Intraocular Tumors of Childhood

Four intraocular locations (ie, structures/tissues)
Intraocular Tumors of Childhood

Four intraocular locations (ie, structures/tissues):

- Iris/Ciliary Body
- Choroid
- RPE
- Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) ?

2) ?

3) ?

4) ?

5) ?

6) ?

Six tumors of the iris/ciliary body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

2) Medulloepithelioma

3) Lisch nodules
4) Brushfield spots
5) Iris mammillations
6) Iris cysts

Six tumors of the iris/ciliary body
Iris/Ciliary Body

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG?
It is a nonneoplastic histiocytic proliferation.

How does JXG usually present? (Hint: It's not ophthalmic)
As orangish skin papules

At what age does it present?
The majority before age 1 year, and almost all by age 2

When JXG iris nodules are present, are they uni-, or bilateral?
Unilateral

In what three ways are the iris nodules clinically significant?
--- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--- They are in the DDx as a 'masquerade syndrome' in peds uveitis
--- If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?
It is self-limited, usually resolving by age 5 years
1) **Juvenile xanthogranuloma**

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It is a…**nonneoplastic histiocytic proliferation**

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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

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1) **Juvenile xanthogranuloma**: Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.

2) **Medulloepithelioma**: Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate.

3) **Lisch nodules**: Strong association with NF1. Lighter on dark irides; darker on light.

4) **Brushfield spots**: Strong association with Down syndrome; 15% of non-Down pop.

5) **Iris mammillations**: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) **Iris cysts**: Can be pupillary, stromal, secondary.

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1) **Juvenile xanthogranuloma**

*In three words, what sort of condition is JXG?*

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JXG: Skin papules. The orangish color is classic
Iris/Ciliary Body

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## Intraocular Tumors of Childhood

### Iris/Ciliary Body

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### Choroid

### RPE

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Intraocular Tumors of Childhood

JXG: Iris lesion
## Iris/Ciliary Body

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**Iris/Ciliary Body**

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### Intraocular Tumors of Childhood

#### Iris/Ciliary Body

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JXG: Spontaneous hyphema
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*Should JXG nodules be removed surgically?*
Only if the glaucoma is uncontrollable

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The presence of **Touton giant cells**
The presence of ‘foamy macrophages’

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Touton giant cells

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JXG
Intraocular Tumors of Childhood

Iris/Ciliary Body

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*What are the two hallmarks of JXG histology?*

The presence of *Touton* giant cells

*’foamy macrophages’*

*What is the natural history of JXG?*

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*This histology—’foamy macrophages’—is often described with other, equivalent terms. What are they?*

Foamy = ‘lipid filled’

Macrophages = ‘histiocytes’

---

*Intraocular Tumors of Childhood*
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*What are the two hallmarks of JXG histology?*  
The presence of  Touton  giant cells  
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*When are the JXG iris nodules present—unilateral or bilateral?*  
Unilateral

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*The point being, the terms ‘foamy macrophages,’ ‘lipid-filled (or -laden) macrophages,’ ‘foamy histiocytes,’ etc, all mean the same thing, so don’t be misled if you see one term when you’re expecting another*
1) Juvenile xanthogranuloma

**In three words, what sort of condition is JXG?**
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**How does JXG usually present? (Hint: It’s not ophthalmic)**
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Speaking of 'foamy macrophages'...

**What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?**

--- First clue—more forthcoming
Iris/Ciliary Body

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The presence of ‘foamy macrophages’

*When unilateral?*  
Unilateral

*Speaking of ‘foamy macrophages’…*  
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?  
And a hx of chronic migratory arthritis?  
Clue #2
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Speaking of ‘foamy macrophages’…

*What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?*

*And a hx of chronic migratory arthritis?***

*Associated with chronic diarrhea? Need another?*

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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And a hx of chronic migratory arthritis?
Associated with chronic diarrhea?
And CNS symptoms--seizures, dementia, coma?

Retina

Choroid

Whipple’s disease

Last chance--answer is next!
Intraocular Tumors of Childhood

Iris/Ciliary Body

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*When are the iris nodules usually unilateral?*
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What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis? And a hx of chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms--seizures, dementia, coma? Whipple’s disease

Broadly speaking, what sort of condition is Whipple’s?
Intraocular Tumors of Childhood

Iris/Ciliary Body

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**Whipple’s disease**
It is infectious

Broadly speaking, what sort of condition is Whipple’s?
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Choroid

Retina
Intraocular Tumors of Childhood

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*Whipple’s disease*

*Broadly speaking, what sort of condition is Whipple’s?*
*It is infectious*

*What infection agent is responsible for Whipple’s?*

---

**Iris/Ciliary Body**

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Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed -> hyphema -> increased IOP -> glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.


