Q

Best Disease: T/F

- Best disease is AD (like most inherited retinal diseases)

A

Best Disease: T/F

- Best disease is AD (~~like~~ unlike most inherited retinal diseases) **F**

Q

Best Disease: T/F

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What is the name of the gene implicated in Best dz?

A

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What is the name of the gene implicated in Best dz?
Best1 (or VMD2)

Q

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What is the name of the gene implicated in Best dz?
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What protein does the Best1 *gene code for?*

A

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What is the name of the gene implicated in Best dz?
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- EOG is normal in adult vitelliform disease ~~and~~ but not in Best carriers **F**

Q

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What does EOG *stand for?*

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What does EOG *stand for?*
Electro-oculogram

Q

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In a nutshell, what does an electro-oculogram measure?

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RPE function

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Again in a nutshell, how does it work?

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The resting potential of the RPE is measured in both the light- and dark-adapted states, and a ratio of the two resting potentials is calculated

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What is this ratio called?
The **Arden ratio**

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The **Arden ratio**

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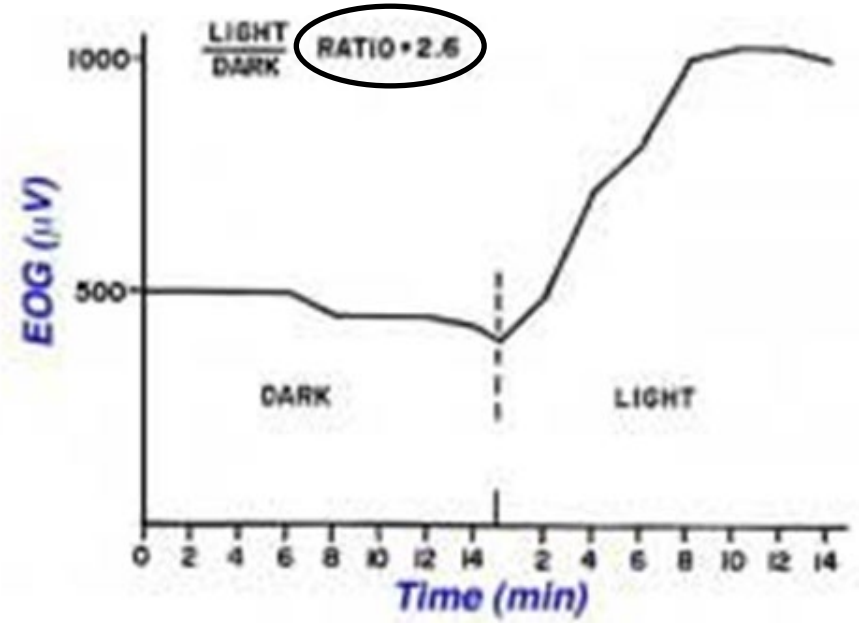
What is this ratio called?
The **Arden ratio**

What is the normal range for the Arden ratio?
1.9-2.8

Best Disease: T/F



Non-Best pts

EOG

Q

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1.9-2.8

At what value is the Arden ratio considered definitely abnormal?

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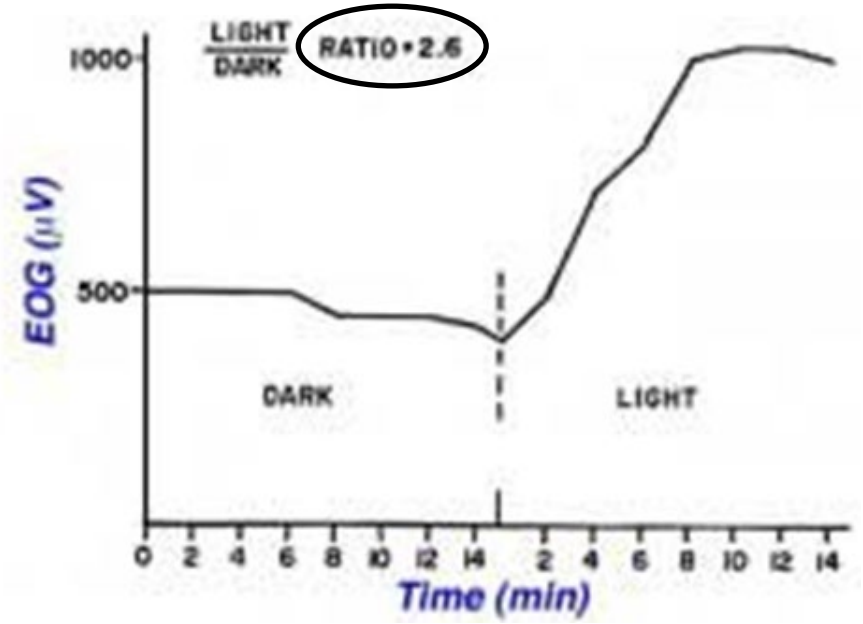
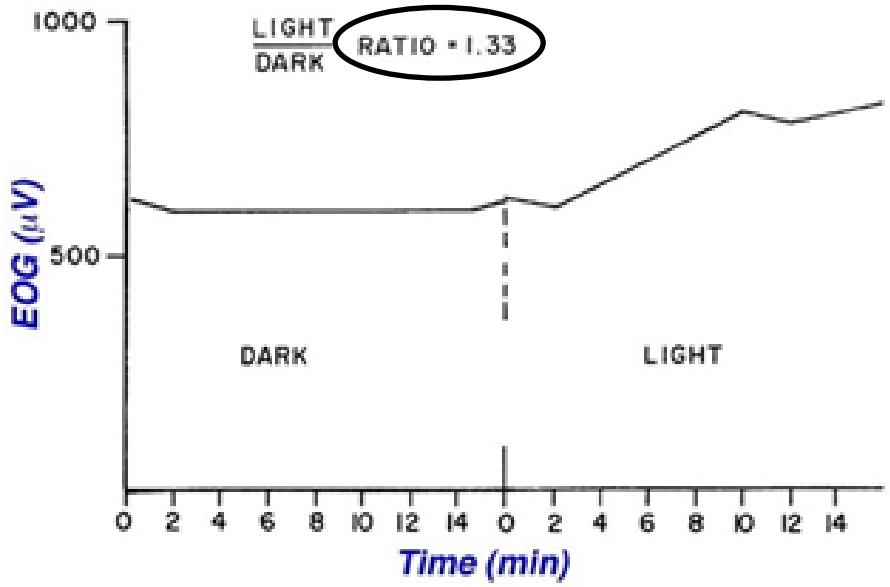
1.9-2.8

