INHERITED RETINAL DISEASES

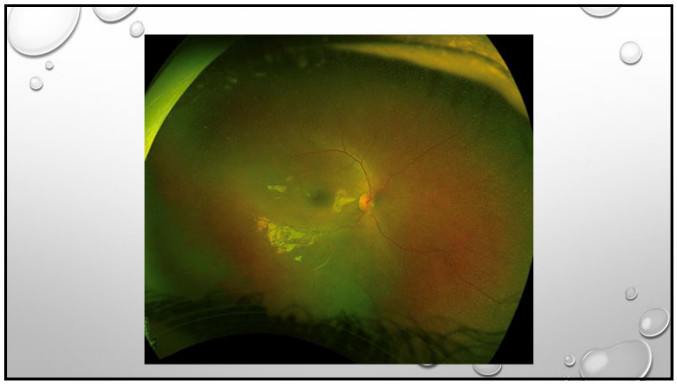
JASON CROSSON MD

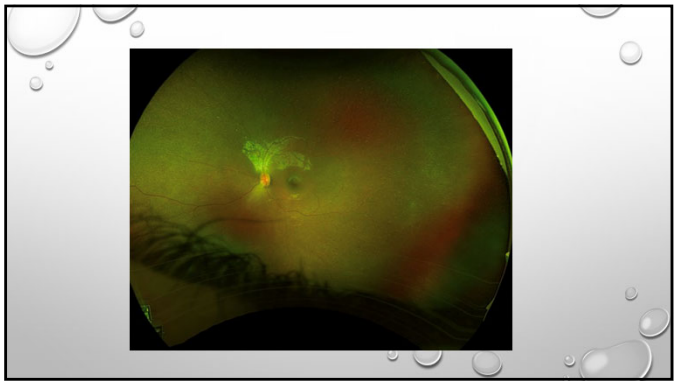
RESOURCES

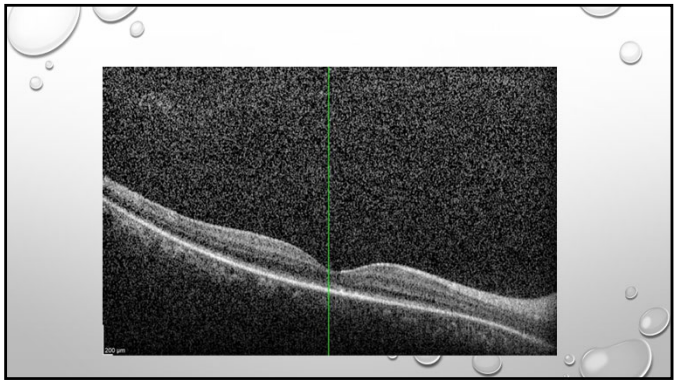
- STONEROUNDS.ORG
- FOUNDATION FOR FIGHTING BLINDNESS CLINICAL TRIALS PIPELINE

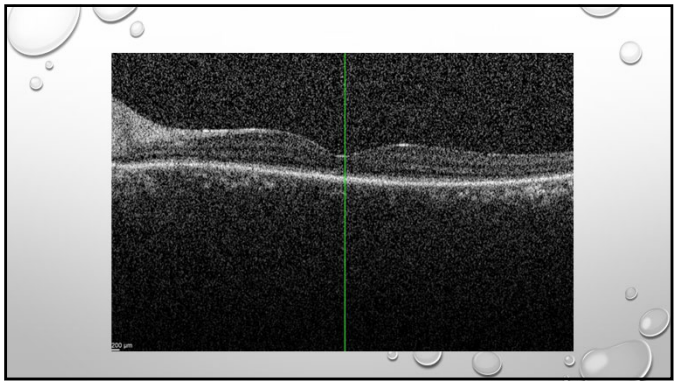
TAKE HOME POINTS

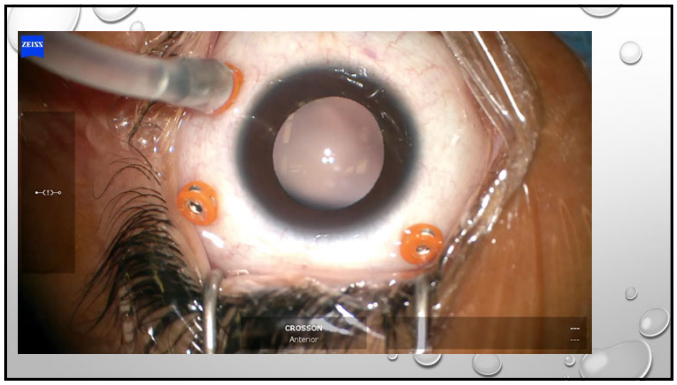
- BASIC FRAMEWORK FOR IRD'S
- DO GENETIC TESTING











DO GENETIC TESTING

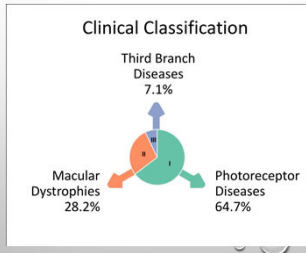
WHY ARE IRD'S DAUNTING?

