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

Best Disease: T/F
Best disease is AD (like most inherited retinal diseases)  \( \text{F} \)

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

**Best Disease: T/F**
Best disease is AD (like most inherited retinal diseases)  \[\text{F}\]

What is the name of the gene implicated in Best dz?
Best1 (or VMD2)
Best disease is AD (like most inherited retinal diseases)  F

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

What protein does the Best1 gene code for?
Best disease is AD (like most inherited retinal diseases) F

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

What protein does the Best1 gene code for?
Bestrophin
Best disease is AD (like most inherited retinal diseases)  F

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?
Lipofuscin
Best disease is AD (like most inherited retinal diseases) **F**

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? Lipofuscin
Best disease is AD (like most inherited retinal diseases) **F**

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

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 **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 **E**R**G** in Best **unlike** but not **F**
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

- 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

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

What does EOG stand for?
Electro-oculogram

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

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 **ERG** in
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

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

Again in a nutshell, how does it work?
Best disease is AD (like most inherited retinal diseases) \( \text{F} \)

**EOG** is normal in adult vitelliform disease and in Best carriers \( \text{F} \)

---

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

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

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**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

What does EOG stand for?
Electro-oculogram

In a nutshell, what does an electro-oculogram measure?
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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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**What does EOG stand for?**
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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?**
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- 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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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

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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

What is this ratio called?
The Arden ratio

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

EOG

Best Disease: T/F
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**

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

**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

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**What is the normal range for the Arden ratio?**
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**At what value is the Arden ratio considered definitely abnormal?**
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**

What does EOG stand for?
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In a nutshell, what does an electro-oculogram measure?
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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**

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 (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

EOG
Q

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

What does EOG stand for?  
Electro-oculogram

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

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

**In a nutshell, what does an electro-oculogram measure?**
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 **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 (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
Best disease is AD (like most inherited retinal diseases) \(\text{F}\)
EOG is normal in adult vitelliform disease and in Best carriers \(\text{F}\)
In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion
- Best disease is AD (like most inherited retinal diseases) \( \text{F} \)
- EOG is normal in adult vitelliform disease and in Best carriers \( \text{F} \)
- In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion \( \text{F} \) (all stages)
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**
- 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
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**
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**
- 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**

*Best dz presents with bilateral symmetric yellow macular lesions in childhood.*

*What is the DDx for a Best-like presentation in an adult?*

--?
--?
--?
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**

Best dz presents with bilateral symmetric yellow macular lesions in childhood. What is the DDx for a Best-like presentation in an adult?
--Adult-onset foveomacular vitelliform dystrophy
--Vitelliform exudative macular detachment
--Drusenoid PED
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
- 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**
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 **T** has a later onset but a worse ultimate visual prognosis than Best disease **F**

Can I assume that, like Best dz, adult-onset foveomacular vitelliform dystrophy (AOFVD) is also AD inheritance? **F**
- Best disease is AD (like most inherited retinal diseases) \( \text{F} \)
- EOG is normal in adult vitelliform disease and in Best carriers \( \text{F} \)
- In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion \( \text{F} \) (all stages)
- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage \( \text{T} \)
- Adult-onset foveomacular vitelliform dystrophy \( \text{T} \) has a later onset but a worse ultimate visual prognosis than Best disease \( \text{F} \)

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

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

Can I assume also that, again like Best dz, the gene implicated in AOFVD is Best1/VMD2?
Best Disease: T/F

- 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? Lipofuscin
- Adult-onset foveomacular vitelliform dystrophy has a later onset but a worse ultimate visual prognosis than Best disease.

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

Can I assume also that, again like Best dz, the gene implicated in AOFVD is Best1/VMD2? Nope. Despite their phenotypic similarities, AOFVD is genetically unrelated to Best dz.
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**
- 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**

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

**Can I assume also that, again like Best dz, the gene implicated in AOFVD is Best1/VMD2?**
Nope. Despite their phenotypic similarities, **AOFVD is genetically unrelated to Best dz.**

**If not Best dz, to what disease(s) is AOFVD related?**
- Best disease is AD (like most inherited retinal diseases) \( \text{F} \)
- EOG is normal in adult vitelliform disease and in Best carriers \( \text{F} \)
- In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion \( \text{F} \) \( \text{(all stages)} \)
- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage \( \text{T} \)
- Adult-onset foveomacular vitelliform dystrophy has a later onset but a worse ultimate visual prognosis than Best disease \( \text{F} \)

**Best Disease: T/F**

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

You can indeed

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

Nope. Despite their phenotypic similarities, **AOFVD is genetically unrelated to Best dz.**

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

The **pattern dystrophies**
Q

- 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

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

Can I assume also that, again like Best dz, the gene implicated in AOFVD is Best1/VMD2? Nope. Despite their phenotypic similarities, AOFVD is genetically unrelated to Best.

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

The pattern dystrophies

What gene is implicated in the pattern dystrophies?
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
- 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

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

Can I assume also that, again like Best dz, the gene implicated in AOFVD is Best1/VMD2? Nope. Despite their phenotypic similarities, AOFVD is genetically unrelated to Best.

