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

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

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

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

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

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

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

- Best disease is AD (like most inherited retinal diseases)
- What is the name of the gene implicated in Best dz? Best1 (or VMD2)
- What protein does the Best1 gene code for? Bestrophin

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

- Best disease is AD (like most inherited retinal diseases)
- What is the name of the gene implicated in Best dz? Best1 (or VMD2)
- What protein does the Best1 gene code for? Bestrophin
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy: Which of the following are true?

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

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)

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
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
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**
Which of the following are true?

- 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?
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**

What does EOG stand for?
Electro-oculogram
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

What does EOG stand for?
Electro-oculogram

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

What does EOG stand for?
 Electro-oculogram

In a nutshell, what does an electro-oculogram measure?
 RPE function
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy: Which of the following are true?

- 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?
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

**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.
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

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?
Arden ratio

The normal range for the Arden ratio is 1.9-2.8.
Below 1.7, the Arden ratio is considered definitely abnormal.
In Best dz (and often in asymptomatic carriers), the Arden ratio is reduced.

Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy:
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?

F EOG RPE function

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

In Best dz (and often in asymptomatic carriers), the Arden ratio is reduced. What is typical status of the E R G in unlike but not
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**

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
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

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

- Non-Best pts

EOG
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

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

- EOG
- Best pts
- Non-Best pts
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**

**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?**
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**

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

- **Best disease is AD** (like most inherited retinal diseases)  
- EOG is normal in adult vitelliform disease and in Best carriers  
- In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion  
- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage  
- 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?
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy: Which of the following are true?

- **Best disease is AD**

- EOG is normal in adult vitelliform disease and in Best carriers

- In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion

- In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage

- **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
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)
- **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?
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)
- 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.
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

**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?**
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**

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**
**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

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**

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

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)
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

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

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.

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

---

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)
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

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

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.

The pattern dystrophies

What gene is implicated in the pattern dystrophies?  PRPH2 (formerly Peripherin/RDS)
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy: Which of the following are true?

- 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** 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't assume that. Despite their phenotypic similarities, AOFVD is genetically unrelated to Best dz.

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

- **Butterfly dystrophy**
- **A** dult-onset foveomacular vitelliform dystrophy
- **R** eticular dystrophy
- **F** undus 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)
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy:

Which of the following are true?

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.

Briefly, what is a pattern dystrophy?

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 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

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)
**Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy (AOFVD):**

- Best disease (BD) is usually inherited as an autosomal dominant (AD) trait, with significant visual impairment usually developing in the vitelliruptive (scrambled egg) stage.
- EOG (Electrooculography) is normal in BD and in BD carriers.
- In BD, the onset of abnormalities on EOG coincides with the development of the vitelliform (fried egg) lesion.
- In BD, significant visual impairment is usually delayed until the vitelliruptive stage.
- Adult-onset foveomacular vitelliform dystrophy (AOFVD) has a later onset but a worse ultimate visual prognosis than BD.
- Unlike BD, AOFVD is genetically unrelated to BD.

---

**Briefly, what is a pattern dystrophy?**

An inherited macular dystrophy that has a characteristic appearance (ie, a particular ‘pattern’).

---

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

You can indeed.

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

Nope. Despite their phenotypic similarities, AOFVD is genetically unrelated to BD.

---

**Concerning adult-onset foveomacular vitelliform dystrophy (AOFVD):**

**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 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

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

The pattern dystrophies

**What gene is implicated in the pattern dystrophies?**

PRPH2 (formerly Peripherin/RDS)
Concerning Best disease and adult-onset foveomacular vitelliform dystrophy (AOFVD):

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, 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.

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, 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.

Can I assume that, like Best dz, 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.

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)
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy (AOFVD):

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

- **What is the inheritance pattern?**
  AD

- **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)

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

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 is AD (like most inherited retinal diseases). EOG is normal in adult vitelliform disease and in Best carriers. In Best disease, onset of EOG abnormalities coincides with the development of the vitelliform (fried egg) lesion. In Best disease, significant visual impairment usually is delayed until the vitelliruptive (scrambled egg) stage. Adult-onset foveomacular vitelliform dystrophy has a later onset but a worse ultimate visual prognosis than Best disease.

Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy:

Which of the following are true?

- 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.

Briefly, what is a pattern dystrophy? 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 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

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

- The pattern dystrophies
- 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

What gene is implicated in the pattern dystrophies?
- PRPH2 (formerly Peripherin/RDS)
Concerning Best disease, and Adult-onset foveomacular vitelliform dystrophy:

Briefly, what is a pattern dystrophy?
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

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.

