupping
- 9) corneal edema

**B) Systemic**

- 1) TIA's
- 2) occipital infarct
- 3) PMR
- 4) MI
- 5) CVA
- 6) ESR normal in 10%
- 7) biopsy + for 2 weeks after steroids started
- 8) biopsy: 3 cm long

**Trisomy 13 (Patau Syndrome)****A) Ocular**

- 1) globe: synophthalmos/cycloopia, coloboma with cartilage, PHPV, microphthalmia
- 2) retina: *retinal dysplasia* (multilayered rosettes)
- 3) lens: cataract (retained nuclei?)
- 4) cornea: clouding
- 5) uvea: coloboma
- 6) *optic nerve hypoplasia*, atrophy
- 7) lids: epicanthal folds, no eyebrows

**B) Systemic**

- 1) polydactily
- 2) cleft lip/palate
- 3) GU abnormalities

### Trisomy 18 (Edwards)

- 1) lids: epicanthal folds, ptosis, blepharophimosis
- 2) glaucoma
- 3) microphthalmos
- 4) coloboma
- 5) corneal opacities
- 6) blue sclera

### Trisomy 21 (Down)

#### A) Systemic

- 1) protruding tongue
- 2) flat nasal bridge
- 3) low malformed ears
- 4) MR
- 5) simian crease

#### B) Ocular findings

- 1) lids: epicanthal fold, blepharitis, mongoloid slant, ectropion, telecanthus, sparse lashes
- 2) conj: conjunctivitis
- 3) cornea: keratoconus with hydrops, prominent nerves
- 4) lens: cataracts
- 5) iris: Brushfield's - hypercellular iris stroma
- 6) angle: glaucoma
- 7) retina: pigmentary retinopathy, macular hypoplasia ?, RD's?, abnormal vessels?
- 8) optic nerve: atrophy
- 9) EOM: strabismus (esp ET), nystagmus
- 10) refraction: hyperopia and myopia

### Turner's (45 XO)

#### A) Ocular

- 1) sclera: blue color
- 2) lens: ant. embryonal cataract
- 3) pupillary heterotropia
- 4) nystagmus
- 5) strabismus

#### B) Systemic

- 1) shield chest
- 2) MR

### Usher's syndrome

- several types ranging in severity
- AR
- 1) pigmentary retinopathy

- 2) deafness

### Vertebrobasilar insufficiency

#### A) Ocular

- 1) horizontal gaze palsy
- 2) vertical gaze palsy
- 3) INO
- 4) skew deviation
- 5) CN 3,4, or 6 palsy
- 6) Horner's
- 7) nystagmus

#### B) Systemic

- 1) drop attacks
- 2) ataxia
- 3) vertigo
- 4) vomiting
- 5) tinnitus
- 6) deafness
- 7) dysarthria
- 8) dysphagia
- 9) hemiparesis
- 10) hemisensory deficit

### VKH

- a panuveitis
- choriocapillaris involved

#### A) Integumentary

- 1) vitiligo
- 2) poliosis
- 3) alopecia areata

#### A) Anterior segment

- 1) mutton-fat KP's
- 2) iritis
- 3) iris nodules
- 4) synechiae

#### B) Posterior Segment

- 1) vitritis
- 2) optic disc swelling
- 3) retinal edema and hemorrhages
- 4) exudative RD
- 5) choroidal infiltrates
- 6) RPE infiltrates
- 7) deep orbital pain
- 8) depigm. of the perilimbus (Sugiura's sign)
- 9) depigm. of the fundus (sunset glow fundus)

#### C) Systemic

- 1) headaches
- 2) nausea
- 3) slight fever
- 4) meningitis

- 5) tinnitus
- 6) deafness
- 7) hyperesthesia of skin

### **Waardenburg's**

- AD
- 1) telecanthus
- 2) confluent eyebrow
- 3) heterochromia
- 4) deafness
- 5) white forelock
- 6) pale fundus

### **Wegener's areas of involvement**

- A) *Ocular*
  - 1) orbit vasculitis
  - 2) scleritis
  - 3) retinal vasculitis
- B) *Systemic*
  - 1) kidney
  - 2) lung
  - 3) sinuses
  - 4) positive C-ANCA

