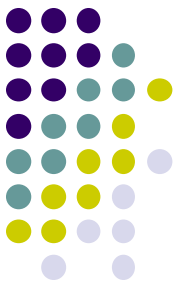


Q

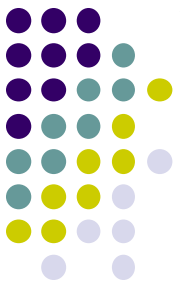
- *Albinism and the eye*

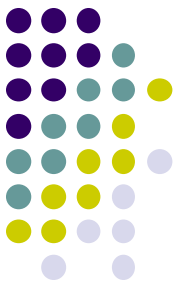
- Defect in and/or



A

- *Albinism and the eye*
 - Defect in **melanin production** and/or **distribution**





Q

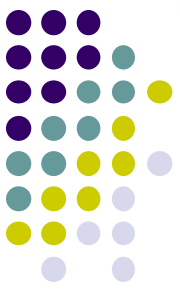
- *Albinism and the eye*

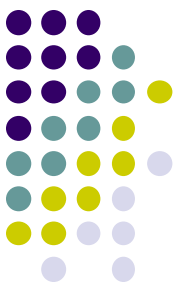
- Defect in **melanin production** and/or **distribution**
- Can be **one class** or **the other class**

A

- *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**





A

- *Albinism and the eye*
 - Defect in **melanin production** and/or **distribution**
 - Can be **ocular** or **oculocutaneous**

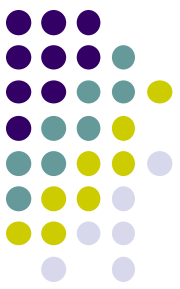
Because the manifestations of albinism may be limited to the eyes, it is not uncommon for the ophthalmologist to be the one to diagnose it. Thus, it is important that you have an index of suspicion for this not-as-rare-as-you-might-think condition!



Q

- *Albinism and the eye*

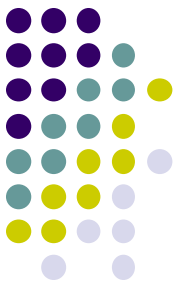
- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **one type** X-linked
 - **the other type** Autosomal recessive



A

- *Albinism and the eye*

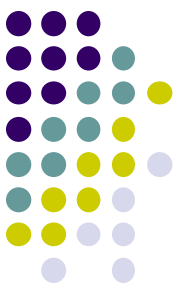
- Defect in **melanin production** and/or **distribution**
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Q

- *Albinism and the eye*
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What is the other, eponymous name for ocular albinism?



A

- *Albinism and the eye*
 - Defect in **melanin production** and/or **distribution**
 - Can be **ocular** or **oculocutaneous**
 - Inheritance:
 - **Ocular:** **X-linked**
 - **Oculocutaneous:** Autosomal recessive

What is the other, eponymous name for ocular albinism?

Nettleship-Falls albinism



Q

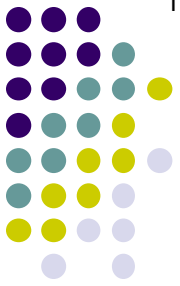
- *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** **X-linked**
 - **Oculocutaneous:** Autosomal recessive

What is the other, eponymous name for ocular albinism?

Nettleship-Falls albinism

Given that ocular albinism is X-linked, what does this imply about its clinical presentation?



A

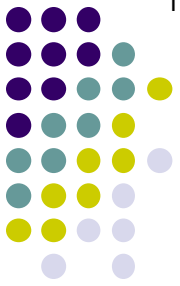
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 - Defect in **melanin production** and/or **distribution**
 - Can be **ocular** or **oculocutaneous**
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What is the other, eponymous name for ocular albinism?

Nettleship-Falls albinism

Given that ocular albinism is X-linked, what does this imply about its clinical presentation?

That it occurs only in males, and that females are carriers



Q

- *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** **X-linked**
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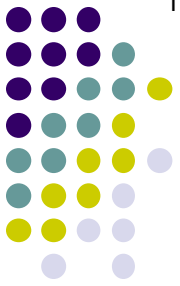
What is the other, eponymous name for ocular albinism?

