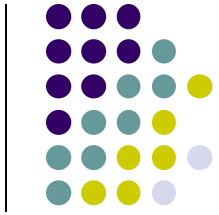
# Q

## Stargardt Disease/Fundus Flavimaculatus



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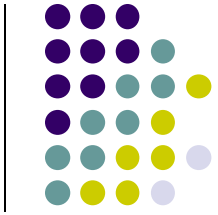

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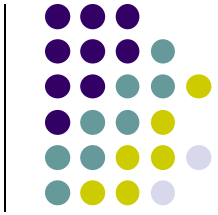

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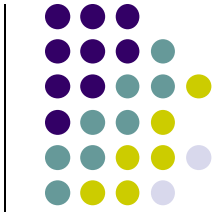

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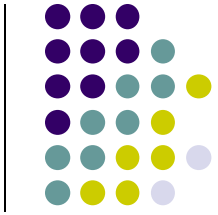

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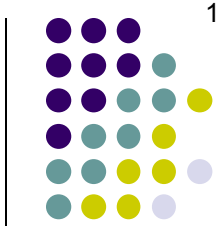

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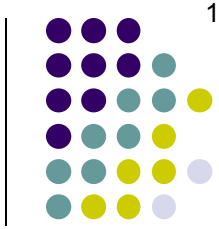

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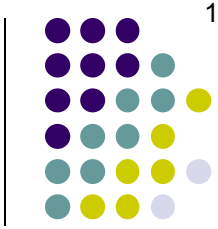

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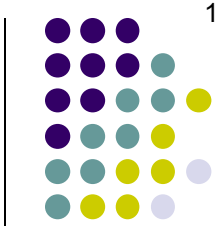

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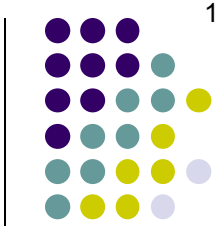

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*The ABCA4 gene is mission-critical to the eye, with implications extending far beyond Stargardt/FF. Given this, let's look at it in some detail…*

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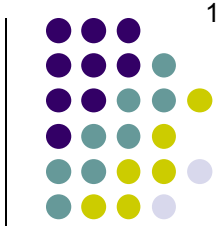

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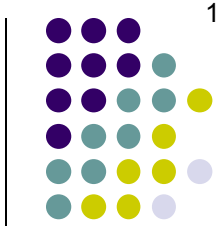

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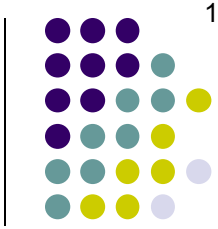

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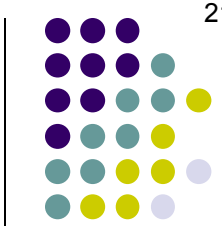

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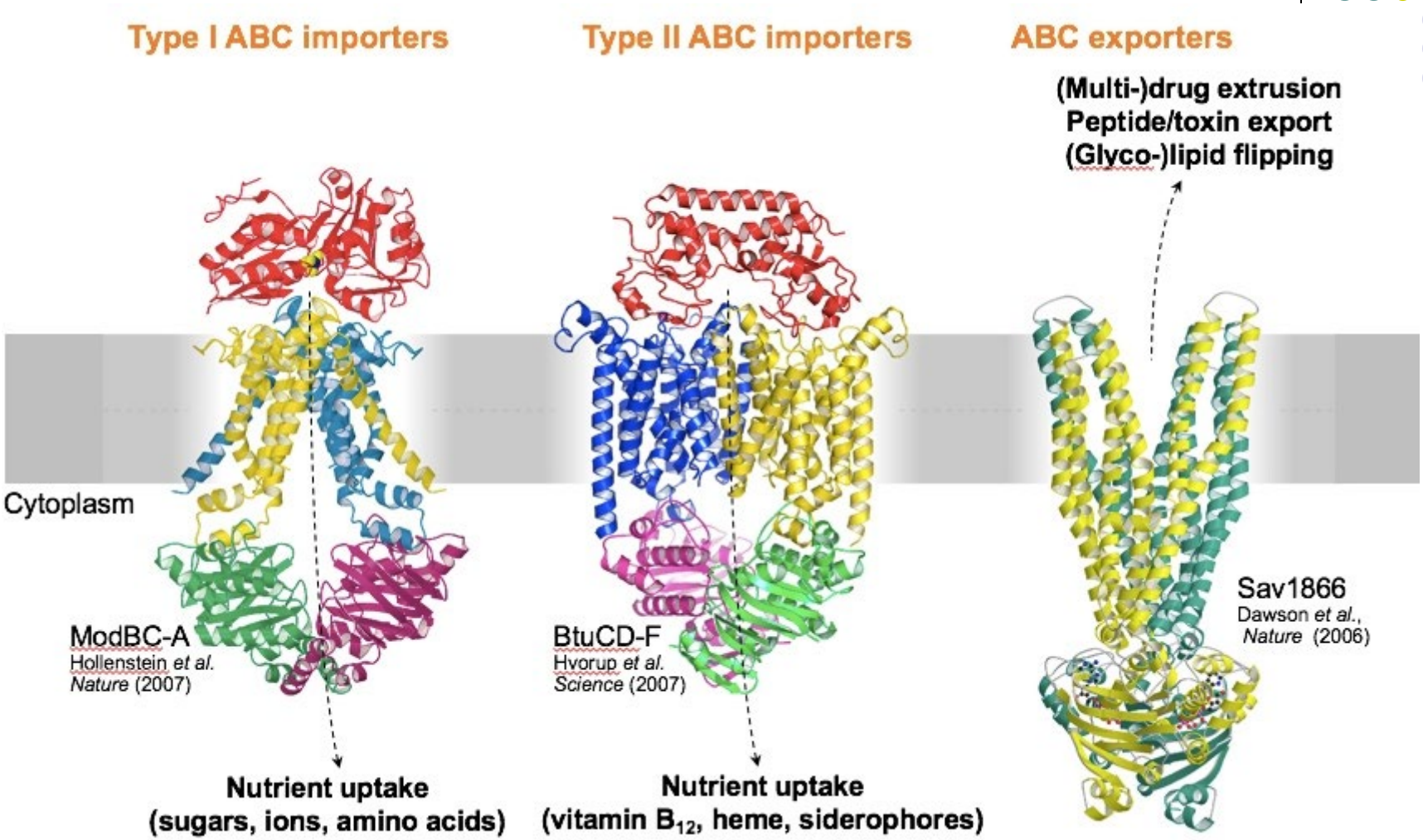
Type I ABC importers
Type II ABC importers
ABC exporters
(Multi-)drug extrusion
Peptide/toxin export
(Glyco-)lipid flipping
Cytoplasm
ModBC-A
Hollenstein et al.
Nature (2007)
BtuCD-F
Hvorup et al.
Science (2007)
Sav1866
Dawson et al.,
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Nutrient uptake
(sugars, ions, amino acids)
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(vitamin B12, heme, siderophores)

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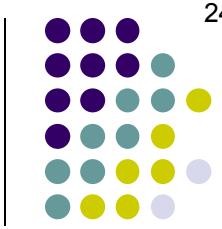

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Name a substrate, and an ABC transporter is probably involved--most nutrients, vitamins, trace elements, etc coming in; metabolic waste, fats, sterols, and drugs going out.