### **Weil-Marchesani (1:100000)**

- A) *Ocular*
  - 1) microspherophakia (→ pupil block and ACG)
- Rx: Meds: dilate; Surgery: PI
- 2) myopia
- B) *Systemic*
  - 1) short stature
  - 2) short stubby hands and feet
  - 3) normal intelligence

### **Wilson's**

- AR
- decreased ceruloplasmin
- Tx: penicillamine, decreased copper intake
- DDx: primary biliary cirrhosis, hepatitis, chalcosis, cholestasis of childhood
- A) *Ocular*
  - 1) K-F ring
  - 2) sunflower cataract
- B) *Systemic*
  - 1) liver cirrhosis
  - 2) CNS: basal ganglia (PD-like)
  - 3) kidney

### **Xeroderma Pigmentosum**

- A) *Ocular*

- 1) entropion, ectropion
- 2) SCC, BCC of lids
- 3) dry eye
- 4) conj. melanosis and melanoma
- B) *Systemic*
  - 1) skin: dryness, scaling, freckling
  - 2) malignant skin tumors
  - 3) neurologic abnormalities (MR)
  - 4) deafness

#### 4) Ocular syndromes

##### Acute angle closure glaucoma

- 1) cornea: endothelial damage
- 2) iris: atrophy, PAS, and post synechia
- 3) lens: glaucomflecken, cataract
- 4) c.b.: hyosecretion
- 5) optic nerve: atrophy, no cupping

##### Aniridia

- AD: 2/3; sporadic: 1/3
- 1/60000
- chromosome 11
- VA < 20/200
- assoc: Wilm's, MR, GU abn.
- U/S or IVP every 3 months

##### Signs

- 1) cornea: pannus (epithelial metaplasia), keratoconus (Duanes')
- 2) angle: angle closure glaucoma
- 3) iris: hypoplasia
- 4) lens: subluxation, cataract
- 5) retina: foveal hypoplasia
- 6) optic nerve: hypoplasia
- 7) neuro: nystagmus
- 8) EOM: strabismus

#### Anterior Segment dysgeneses

##### A) Post. embryotoxon

- AD
- anterior displaced Schwalbe's line

##### B & C) Axenfeld-Rieger Syndrome

- new terminology
- AD (75%); 25% sporadic
- bilateral?

##### B) Axenfeld Anomaly

###### i) Ocular

- 1) posterior embryotoxin
- 2) iris process to SL
- 3) glaucoma ("Axenfeld's syndrome")

###### ii) Systemic

- 1) MSK abnormalities

##### C) Rieger's

###### i) Ocular

- 1) Axenfeld's
- 2) iris stroma defects
- 3) glaucoma (50%)
- 4) corectopia

- 5) ectropion uvea

###### 6) PAS

###### 7) pseudopolicoria

###### ii) Systemic ("Rieger's syndrome")

- 1) facial, skeletal, dental abnormalities
- 2) umbilical hernia

##### D) Peter's

- AD, AR or sporadic
- bilateral: 80%

###### A) Ocular

- 1) loss of central Decemet's and endothelium
- 2) +/- lens sticking to stroma
- 3) glaucoma (50%)

###### B) Systemic ("Peter's plus" syndrome)

- 1) cardiac defects
- 2) cleft lip/palate
- 3) MSK defects
- 4) craniofacial anomalies

##### E) Von-Hippels internal ulcer

- Peter's associated with intrauterine inflammation
- may see uveitis, KP's, pannus after birth

##### F) circumscribed posterior keratoconus

- localized indentation of posterior cornea
- unilateral
- sporadic

##### Benign papillophlebitis

- 1) unilateral
- 2) normal field
- 3) normal acuity
- 4) like partial CRVO
- 5) peripheral retinal NFL heme
- 6) resolves on its own
- 7) young people

##### Best's Disease

- AD
- EOG abnormal with normal ERG
- lipofuscin deposits

##### Stages:

- 1) RPE changes
- 2) yolk
- 3) scrambled or fried egg
- 4) pseudohypopion
- 5) atrophy

**Blepharitis (Rosacea)**

- type IV response

A) *Systemic*

- 1) skin telangiectasias
- 2) rhinophyma
- 3) skin pustules
- 4) fair skinned
- 5) skin erythema
- 6) skin papules
- 7) hypertrophic sebaceous glands
- 8) malar rash
- 9) flushing with alcohol, coffee

B) *Lid*

- 1) m.g. plugging
- 2) debris (scurf)
- 3) lid thickening (tylosis)
- 4) chalazia

