

Q

*Concerning Best disease, and
adult-onset foveomacular vitelliform dystrophy:
Which of the following are true?*

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

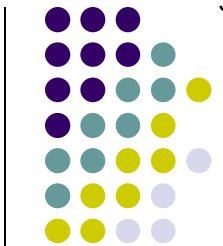




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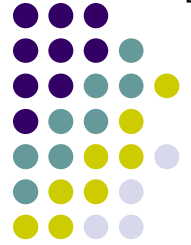


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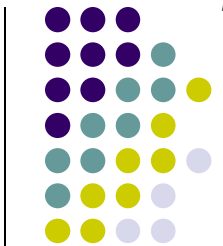
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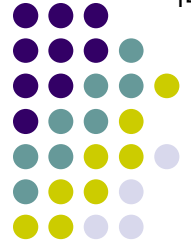
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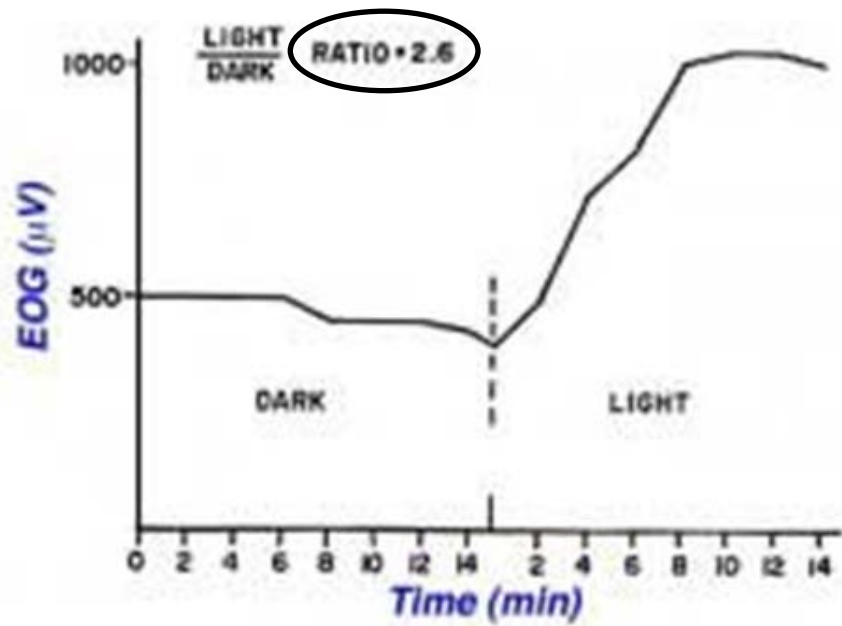
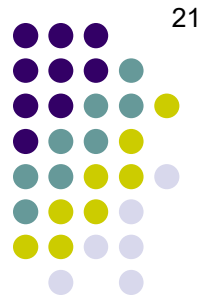
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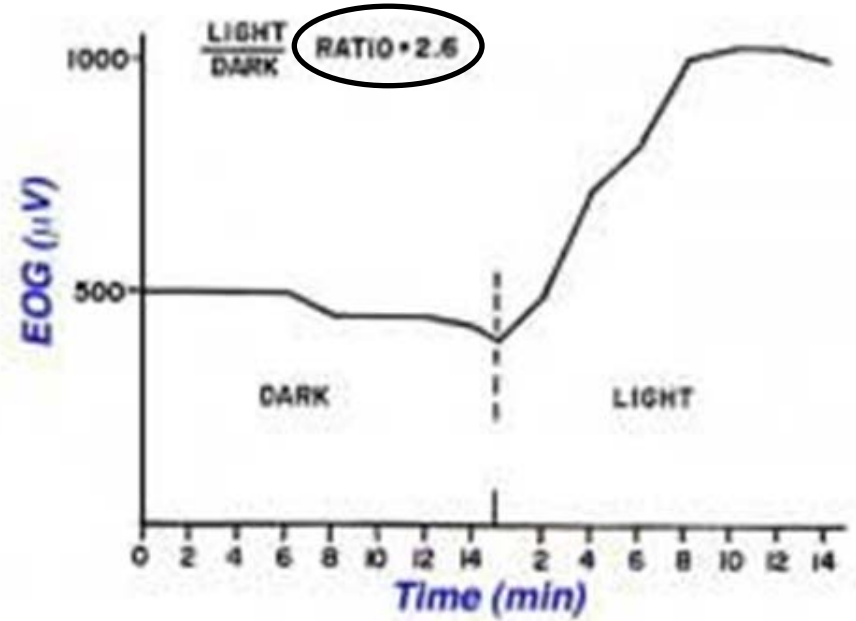
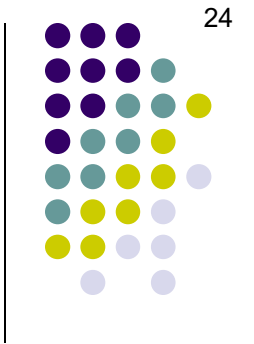
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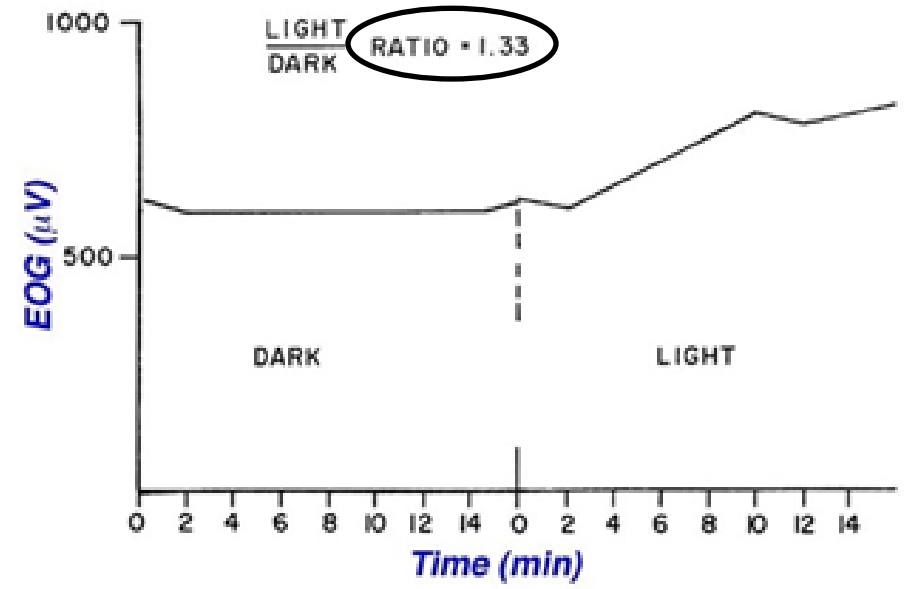
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It is normal, or even supranormal