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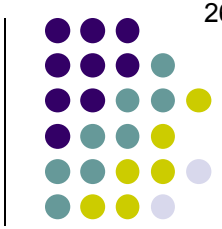

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It means exactly what it says--ABC transporters are how cells rid themselves of therapeutic compounds. For example, bacterial drug resistance is often 2ndry to the development of ABC transporters. Likewise, when a previously effective cancer drug loses efficacy for an individual, it can often be attributed to the appearance in the cancer line of an ABC transporter that effluxes the drug.

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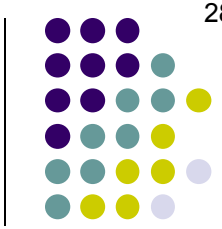

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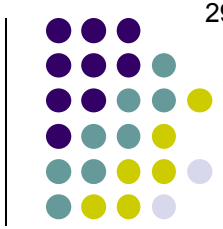

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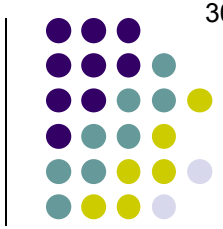

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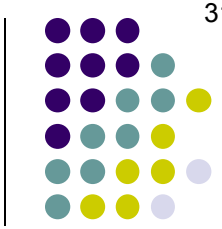

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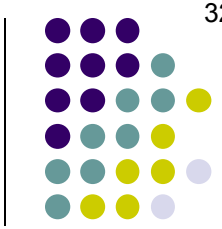

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It is responsible for exporting a potentially toxic metabolic byproduct of the visual cycle. When ABCA4 is defective, this byproduct accumulates within the segment. Then, when the outer segments are shed and 'swallowed' by the underlying RPE (as part of the normal retinal renewal process), the metabolic byproduct is incorporated into the RPE cell's wear-and-tear granule.

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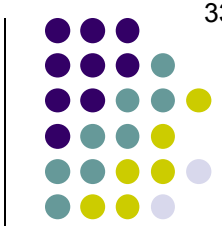

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# Q/A

## Stargardt Disease/Fundus Flavimaculatus

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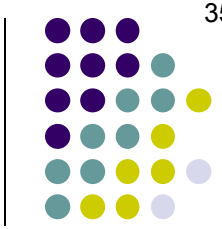

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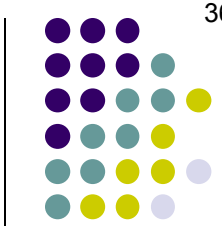

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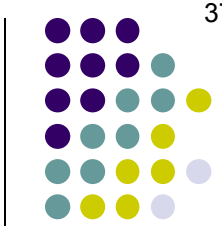

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*And death of the RPE leads to…?*

# A

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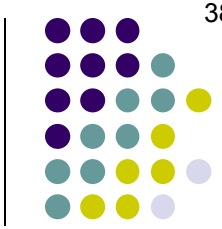

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*And death of the RPE leads to…?*
Changes in the appearance of the posterior pole, as well as (far more importantly) death of overlying photoreceptors, with subsequent decreased vision

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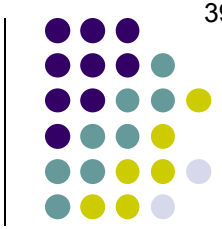

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*The pathophysiology of Stargardt/FF* ***tl;dr***
--Defective ABCA4 alleles inherited; defective ABCA4 transporters expressed in rod outer segments
--Defective ABCA4 transporters can't export metabolic byproducts of the visual cycle, leading to their accumulation within the segments
-- As part of the normal retinal renewal process, byproduct-laden outer segments are shed, then phagocytized by RPE cells
--Within RPE cells, the byproduct is converted to A2E, which eventually kills the cell
--When the RPE cell dies, photoreceptors that depend on it die as well

*And death of the RPE leads to…?*
Changes in the appearance of the posterior pole, as well as (far more importantly) death of overlying photoreceptors, with subsequent decreased vision

# Q

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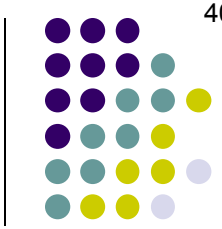

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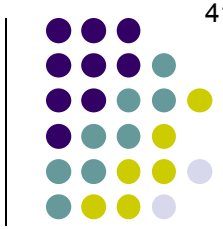

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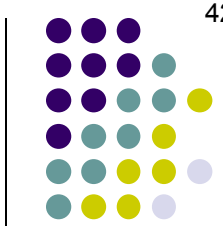

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*At what level of the retina do the flecks occur?*

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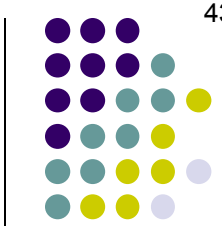