At what value is the Arden ratio considered definitely abnormal?
Below 1.7 (it's usually <1.5 in Best dz, and ratios as low as 1.1 are not uncommon)

Best Disease: T/F



Non-Best pts



Best pts

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What is typical status of the E**R**G *in Best dz?*

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Below 1.7 (it's usually <1.5 in Best dz, and ratios as low as 1.1 are not uncommon)

What is typical status of the E**R**G *in Best dz?*
It is normal, or even supranormal

Q

Best Disease: T/F

- Best disease is AD (~~like~~ unlike most inherited retinal diseases) **F**
- EOG is normal in adult vitelliform disease ~~and~~ but not in Best carriers **F**
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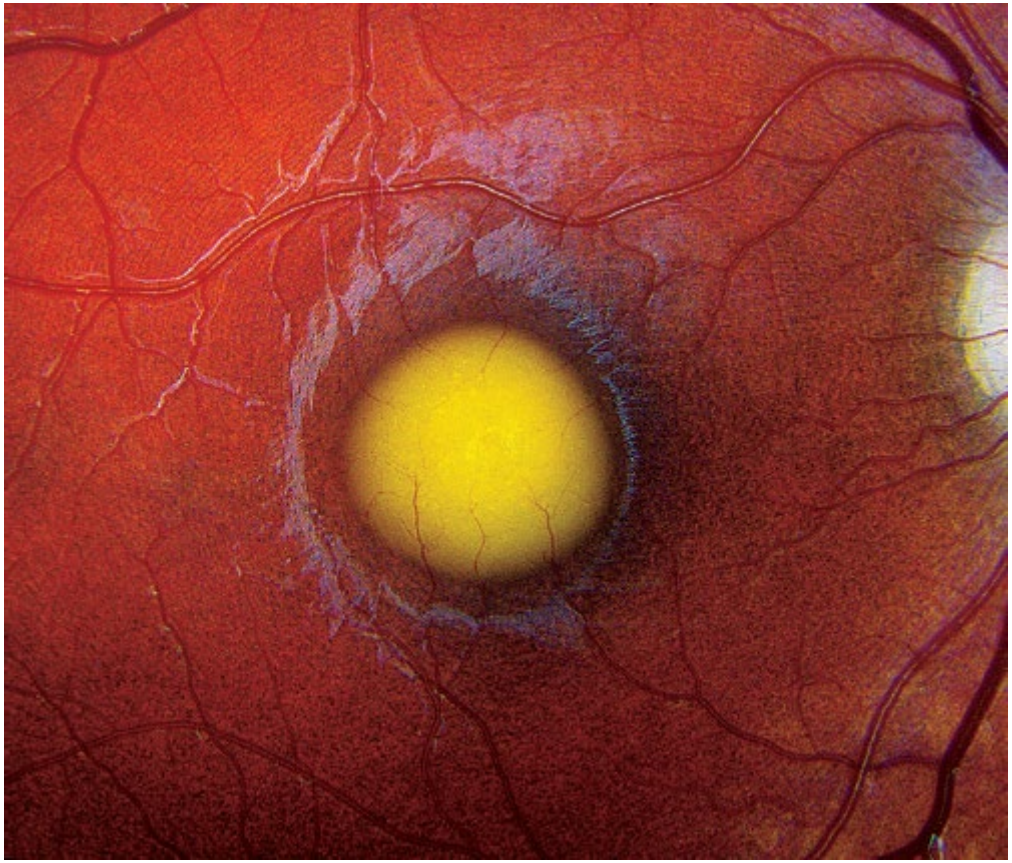

Best dz: ‘Fried egg’ lesion

Q

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Best dz presents with bilateral symmetric yellow macular lesions in childhood.
What is the DDx for a Best-like presentation in an ***adult****?*
--?
--?
--?