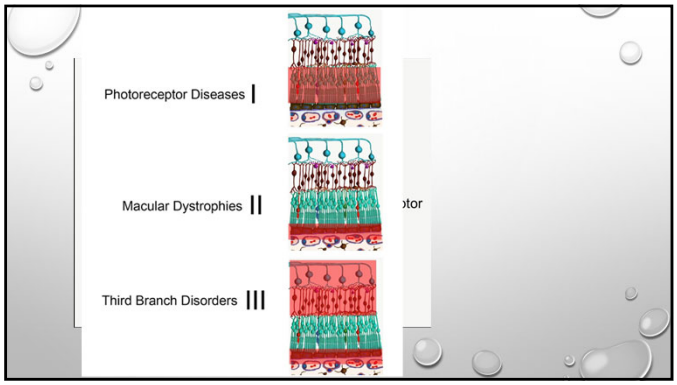
- THEY ARE RARE
- THERE ARE SO MANY
- OVERLAPPING FEATURES

WHAT SHOULD MAKE US SUSPECT AN IRD?

- ONSET BY THE AGE OF 40
- USUALLY BILATERAL
- SYMMETRIC (1 EYE MIRROR IMAGE OF THE OTHER)
- AFFECTED FAMILY MEMBERS

IRD'S MADE SIMPLE





PHOTORECEPTOR DISEASE

- IS IT CONGENITAL/STATIONARY OR AQUIRED/PROGRESSIVE?
- ARE THERE FEATURES OUTSIDE THE EYE?
  - SYNDROMIC VS. NON SYNDROMIC

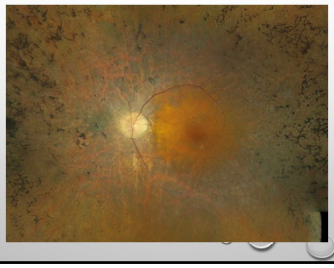
I - Photoreceptor Disease
- A – Isolated
  - 1 – Acquired/Progressive
    - a – Retinitis Pigmentosa
    - b – Cone & C/R Dystrophy
  - 2 – Congenital/Stationary
    - a – LCA
    - b – SECORD
    - c – ECORD
    - d – Achromatopsia
- B – Syndromic
  - 1 – Usher Syndrome
  - 2 – Bardet-Biedl Syndrome

II – Macular Diseases
- A – AR Stargardt Disease
- B – Best Disease
- C - Pattern Dystrophy

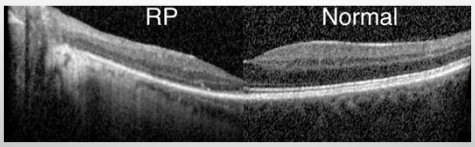
III – Third Branch Disorders
- A1 – Choroideremia
- B1 – XL Retinoschisis
- C1a – Dominant Optic Atrophy

NONSYNDROMIC PHOTORECEPTOR DISEASE

60 YO M WITH POOR NIGHT VISION AND CONSTRICTED VISUAL FIELD



LOSS OF ELLIPSOID ZONE AND OUTER NUCLEAR LAYER



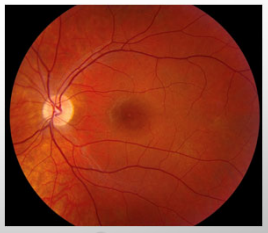
FEATURES OF NONSYNDROMIC RP

- NIGHT BLINDESS
- CONSTRICTED VF
- BONE SPICULES
- ARTERIOLAR ATTENUATION
- WAXY PALLOR
- LOSS OF ONL AND ELLIPSOID ZONE ON OCT

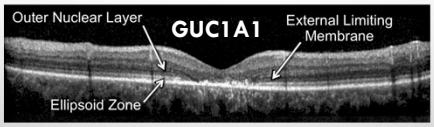
USH2A

28 YO WITH 20/100 VA

- Normal vision as a child
- Gradually lost vision after age 10
- Photophobic
- Poor color vision
- 2 siblinngs with poor vision along with his mother and maternal grandfather