Nettleship-Falls albinism

Given that ocular albinism is X-linked, what does this imply about its clinical presentation?

That it occurs only in males, and that **females are carriers**

Do female carriers evidence any signs of the disease on exam?



A

- *Albinism and the eye*
 - Defect in **melanin production** and/or **distribution**
 - Can be **ocular** or **oculocutaneous**
 - Inheritance:
 - **Ocular:** **X-linked**
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What is the other, eponymous name for ocular albinism?

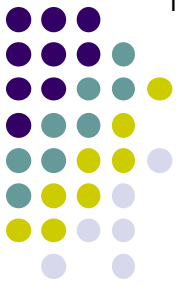
Nettleship-Falls albinism

Given that ocular albinism is X-linked, what does this imply about its clinical presentation?

That it occurs only in males, and that **females are carriers**

Do female carriers evidence any signs of the disease on exam?

Yes, subtle subclinical findings c/w albinism are common

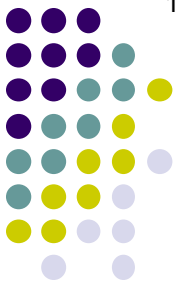


Q

- *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** X-linked
 - **Oculocutaneous: Autosomal recessive**

Given that oculocutaneous albinism is AR, is it commonly associated with consanguinity?

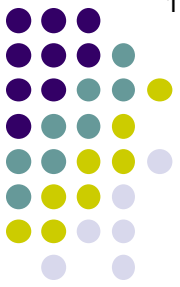


A

- *Albinism and the eye*

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- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** X-linked
 - **Oculocutaneous:** **Autosomal recessive**

Given that oculocutaneous albinism is AR, is it commonly associated with consanguinity?
No, because the responsible genes are relatively common

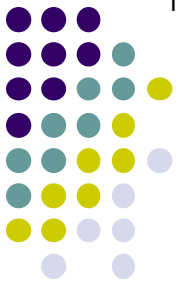


Q

- *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular**: X-linked
 - **Oculocutaneous**: Autosomal recessive

Which group is affected more frequently, blacks or whites?



A

- *Albinism and the eye*

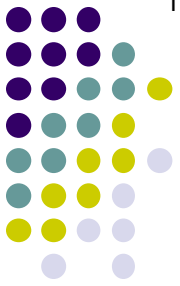
- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** X-linked
 - **Oculocutaneous:** Autosomal recessive

Which group is affected more frequently, blacks or whites?

Blacks, although they are more likely to manifest incomplete penetrance



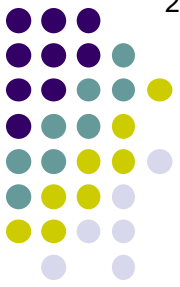
Oculocutaneous albinism



Q

- *Albinism and the eye*

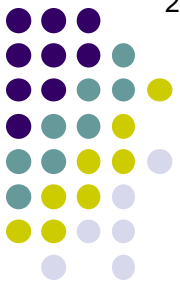
- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** X-linked
 - **Oculocutaneous:** Autosomal recessive
- Complain of:
 -
 -
 -



A

● *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** X-linked
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- Complain of:
 - **Nystagmus**
 - **Photophobia**
 - **Poor central vision**