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4) Brushfield spots: Strong association with Down syndrome; 15% of non-Down pop.

5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) Iris cysts: Can be pupillary, stromal, secondary.

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation.

How does JXG usually present? (Hint: It’s not ophthalmic) As orangish skin papules.

At what age does it present?
The majority before age 1 year, and almost all by age 2.

When JXG iris nodules are present, are they uni-, or bilateral? Unilateral.

In what three ways are the iris nodules clinically significant?
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG? It is self-limited, usually resolving by age 5 years.

What are the two hallmarks of JXG histology? The presence of Touton giant cells.
The presence of ‘foamy macrophages’.

Speaking of ‘foamy macrophages’...
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis? And a hx of chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms--seizures, dementia, coma?

Broadly speaking, what sort of condition is Whipple’s? It is infectious.

What infection agent is responsible for Whipple’s? The bacterium *Tropheryma whipplei*.
1) Juvenile xanthogranuloma

**In three words, what sort of condition is JXG?**
It is a...nonneoplastic histiocytic proliferation

**How does JXG usually present?** (Hint: It’s not ophthalmic)
As orangish skin papules

**At what age does it present?**
The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?
Unilateral

In what three ways are the iris nodules clinically significant?
-- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
-- They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
-- If enough nodules are present, heterochromia iridis will result

**What is the natural history of JXG?**
It is self-limited, usually resolving by age 5 years

**What are the two hallmarks of JXG histology?**
The presence of Touton giant cells
The presence of ‘foamy macrophages’

Speaking of ‘foamy macrophages’...

What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with panuveitis, a hx of chronic migratory arthritis, associated with chronic diarrhea, and CNS symptoms--seizures, dementia, coma?

**Whipple’s disease**

**Broadly speaking, what sort of condition is Whipple’s?**
It is infectious

**What infection agent is responsible for Whipple’s?**
The bacterium *Tropheryma whipplei*
Iris/Ciliary Body

Choroid

Retina

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?*  
It is a… **nonneoplastic histiocytic proliferation**

*How does JXG usually present? (Hint: It’s not ophthalmic)*  
As orangish skin papules

*At what age does it present?*  
The majority before age 1 year, and almost all by age 2

*When JXG iris nodules are present, are they uni-, or bilateral?*  
Unilateral

*In what three ways are the iris nodules clinically significant?*  
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma  
--They are in the DDx as a ‘masquerade syndrome’ in peds uveitis  
--If enough nodules are present, heterochromia iridis will result

*What is the natural history of JXG?*  
It is self-limited, usually resolving by age 5 years

*What are the two hallmarks of JXG histology?*  
The presence of Touton giant cells  
The presence of *‘foamy macrophages’*

Speaking of ‘foamy macrophages’…

*What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?*  
And a hx of chronic migratory arthritis?  
And associated with chronic diarrhea?  
And CNS symptoms—seizures, dementia, coma?  
**Whipple’s disease**

Broadly speaking, what sort of condition is Whipple’s?  
It is infectious

*What infection agent is responsible for Whipple’s?*  
The bacterium *Tropheryma whipplei*
Whipple’s disease: Duodenal biopsy, high mag. The image shows the characteristic feature of foamy macrophages in the lamina propria.
**Intraocular Tumors of Childhood**

1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. 
   - Typically presents before age 2 years.
   - Characterized by skin papules that can bleed.
   - Iris nodules can lead to hyphema, increased IOP, and secondary glaucoma.
   - Self-limited; regresses by age 5.
   - Treatment focuses on inflammation and IOP management.
   - Pathological hallmark: Touton giant cells.

2) **Medulloepithelioma:** Benign but locally aggressive neoplasia of nonpigmented epithelium of Ciliary Body.
   - Presents as an iris mass before age 10 years.
   - Can cause bleeding, hyphema, and increased IOP leading to glaucoma.
   - Locally invasive and can be fatal.
   - Treatment: Enucleation.

3) **Lisch nodules:** Strong association with Neurofibromatosis Type 1 (NF1).
   - Lighter on dark irides; darker on light.

4) **Brushfield spots:** Strong association with Down syndrome; 15% of non-Down population.

5) **Iris mammillations:** Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) **Iris cysts:** Can be pupillary, stromal, or secondary.