- Loss of ONL, ELM, and Ellipsoid zone
- Centered on the fovea where the cones are



AD Cone Dystrophy

FEATURES OF AD CONE DYSTROPHY

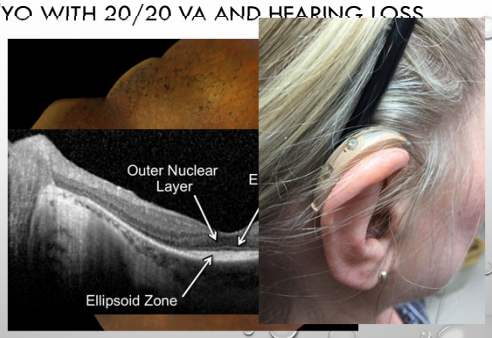
- FAIRLY NORMAL FUNDUS
- REDUCED ACUITY
- POOR COLOR VISION
- PHOTOPHOBIA
- LOSS OF CONE PHOTORECEPTORS ON OCT
- AD FAMILY HISTORY

SYNDROMIC PHOTORECEPTOR DISEASE

45 YO WITH 20/20 VA AND HEARING LOSS

Usher Syndrome

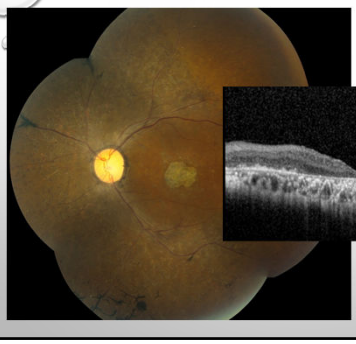
USH2A

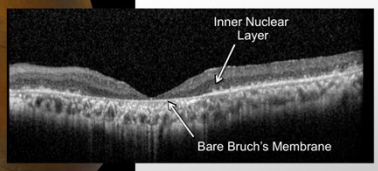


FEATURES OF USHER SYNDROME

- RP
- HEARING LOSS

45 YO F WITH 20/400 VA





- BOTH PARENTS NORMAL VISION
- COGNITIVELY BELOW NORMAL
- BMI ABOVE NORMAL
- DM TYPE 2

FEATURES OF BARDET BIEDL

- CONE-ROD DYSTROPHY
- POOR VISION
- REDUCED COGNITION
- HIGH BMI
- DMII
- POLYDACTYLY

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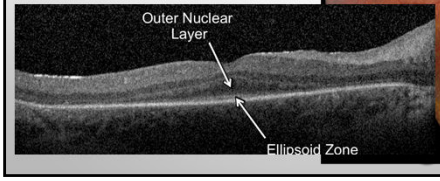
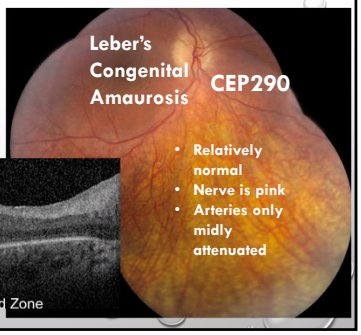
CONGENITAL PHOTORECEPTOR DISEASES

15 YEAR OLD MALE

- POOR VISION SINCE 3 MONTHS OF AGE
- NYSTAGMUS
- PUPILS CONSTRICT IN DARK, DILATE IN LIGHT
- 7D HYPEROPE
- VA LP
- PARENTS HAVE NORMAL VISION





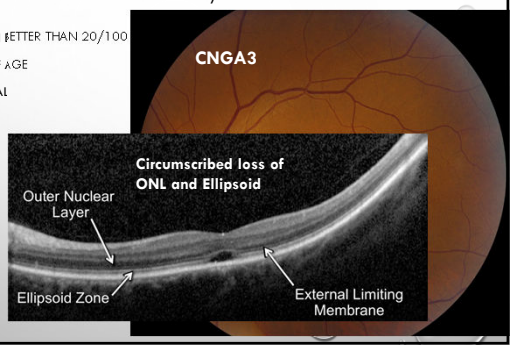
FEATURES OF LCA

- POOR VISION NOTED SHORTLY AFTER BIRTH
- NYSTAGMUS
- PARADOXICAL PUPILLARY RESPONSE
- HIGH HYPEROPIA
- RELATIVELY NORMAL APPEARING FUNDUS

Remember RPE-65 can cause LCA

46 YO F WITH 20/150 VA

- VISION HAS NEVER BEEN BETTER THAN 20/100
- NOTED AT 12 WEEKS OF AGE
- PARENTS VISION NORMAL
- POOR COLOR VISION
- PHOTOPHOBIA
- SEES WELL IN THE DARK