A

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--Vitelliform exudative macular detachment
--Drusenoid PED

Q

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Can I assume that, like Best dz, adult-onset fovemacular vitelliform dystrophy (AOFVD) is also AD inheritance?

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- ***What is the name of the gene implicated in Best dz?***
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- *What protein does the Best1 gene code for?*
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Nope. Despite their phenotypic similarities, AOFVD is genetically **unrelated** to Best dz.

Q

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The **pattern dystrophies**

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You can indeed

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The **pattern dystrophies**

What gene is implicated in the pattern dystrophies?

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You can indeed

*Can I assume also that, again like Best dz, the gene implicated in AOFVD **is** Best[illegible]D2?*
Nope. Despite their phenotypic similarities, AOFVD is genetically **unrelated** to Best[illegible].

If not Best dz, to what disease(s) is AOFVD related?
The **pattern dystrophies**

What gene is implicated in the pattern dystrophies?
PRPH2 (formerly Peripherin/RDS)

Q

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Can I as
Nope. D

The BCSC Retina *book identifies four pattern dystrophies by name--what are the other three?*
--
--**A**dult-onset foveomacular vitelliform dystrophy
--
--

Mnemonic is…

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Q/A

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--**B**
--**A**dult-onset foveomacular vitelliform dystrophy
--**R**
--**F**

Mnemonic is…BARF

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The **pattern dystrophies**

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Briefly, what is a pattern dystrophy?

Can I
You can

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The **pattern dystrophies**

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Briefly, what is a pattern dystrophy?
An inherited macular dystrophy that has a characteristic appearance (ie, a particular 'pattern')

-
-
-
-
-

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AD

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Are pattern dystrophies associated with severe vision loss?

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CNVM **GA**

What do CNVM *and* GA *stand for in this context?*
CNVM: ?
GA: ?

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CNVM: Choroidal neovascular membrane
GA: Geographic atrophy

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Cuticular drusen are aka as…

Q/A

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The qualifier 'basal laminar' is intended to convey something about such drusen—what?
It conveys that they are located between the two words of the RPE and the two different words (aka the basal **lamina**) of the RPE cells themselves

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Basal ***linear*** drusen

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Where are basal linear drusen located?
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- ~~Optic nerve head~~ Cuticular drusen are a strong risk factor for development of vitelliform exudative macular detachment **F**
- Pts with **vitelliform exudative macular detachment** are at risk for per

What is the DFE appearance of vitelliform exudative macular detachment (VEMD)?
It's right there in the name—a dome-shaped detachment of the macula containing a yellowish exudate. It looks like a large Best dz lesion.

Best Disease: T/F

VEMD

Q

Best Disease: T/F

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How would one distinguish between a large Best lesion and a VEMD?

Q/A

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By the company they keep—the VEMD lesion with be surrounded by

two words

A

Best Disease: T/F

- Best disease is AD (~~like~~ unlike most inherited retinal diseases) F
- EOG is normal in adult vitelliform disease ~~and~~ but not in Best carriers F
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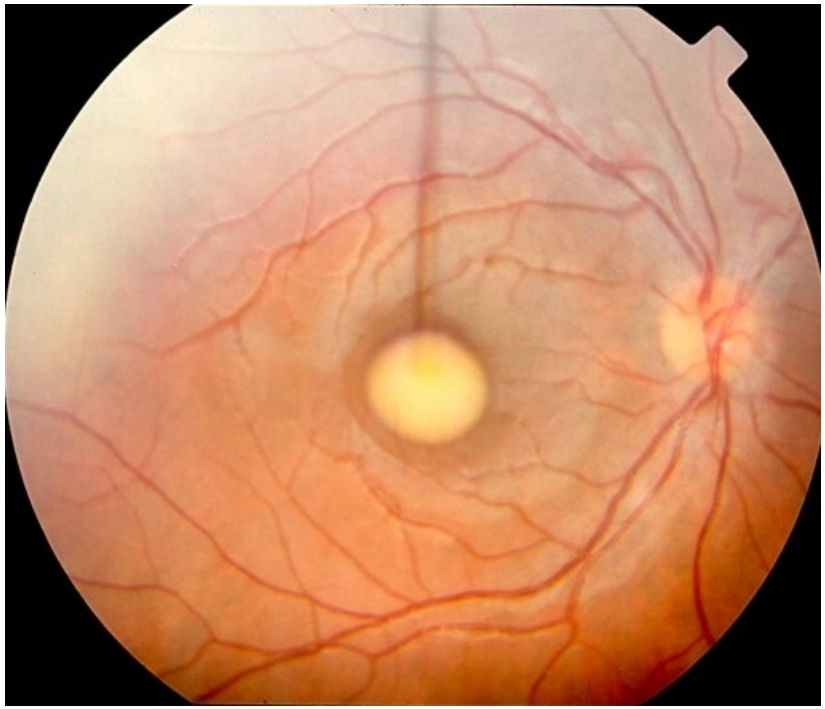
It's right there in the name—a dome-shaped detachment of the macula containing a yellowish exudate. **It looks like a large Best dz lesion**.

How would one distinguish between a large Best lesion and a VEMD?
By the company they keep—the VEMD lesion with be surrounded by cuticular drusen

Best Disease: T/F



Best dz lesion: No cuticular drusen

VEMD lesion: Lotsa cuticular drusen

Q

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What two VEMD sequelae result in permanent vision loss?
--?
--?