Q

● *Albinism and the eye*

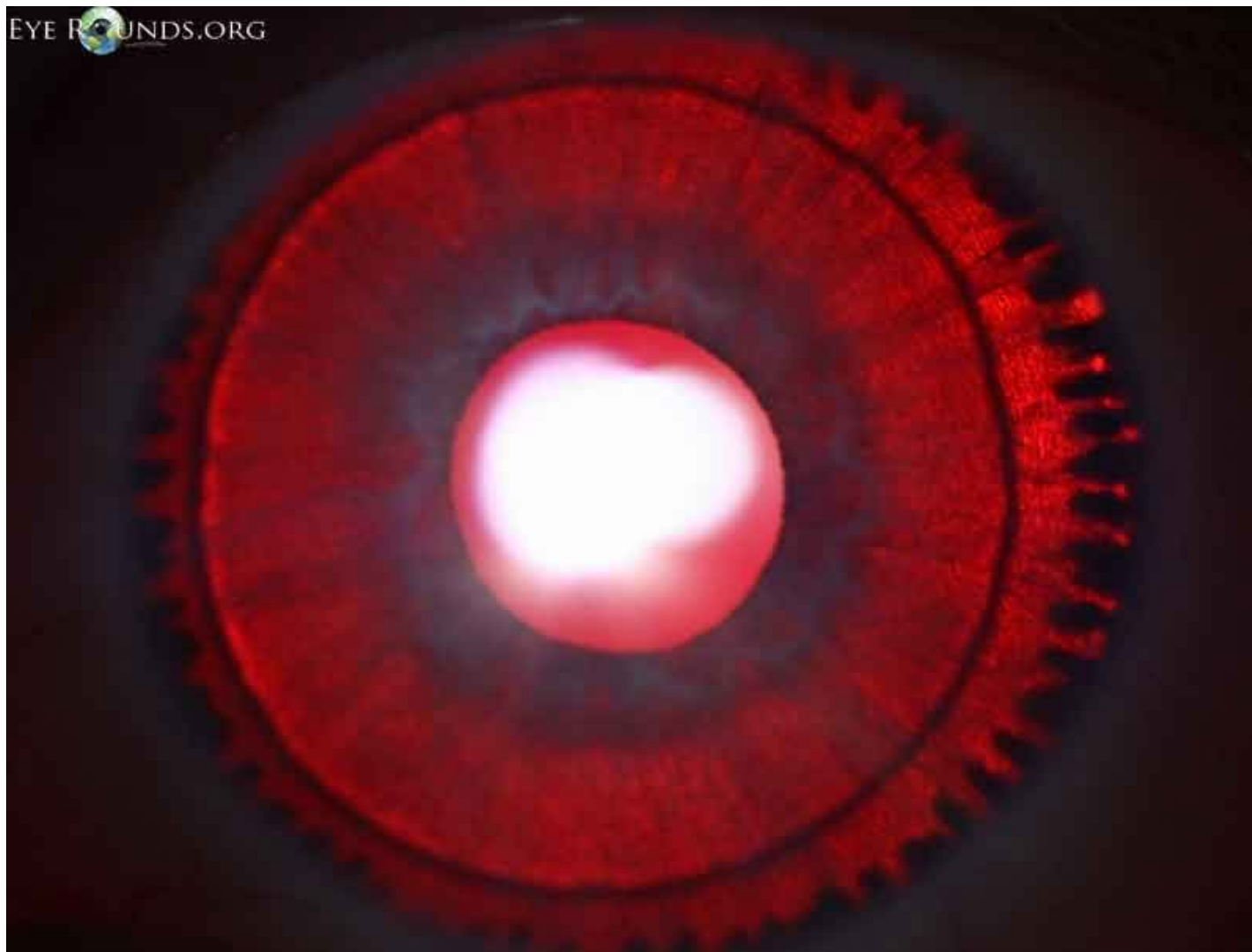
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- Complain of:
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 - **Photophobia**
 - **Poor central vision**
- Exam findings:
 - iris finding
 - retina finding
 - refractive finding



