Albinism and the eye

- Defect in basic pathology
- and/or ditto
Albinism and the eye

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

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

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

- Defect in melanin production and/or distribution
- Can be ocular or oculocutaneous
- Inheritance:
  - Ocular: X-linked
  - Oculocutaneous: Autosomal recessive
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?
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
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?**
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? That it occurs only in males, and that females are carriers
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?
Netleship-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?
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?**
Netleship-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
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?
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?
No, because the responsible genes are relatively common
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?
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
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
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
**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 finding
- Retina finding
- Refractive finding
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
- Foveal hypoplasia
- High refractive error
Albinism: Iris transillumination
Albinism: Foveal hypoplasia
Albinism: Foveal hypoplasia
Albinism and the eye

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- Complain of:
  - Nystagmus
  - Photophobia
  - Poor central vision
- Exam findings:
  - Iris transillumination
  - Foveal hypoplasia
  - High refractive error

What (mis)diagnosis are albinotic patients with incomplete penetrance likely to receive?
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
  - Foveal hypoplasia
  - High refractive error

What (mis)diagnosis are albinotic patients with incomplete penetrance likely to receive? Congenital motor nystagmus
Albinism and the eye
- Defect in \textit{melanin production} and/or \textit{distribution}
- Can be \textit{ocular} or \textit{oculocutaneous}
- Inheritance:
  - Ocular: X-linked
  - Oculocutaneous: Autosomal recessive
- Complain of:
  - \textbf{Nystagmus}
  - Photophobia
  - Poor central vision
- Exam findings:
  - Iris transillumination
  - Foveal hypoplasia
  - High refractive error

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

How can you avoid such an embarrassing error?
By carefully screening all children with nystagmus for iris transillumination and foveal hypoplasia.
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
- Exam findings:
  - Iris transillumination
  - Foveal hypoplasia
  - High refractive error

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  - Ocular: X-linked
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- Complain of:
  - Nystagmus
  - Photophobia
  - Poor central vision

Exam findings:
- Iris transillumination
- Foveal hypoplasia
- High refractive error
- Poor stereopsis

They also have poor stereopsis--why?
Albinism and the eye

- Defect in melanin production and/or distribution
- Can be ocular or oculocutaneous
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  - Ocular: X-linked
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  - Nystagmus
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- Exam findings:
  - Iris transillumination
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They also have poor stereopsis--why? An abnormal percentage of retinal fibers decussate, severely limiting stereopsis.
Albinism and the eye

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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
Albinism and the eye

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What does decussate mean?
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What crossing are we talking about here?

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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
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    - Poor central vision
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    - Iris transillumination
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What does decussate mean? To cross

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They also have poor stereopsis--why? An abnormal percentage of retinal fibers decussate, severely limiting stereopsis
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  - Iris transillumination
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  - 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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- 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
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Exam findings:
- Iris transillumination
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What percentage of the fibers should decussate?
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Is the percentage in albinism higher or lower?
Higher (much)

What does decussate mean?
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What crossing are we talking about here?
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They also have poor stereopsis--why?
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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:

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

Albinoidism
- A variant of albinism (either ocular or oculocutaneous)
- How does it differ from albinism?
  - It differs in that three eye manifestations are missing:
    - Nystagmus
    - Poor visual acuity
    - Foveal hypoplasia
- What is 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.

What is albinoidism?
Albinism and the eye

- Defect in melanin production and/or distribution
- Can be ocular or oculocutaneous
- Inheritance:
  - Ocular: X-linked
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A variant of albinism (either ocular or oculocutaneous)

How does it differ from albinism?
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  - --
Albinism and the eye

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  - **Iris transillumination**
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---

**What is albinoidism?**
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- Complain of:
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- Exam findings:
  - Iris transillumination
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What is albinoidism?
A variant of albinism (either ocular or oculocutaneous)

How does it differ from albinism?
It differs in that three eye manifestations are missing:
--Nystagmus
--Poor visual acuity
--Foveal hypoplasia

Which of these constitutes the fundamental difference between albinism and 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**
- **Foveal hypoplasia**
- **High refractive error**

**What is albinism?**
A variant of albinism (either ocular or oculocutaneous)

**How does it differ from albinism?**
It differs in that three eye manifestations are missing:
--Nystagmus
--Poor visual acuity
--Foveal hypoplasia

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

- Chediak-Higashi syndrome
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

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

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

- Chediak-Higashi syndrome
  - Oculocutaneous albinism + systemic problem

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

- **Chediak-Higashi syndrome**
  Oculocutaneous albinism + immune dysfunction

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

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

- **Chediak-Higashi syndrome**
  Oculocutaneous albinism + **immune dysfunction**
  Very susceptible to infection → death **in childhood**

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

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

What proportion of Chediak-Higashi children will succumb to a pyogenic infection by age 10?
Two potentially lethal syndromes are associated with albinism—what are they?

- Chediak-Higashi syndrome
  Oculocutaneous albinism + immune dysfunction
  Very susceptible to infection → death in childhood

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

- Two potentially lethal syndromes are associated with albinism—what are they?
  - **Chediak-Higashi syndrome**
    - Oculocutaneous albinism + immune dysfunction
    - Very susceptible to infection \( \rightarrow \) death in childhood

*What is the classic hair color of patients with Chediak-Higashi syndrome?*
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

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

Silver-gray
Chediak-Higashi syndrome: Silver-gray hair
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak syndrome**
  - Oculocutaneous albinism + pulmonary interstitial fibrosis
  - Organ problem (three words)
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi** syndrome
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

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

- **Chediak-Higashi** syndrome
  - Oculocutaneous albinism + **immune dysfunction**
  - Very susceptible to infection → death **in childhood**

- **Hermansky-Pudlak** syndrome
  - Oculocutaneous albinism + **platelet dysfunction** + **pulmonary interstitial fibrosis**
  - Two words **ethnicity**
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi** syndrome
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak** syndrome
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity
Two potentially lethal syndromes are associated with albinism—what are they?

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  - Puerto Rican ethnicity
Two potentially lethal syndromes are associated with albinism—what are they?

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  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak syndrome**
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

If a child of Puerto Rican ancestry has oculocutaneous albinism, how likely is that child to have Hermansky-Pudlak syndrome?
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi** syndrome
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak** syndrome
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

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

**Very** likely
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi** syndrome
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak** syndrome
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

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

What is the classic presenting complaint in Hermansky-Pudlak syndrome (other than the typical stigmata of oculocutaneous albinism)?
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak syndrome**
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

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

**Very likely**

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

The child will have a history of recurrent **epistaxis**
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak syndrome**
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

If either is suspected, get another service consult
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak syndrome**
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

If either is suspected, get **Heme-Onc consult**
Two potentially lethal syndromes are associated with albinism—what are they?

- **Chediak-Higashi syndrome**
  - Oculocutaneous albinism + immune dysfunction
  - Very susceptible to infection → death in childhood

- **Hermansky-Pudlak syndrome**
  - Oculocutaneous albinism + platelet dysfunction + pulmonary interstitial fibrosis
  - Puerto Rican ethnicity

If either is suspected, get Heme-Onc consult

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!