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Adult-onset foveomacular vitelliform dystrophy *Mnemonic is...*
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--B
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Mnemonic is...BARF

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Can I assume that, like Best dz, adult-onset foveomacular vitelliform dystrophy (AOFVD) is also AD inheritance?
 You can't. The BCSC Retina book identifies four pattern dystrophies by name--what are the other three?
 --Butterfly dystrophy
 --Adult-onset foveomacular vitelliform dystrophy
 --Reticular dystrophy
 --Fundus pulverulentus

If not Best dz, to what disease(s) is AOFVD related?
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 What gene is implicated in the pattern dystrophies?
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Briefly, what is a pattern dystrophy?

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Can I

You can

Can I as

Nope. D

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CNVM = choroidal neovascular membrane

GA = Geographic atrophy

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Concerning Best disease, and
adult-onset foveomacular vitelliform dystrophy:
Which of the following are true?

- 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**
- In Best disease, onset of EOG abnormalities coincides with the development of ~~the vitelliform (fried egg) lesion~~ **F** (*all stages*)
- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage **T**
- Adult-onset foveomacular vitelliform dystrophy has a later onset but a ~~worse~~ ^{better} ultimate visual prognosis than Best disease **F**
- End-stage Best disease can look like ARMD



Concerning Best disease, and
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Which of the following are true?