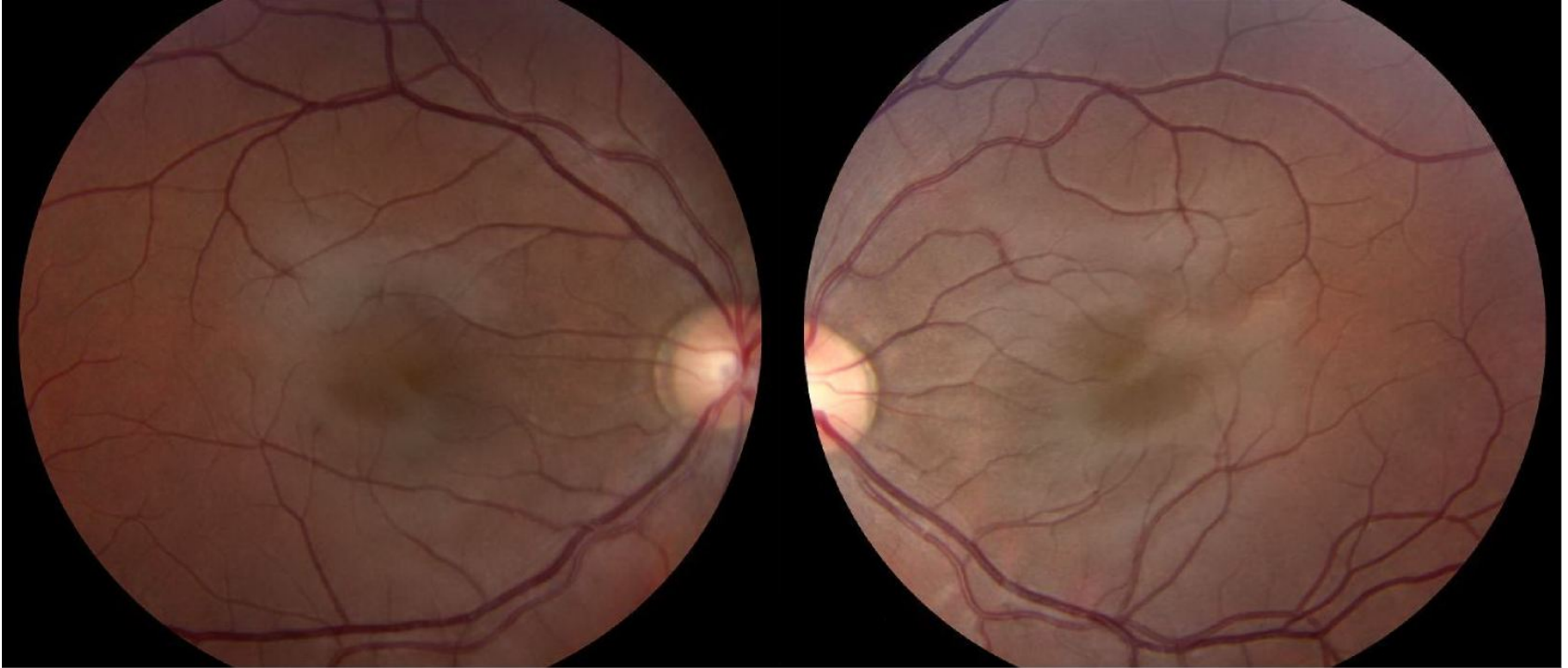
A

● *Albinism and the eye*

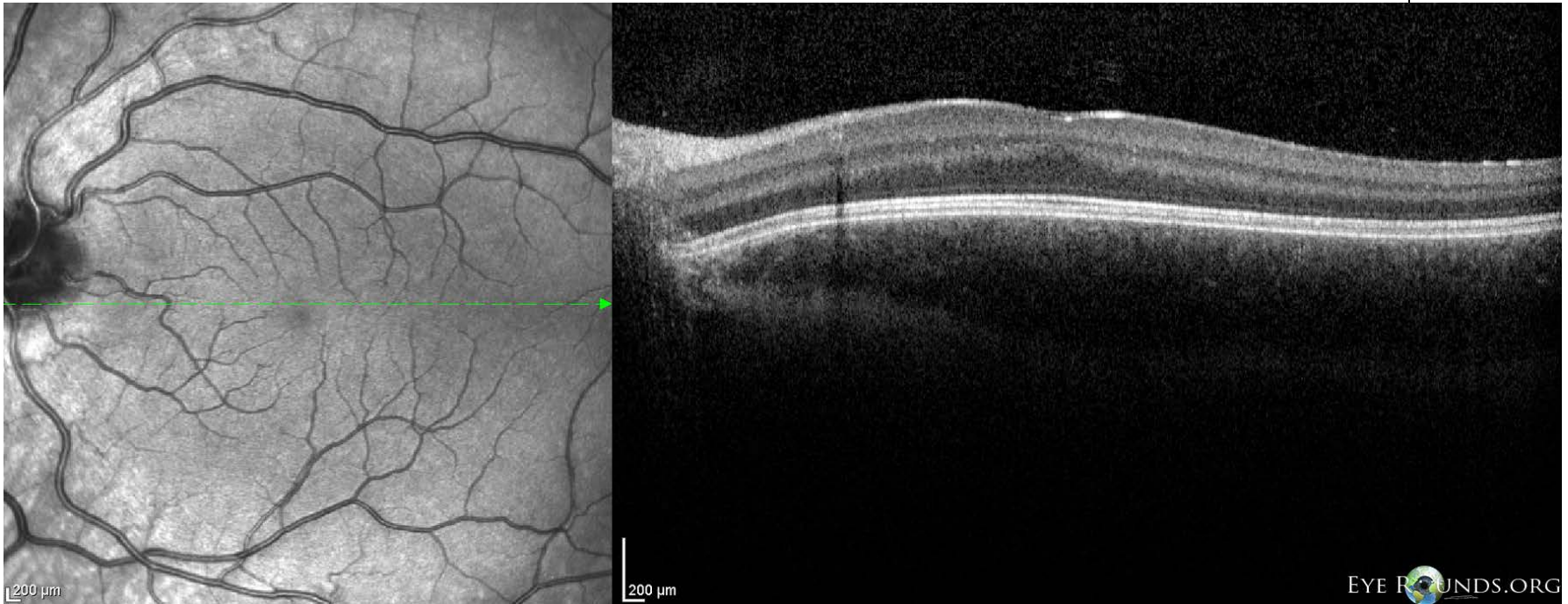
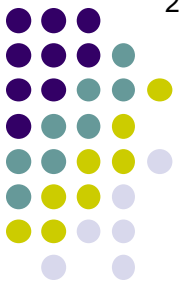
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- Exam findings:
 - **Iris transillumination**
 - **Foveal hypoplasia**
 - **High refractive error**



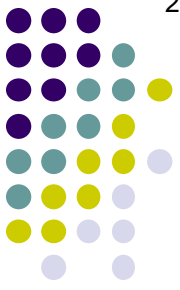
Albinism: Iris transillumination



Albinism: Foveal hypoplasia



Albinism: Foveal hypoplasia

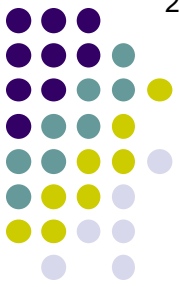


Q

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What (mis)diagnosis are albinotic patients with incomplete penetrance likely to receive?

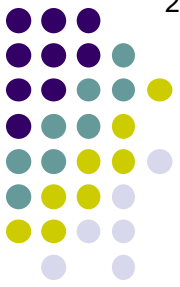


A

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What (mis)diagnosis are albinotic patients with incomplete penetrance likely to receive?
Congenital motor nystagmus



Q

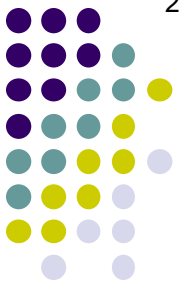
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What (mis)diagnosis are albinotic patients with incomplete penetrance likely to receive?

Congenital motor nystagmus

How can you avoid such an embarrassing error?



A

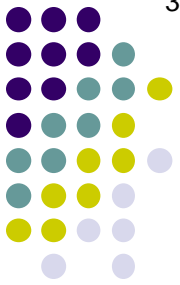
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What (mis)diagnosis are albinotic patients with incomplete penetrance likely to receive?

Congenital motor nystagmus

How can you avoid such an embarrassing error?
By carefully screening all children with nystagmus for **iris transillumination** and **foveal hypoplasia**

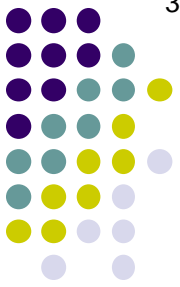


Q

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 - **Photophobia**
 - **Poor central vision**
- **Exam findings:**
 - **Iris transillumination**
 - **Foveal hypoplasia**
 - **High refractive error**
 - **Poor stereopsis**

They also have poor stereopsis--why?