FEATURES OF ACHROMATOPSIA

- POOR VISION SINCE BIRTH
- FAIRLY NORMAL FUNDUS
- PHOTOPHOBIA
- NEAR NORMAL NIGHT VISION
- ABSENT COLOR VISION

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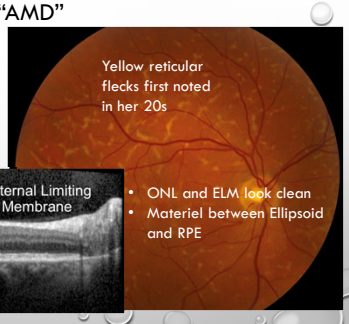
MACULAR DYSTROPHIES

62 YO F WITH 20/25 VA AND MANY RELATIVES WITH "AMD"

Pattern Dystrophy

PRPH2/RDS



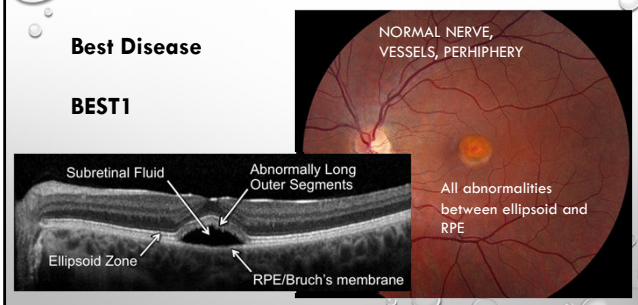
FEATURES OF PATTERN DYSTROPHY

- BRANCHING OR RETICULAR YELLOW DEPOSITS
- ONSET IN EARLY ADULTHOOD
- AD INHERITANCE
- OCT SHOWS DEPOSITS UNDER THE PHOTORECEPTOR LAYERS (DEEPER THAN PHOTORECEPTOR DISEASES)

18 YO MALE WITH 20/20 VA, NORMAL PERIPHERAL VA

Best Disease

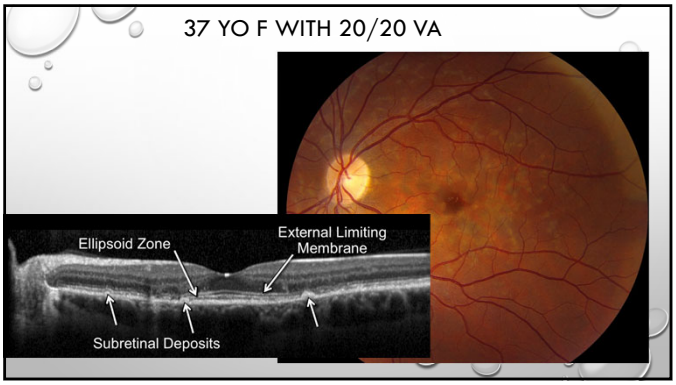
BEST1



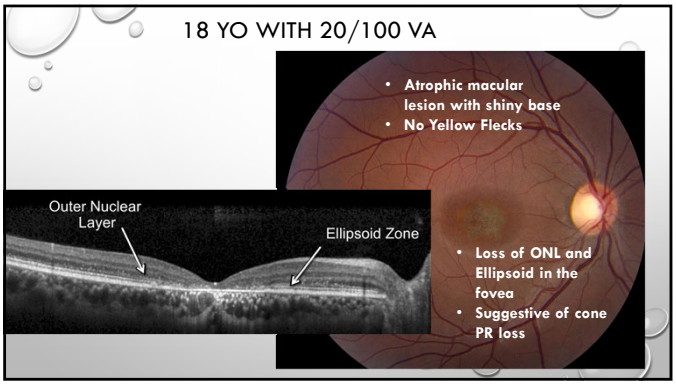
FEATURES OF BEST DISEASE

- OFTEN GOOD VISUAL ACUITY
- AD FAMILY HISTORY
- YELLOW EGG YOLK-LIKE LESION CENTERED ON THE FOVEA
- OCT SHOWS LESION BETWEEN ELLIPSOID AND RPE