A

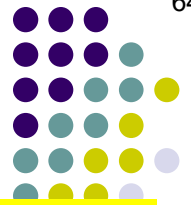
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*Speaking of stages in Best disease,
let's take a look at them in more depth*

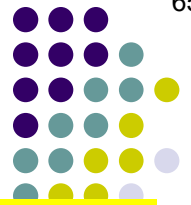


Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy: Which of the following are true?

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

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

Speaking of stages in Best disease, let's take a look at them in more depth
 Provide the name and appearance of each stage, as well as an estimation of the vision at the stage

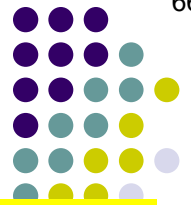


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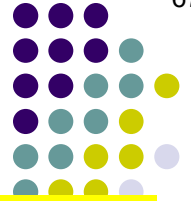
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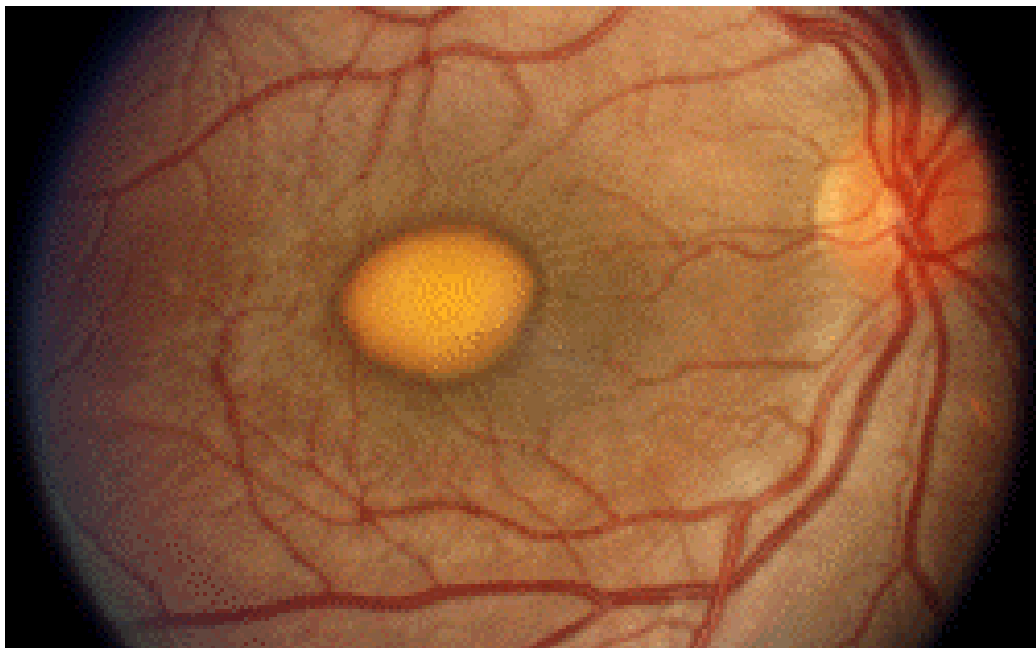
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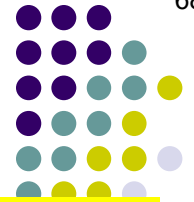