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--GA

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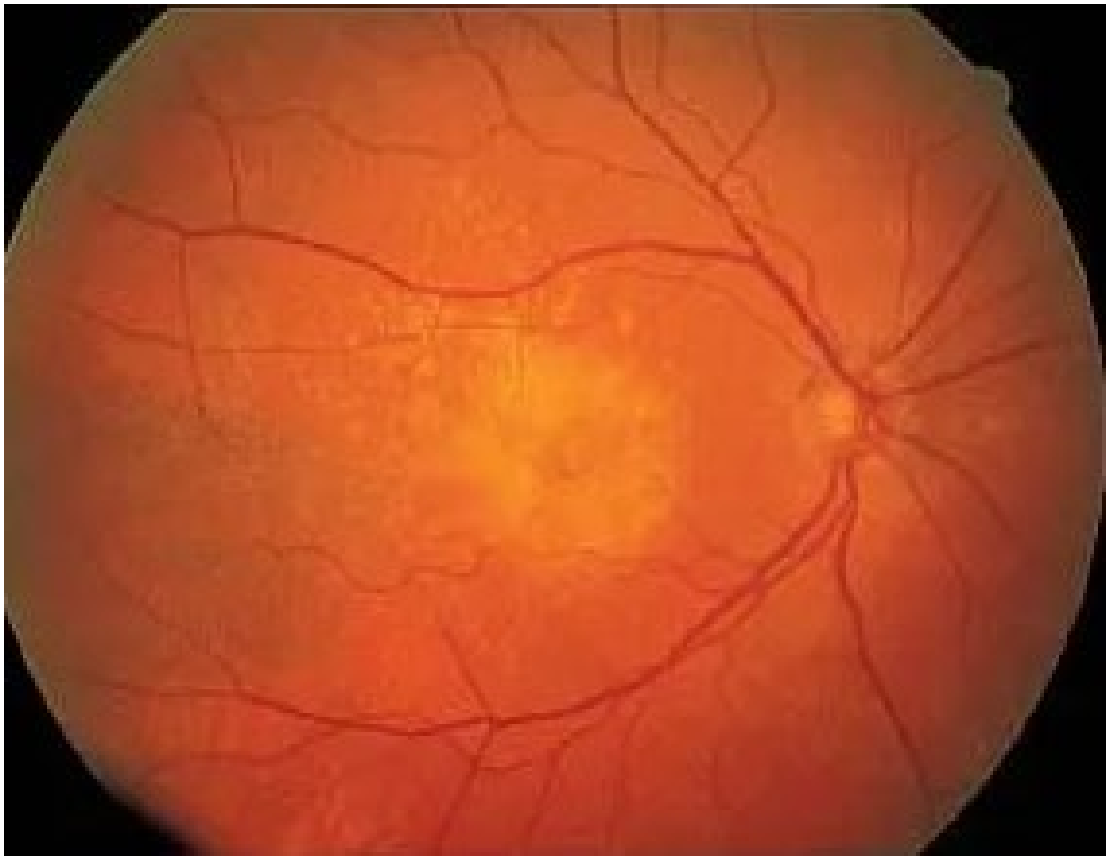
Best Disease: T/F



Fundus photo demonstrating central coalescence of large drusen simulating a macular vitelliform lesion

Q

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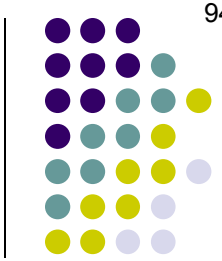

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- Drusenoid PED are strongly associated with ARMD **T**
- **End-stage** Best disease can look like ARMD **T**

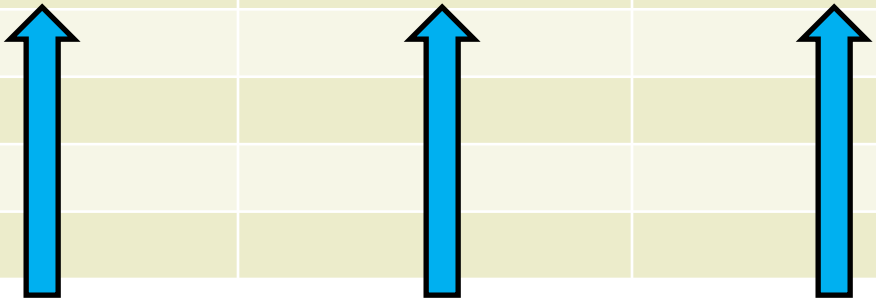
Speaking of stages in Best disease,
let's take a look at them in more depth

Q

Best Disease: T/F

Name the stages of Best dz, and describe the fundus appearance and vision

Stage	Name	Appearance	Vision
I	?	?	?
II			
III			
IV			
V			



Provide the name and appearance of each stage, as well as an estimation of the vision at the stage

A

Best Disease: T/F

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Stage	Name	Appearance	Vision
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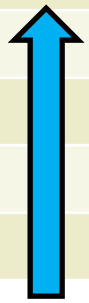
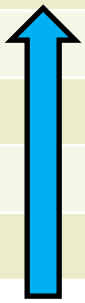
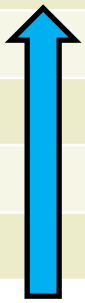
IV			
V			