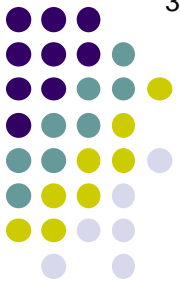


A

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 - **High refractive error**
 - **Poor stereopsis**

*They also have poor stereopsis--why?
An abnormal percentage of retinal fibers
decussate, severely limiting stereopsis*



Q

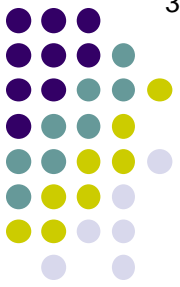
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What does decussate mean?

*They also have poor stereopsis--why?
An abnormal percentage of retinal fibers **decussate**, severely limiting stereopsis*



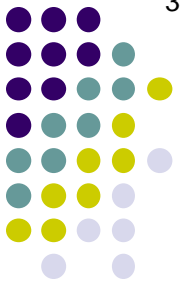
A

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What does decussate mean?
To cross

They also have poor stereopsis--why?
An abnormal percentage of retinal fibers **decussate**, severely limiting stereopsis



Q

● *Albinism and the eye*

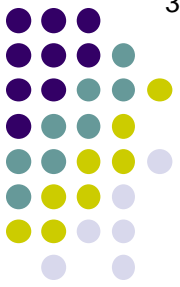
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*What does decussate mean?
To cross*

What crossing are we talking about here?

*They also have poor stereopsis--why?
An abnormal percentage of retinal fibers
decussate, severely limiting stereopsis*



A

● *Albinism and the eye*

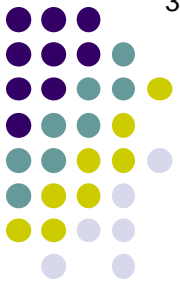
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 - **Poor central vision**
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 - **Foveal hypoplasia**
 - **High refractive error**
 - **Poor stereopsis**



What does decussate mean?
To cross

What crossing are we talking about here?
The contralateral path taken by nasal retinal fibers at the optic chiasm

They also have poor stereopsis--why?
An abnormal percentage of retinal fibers **decussate**, severely limiting stereopsis



Q

● *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:

- **Ocular:** X-linked

- **Oculocutaneous:** Autosomal

● Complain of:

- **Nystagmus**
- **Photophobia**
- **Poor central vision**

● **Exam findings:**

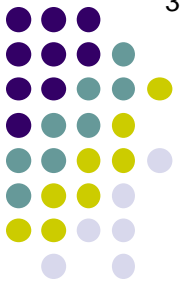
- **Iris transillumination**
- **Foveal hypoplasia**
- **High refractive error**
- *Poor stereopsis*

What percentage of the fibers should decussate?

What does decussate mean?
To cross

What crossing are we talking about here?
The contralateral path taken by nasal retinal fibers at the optic chiasm

They also have poor stereopsis--why?
An **abnormal percentage of retinal fibers decussate**, severely limiting stereopsis



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- Can be **ocular** or **oculocutaneous**
- Inheritance:

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● Complain of:

- **Nystagmus**
- **Photophobia**
- **Poor central vision**

● **Exam findings:**

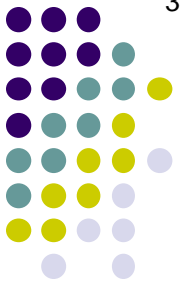
- **Iris transillumination**
- **Foveal hypoplasia**
- **High refractive error**
- *Poor stereopsis*

*What percentage of the fibers should decussate?
50, maybe a little higher*

*What does decussate mean?
To cross*

*What crossing are we talking about here?
The contralateral path taken by nasal retinal fibers at the optic chiasm*

*They also have poor stereopsis--why?
An **abnormal percentage of retinal fibers decussate**, severely limiting stereopsis*



Q

● *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:

- **Ocular:** X-linked

- **Oculocutaneous:** Autosomal

● Complain of:

- **Nystagmus**
- **Photophobia**
- **Poor central vision**

● **Exam findings:**

- **Iris transillumination**
- **Foveal hypoplasia**
- **High refractive error**
- *Poor stereopsis*

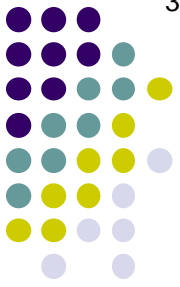
*What percentage of the fibers should decussate?
50, maybe a little higher*

Is the percentage in albinism higher or lower?

