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

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

Best1 (or VMD2)

What protein does the Best1 gene code for?

Bestrophin

Abnormalities in bestrophin lead to accumulation of what material in RPE cells?

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In a nutshell, what does an electro-oculogram measure?
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Again in a nutshell, how does it work?
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.

Below 1.7
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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

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

The normal range for the Arden ratio is 1.9-2.8
At what value is the Arden ratio considered definitely abnormal?
Below 1.7

In Best dz (and often in asymptomatic carriers), the Arden ratio is reduced. What is typical status of the **ERG**?
A

Best disease is AD (like most inherited retinal diseases) F
EOG is normal in adult vitelliform disease and in Best carriers F

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

- Unlike
- but not

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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)

What is typical status of the ERG in Best dz?
It is normal, or even supranormal

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

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- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage ✗
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Can I assume that, like Best dz, adult-onset foveomacular vitelliform dystrophy (AOFVD) is also AD inheritance?
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Can I assume that, like Best dz, adult-onset foveomacular vitelliform dystrophy (AOFVD) is also AD inheritance? You can indeed
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy: Which of the following are true?

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

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Can I assume that, like Best dz, adult-onset foveomacular vitelliform dystrophy (AOFVD) is also AD inheritance? You can... **F**

Can I assume also that, again like Best dz, the gene implicated in AOFVD is **Best1/VMD2**? **Nope. D**

The BCSC Retina book identifies four pattern dystrophies by name--what are the other three? **--**
--Adult-onset foveomacular vitelliform dystrophy **--**
--Mnemonic is... **--**

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Briefly, what is a pattern dystrophy?

- It is an inherited macular dystrophy that has a characteristic appearance (i.e., a particular pattern).

What is the inheritance pattern?

- AD

Are pattern dystrophies associated with severe vision loss?

- Generally no—vision is only slightly affected (although patients are at a mildly increased risk of developing CNVM and/or GA late in life, either of which can significantly impact acuity).

Do the macular 'patterns' appear early in life?

- Generally no—they usually show up in middle adulthood.

Are the macular 'patterns' stable?

- Generally no—three sorts of instability are common:
  - Members of the same family can present with different patterns
  - The same individual can have different patterns in their two eyes
  - The pattern in a given eye can evolve over time from one pattern to another

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- The pattern dystrophies

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**Are pattern dystrophies associated with severe vision loss?**

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**Concerning Best disease, and AOFVD:**

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- 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 ultimate visual prognosis than Best disease  F
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What percent of cases present with multifocal lesions?
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What dreaded complication occurs in ~20% of Best pts, and is sometimes referred to as Stage VI disease?
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**What dreaded complication occurs in ~20% of Best pts, and is sometimes referred to as Stage VI disease?**

**CNVM**
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 is in the 30s – 40s. Lesions are smaller than those of Best disease and do not evolve. EOG is normal throughout. Acuity tends to remain quite good.

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

- Best disease is AD (like most inherited retinal diseases)  F
- EOG is normal in adult vitelliform disease and 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 ultimate visual prognosis than Best disease  F