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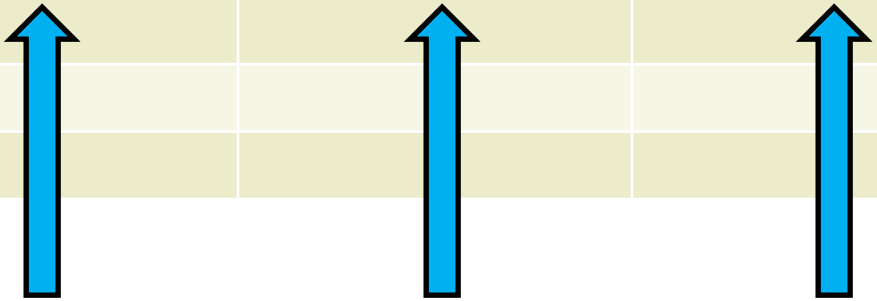

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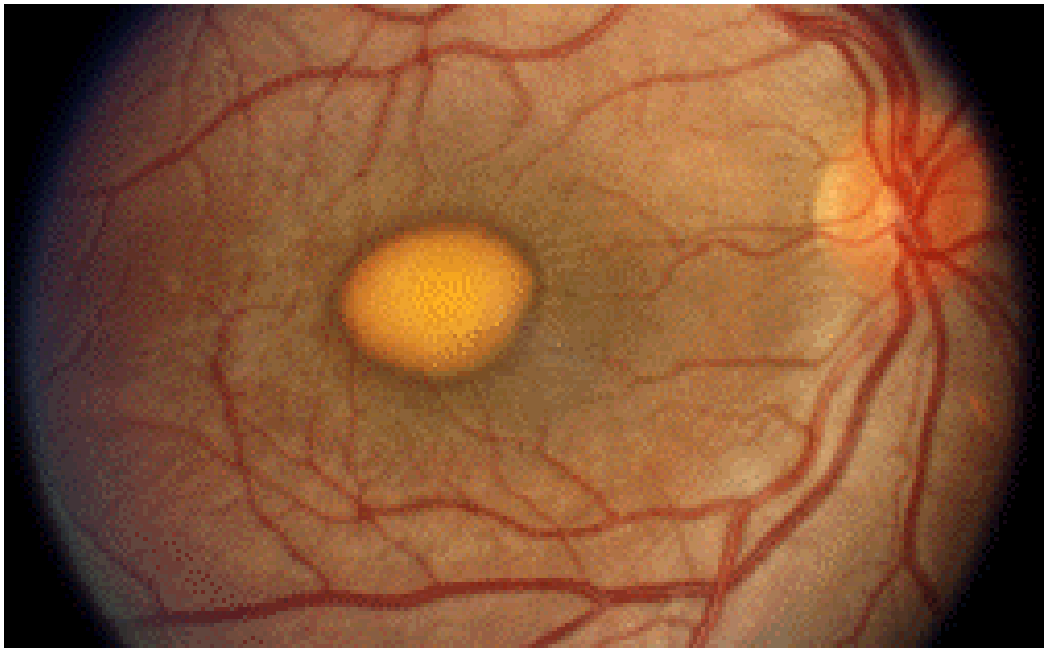

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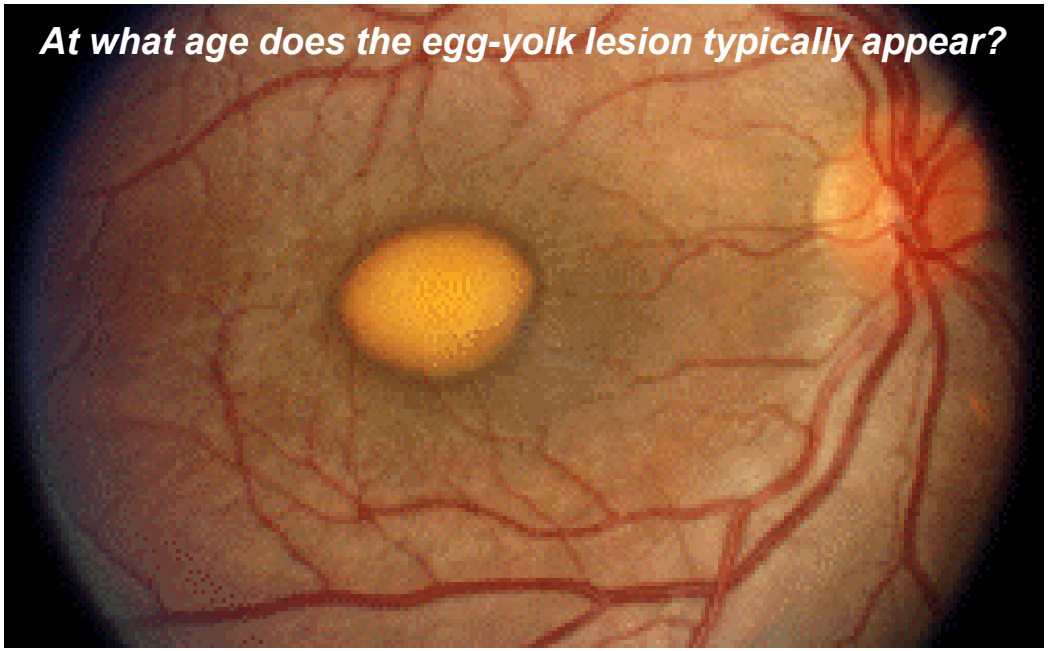