C) *Ocular*

- 1) conjunctivitis
- 2) corneal pannus
- 3) punctate epithelial erosions
- 4) dry eye
- 5) peripheral infiltrates (10:00, 2:00, 4:00, 8:00)
- 6) sterile ulcer
- 7) episcleritis
- 8) iritis

**Blepharitis (Staph)**

- F>M

A) *Lid*

- 1) poliosis, trichiasis, madaurosis
- 2) ant. blepharitis
- 3) collarettes
- 4) lid ulceration
- 5) lid thickening (tylosis)

B) *Ocular*

- 1) phlyctenules (rxn to staph cell wall Ag)
- 2) marginal infiltrates (spade-shape)
- 3) pannus
- 4) conjunctivitis
- 5) dry eye

**Carotid-Cavernous fistula**

- head trauma: 75%

- spontaneous: 25%

## A) anterior segment

- 1) injected conjunctival and episcleral vessels (corkscrew)
- 2) elevated IOP

3) anterior segment ischemia in 20% (cells, flare, corneal edema, cataract, rubeosis)

## B) Posterior segment

- 1) CRVO
  - 2) ocular ischemic syndrome?
- C) Orbit
- 1) pulsating proptosis
  - 2) enlarged EOM's
  - 3) CN 6 palsy (compression in cav. sinus): 50%
  - 4) CN 3, 4 palsy less common
  - 8) bruit

**Central Areolar Dystrophy**

- 1) AD
- 2) symptoms start at 40-50
- 3) decreased ERG and EOG

**Ciaccia syndrome**

- 1) large ET
- 2) cross fixate
- 3) nystagmus when fixing eye abducts
- 4) often undercorrected
- 5) need large recessions

**CSNB**

- 3 types: XL, AD, AR

- 1) vision 20/20 to 20/200
- 2) no nystagmus
- 3) poor nite vision
- 4) problem with on/off connections

**Diabetes ocular findings (less known)**A) *Ant segment*

- 1) decreased corneal sensation
- 2) dry eye
- 3) corneal opacity
- 4) epithelial abnormalities
- 5) Descemet's folds ("wrinkles")
- 6) ectropion uvea
- 7) xanthelasma

B) *Posterior segment*

- 1) sheathing
- 2) Roth spots
- 3) macular dragging
- 4) diabetic papillopathy
- 5) lipemia retinalis (white vessels from fat)
- 6) c.b. b.m. thickening
- 7) choriocapillaris b.m. thickening
- 8) asteroid hyalosis

**Ectopia lentis et pupillae**

- AR
- 1) large cornea
- 2) lens and pupil move opposite dir
- 3) cataract
- 4) iris transillumination
- 5) poor dilation
- 6) slit pupil
- 7) RD
- 8) high myopia

**Esotropia (Infantile, congenital, essential)**

- 1) deviation > 30 PD
- 2) FH common
- 3) DVD (75%)
- 4) latent nystagmus (50%) - horiz or rotat.
- 5) IOOA (70%)
- 6) amblyopia (50%)
- 7) cross-fixating
- 8) assymetric OKN's
- 9) monocular smooth pursuit asymmetry

**Esotropia (Accomodative)**

- 1) onset 6 months - 7 years
- 2) usual 2 - 3 years
- 3) hereditary
- 4) hyperopes (+3 to +10; average = +4)
- 5) amblyopia
- 6) most common ET
- 7) ET moderate (20-30 PD)
- 8) near-distance difference < 10 PD

**Exfoliation syndrome**

- 1) peripupillary atrophy
- 2) deposits: lens, cornea, iris, conj, c.b., zonules, angle
- 3) increased pigment dispersion with dilation
- 4) weak zonules
- 5) may result in CACG as well as COAG
- 6) prevalence of glaucoma: 7% and OHT 15% (Yanoff study)
- 7) normal response to steroids (25% vs 90% in COAG)
- 8) W > B
- 9) often unilateral

**Fuch's iridocyclitis**

- 1) stellate KP's
- 2) unilateral (usually)
- 3) minimal AC reaction

- 4) vitritis
- 5) heterocheomia
- 6) no synechia
- 7) cataract
- 8) glaucoma

**Giant Papillary Conjunctivitis**

- 1) CL intolerance
- 2) discharge
- 3) blurred vision
- 4) conj injection
- 5) papillae > 0.3 mm
- 6) bloody tears
- 7) ptosis
- 8) lens displaced