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**Questions:***

**In three words, what sort of condition is JXG?**

**nonneoplastic histiocytic proliferation**

**How does JXG usually present? (Hint: It’s not ophthalmic)**

As orangish skin papules

**At what age does it present?**

The majority before age 1 year, and almost all by 2 years.

When JXG iris nodules are present, are they uni-, or bilateral?

Unilateral

In what three ways are the iris nodules clinically significant?

-- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma.
-- They are in the differential diagnosis (DDx) for uveitis in children.
-- If enough nodules are present, heterochromia iridis will result.

**What is the natural history of JXG?**

It is self-limited, usually resolving by age 5 years.

**What are the two hallmarks of JXG histology?**

The presence of Touton giant cells.

The presence of 'foamy macrophages'.

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**Speaking of ‘foamy macrophages’…**

What disease comes to mind if, instead of a young child with iris nodules, the patient was a middle-aged white guy with chronic migratory arthritis, associated with chronic diarrhea and CNS symptoms—seizures, dementia, coma?

**Whipple’s disease**

**Broadly speaking, what sort of condition is Whipple’s?**

It is infectious.

**What infection agent is responsible for Whipple’s?**

The bacterium *Tropheryma whipplei*.

**When foamy macrophages are found in a biopsy performed on a Whipple’s pt, from what site was the biopsy collected?**

The duodenum (remember, they have GI issues).

**What other finding will a duodenal biopsy reveal?**

The presence of acid-fast bacteria within macrophages located in intestinal villi.
Iris/Ciliary Body

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a…nonneoplastic histiocytic proliferation

How does JXG usually present? (Hint: It’s not ophthalmic) As orangish skin papules

At what age does it present?
The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral? Unilateral

In what three ways are the iris nodules clinically significant?
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG? It is self-limited, usually resolving by age 5 years

What are the two hallmarks of JXG histology? The presence of Touton giant cells

When foamy macrophages are found in a biopsy performed on a Whipple’s pt, from what site was the biopsy collected? The duodenum (remember, they have GI issues)

What other finding will a duodenal biopsy reveal? The presence of bacteria within macrophages located in intestinal villi

Broadly speaking, what sort of condition is Whipple’s? It is infectious

What infection agent is responsible for Whipple’s? The bacterium Tropheryma whippelii
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- In three words, what sort of condition is JXG? It is a…nonneoplastic histiocytic proliferation
- How does JXG usually present? (Hint: It’s not ophthalmic) As orangish skin papules
- At what age does it present?
  - The majority before age 1 year, and almost all by 2
- When JXG iris nodules are present, are they uni-, or bilateral? Unilateral
- In what three ways are the iris nodules clinically significant?
  - They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
  - They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
  - If enough nodules are present, heterochromia iridis will result
- What is the natural history of JXG?
  - It is self-limited, usually resolving by age 5 years
- What are the two hallmarks of JXG histology?
  - The presence of Touton giant cells
  - The presence of ‘foamy macrophages’

Speaking of ‘foamy macrophages’…

- When foamy macrophages are found in a biopsy performed on a Whipple’s pt, from what site was the biopsy collected? The duodenum (remember, they have GI issues)
- What other finding will a duodenal biopsy reveal? The presence of PAS+ bacteria within macrophages located in intestinal villi
- Broadly speaking, what sort of condition is Whipple’s? It is infectious
- What infection agent is responsible for Whipple’s? The bacterium Tropheryma whippelii
Small-intestine biopsy stained with periodic acid-Schiff. Note the numerous macrophages in the lamina propria (arrows).
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?*
It is a... **nonneoplastic histiocytic proliferation**

*How does JXG usually present? (Hint: It’s not ophthalmic)*
As orangish skin papules

*At what age does it present?*
The majority before age 1 year, and almost all by 2

*What are the two hallmarks of JXG histology?*
The presence of **Touton** giant cells
The presence of 'foamy macrophages'

2) Medulloepithelioma

*Benign but locally aggressive neoplasia of nonpigmented epithelium of CB.*

3) Lisch nodules

4) Brushfield spots

5) Iris mammillations

6) Iris cysts

In three words, what sort of condition is JXG?
It is a... **nonneoplastic histiocytic proliferation**

How does JXG usually present? (Hint: It’s not ophthalmic)
As orangish skin papules

At what age does it present?
The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?
Unilateral

In what three ways are the iris nodules clinically significant?
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--They are in the DDx as a 'masquerade syndrome' in peds uveitis
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?
It is self-limited, usually resolving by age 5 years

Whipple’s disease

Speaking of ‘foamy macrophages’...