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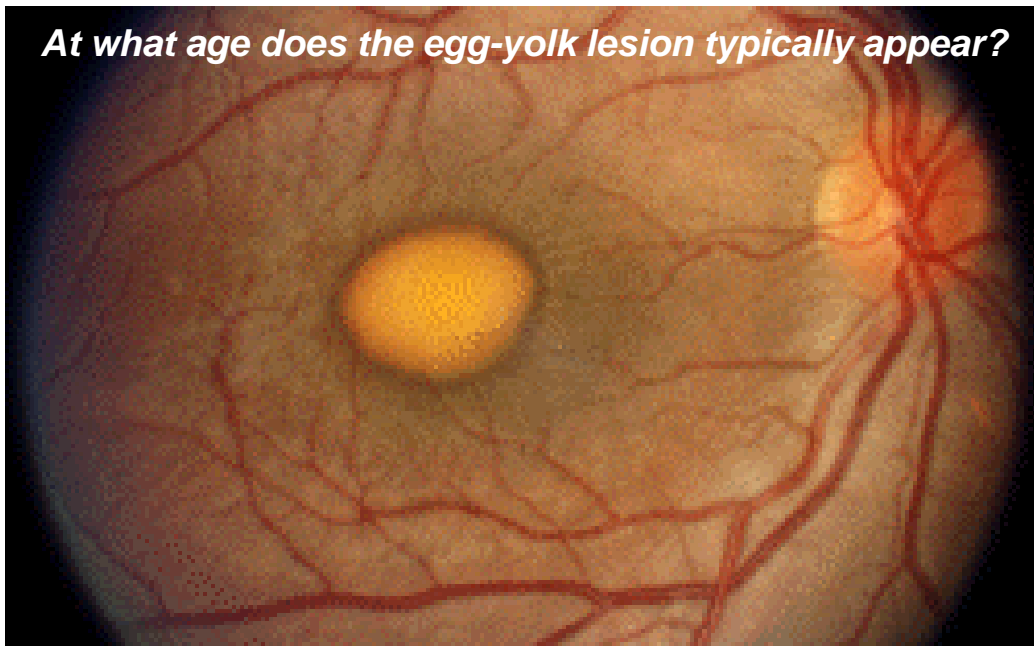
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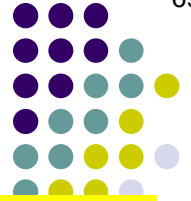
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At what age does the egg-yolk lesion typically appear?



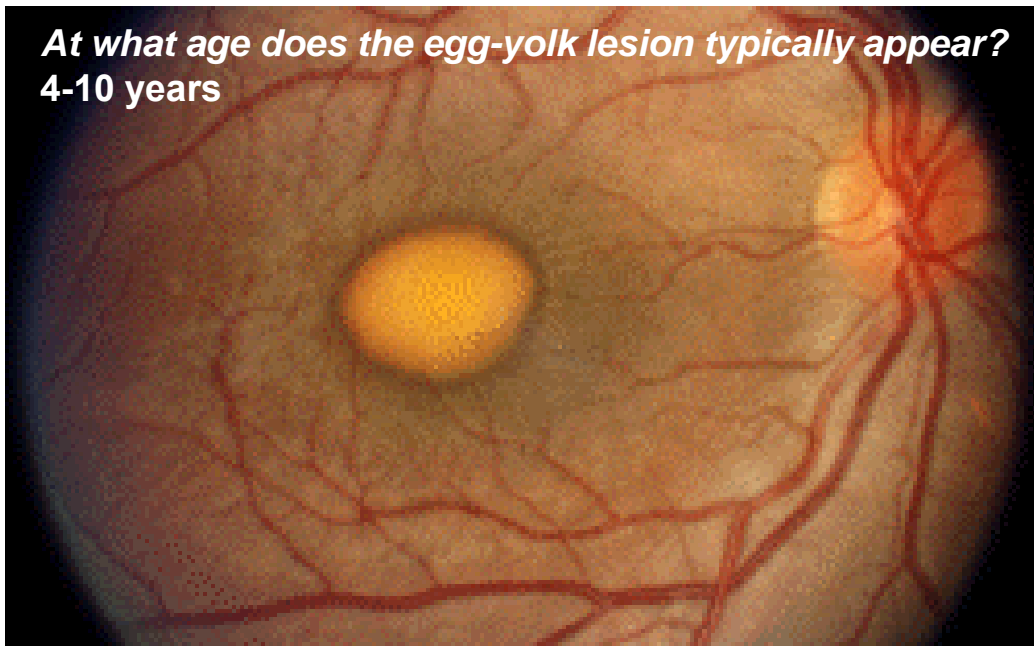
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4-10 years