Briefly, what is a pattern dystrophy?
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

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.

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)
Concerning Best disease, and...

**Briefly, what is a pattern dystrophy?**
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)

**CNVM** = choroidal neovascular membrane
**GA** = Geographic atrophy

---

**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.

---

**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)
Concerning Best disease, and

Briefly, what is a pattern dystrophy?
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?

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.

Briefly, what is a pattern dystrophy?
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?

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.

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?**
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 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

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)
Concerning Best disease and Adult-onset foveomacular vitelliform dystrophy (AOFVD):

**Briefly, what is a pattern dystrophy?**
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 present with different patterns
---
---
---

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)
**Concerning Best disease,** and adult-onset foveomacular vitelliform dystrophy:

**Briefly, what is a pattern dystrophy?**
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 present with…different patterns

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

The pattern dystrophies

---Butterfly dystrophy
---Adult-onset foveomacular vitelliform dystrophy
---Reticular dystrophy
---Fundus pulverulentus

---What gene is implicated in the pattern dystrophies?

PRPH2 (formerly Peripherin/RDS)
**Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy (AOFVD):**

*What is a pattern dystrophy?*
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 present with…

---

### 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.

---

**Briefly, what is a pattern dystrophy?**
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 present with…

---

### 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)
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy:

**Briefly, what is a pattern dystrophy?**
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 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 present with...different patterns

---

**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.

**What is a pattern dystrophy?**
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 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 present with...different patterns

---

**Which of the following are true?**

- 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)
Concerning Best disease, and adult-onset foveomacular vitelliform dystrophy (AOFVD):

**Briefly, what is a pattern dystrophy?**
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 present with different patterns
--The same individual can have different patterns…

**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.

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

**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)
Concerning Best disease and adult-onset foveomacular vitelliform dystrophy (AOFVD):

**Briefly, what is a pattern dystrophy?**
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 present with different patterns
--The same individual can have different patterns in their two eyes

---

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.

---

**The BCSC Retina book identifies four pattern dystrophies by name--what are the other three?**
--B*utterfly dystrophy*
--A*dult-onset foveomacular vitelliform dystrophy*
--R*eticular dystrophy*
--F*undus 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)
Concerning Best disease... and...

**Briefly, what is a pattern dystrophy?**
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 present with...different patterns
--The same individual can have different patterns...in their two eyes
--The pattern in a given eye can...

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.

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

**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)
Concerning Best disease... and...

Briefly, what is a pattern dystrophy?
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 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 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

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.

Briefly, what is a pattern dystrophy?
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 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 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

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.

What gene is implicated in the pattern dystrophies?
PRPH2 (formerly Peripherin/RDS)
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 \( F \)
- End-stage Best disease can look like ARMD
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
- End-stage Best disease can look like ARMD  T
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
- **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
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

<table>
<thead>
<tr>
<th>Stage</th>
<th>Name</th>
<th>Appearance</th>
<th>Vision</th>
</tr>
</thead>
<tbody>
<tr>
<td>I</td>
<td>?</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>II</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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.
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

<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></td>
<td></td>
<td></td>
</tr>
<tr>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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.
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

<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>?</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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.
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**

<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>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
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

<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>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

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

<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>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

At what age does the egg-yolk lesion typically appear? 4-10 years
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

<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>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

What percent of cases present with multifocal lesions?
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**

<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>
<td>III</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

What percent of cases present with multifocal lesions? ~30%
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>
<td>III</td>
<td>?</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td>?</td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td>?</td>
</tr>
</tbody>
</table>
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**

<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>
<td>III</td>
<td>Pseudo-hypopyon</td>
<td>Layered yolk</td>
<td>+/- mild loss</td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**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

<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>
<td>III</td>
<td>Pseudo-hypopyon</td>
<td>Layered yolk</td>
<td>+/- mild loss</td>
</tr>
<tr>
<td>IV</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>V</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
### 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**

<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>
<td>III</td>
<td>Pseudo-hypopyon</td>
<td>Layered yolk</td>
<td>+/- mild loss</td>
</tr>
<tr>
<td>IV</td>
<td>?</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>V</td>
<td>?</td>
<td>?</td>
<td>?</td>
</tr>
</tbody>
</table>
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

<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>
<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></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
**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**

<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>
<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>?</td>
<td>?</td>
<td>?</td>
</tr>
</tbody>
</table>
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**

<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>
<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>
</tbody>
</table>
### 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>
<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?
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>
<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>

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

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 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.

*(Summary slide—no questions)*