*What does decussate mean?
To cross*

*What crossing are we talking about here?
The contralateral path taken by nasal retinal fibers at the optic chiasm*

*They also have poor stereopsis--why?
An **abnormal percentage of retinal fibers decussate**, severely limiting stereopsis*



A

● *Albinism and the eye*

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:

- **Ocular:** X-linked

- **Oculocutaneous:** Autosomal

● Complain of:

- **Nystagmus**
- **Photophobia**
- **Poor central vision**

● **Exam findings:**

- **Iris transillumination**
- **Foveal hypoplasia**
- **High refractive error**
- *Poor stereopsis*

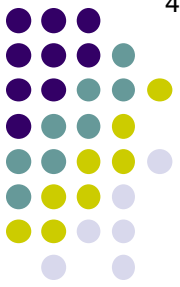
*What percentage of the fibers should decussate?
50, maybe a little higher*

*Is the percentage in albinism higher or lower?
Higher (much)*

*What does decussate mean?
To cross*

*What crossing are we talking about here?
The contralateral path taken by nasal retinal fibers at the optic chiasm*

*They also have poor stereopsis--why?
An **abnormal percentage of retinal fibers decussate**, severely limiting stereopsis*



Q

Albinoidism

● ~~Albinism~~ and the eye

- Defect in **melanin production** and/or **distribution**
- Can be **ocular** or **oculocutaneous**
- Inheritance:
 - **Ocular:** X-linked
 - **Oculocutaneous:** Autosomal recessive
- Complain of:
 - **Nystagmus**
 - **Photophobia**
 - **Poor central vision**
- Exam findings:
 - **Iris transillumination**
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A

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A variant of albinism (either ocular or oculocutaneous)



Q

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How does it differ from albinism?



A/Q

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It differs in that three eye manifestations are missing:

--
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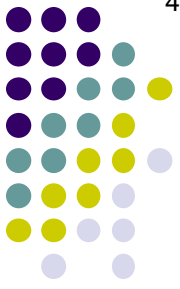
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Which of these constitutes the fundamental difference between albinism and albinoidism?

The status of the fovea. In albinism, the fovea is hypoplastic, which in turn results in poor acuity, which in turn results in nystagmus. In albinoidism, it is normal.



Q

- *Two potentially lethal syndromes are associated with albinism—what are they?*
 - *syndrome*
 - *syndrome*



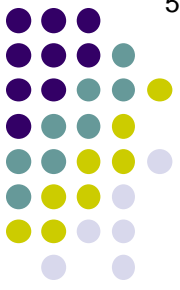
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- *Two potentially lethal syndromes are associated with albinism—what are they?*
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 - *Hermansky-Pudlak syndrome*



Q

- *Two potentially lethal syndromes are associated with albinism—what are they?*
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A

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age range



A

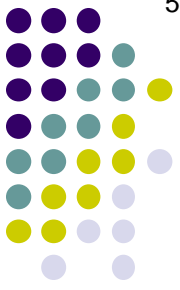
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What proportion of Chediak-Higashi children will succumb to a pyogenic infection by age 10?



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What proportion of Chediak-Higashi children will succumb to a pyogenic infection by age 10?
About half



Q

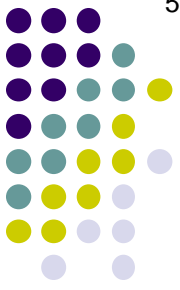
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What is the classic hair color of patients with Chediak-Higashi syndrome?