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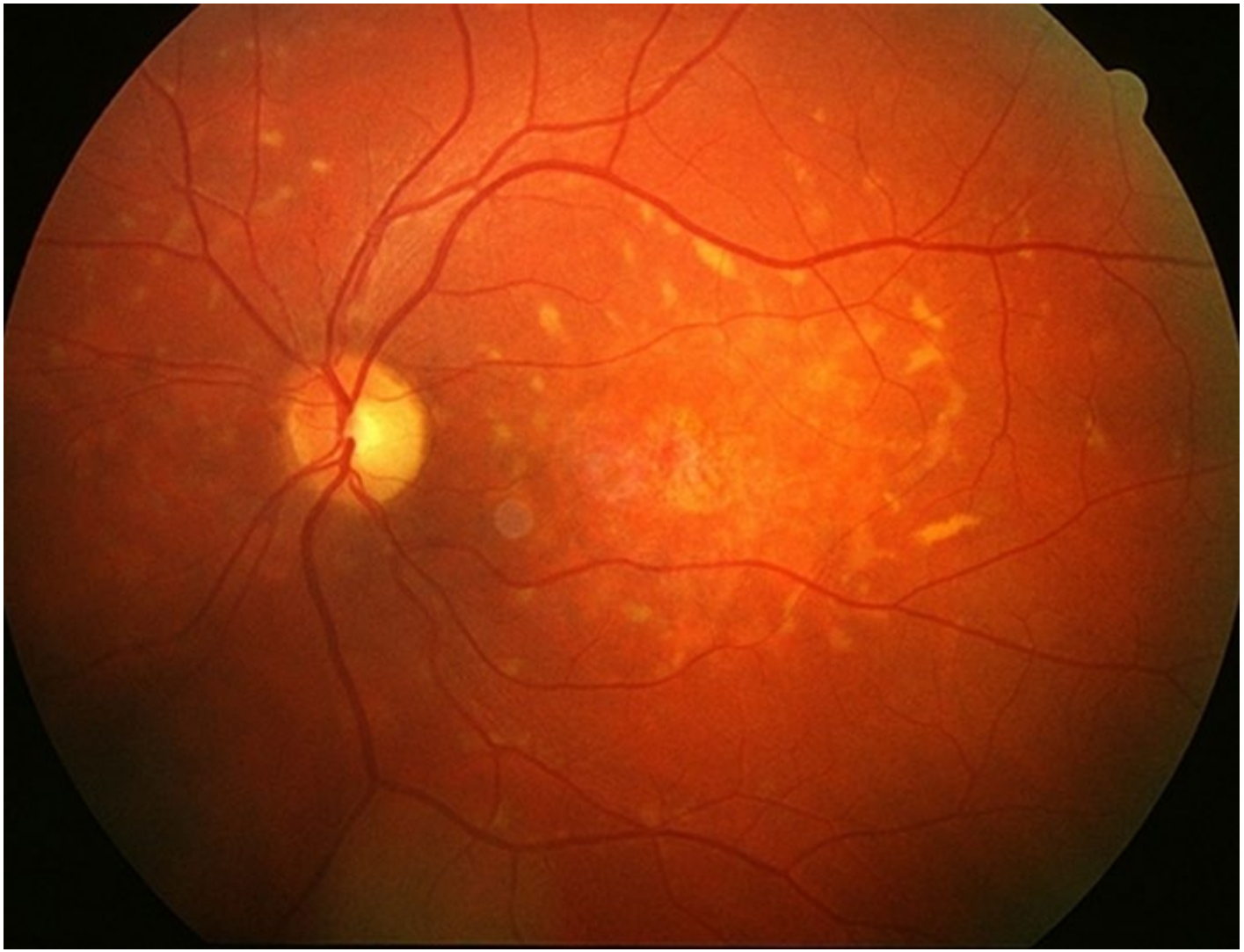
*At what level of the retina do the flecks occur?*
The RPE

# Stargardt Disease/Fundus Flavimaculatus



Stargardt: RPE-level flecks

# Q

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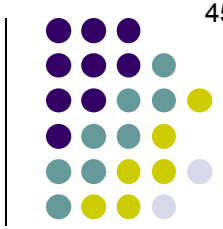

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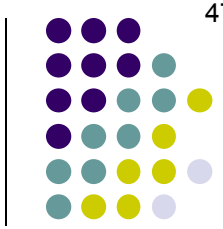

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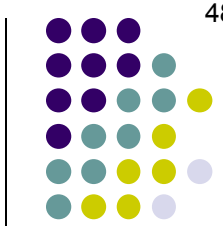

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*How does that come about?*
If two of these elongated flecks touch one another at just the right angle, their appearance will be reminiscent of a fish's tail

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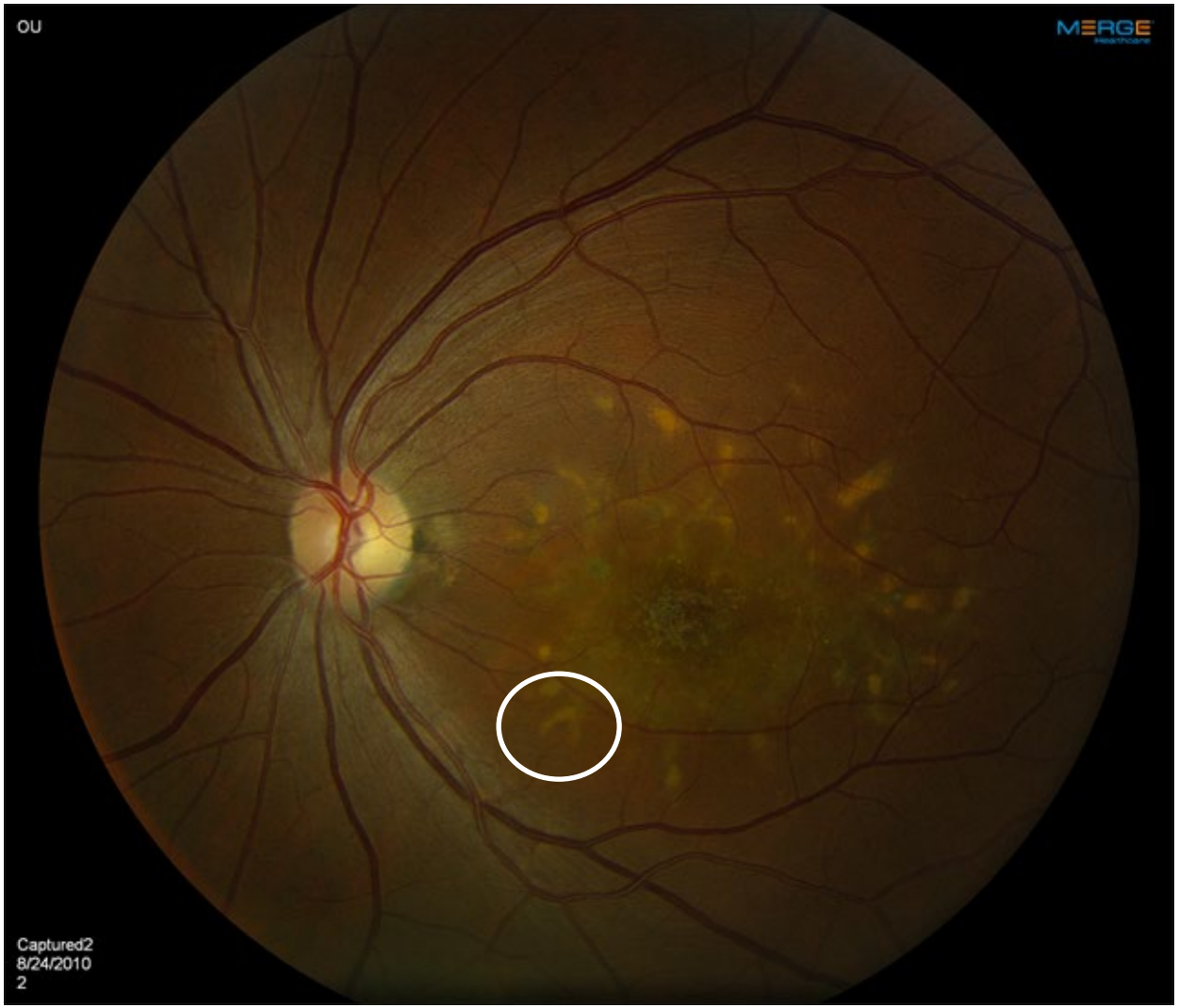


