IRD'S FOR THE OPHTHALMIC TECHNICIAN

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OVERVIEW

- IRD'S REQUIRE A TEAM
- HISTORY IS IMPORTANT
- IMAGING IS VITAL
- WHY GENETIC TESTING IS IMPORTANT
- EXAMPLES OF MOST IMPORTANT IRD'S

IRD'S ARE CHALLENGING

- RARE
- OVERLAPPING FEATURES
- OFTEN IN CHILDREN THAT ARE HARD TO EXAMINE
- CLINIC IS ALWAYS BUSY
- YOUR OPHTHALMOLOGIST NEEDS YOUR HELP!

IRD'S REQUIRE A TEAM

- HISTORY
- IMAGING
- RECOGNIZING FEATURES OF SYNDROMIC IRD'S
- FOUNDATION FOR FIGHTING BLINDNESS CLINICAL TRIALS PIPELINE

HISTORY

- WHEN VISION LOSS STARTED
- LOSS OF NIGHT VISION? PHOTOPHOBIA?
- FAMILY HISTORY OF VISION LOSS
- RECOGNIZING DIFFERENT INHERITANCE PATTERNS

AUTOSOMAL DOMINANT

- DOES NOT SKIP GENERATIONS
- CAN BE MALES OR FEMALES

AUTOSOMAL RECESSIVE

- PARENTS MAY HAVE NORMAL VISION (CARRIERS)
- AFFECTED PATIENT INHERITS 2 COPIES OF THE MUTATED GENE
- 1 FROM EACH PARENT
- CAN SKIP GENERATIONS

X-LINKED

- MEN HAVE 1 X CHROMOSOME
- WOMEN HAVE 2
- IN X-LINKED CONDITIONS, MEN ARE AFFECTED, INHERIT THE GENE FROM THEIR MOTHERS
- WOMEN ARE CARRIERS AND MAY HAVE SLIGHTLY ABNORMAL FUNDUS APPEARANCE

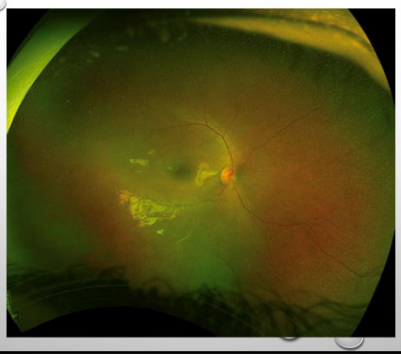
IMAGING

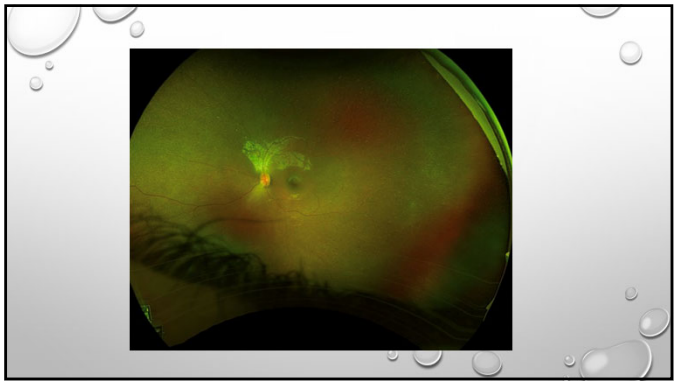
CASE

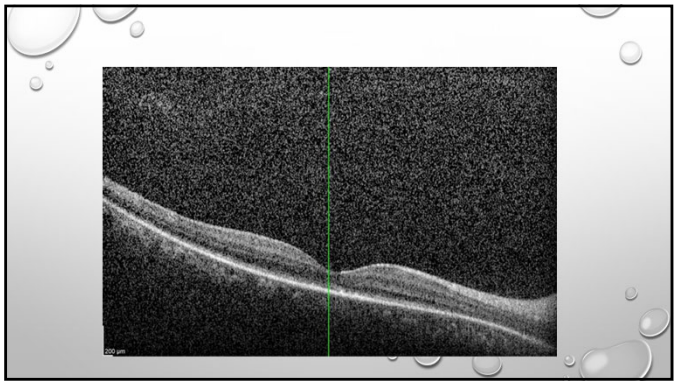
- 7 YEAR OLD
- 20/200 VISION
- NYSTAGMUS