**Goldman Favre**

- AR
- 1) cataracts
- 2) peripheral and foveal retinoschisis
- 3) RP-like changes
- 4) optic atrophy

**Gyrate Atrophy**

- AR
- classic scalloped RPE lesions
- 1) myopia
- 2) cataract (PSCC)
- 3) night blindness at 10 y.o.
- 4) present to MD at 20-30 y.o.
- 5) decreased VA
- 6) decreased VF
- 7) extinguished ERG
- 8) ornithine levels increased 10 fold
- 9) B6, diet control

**ICE syndromes**

- 1) unilateral
- 2) F > M; W > B
- 3) epithelialization of endothelium (seen with specular microscopy)
- 4) PAS
- 5) corectopia
- 6) iris atrophy (greatest in progressive iris atrophy)
- 7) corneal edema (greatest in Chandler's)
- 8) young age

**Keratoconus**

signs

- 1) Munson's sign
- 2) Rizzuti's sign
- 3) Fleisher ring
- 4) Vogt's stria
- 5) breaks in Bowman's
- 6) enlarged corneal nerves
- 7) hydrops

### **Lattice Dystrophy**

- 1) AD
- 2) recurrence in graft: lattice > granular > macular

### **Leber's Congenital Amaurosis**

- AR
- 2-3 months of age when noticed
- absence of rods and cones
- oculodigital reflex

#### A) *Ocular*

- 1) cataracts
- 2) glaucoma
- 3) paradoxical pupils
- 4) RP-like changes
- 5) blond fundus
- 6) macular atrophy
- 7) vessel attenuation
- 8) pigmentary retinopathy
- 9) optic nerve pallor
- 10) sensory nystagmus
- 11) hyperopia

#### B) *Systemic*

- 1) renal anomalies
- 2) MSK anomalies
- 3) brain anomalies

### **Megalocornea**

- XL (90% male)

#### A) *Systemic Associations*

- 1) MR
- 2) craniosyntosis
- 3) Down's
- 4) Alport's
- 5) Marfan's
- 6) short stature
- 7) frontal bossing

#### B) *Ocular associations*

- 1) 13-16 mm corneas
- 2) cataract
- 3) ectopia lentis
- 4) goniodysgenesis

- 5) microcoria
- 6) miosis
- 7) glaucoma
- 8) lipid arcus

### **Microcornea**

- AD and AR

#### A) *Systemic Associations*

- 1) myotonic dystrophy
- 2) fetal alcohol
- 3) Ehlers Danlos

#### B) *Ocular associations*

- 1) PHPV !
- 2) cataracts
- 3) goniodysgenesis
- 4) ON hypoplasia
- 5) ACG
- 6) COAG

### **Monofixation (microtropia) Syndrome**

#### A) *Findings*

- 1) ET less than 8 PD on cover-uncover
- 2) deviation may be > 10 PD on alternate cover
- 3) amblyopia (usually slight)
- 4) macular scotoma on binocular testing
- 5) poor stereopsis
- 6) central scotoma, peripheral fusion
- 7) ARC (extramacular)
- 8) normal cover/ uncover
- 9) usually abnormal 4 base out prism test

#### B) *Causes*

- 1) strabismus (corrected)
- 2) anisometropia
- 3) amblyopia
- 4) macular scar

### **Mooren's ulcer**

precipitating factors: surgery, trauma, or parasite

#### A) *Limited Type*

- 1) unilateral (75%)
- 2) older patients
- 3) M=F
- 4) slow course
- 5) mild pain
- 6) responds to treatment

#### B) *Progressive Type*

- 1) bilateral (75%)
- 2) young, black (nigerian)
- 3) males
- 4) rapidly progressive

- 5) severe pain
- 6) poor response to treatment

#### Nonarteritic AION

- 1) older
- 2) visual loss less severe
- 3) 2<sup>nd</sup> eye affected in 25%
- 4) hypertension and DM associated
- 5) 5-10% progress over 2-3 weeks

#### Oculomotor Apraxia

##### A) Findings

- 1) M > F
- 2) inability to generate horizontal saccades
- 3) vertical saccades normal
- 4) head thrust past target and then refixate
- 5) VOR impaired
- 6) OKN abnormal
- 7) congenital type may improve with age

##### B) Associations and Causes

- 1) idiopathic
  - 2) corpus callosum agenesis
  - 3) hydrocephalus
  - 4) cerebellar lesion
  - 5) bilateral lesions of frontoparietal cortex
- need CT!