Pisciform lesions

# Q

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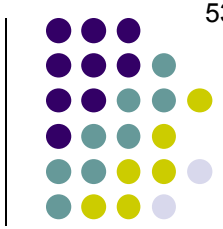

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*Which appears first--foveal atrophy, or the flecks?*

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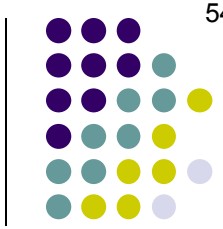

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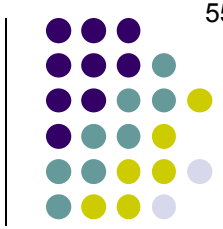

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*The classic appearance of the fovea in Stargardt is described with a two-word alliteration. What is it?*

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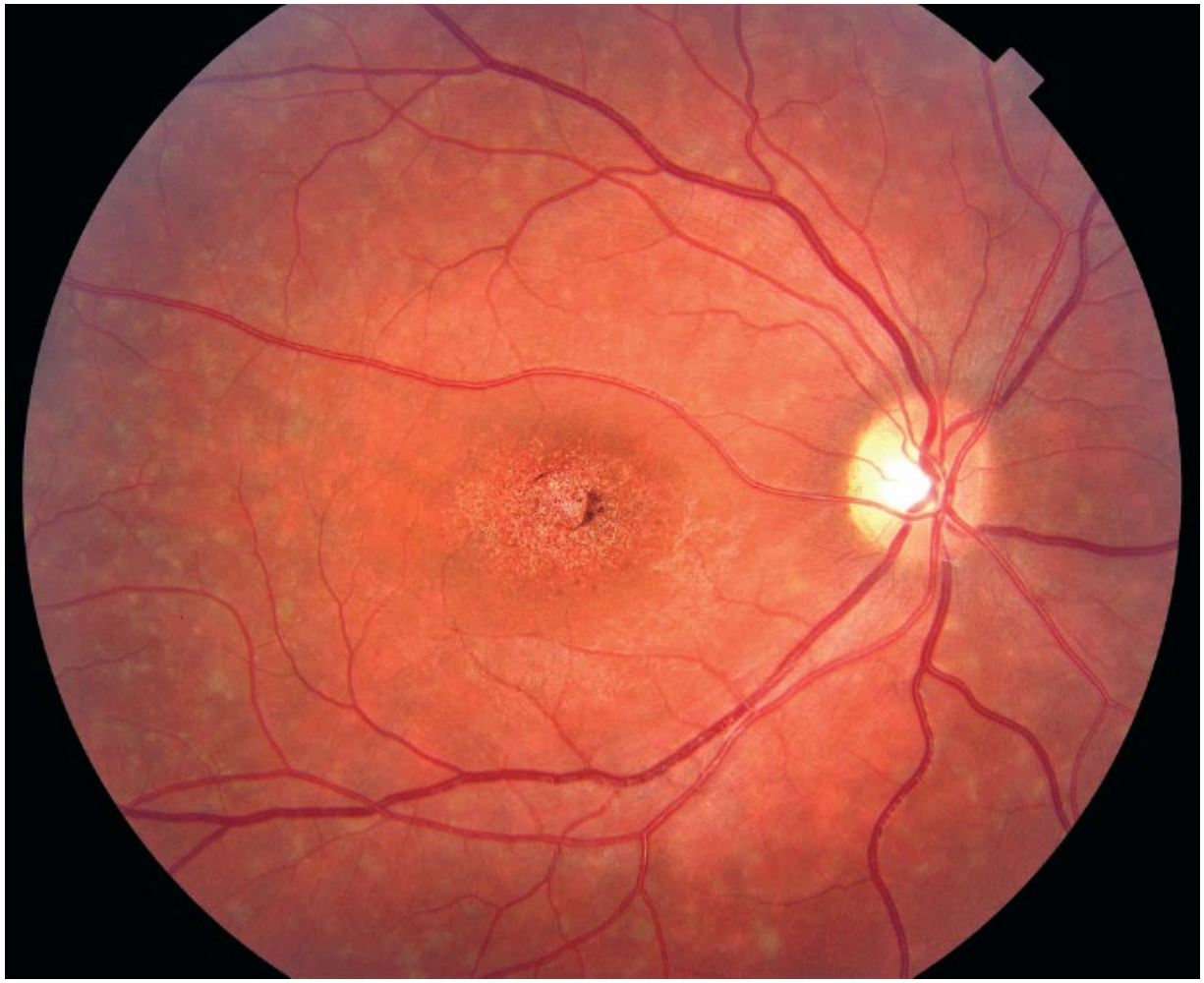
**'Beaten bronze'**

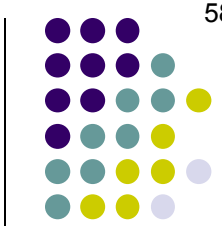
# Stargardt Disease/Fundus Flavimaculatus



Stargardt disease. Macular atrophy, pisciform yellow-white flecks, and a beaten-bronze appearance. Note the peripapillary sparing of retina

# Q

## Stargardt Disease/Fundus Flavimaculatus



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# A

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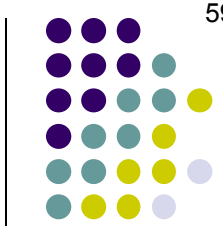