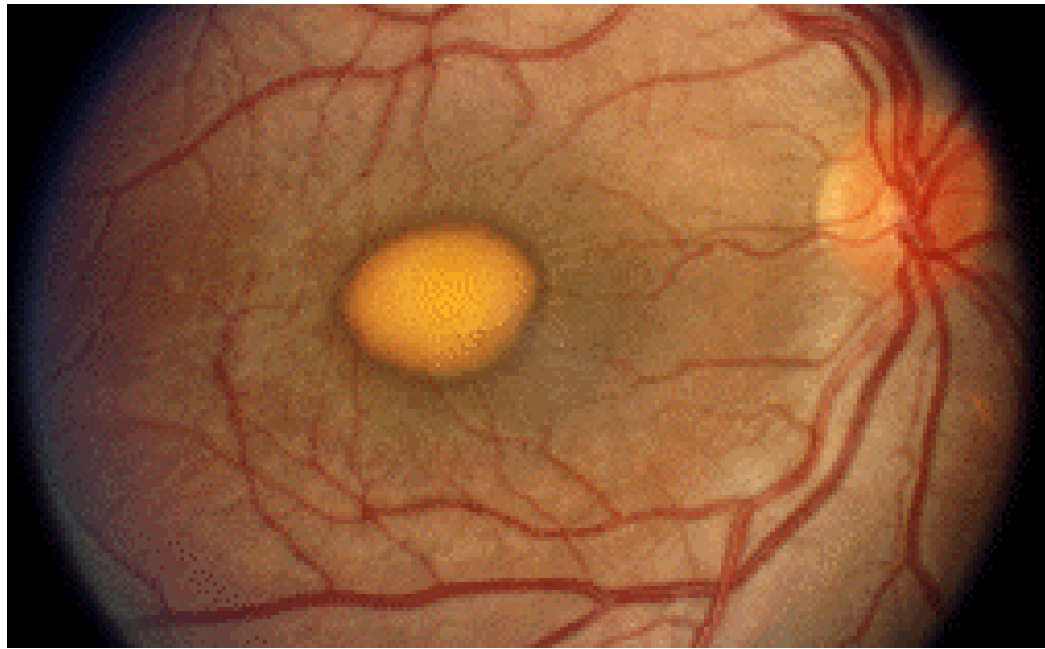




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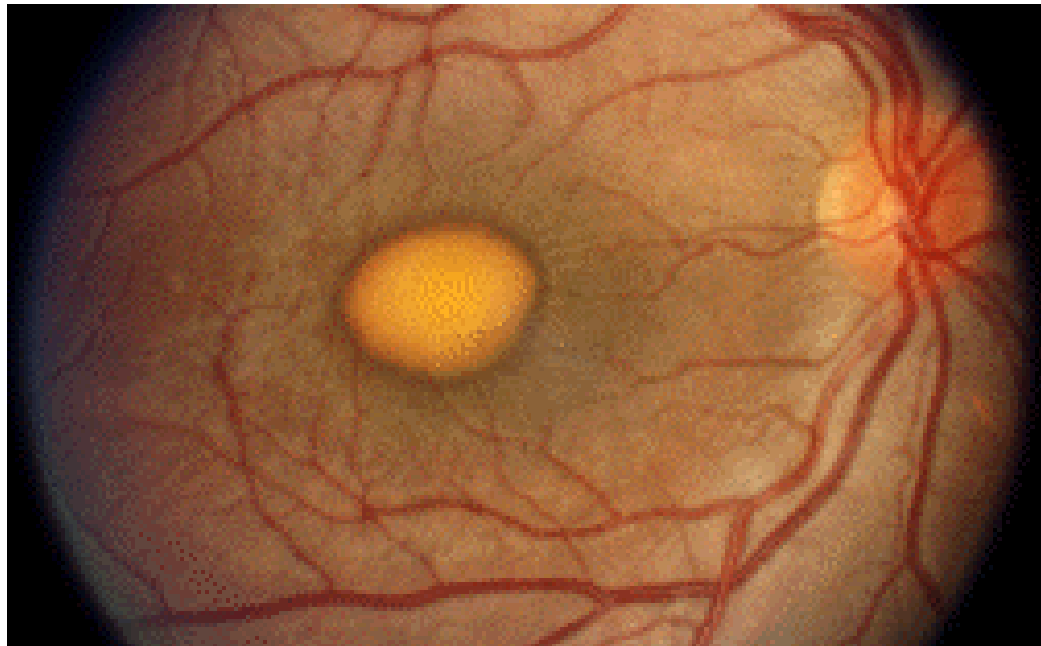
What percent of cases present with multifocal lesions?