A

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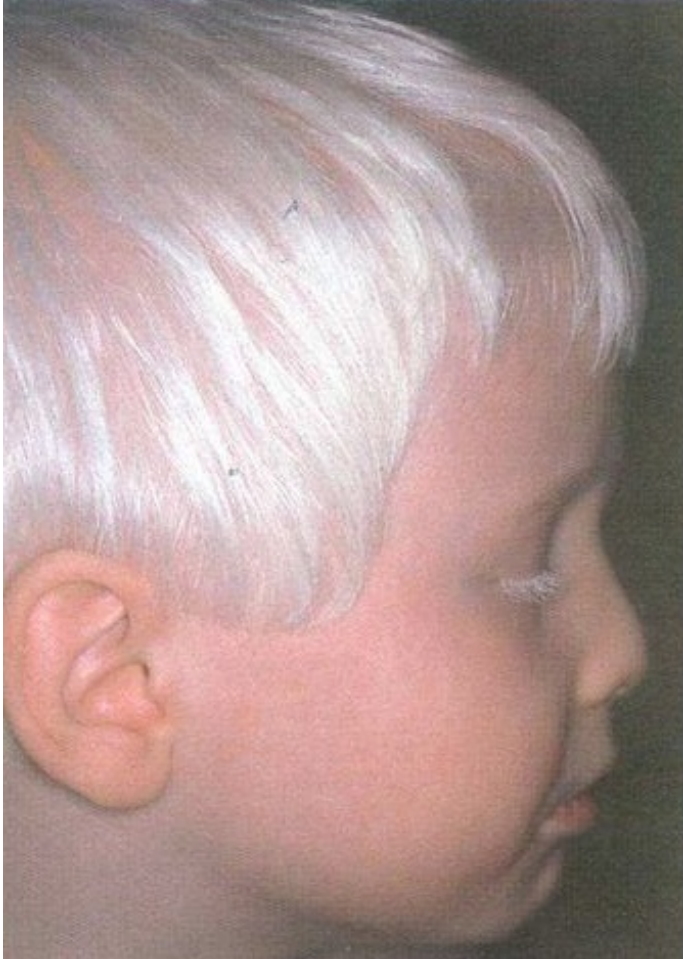
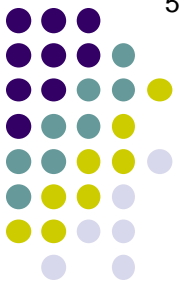
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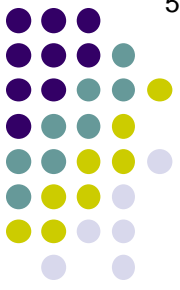
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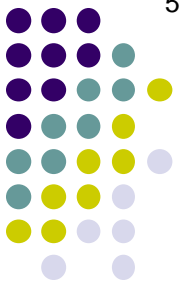


Chediak-Higashi syndrome: **Silver-gray** hair



Q

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 - Oculocutaneous albinism + systemic problem + organ problem (three words)



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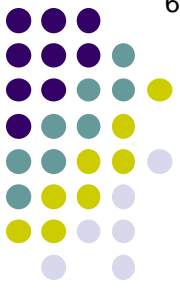
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Pudlak
 Puerto Rican
 platelet dysfunction
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If a child of Puerto Rican ancestry has oculocutaneous albinism, how likely is that child to have Hermansky-Pudlak syndrome?

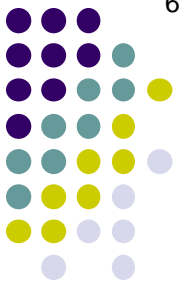


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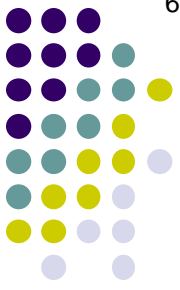
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What is the classic presenting complaint in Hermansky-Pudlak syndrome (other than the typical stigmata of oculocutaneous albinism)?



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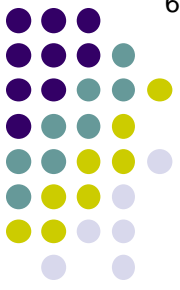
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The child will have a history of recurrent **epistaxis**



Q

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 - **Puerto Rican** ethnicity
 - If either is suspected, get **another service** consult



A

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Note that this would make a great Boards ‘trap case’ in which you make the right diagnosis (‘This child has oculocutaneous albinism’) but fail to consider whether s/he has one of these potentially lethal syndromes. Thus, be sure to mention these conditions if you encounter an albinism case on the Boards!