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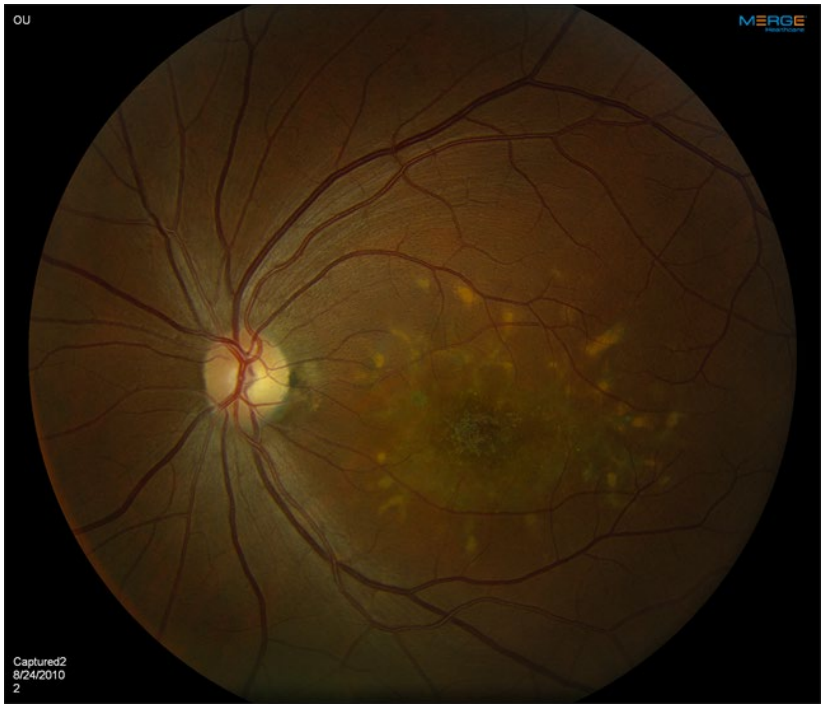
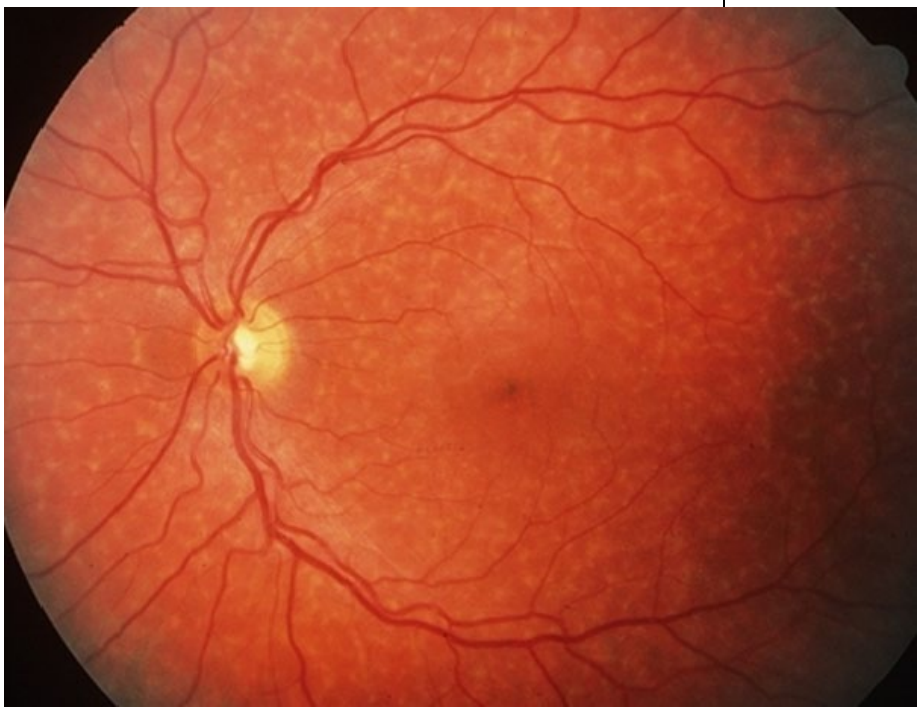



Stargardt



Fundus flavimaculatus

# Q

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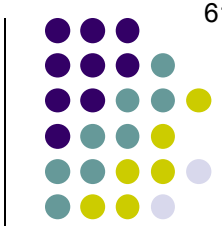

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- Pts present with c/o [ ], usually in [life period]

# A

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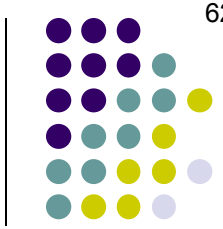

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# Q

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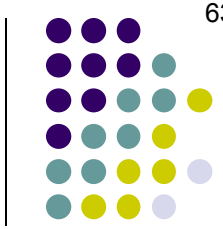

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# A

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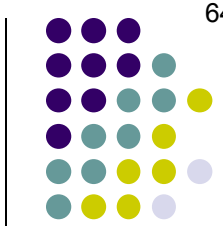

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*Which appears first--changes in the fundus, or decreased vision?*
Usually the decreased vision (especially in childhood onset cases)

# Q

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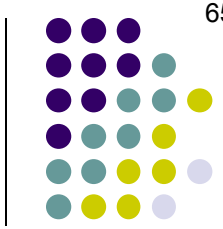

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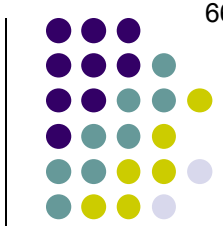

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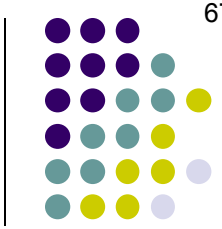

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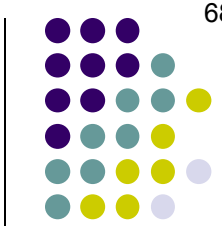

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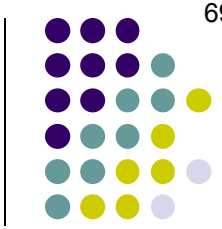

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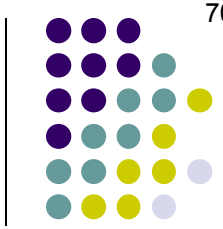