What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt had chronic migratory arthritis, associated with chronic diarrhea, and CNS symptoms--seizures, dementia, coma?

Whipple’s disease

When foamy macrophages are found in a biopsy performed on a Whipple’s pt, from what site was the biopsy collected?
The duodenum (remember, they have GI issues)

What other finding will a duodenal biopsy reveal?
The presence of PAS+ bacteria within macrophages located in intestinal villi

Broadly speaking, what sort of condition is Whipple’s?
It is infectious

What infection agent is responsible for Whipple’s?
The bacterium **Tropheryma whipplei**
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?*
It is a nonneoplastic histiocytic proliferation

**Speaking of foamy macrophages part deaux…**
*What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an Adult with bilateral upper-lid yellow lesions?*

*(Pic forthcoming—give the dx after seeing it)*

**Choroid**

The presence of Touton giant cells

The presence of ‘foamy macrophages’

When is it unilateral? When is it bilateral?

In what three ways are the iris nodules clinically significant?

--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
--If enough nodules are present, heterochromia iridis will result

**RPE**

What is the natural history of JXG?

It is self-limited, usually resolving by age 5 years

**Retina**

In what age does it present?

The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?

Unilateral

In what three ways are the iris nodules clinically significant?

--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma

--They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?

It is self-limited, usually resolving by age 5 years
Intraocular Tumors of Childhood

Condition?
1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux...
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions?
Xanthelsasma

When the presence of 'foamy macrophages' is unilateral?
Unilateral

In what three ways are the iris nodules clinically significant?
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--They are in the DDx as a 'masquerade syndrome' in peds uveitis
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?
It is self-limited, usually resolving by age 5 years
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux… What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an Adult with bilateral upper-lid yellow lesions? Xanthelasmas

Are xanthelasmas a harbinger of elevated serum lipids?

What is the natural history of JXG? It is self-limited, usually resolving by age 5 years

- Intraocular Tumors of Childhood
  - Iris/Ciliary Body
  - Choroid
  - RPE
  - Retina

1) Juvenile xanthogranuloma

- Nonneoplastic histiocytic proliferation
- <2 years old
- +/- skin papules
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Self-limited; regresses by age 5
- Treat inflammation and IOP
- Path: Touton giant cells

2) Medulloepithelioma

- Benign but locally aggressive neoplasia of nonpigmented epithelium of CB
- Presents: Iris mass before age 10 years
- Can bleed → hyphema → increased IOP → glaucoma
- Locally invasive → death
- Tx: Enucleate

3) Lisch nodules

- Strong association with NF1
- Lighter on dark irides; darker on light

4) Brushfield spots

- Strong association with Down syndrome; 15% of non-Down pop

5) Iris mammillations

- Tiny, numerous
- Same color as iris
- Weak association with NF1, Nevus of Ota

6) Iris cysts

- Can be pupillary, stromal, secondary

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation

How does JXG usually present? (Hint: It's not ophthalmic)

As orangish skin papules

At what age does it present?

The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?

Unilateral

In what three ways are the iris nodules clinically significant?

-- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
-- They are in the DDx as a 'masquerade syndrome' in peds uveitis
-- If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?

It is self-limited, usually resolving by age 5 years

What are the two hallmarks of JXG histology?

The presence of Touton giant cells
The presence of 'foamy macrophages'

Speaking of foamy macrophages part deaux… What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an Adult with bilateral upper-lid yellow lesions? Xanthelasmas

Are xanthelasmas a harbinger of elevated serum lipids?

They can be, but in most cases the individual has normal lipid panels

Can they be congenital?

Yes, and when they are, they usually are a sign of lipid derangement

Xanthelasmas
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?*
It is a...nonneoplastic histiocytic proliferation

**Speaking of foamy macrophages part deaux...**
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a **Adult with bilateral upper-lid yellow lesions?**

**Xanthelasmas**

**Choroid**

*Are xanthelasmas a harbinger of elevated serum lipids?*
They can be, but in most cases the individual has normal lipid panels

**RPE**

*What is the natural history of JXG?*
It is self-limited, usually resolving by age 5 years

**Retina**

1) Juvenile xanthogranuloma

-Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old.
+/- skin papules. Iris nodules bleed -> hyphema -> increased IOP -> glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.


3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light.

4) Brushfield spots: Strong association with Down syndrome; 15% of non-Down pop.

5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) Iris cysts: Can be pupillary, stromal, secondary.
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?* It is a...nonneoplastic histiocytic proliferation

**Speaking of foamy macrophages part deaux…** What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an Adult with bilateral upper-lid yellow lesions?