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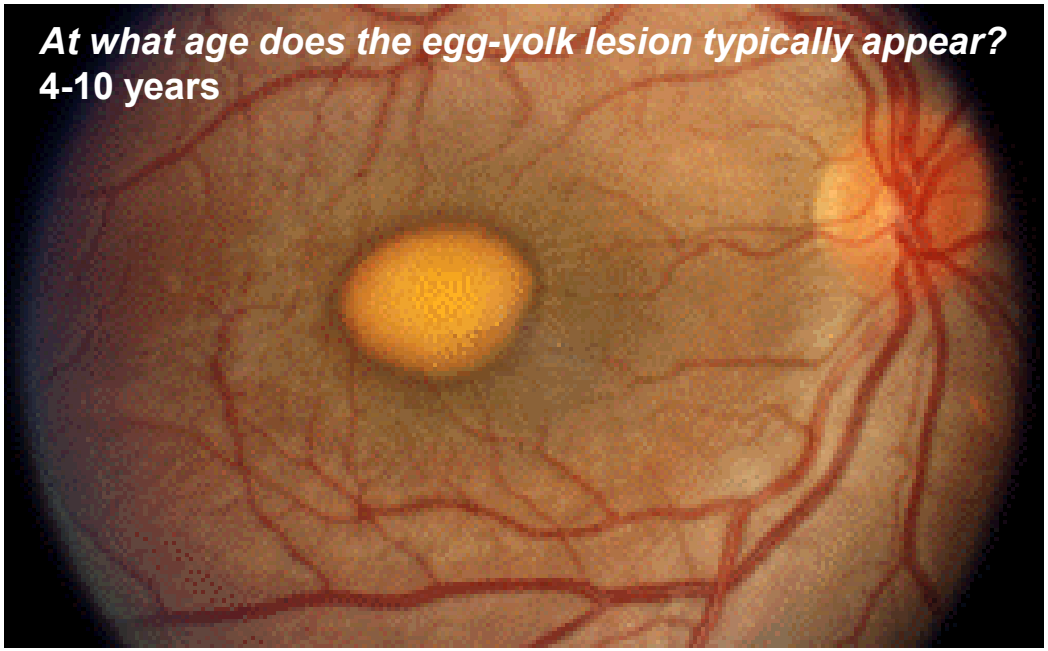

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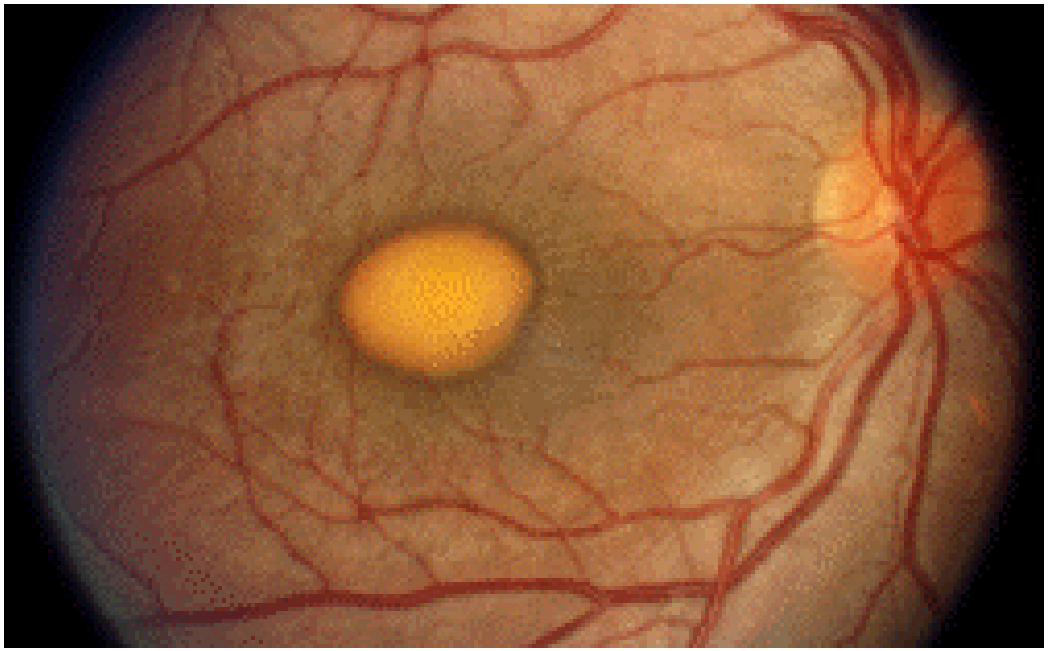