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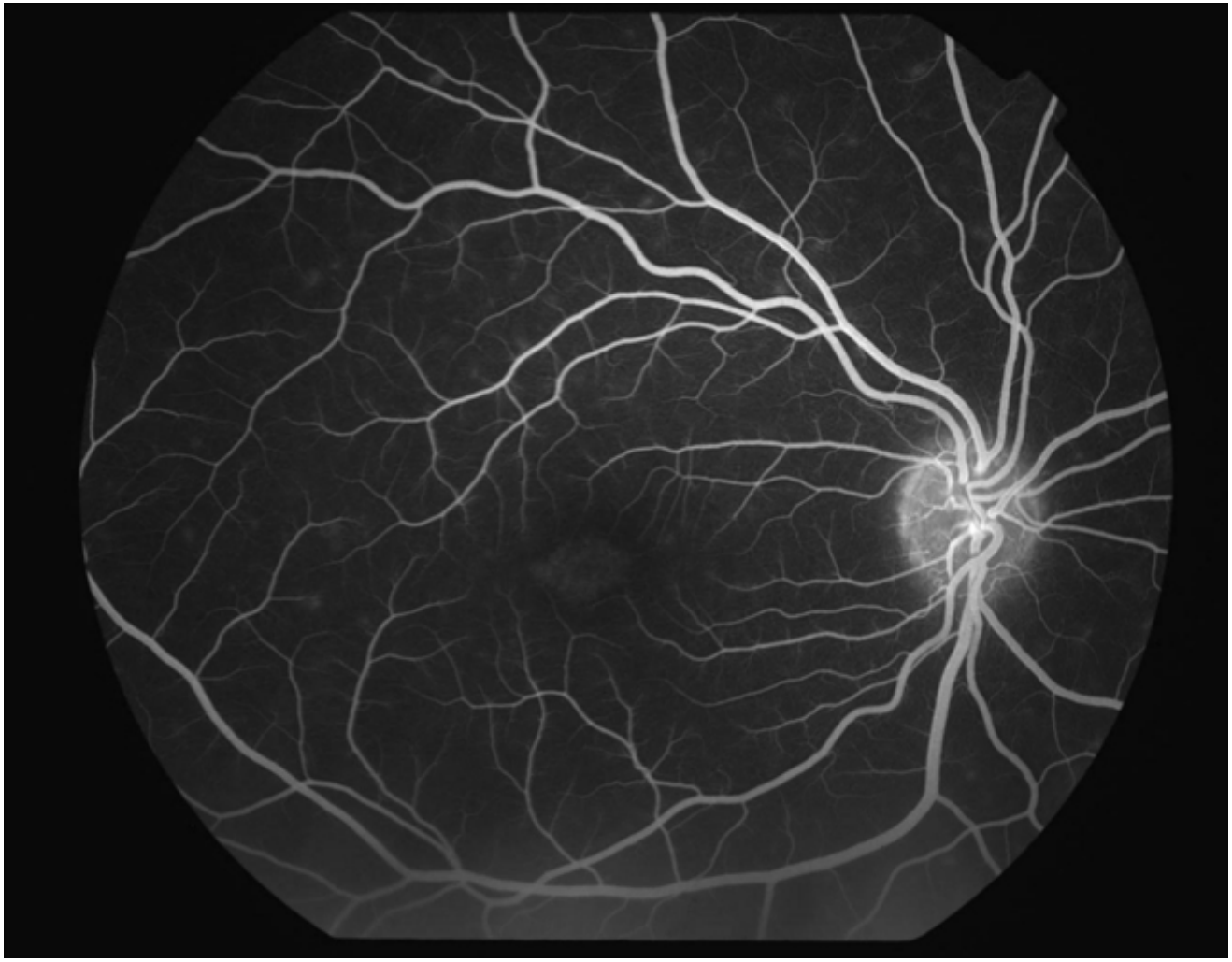

Stargardt—*dark choroid* appearance on FA

# Q

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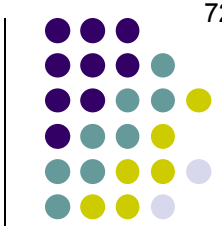

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# Q/A

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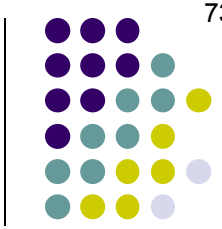

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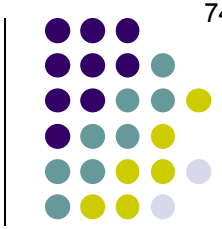

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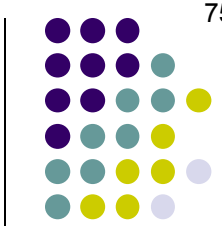

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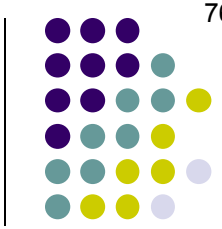

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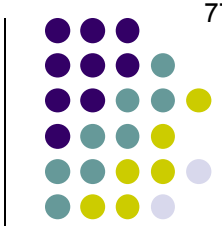

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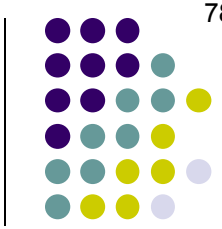

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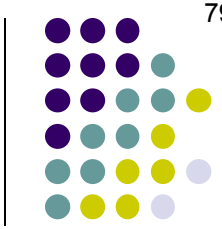

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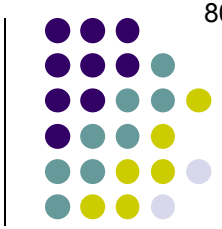

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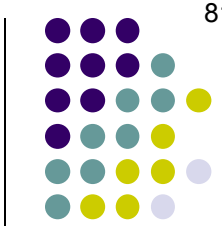

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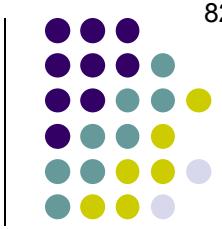

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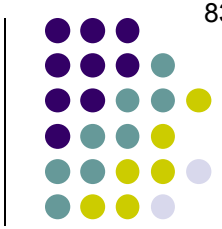

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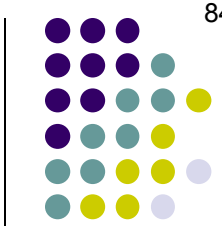

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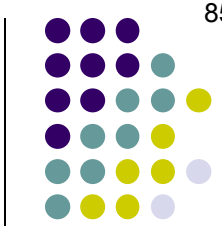

