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Abnormalities in bestrophin lead to accumulation of what material in RPE cells?
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**What does \textit{EOG} stand for?**

- \textit{Electro-oculogram}

In a nutshell, what does an electro-oculogram measure?
- An electrophysiology test that measures RPE function

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

What is this ratio called?
- The \textit{Arden ratio}

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

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 \textit{E}R\textit{G} in Best?
● Best disease is AD (like most inherited retinal diseases)  F
● EOG is normal in adult vitelliform disease and in Best carriers  F

What does EOG stand for?  
Electro-oculogram
Best Disease: T/F

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**What does EOG stand for?**
Electro-oculogram

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

- Best disease is AD (like most inherited retinal diseases)  F
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**What does EOG stand for?**
Electro-oculogram

**In a nutshell, what does an electro-oculogram measure?**
RPE function
Best disease is AD (like most inherited retinal diseases) \( \text{F} \)

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Non-Best pts

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Best Disease: T/F

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What is typical status of the ERG in Best dz?
It is normal, or even supranormal
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In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion
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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?

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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?*

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--Vitelliform exudative macular detachment
--Drusenoid PED
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What protein does the Best1 gene code for? 
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Abnormalities in bestrophin lead to accumulation of what material in RPE cells? 
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What gene is implicated in the pattern dystrophies? PRPH2 (formerly Peripherin/RDS)
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The BCSC Retina book identifies four pattern dystrophies by name—what are the other three?

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Mnemonic is…

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---B
---A
---R
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The BCSC Retina book identifies four pattern dystrophies by name—what are the other three?
- **Butterfly dystrophy**
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Generally no--vision is only slightly affected (although pts are at a mildly increased risk of developing and/or late in life, either of which can significantly impact acuity)

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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 demonstrate 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

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

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

What gene is implicated in the pattern dystrophies?
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An inherited macular dystrophy that has a characteristic appearance (ie, 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 pts 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 demonstrate different patterns
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• 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**
• Optic nerve head drusen are a strong risk factor for development of vitelliform exudative macular detachment  **F**
Best Disease: T/F

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Cuticular drusen are aka as…
Q/A

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Cuticular drusen are aka as…basal laminar drusen
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The qualifier ‘basal laminar’ is intended to convey something about such drusen—what?
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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 different words of the RPE and the  two different words (aka the basal lamina) of the RPE cells themselves

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Another sort of drusen is also ‘basal [something] drusen.’ What is the word in the middle?
Best Disease: T/F

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Where are basal linear drusen located?
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Where are basal linear drusen located?
Within Bruch’s membrane
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What is the DFE appearance of vitelliform exudative macular detachment (VEMD)?
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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.
Best Disease: T/F

VEMD
Best Disease: T/F

- Best disease is AD (like most inherited retinal diseases)  **F**
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- 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**
- Optic nerve head drusen are a strong risk factor for development of vitelliform exudative macular detachment  **F**
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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.**

How would one distinguish between a large Best lesion and a VEMD?
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How would one distinguish between a large Best lesion and a VEMD? By the company they keep—the VEMD lesion with be surrounded by two words.
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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 will be surrounded by cuticular drusen
Best disease: T/F

Best dz lesion: No cuticular drusen

VEMD lesion: Lotsa cuticular drusen
Best disease is AD (like most inherited retinal diseases)  F
EOG is normal in adult vitelliform disease and in Best carriers  F
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Fundus photo demonstrating central coalescence of large drusen simulating a macular vitelliform lesion
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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
Name the stages of Best dz, and describe the fundus appearance and vision

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Provide the name and appearance of each stage, as well as an estimation of the vision at the stage.
Name the stages of Best dz, and describe the fundus appearance and vision

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At what age does the egg-yolk lesion typically appear?
### Best Disease: T/F

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**At what age does the egg-yolk lesion typically appear?**
4-10 years
**Name the stages of Best dz, and describe the fundus appearance and vision**

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**Best Disease: T/F**

What percent of cases present with multifocal lesions?
### Name the stages of Best dz, and describe the fundus appearance and vision

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- **Best Disease: T/F**

*What percent of cases present with multifocal lesions? ~30%*
Name the stages of Best dz, and describe the fundus appearance and vision

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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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<td>Vitelliform</td>
<td>Egg yolk</td>
<td>+/- mild loss</td>
</tr>
<tr>
<td>III</td>
<td>Pseudo-hypopyon</td>
<td>Layered yolk</td>
<td>+/- mild loss</td>
</tr>
<tr>
<td>IV</td>
<td>Vitelliruptive</td>
<td>Scrambled eggs</td>
<td>A little worse</td>
</tr>
<tr>
<td>V</td>
<td>Atrophic</td>
<td>Dry ARMD-like</td>
<td>20/50 - 20/200</td>
</tr>
<tr>
<td>VI</td>
<td>CNVM</td>
<td>Wet-ARMD-like</td>
<td>&lt;20/200</td>
</tr>
</tbody>
</table>

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 most inherited retinal diseases)  F
- EOG is normal in adult vitelliform disease 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.

*(tl;dr slide—no questions)*