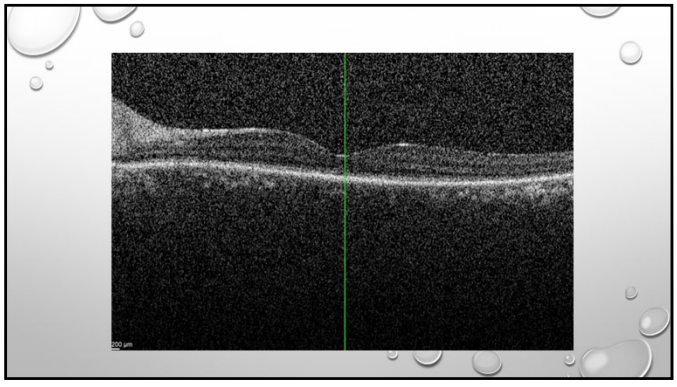
IMAGING REALLY HELPS

- KIDS ARE HARD TO EXAMINE
- WIDEFIELD IMAGING CAN BE BETTER THAN AN IDO EXAM
- OCT IS SUPER HELPFUL
- EVEN AN SINGLE LINE CUT THROUGH THE MACULA CAN HELP MAKE THE DIAGNOSIS







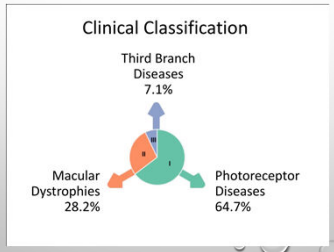


GENTIC TESTING

- POSITIVE FOR RPE-65

LUXTURNA VIDEO

IRD'S MADE SIMPLE



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

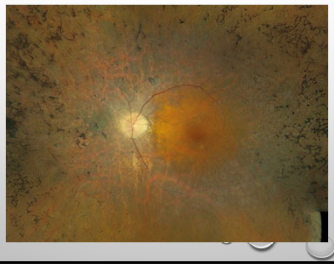
II – Macular Diseases
- A – AR Stargardt Disease
- B – Best Disease
- C - Pattern Dystrophy