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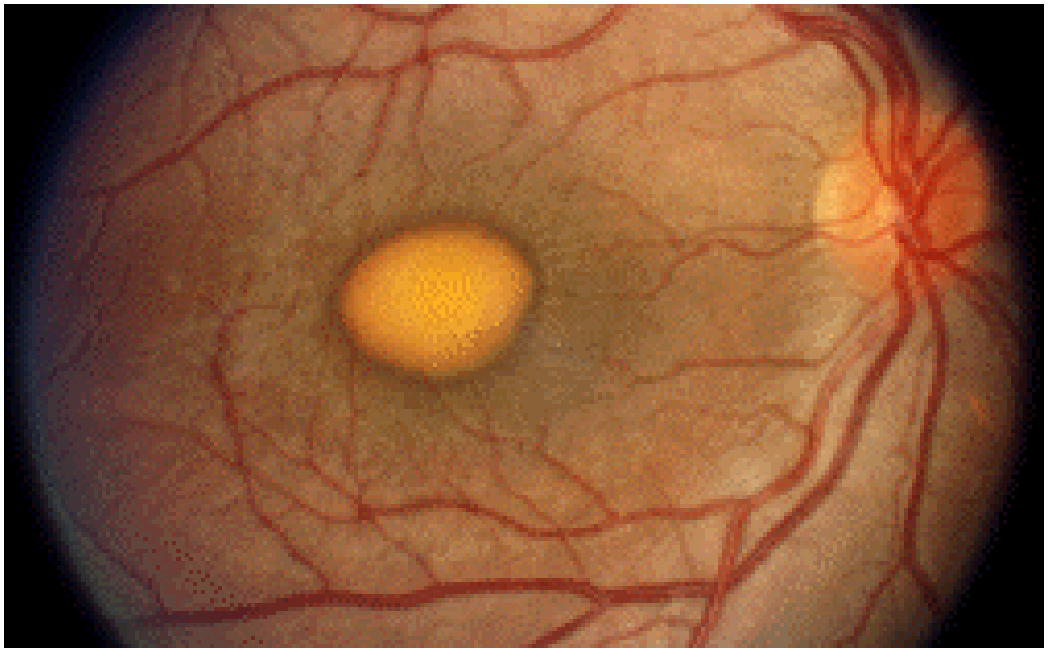

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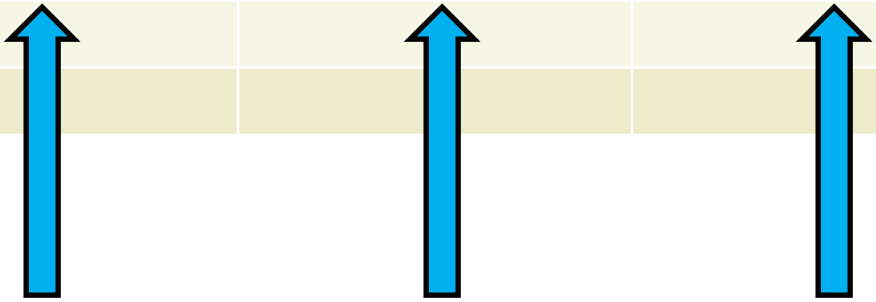

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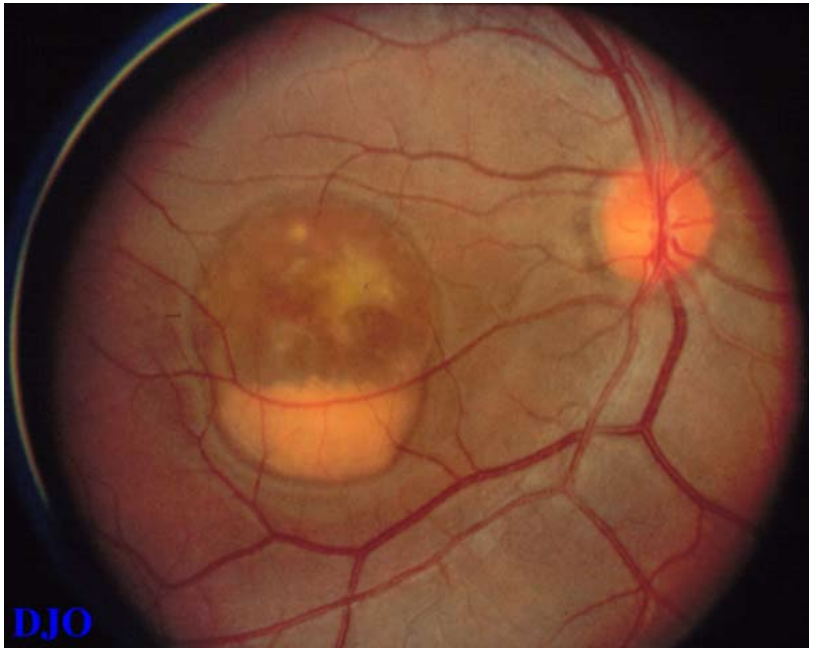
IV			
V			