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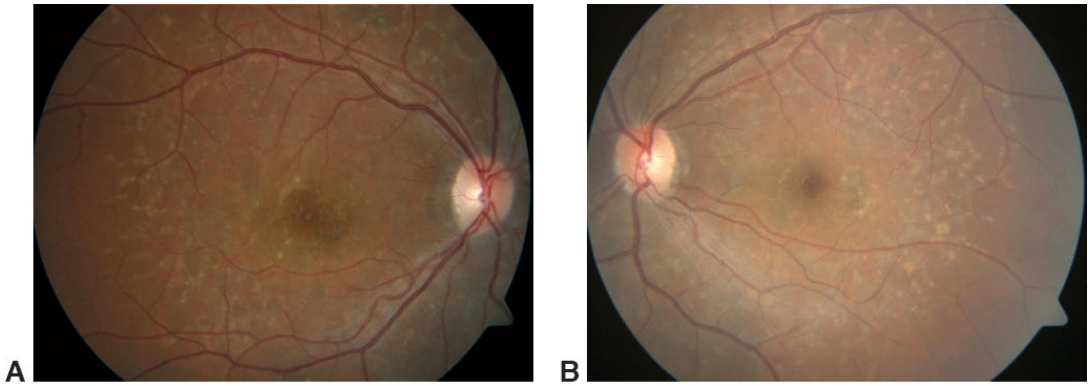
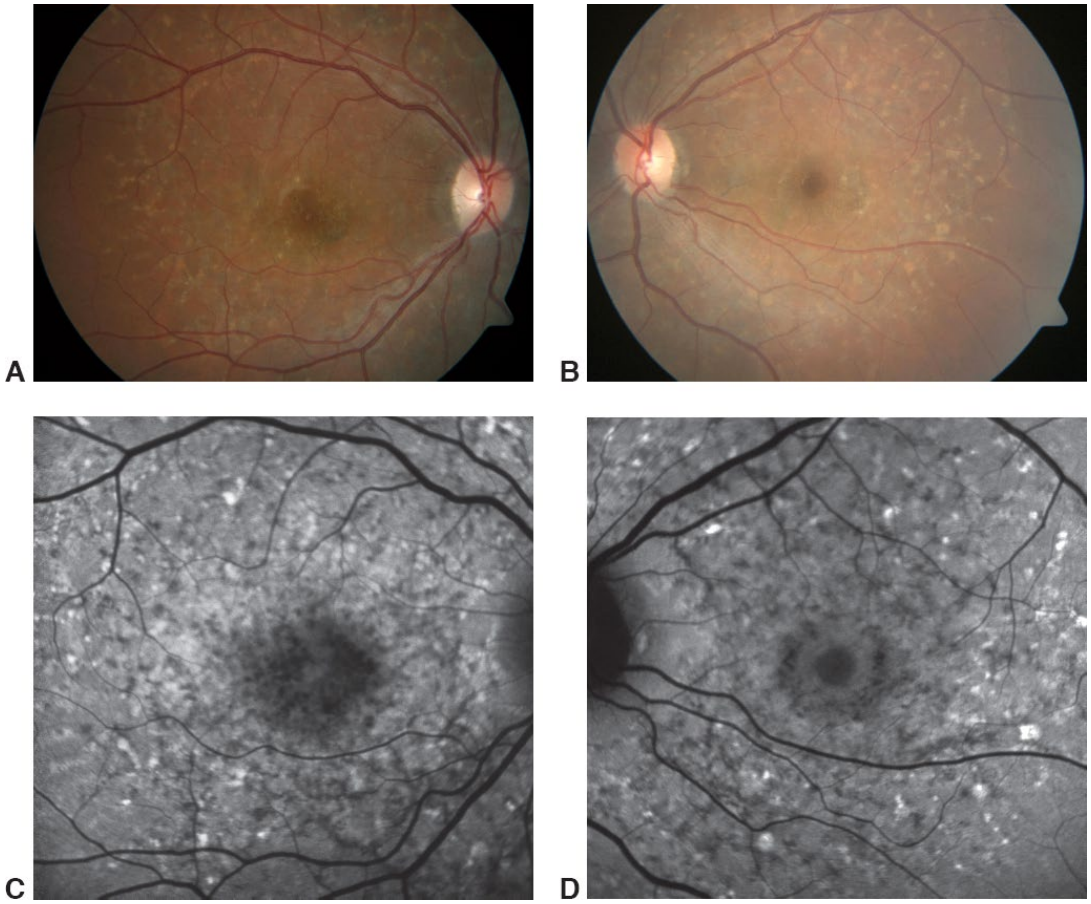



Stargardt disease. The right eye (A) and left eye (B) demonstrate classic pisciform yellow-white flecks throughout the macula, with mottling of the central retinal pigment epithelium (RPE).

# Stargardt Disease/Fundus Flavimaculatus




Stargardt disease. The right eye (A) and left eye (B) demonstrate classic pisciform yellow-white flecks throughout the macula, with mottling of the central retinal pigment epithelium (RPE).
Corresponding right (C) and left (D) FAF images reveal mottled hypo- and hyperautofluorescence with hyperautofluorescent flecks (corresponding to the pisciform flecks) and a bull's-eye maculopathy, greater in the left eye than in the right eye

# Q

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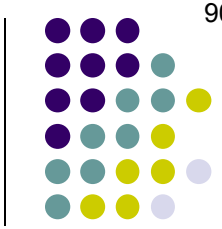

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  - If they are widely scattered, is fundus flavimaculatus
- Pts present with c/o decreased vision, usually in childhood
- Classic ~~FA~~ FAF appearance: ~~dark choroid~~ Bull's eye

*What is the classic FAF appearance of Stargardt?*
A bull's eye maculopathy--a ring of **perifoveal hyperfluorescence** surrounding a central foveal area of hypofluorescence

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*Why is FAF preferred?*
It is more reliable (not all Sta

*Why does the perifoveal macula hyperfluoresce?*

ommonly,
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*What should you do if/when encountering such a child (either on the OKAP/Boards or IRL)?*
The answer **used** to be '**order an** FAF FA,' so we'll address that first…

# A

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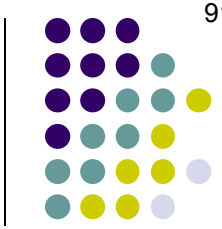

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*Why does the perifoveal macula hyperfluoresce?*
Because its RPE cells are stuffed with lipofuscin-containing A2E, a substance that autofluoresces particularly well

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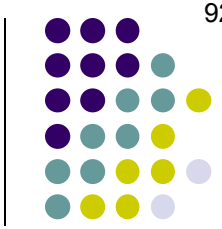

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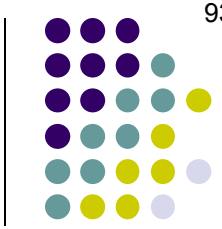

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*OK, but then why does the central foveal area hyp**O**fluoresce?*
Because its RPE cells are dead and gone, leaving little lipofuscin in that area to fluoresce

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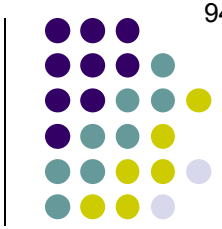