#### Optic pit

- 1) usually unilateral (85%)
- 2) serous RD (40%)
- 3) disc may be large
- 4) macular holes
- 5) retinal hemorrhages
- 6) presents age 20-40
- 7) FA leakage
- 8) theories on source of fluid:
  - i) vitreous (most likely)
  - ii) choriocapillaris
  - iii) CSF
  - iv) macular hole

#### 3 Types of perifoveal telangiectasias

##### I) unilateral parafoveal

- congenital and acquired
- males more often
- like local Coats'
- laser helps in type 1 only

##### II) bilateral parafoveal

- SRNV as complication
- most common

associations:

- 1) small, yellow lesions within the FAZ
- 2) right-angle retinal venules
- 3) stellate plaques of RPE hyperplasia
- III) *bilateral perifoveal with capillary obliteration*

#### Pars Planitis sequelae

- 1) cataract
- 2) glaucoma
- 3) CME
- 4) RD
- 5) VH
- 6) NV
- 7) band
- 8) phthisis
- 9) disc edema

#### PHPV

(retrol. mass of fat, SM, collagen, cartil.)

- 1) microphthalmos, microcornea
- 2) cataract
- 3) angle closure glaucoma
- 4) usually unilateral
- 5) elongated ciliary processes

#### Pigment dispersion syndrome

- 1) M > F; W > B
- 2) young
- 3) myope
- 4) Krukenberg spindle
- 5) lens, angle, iris, zonule pigment deposits
- 6) midperipheral atrophic spokes
- 7) worse after exercise (dilation)
- 8) glaucoma develops in 33% (one third)
- 9) may be steroid responders (1 study yes, 1 no)
- 10) may be related to COAG (1 study yes, 1 no)
- 11) blinking causes increased IOP (pupil block?)
- 12) Tx: PI, pilo (usually not tolerated), IOP control, ALT

#### Plateau Iris

- 1) deep central AC
- 2) narrow angle
- 3) configuration: PI curative
- 4) syndrome: PI not curative

#### Posner Schlossman

- 1) recurrent
- 2) IOP 40-60
- 3) due to PG abnormality

- 4) ciliary flush
- 5) mild AC rxn
- 6) sluggish pupil
- 7) epithelial edema
- 8) flare and a few KP's
- 10) no PAS
- 11) some have steroid response like COAG
- 12) Tx. steroids, NSAIDs (when to treat?)

### Posterior Polymorphous Dystrophy

- AD or AR
- 1) broad bands with scalloped edge
- 2) gray geographic lesions
- 3) stromal edema
- 4) corectopia
- 5) iridocorneal adhesions

### Posterior scleritis

- 1) pain
- 2) proptosis
- 3) visual loss
- 4) restricted EOM movements
- 5) lower lid retraction on upgaze
- 6) choroidal folds
- 7) exudative RD
- 8) papilledema
- 9) ACG
- 10) posterior uveitis (vitritis)
- 11) thickening on CT/MRI
- 12) sometimes associated TB or c.t. disease

### Reis Buckler's

- AD
- 1) areas of absent epith b.m
- 2) recurrent erosions
- 3) recurrence in graft
- 4) subepithelial honeycomb pattern
- 5) scarring

### Retinopathy of Prematurity

- 1) myopia
- 2) RD
- 3) glaucoma
- 4) cataract
- 5) phthisis bulbi

### Retinoschisis - Congenital

- 1) macular schisis (100%)
- 2) peripheral schisis (50%)
- 3) vitreous vascular veils

- 4) poor vision
- 5) nystagmus
- 6) strabismus
- 7) vitreous hemorrhage
- 8) normal EOG, decreased b wave on ERG

### Shaken baby syndrome (order of frequency)

- 1) intraocular hemorrhage (preretinal and retinal heme)
- 2) lid ecchymosis
- 3) RD, retinal dialysis
- 4) cataract/subluxated lens
- 5) papilledema / optic atrophy
- 6) subconj. heme
- 7) esotropia