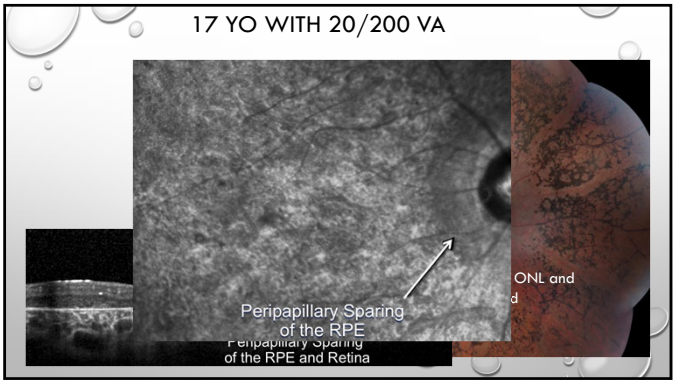
37 YO F WITH 20/20 VA



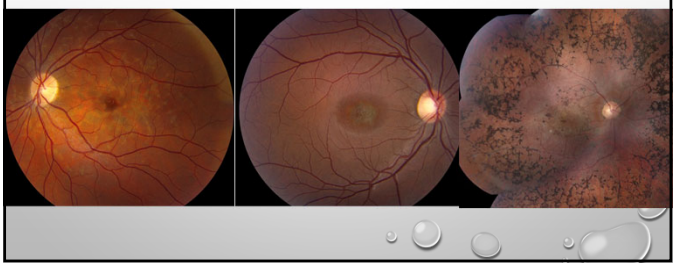
18 YO WITH 20/100 VA



17 YO WITH 20/200 VA



ABCA4 SPECTRUM



STARGART DISEASE

- ABCA4, PRPH2, ELOVL4, PROM1
- ABCA4 MOST COMMON AND CAN CAUSE PHENOTYPES RANGING FROM PATTERN DYSTROPHY TO CONE DYSTROPHY TO RP
- ABCA4 MUTATIONS CAUSE A2PE TO ACCUMULATE (TOXIC)
- MILDER MUTATIONS = MILDER DISEASE AND VICE VERSA

FEATURES OF STARGART DISEASE

- AR INHERITANCE
- PISCIFORM FLECKS
- MACULAR ATROPHY
- PERIPAPILLARY SPARING
- DARK CHOROID ON FA
- HIGHLY VARIABLE PHENOTYPE

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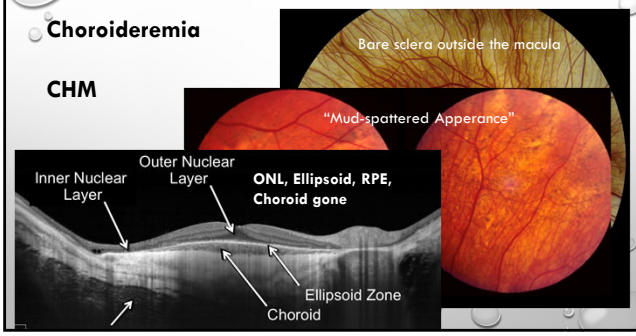
EASES

20 YO WITH 20/20 VA

Choroideremia

CHM



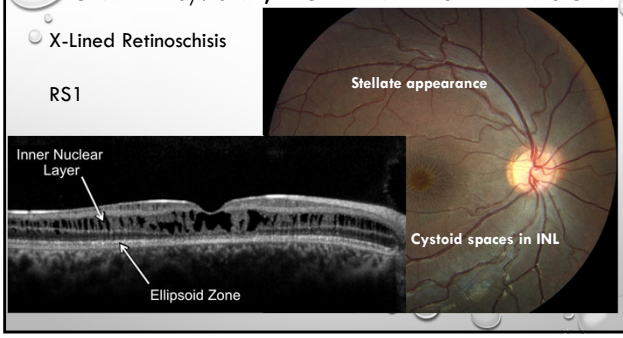
FEATURES OF CHOROIDEREMIA

- X-LINKED INHERITANCE
- FEMALE CARRIERS WITH "MUD-SPATTERED" FUNDUS
- PROFOUND CHORIORETINAL ATROPHY
- LOSS OF CHOROID, RPE, ELLIPSOID, AND ONL ON OCT

12 YO WITH 20/70 VA, BROTHER WITH SIMILAR VISION

- X-Lined Retinoschisis

RS1



FEATURES OF JUVENILE XLRS

- X-LINKED INHERITANCE
- STELLATE MACULAR APPEARANCE
- CYSTOID SPACES IN THE INL ON OCT
- PERIPHERAL RETINOSCHISIS, VH, AND RD

TAKE HOME POINTS

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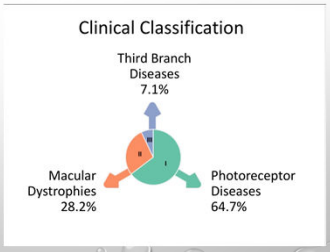

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