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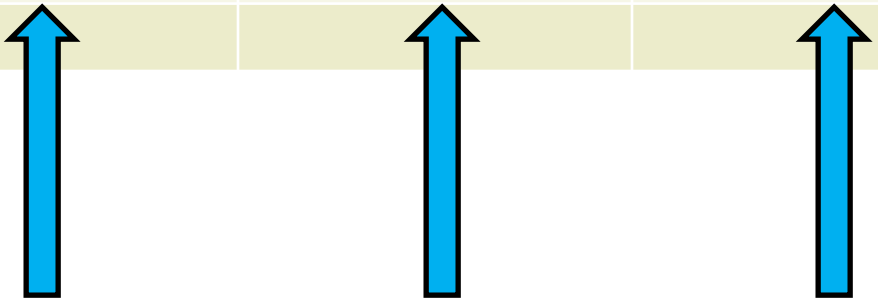
IV	*?*	*?*	*?*
V			



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V			

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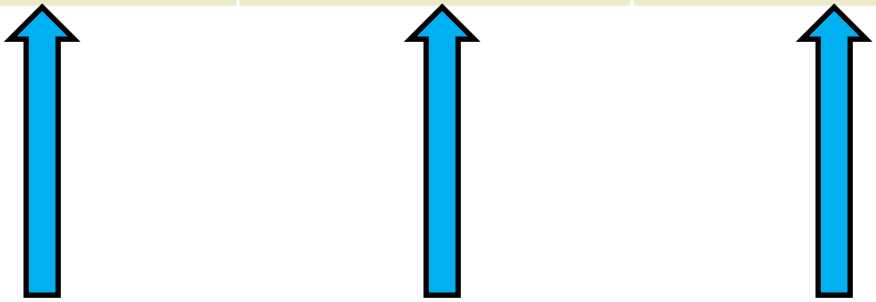
V	***?***	***?***	***?***



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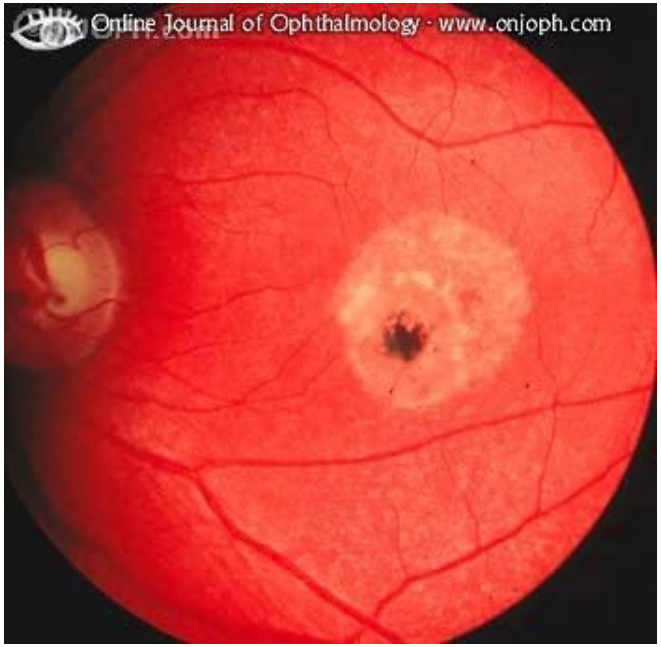
IV	Vitelliruptive	Scrambled eggs	A little worse
V	Atrophic	Dry ARMD-like	20/50 - 20/200



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V	Atrophic	Dry ARMD-like	20/50 - 20/200
VI	*?*	*?*	*?*

What dreaded complication occurs in ~20% of Best pts, and is sometimes referred to as Stage VI disease*?*

A

Best Disease: T/F

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Stage	Name	Appearance	Vision
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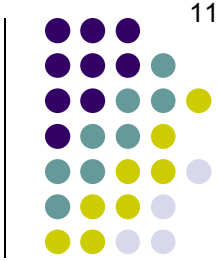
IV	Vitelliruptive	Scrambled eggs	A little worse
V	Atrophic	Dry ARMD-like	20/50 - 20/200
VI	CNVM	Wet-ARMD-like	<20/200

What dreaded complication occurs in ~20% of Best pts, and is sometimes referred to as Stage VI disease*?*
CNVM



Best Disease: T/F

- Best disease is AD (~~like~~ unlike most inherited retinal diseases) **F**
- EOG is normal in adult vitelliform disease ~~and~~ but not in Best carriers **F**

- *Best vitelliform macular dystrophy* is transmitted in an AD fashion (unlike the AR transmission of the majority of inherited retinal diseases). It progresses through a number of well-described stages. In the *pre-vitelliform stage* the fundus appearance is normal, but the EOG is abnormal (as it is in all stages, and carriers). The *vitelliform stage* is marked by the appearance of the classic 'egg yolk' lesion in the macula. A single lesion 1/3 -1/2 DD is typical, but multifocal lesions can occur. Despite all appearances, acuity is usually only minimally affected at this stage. In the *pseudohypopyon stage*, the yellow contents of the egg yolk sink inferiorly and layer out. The *vitelliruptive* (or 'scrambled egg') *stage* is marked by the onset of significant decline in acuity. *End-stage* Best disease is characterized by a disciform scar often similar in appearance to that of late ARMD.

  Because EOG is specific for Best disease, it is a useful adjunct in the work-up for central macular lesions of uncertain etiology.

  Adult-onset foveomacular vitelliform dystrophy is also AD. Onset typically occurs in the fourth or fifth decades. Lesions are smaller than those of Best disease and do not evolve. EOG is normal throughout. Acuity tends to remain quite good.

(tl;dr slide—no questions)