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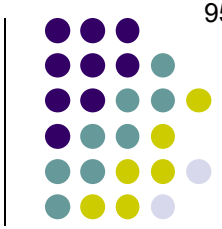

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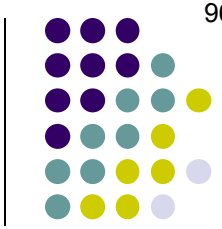

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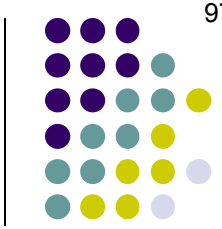

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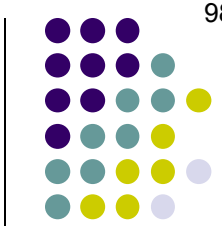

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*How can one dysfunction of a single protein cause such a variety of pathology?*

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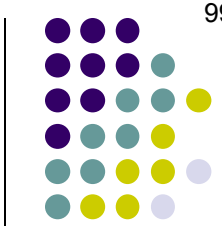

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*How can one dysfunction of a single protein cause such a variety of pathology?*
Because it is not the case that the ABCA4 transporter is either fully functional or completely dysfunctional.

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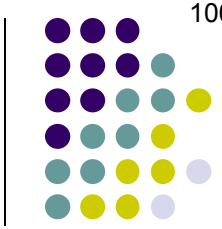

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- Classic FA appearance: *dark choroid*
- Ultimate vision is usually in the 20/50 – 20/200 range
- Other diseases associated with ABCA4 dysfunction include **cone dystrophy**, **cone-rod dystrophy** and **RP**

*How can one dysfunction of a single protein cause such a variety of pathology?* Because it is not the case that the ABCA4 transporter is either fully functional or completely dysfunctional. Rather, ABCA4 function exists on a *continuum*, from completely intact (= normal/non-diseased) to mildly impaired (= mild Stargardt) to moderately impaired (= worse Stargardt) to severely impaired (= cone-rod dystrophy) to completely nonfunctional (= RP).

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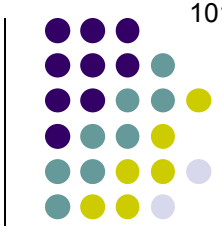

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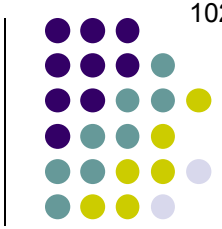

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*So, all of these conditions are caused by mutations on ABCA4?*
Yes and no. All of these conditions can be caused by ABCA4 mutations. But all of them can be caused by mutations to *other* genes as well.

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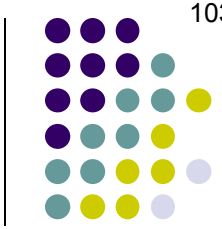

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*For each condition, what percent of cases are caused by ABCA4 mutation?*
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*--Cone dystrophy:*
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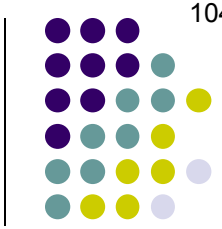

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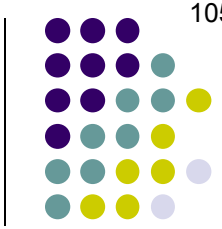

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*What is the $2 term for this situation, ie, when mutations in a single gene can produce multiple phenotypes?*

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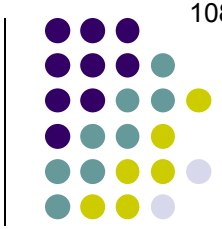

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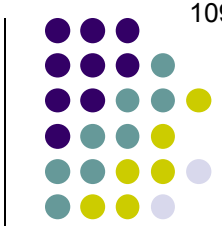

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**Genetic heterogeneity**

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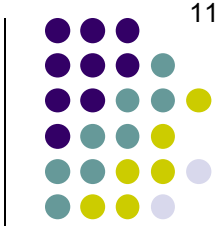
## Stargardt Disease/Fundus Flavimaculatus



- Inheritance? AR (in most cases; a small % are AD)
- Gene responsible for most cases? ABCA4
- Fundus appearance: Atrophic fovea surrounded by white-yellow pisciform
  - If pisci
  - If they
- Pts prese od
- Classic F
- Ultimate vision is usually in the 20/50 – 20/200 range
- Other diseases associated with ABCA4 dysfunction include **cone dystrophy , cone-rod dystrophy** and **RP** & Stargardt/FF

*What is the $2 term for this situation, ie, when mutations in a single gene can produce multiple phenotypes?*
**Pleiotropy**. The *ABCA4* gene is an excellent example of it.

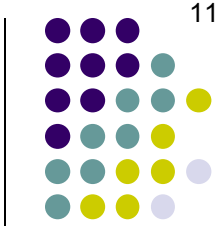
*What is the term for the opposite situation, ie, when mutations to* multiple *different genes can give rise to the* same *phenotype?*
**Genetic heterogeneity**. LCA is an excellent example—it can be caused by mutations to at least 17 different genes.

*So, all of th* gy?
Yes and no al
But all of th
from co
Stargar
(= cone-rod dystrophy) to completely nonfunctional (= RP).

*For each condition, what percent of cases are caused by ABCA4 mutation?*
*--Stargardt:* **>95**
*--Cone dystrophy:* don't have a number for this
*--Cone-rod dystrophy:* **30-50**
*--RP:* **5-10**

# Q

## Stargardt Disease/Fundus Flavimaculatus



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Leber's congenital amaurosis

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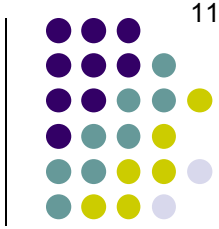

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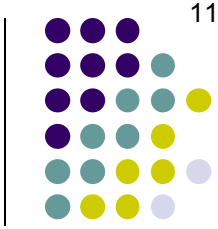
- Fundus appearance: Atrophic fovea surrounded by white-yellow pisciform flecks
  - If pisciform lesions are in macula only, is Stargardt
  - If they are widely scattered, is fundus flavimaculatus
- Pts present with c/o decreased vision, usually in childhood
- Classic FA appearance: *dark choroid*
- Ultimate vision is usually in the 20/50 – 20/200 range
- Other diseases associated with ABCA4 dysfunction include cone dystrophy, cone-rod dystrophy and RP
- Treatment?

# A

## Stargardt Disease/Fundus Flavimaculatus



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- Other diseases associated with ABCA4 dysfunction include cone dystrophy, cone-rod dystrophy and RP
- Treatment? No effective treatments are available