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What percent of cases present with multifocal lesions? ~30%

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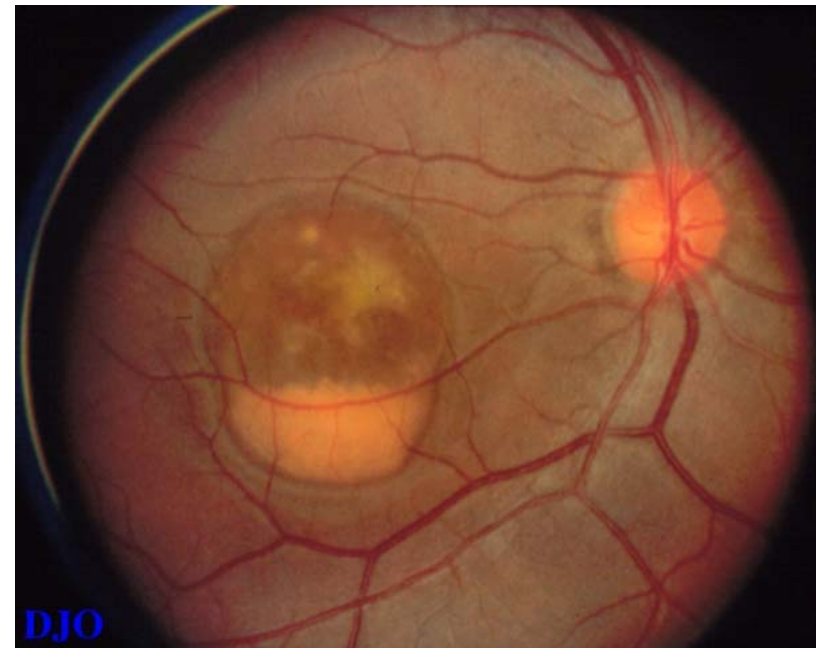
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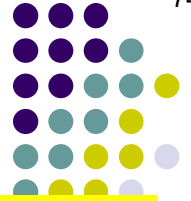


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V			

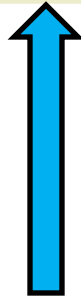


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adult-onset foveomacular vitelliform dystrophy:
Which of the following are true?



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

Stage	Name	Appearance	Vision
I	Pre-vitelliform	Essentially normal	Normal
II	Vitelliform	Egg yolk	+/- mild loss
III	Pseudo-hypopyon	Layered yolk	+/- mild loss
IV	Vitelliruptive	Scrambled eggs	A little worse
V	?	?	?

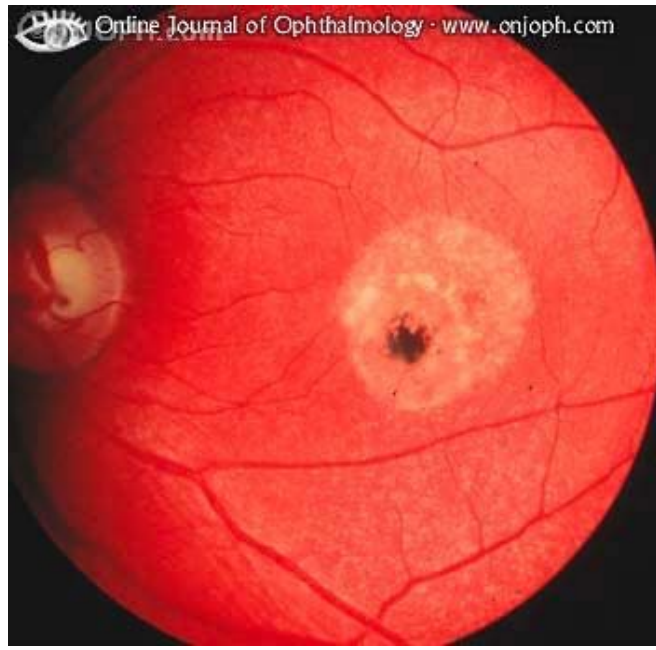




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VI	?	?	?

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



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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



Concerning Best disease, and
adult-onset foveomacular vitelliform dystrophy:
Which of the following are true?

- Best disease is AD (~~like~~ ^{unlike} most inherited retinal diseases) F
- EOG is normal in adult vitelliform disease ^{but not} and 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.

(Summary slide—no questions)