### Spasmus Nutans

- 1) unilateral or bilateral nystagmus
- 2) begins before 1 year
- 3) usually finishes prior to 3 years
- 4) no nystagmus during sleep
- 5) torticollis
- 6) head nodding
- 7) begins between 4 and 18 months of age
- 8) horizontal, torsional, or vertical nystagmus

### Spherophakia

Associations: Marfan's, Weil Marchesani

Findings

- 1) myopia
- 2) pupil block, worse with miotics
- 3) Tx: mydriatics

### Stargardt's ("juvenile mac deg.")

- most common hereditary maculopathy

- AR

- 1) present as young children
- 2) vision decreases to 20/200 by age 20, but one eye may remain 20/70 -20/100
- 3) pattern ERG: abnormal
- 4) full field ERG: ?
- 5) EOG: normal ?

### Superior limbic keratitis

- 1) F > M
- 2) recurs over 1-10 years
- 3) often bilateral
- 4) papillary rxn
- 5) thickened superior limbus
- 6) staining of superior conj.

- 7) filaments
- 8) associated with thyroid (50%)

### **Therrien's marginal degeneration**

- 1) unil or bilateral
- 2) 20-40 y.o.
- 3) begin's at 12:00
- 4) epithelium intact
- 5) pannus over area of thinning
- 6) perforation with mild trauma
- 7) lipid line at anterior edge
- 8) against the rule astigmatism (like wound gape)
- 9) Tx: lamellar graft

### **Thygeson's SPK (1950)**

- 1) multiple gray/granular epithelial opacities
- 2) lesions stain with both fluorescein and rose bengal (mild fluor. stainig?) - minimal staining with fluo.
- 3) quiet eye
- 4) rare subepithelial opacity
- 5) usually bilateral
- 6) symptoms: f.b. sensation, reduced vision, photophobia
- 7) recurrent episodes
- 8) respond to steroids or CL

## 5) Phacomatoses

### 1) Neurofibromatosis (Von Recklinhausen's)

#### A) NF type I

- 1 per 3500

chromosome 17 (AD)

##### A) Systemic features

- 1) neurofibroma
- 2) neurofibroma (Schwannoma)
- 3) axillary and inguinal freckling
- 4) cafe au lait spots
- 5) sphenoid hypoplasia
- 6) skeletal deformities
- 7) seizures
- 8) hemiparesis
- 9) MR
- 10) hemiatrophy
- 11) hemifacial hypertrophy
- 12) pheochromocytoma
- 13) molluscum fibrosa?

##### B) Ocular features

- 1) neurofibromas: eyelids, conjunctiva, iris, c.b., and choroid
- 2) enlarged corneal nerves
- 3) glaucoma (50% if lid tumor)
- 4) iris melanocytic hamartoma
- 4) retinal glial hamartoma
- 5) optic nerve glial hamartoma
- 6) optic nerve gliomas, meningioma
- 7) lid plexiform neurofibromas
- 8) ectropion uvea
- 9) conj. melanomas
- 10) uveal melanoma (some studies)
- 11) thickening of fibrous elements of choroid

##### Criteria for NF-1

- two or more of following :

- 1) 6 or more cafe au lait spots (>5 mm <13 years old; >15 mm > 13 years old)
- 2) two or more neurofibromas (or 1 if plexiform)
- 3) axillary or inguinal freckling
- 4) optic nerve glioma (NF 1)
- 5) two or more Lisch nodules (melanocytic hamartomas); by age 6 >90% have them
- 6) osseous lesion (eg. sphenoid dysplasia)
- 7) first degree relative with NF

#### B) NF type II

- 1 in 50,000

chromosome 22 (AD)

##### A) Systemic findings

- 1) acoustic neuroma (Schwannoma)
  - 2) neurofibroma
  - 3) schwannoma
  - 4) meningioma
  - 5) deafness
  - 6) café au lait
  - 7) axillary freckling - uncommon
- ##### B) Ocular
- 1) PSCC - 50%
  - 2) Lisch nodules - uncommon
  - 3) combined hamartoma of retina
  - 4) optic nerve glioma not seen (or rare)

##### Criteria for NF-2

- 1) bilat. CN 8 masses
- or
- 2) 1<sup>st</sup> degree relative with NF-2 with either
- a) unilateral CN 8 mass
- or
- b) two of
- 1) neurofibroma
  - 2) schwannoma
  - 3) meningioma
  - 4) PSCC (not glioma)
- gliomas not seen in NF 2