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)
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**
- 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**

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

The BCSC Retina book identifies four pattern dystrophies by name—what are the other three?

- Adult-onset foveomacular vitelliform dystrophy  **Mnemonic is…**
- 
- 

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)
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)
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- Adult-onset foveomacular vitelliform dystrophy has a later onset but a worse ultimate visual prognosis than Best disease F

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

The BCSC Retina book identifies four pattern dystrophies by name--what are the other three?

- B
- A
- R
- F

Mnemonic is...BARF

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

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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? The pattern dystrophies

What gene is implicated in the pattern dystrophies? PRPH2 (formerly Peripherin/RDS)
Briefly, what is a pattern dystrophy?

The BCSC Retina book identifies four pattern dystrophies by name—what are the other three?

- Butterfly dystrophy
- Adult-onset foveomacular vitelliform dystrophy
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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)
Briefly, what is a pattern dystrophy?  
An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’)

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

Can I assume also that, again like Best dz, the gene implicated in AOFVD is Best1/VMD2?  
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---Reticular dystrophy  
---Fundus pulverulentus

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

What is the inheritance pattern?

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

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Are pattern dystrophies associated with severe vision loss?

The BCSC Retina book identifies four pattern dystrophies by name--what are the other three?
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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 and/or late in life, either of which can significantly impact acuity)

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

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

What gene is implicated in the pattern dystrophies?
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Do the macular ‘patterns’ appear early in life?

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

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What do CNVM and GA stand for in this context?

CNVM: Choroidal neovascular membrane
GA: Geographic atrophy

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- In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion \(\text{F}\)
- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage \(\text{T}\)
- Adult-onset foveomacular vitelliform dystrophy has a later onset but a worse ultimate visual prognosis than Best disease \(\text{F}\)
- Optic nerve head drusen are a strong risk factor for development of vitelliform exudative macular detachment \(\text{F}\)
Best disease is AD (like most inherited retinal diseases)  F
EGO is normal in adult vitelliform disease and in Best carriers  F
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**Cuticular drusen** are aka as…
Q/A

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Cuticular drusen are aka basal drusen

Best Disease: T/F
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The qualifier ‘basal laminar’ is intended to convey something about such drusen—what?
Best Disease: T/F

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**Cuticular drusen** are aka as **basal laminar drusen**

*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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**Cuticular drusen are aka as…** \( \text{basal laminar} \) drusen

The qualifier ‘basal laminar’ is intended to convey something about such drusen—what?

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

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Basal linear drusen

Where are basal linear drusen located?
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*Where are basal linear drusen located? Within Bruch’s membrane*
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- Optic nerve head drusen are a strong risk factor for development of vitelliform exudative macular detachment  **F**
- Pts with **vitelliform exudative macular detachment** are at risk for permanent significant vision loss  **F**

What is the DFE appearance of vitelliform exudative macular detachment (VEMD)?

Unlike but not better

Cuticular optic nerve

Worse
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  \( F \)
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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.
Best Disease: T/F

VEMD
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
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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?
Best Disease: T/F

- Best disease is AD (like most inherited retinal diseases)  F
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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 cuticular drusen.
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It's right there in the name—a dome-shaped detachment of the macula containing a yellowish exudate. \textbf{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 dz lesion: No cuticular drusen

VEMD lesion: Lotsa cuticular drusen

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**

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**

Pts with **vitelliform exudative macular detachment** are at risk for permanent significant vision loss **T**

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.

What two VEMD sequelae result in permanent vision loss?
--?
--?
Best Disease: T/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.

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

--CNVM
--GA
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• Pts with vitelliform exudative macular detachment are at risk for permanent significant vision loss \textbf{T}
• Drusenoid PED are strongly associated with ARMD \textbf{T}
Color fundus photograph shows central coalescence of large drusen simulating a macular vitelliform lesion.
Best Disease: T/F

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- End-stage Best disease can look like ARMD
Best Disease: T/F

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- **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**
**Best Disease: T/F**

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

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

![Image of eye with egg-yolk lesion]
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?
4-10 years
### Name the stages of Best dz, and describe the fundus appearance and vision

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**Best Disease: T/F**
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What percent of cases present with multifocal lesions? ~30%

Best Disease: T/F
### Name the stages of Best dz, and describe the fundus appearance and vision

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**Best Disease: T/F**

Q
**Best Disease: T/F**

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

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<td>Egg yolk +/- mild loss</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>?</td>
<td>?</td>
<td>?</td>
</tr>
</tbody>
</table>

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

Best Disease: T/F
### Name the stages of Best dz, and describe the fundus appearance and vision

<table>
<thead>
<tr>
<th>Stage</th>
<th>Name</th>
<th>Appearance</th>
<th>Vision</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>Pre-vitelliform</td>
<td>Essentially normal</td>
<td>Normal</td>
</tr>
<tr>
<td>II</td>
<td>Vitelliform</td>
<td>Egg yolk</td>
<td>+/- mild loss</td>
</tr>
<tr>
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<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.