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

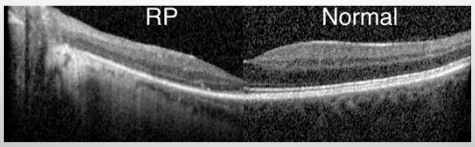
NON-SYNDROMIC PHOTORECEPTOR DISEASE

- JUST THE EYE IS AFFECTED

60 YO M WITH POOR NIGHT VISION AND CONSTRICTED VISUAL FIELD



LOSS OF ELLIPSOID ZONE AND OUTER NUCLEAR LAYER



FEATURES OF NON-SYNDROMIC RP

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

USH2A, but can be caused by RPE65

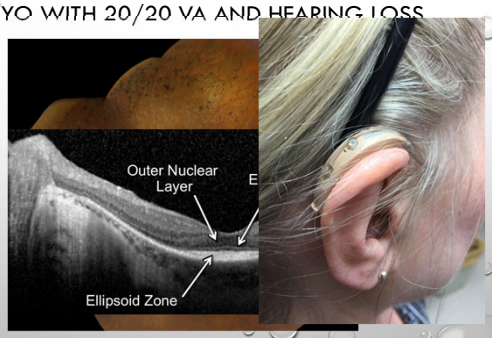
SYNDROMIC PHOTORECEPTOR DISEASE

- OTHER ORGAN SYSTEMS ARE AFFECTED

45 YO WITH 20/20 VA AND HEARING LOSS

Usher Syndrome

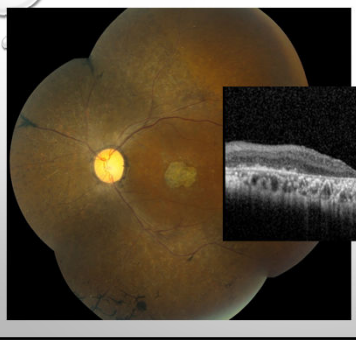
USH2A

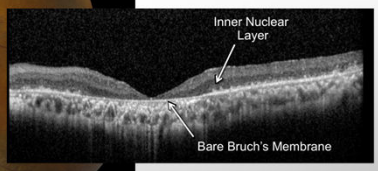


FEATURES OF USHER SYNDROME

- RP
- HEARING LOSS

45 YO F WITH 20/400 VA





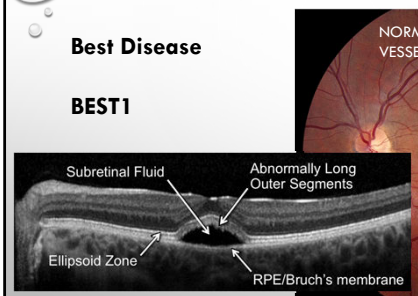
- BOTH PARENTS NORMAL VISION
- COGNITIVELY BELOW NORMAL
- BMI ABOVE NORMAL
- DM TYPE 2

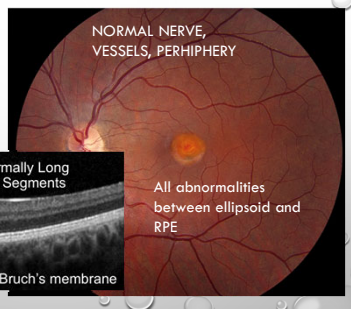
MACULAR DYSTROPHIES

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

Best Disease

BEST1

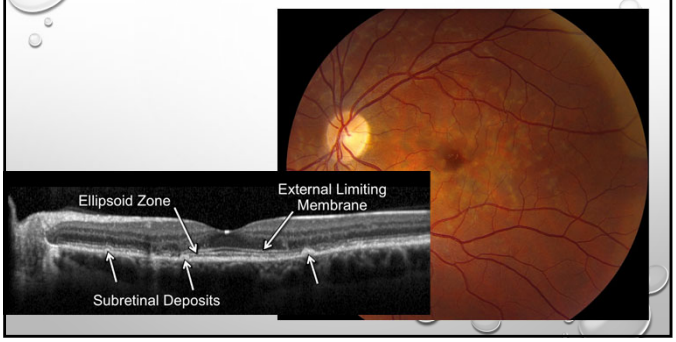




FEATURES OF BEST DISEASE

- 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



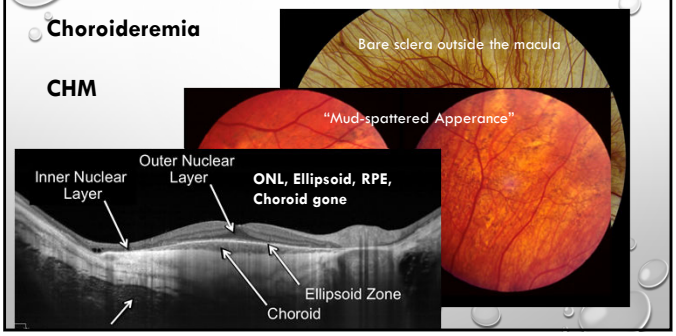
FEATURES OF STARGART DISEASE

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

20 YO WITH 20/20 VA

Choroideremia

CHM



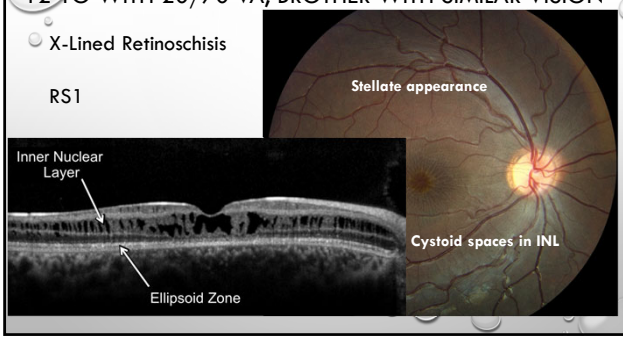
FEATURES OF CHOROIDEREMIA

- 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

- TEAM APPROACH
- HISTORY (WHEN VISION LOSS STARTED, FAMILY HISTORY)
- IMAGING IS KEY (WIDEFIELD PHOTOS, OCT)
- GENETIC TESTING IS IMPORTANT
- FOUNDATION FOR FIGHTING BLINDNESS CLINICAL TRIALS PIPELINE