### Associated Malignancies?? someone's notes

- 1) pit tumors
- 2) melanoma
- 3) breast ca
- 4) pheo
- 5) GI cancer
- 6) GU cancer

### 2) Tuberosus sclerosis (Bourneville's)

- AD

- chromosome 9

#### A) Systemic

- 1) MR (60%)
- 2) seizures (93%) - EEG abnormal?
- 3) "adenoma sebaceum" (80%) - angiofibroma
- 4) ash leaf (80%)
- 5) shagreen patch
- 6) astrocytic hamartoma of brain ("brain tubers")
- 7) cardiac rhabdomyoma
- 8) renal angioliipoma
- 9) subungual fibroma
- 10) periventricular calcifications
- 11) bone lesions

#### B) Ocular

- 1) astrocytic hamartoma (retina or ON head) = "mulberry" or "fish eggs" and calcified
- 2) hypopigmented uveal lesions

### 3) Angiomatosis Retinae (Von-Hippel Lindau)

- AD (20% are); variable penetrance
- chromosome 3
- 1/100000
- A) *Systemic*
  - 1) cerebellar hemangioblastoma (-Lindau)
  - 2) pheochromocytoma
  - 3) renal cell carcinoma assoc.
  - 4) cysts - pancreas, epididymis
- B) *Ocular*
  - 1) retinal capil. hemangioma (leaks) with large feeder vessels
  - 2) bilateral in 50%
  - 3) leaky vessels in lesion
  - 4) serous RD common
  - 5) cataract and glaucoma secondary to RD
  - 6) Tx. Lesions with cryo or laser
  - 7) annual exams for children

### 4) Encephalotrigeminal angiomatosis (Sturge Weber)

- sporadic
- A) *Systemic*
  - 1) "port wine" stain (cavernous hemangioma) of face
  - 2) seizures (80%)
  - 3) MR (60%)
  - 4) intracranial calcifications ("railroad track")
  - 5) treat skin with pulsed dye laser
- B) *Ocular*
  - 1) choroidal cavernous hemangioma (40%)
  - 2) glaucoma (30%)
  - 3) iris heterochromia (darker on involved side)
  - 4) serous RD

### 5) Racemose Hemangioma (Wyburn Mason)

- non hereditary
- 1) A-V malformation of brain
- Ocular
  - 1) A-V malformation
  - 2) unilateral
  - 3) secondary NV glaucoma
  - 4) hemorrhage

### 6) Ataxia Telangiectasias (Louis Bar)

- AR
- 1 per 40000
- chromosome 11; related to DNA repair
- A) *Systemic*
  - 1) CNS: dysarthria, chorea
  - 2) testicular/ovarian atrophy
  - 3) skin telangiectasias
  - 4) hypo and hyper pigmentation skin
  - 5) poor saccades (like cong. OM apraxia)
  - 6) dystonia
  - 7) hypoplastic thymus and T cell malfunction
  - 8) decreased IgA and prone to infections
  - 9) increased lymphoma and leukemia
  - 10) women carriers have 7 fold increased risk of breast CA (10% of all breast CA)

#### B) *Ocular*

- 1) telangiectasias of conjunctiva (not retina) and lid
- 2) nystagmus
- 3) strab
- 4) oculomotor apraxia
- 5) decreased OKN

### 7) Retinal cavernous hemangioma

- "retinal-neuro-cutaneous cavernous hemangioma" aka "Weskamp Cotlier" syndrome
- few AD families
- 1) CNS cavernous hemaingiomas
- 2) skin cavernous hemangiomas
- refer to neurology because at risk for bleeds from cerebral hemangiomas

### 8) Incontinentia Pigmenti (Yanoff p.692)

- incontinence of melanin
- X-linked
- A) *Systemic*
  - 1) skeletal
  - 2) teeth
  - 3) CNS (MR, seizures)
- B) *Ocular*
  - often unilateral or assymetric
  - most signs (3-8) are sequelae of RD
  - 1) corneal scarring
  - 2) conj. pigmentation
  - 3) cataracts
  - 4) strabismus
  - 5) nystagmus
  - 6) microphthalmos
  - 7) glaucoma

8) uveitis

9) blue sclera

10) retina: retrolental mass (from ischemia or inflammation - unknown); like ROP; leads to RD

- Tx: laser or cryo to avascular retina