**Xanthelasma**

- The presence of Touton giant cells
- The presence of foamy macrophages
  - [‘**foamy macrophages**’]
- Unilateral?
- Bilateral?

**Choroid**

Are xanthelasmamas a harbinger of elevated serum lipids? They can be, but in most cases the individual has normal lipid panels

**Can they be congenital?**

**RPE**

What is the natural history of JXG? It is self-limited, usually resolving by age 5 years

**Retina**

1) Juvenile xanthogranuloma

- profitable histiocytic proliferation
- +/- skin papules
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Self-limited; regresses by age 5
- Treat inflammation and IOP
- Path: Touton giant cells


3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light.

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6) Iris cysts: Can be pupillary, stromal, secondary.
**Intraocular Tumors of Childhood**

1) **Juvenile xanthogranuloma**

*In three words, what sort of condition is JXG?*
It is a... nonneoplastic histiocytic proliferation

**Speaking of foamy macrophages part deaux...**
What **dz** comes to mind if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions? **Xanthelasmas**

**Choroid**

Are xanthelasmas a harbinger of elevated serum lipids?
They can be, but in most cases the individual has normal lipid panels

Can they be congenital?
Yes, and when they are, they usually are a sign of lipid derangement

**RPE**

What is the natural history of JXG?
It is self-limited, usually resolving by age 5 years
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

It is a... nonneoplastic histiocytic proliferation

In three words, what sort of condition is JXG?

Speaking of foamy macrophages part deaux deaux...

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a
Adult with bilateral upper-lid yellow lesions?

Xanthelsasma

The presence of 'foamy macrophages'

When Unilateral? The presence of Touton giant cells

Speaking of foamy macrophages part deaux deaux...

What if the adult has what could only be described as crazy-bad xanthelsasma (pic next slide)

(No question yet—advance to the pic)

Retina
Intraocular Tumors of Childhood
Iris/Ciliary Body

Choroid

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?*
It is a... **nonneoplastic histiocytic proliferation**

**Speaking of foamy macrophages part deaux deaux…**
What *dz* comes to mind if, instead of a toddler with *iris nodules*, the pt in question was a Adult with bilateral upper-lid yellow lesions? **Xanthelsasma**

**When** The presence of *Touton giant cells*  
**Unilateral**

**Speaking of foamy macrophages part deaux deaux…**
What if the adult has what could only be described as *crazy-bad xanthelasma* (pic next slide), *and*: No other issues whatsoever? Give the diagnosis

Retina

1) Juvenile xanthogranuloma

**Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old.

+/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.


3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light.

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5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) Iris cysts: Can be pupillary, stromal, secondary.
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG?
It is a…nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux deaux…
What dz comes to mind if, instead of a toddler
with iris nodules, the pt in question was a
Adult with bilateral upper-lid yellow lesions?
Xanthelsasma

When
The presence of Touton giant cells

Unilateral?

Lateral?

'foamy macrophages'

Speaking of foamy macrophages part deaux deaux…
What if the adult has what could only be described
as crazy-bad xanthelasma (pic next slide), and:
No other issues whatsoever? Adult-onset xanthogranuloma

Retina
1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a… **nonneoplastic histiocytic proliferation**

Speaking of foamy macrophages part deaux…
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an Adult with bilateral upper-lid yellow lesions?

**Xanthelasma**

The presence of **Touton giant cells**

Unilateral or bilateral?

When

The presence of **'foamy macrophages'**

Speaking of foamy macrophages part deaux deaux…
What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), *and*: No other issues whatsoever? **Adult-onset xanthogranuloma**

Recently diagnosed asthma?

Ditto

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux deaux…
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions?
Xanthelasma

When 'foamy macrophages'

Unilateral?

Speaking of foamy macrophages part deaux deaux…
What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:
No other issues whatsoever? Adult-onset xanthogranuloma
Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma
1) **Juvenile xanthogranuloma**

*In three words, what sort of condition is JXG?*
It is a...nonneoplastic histiocytic proliferation

_Speaking of foamy macrophages part deaux deaux…_

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions? **Xanthelasma**

- The presence of Touton giant cells – unilateral?
- 'foamy macrophages'

_Speaking of foamy macrophages part deaux deaux…_

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), _and:_

No other issues whatsoever? **Adult-onset xanthogranuloma**
Recently diagnosed asthma? **Adult-onset asthma with periocular xanthogranuloma**
The ‘xanthelasma’ are ulcerated? Ditto

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a non-neoplastic histiocytic proliferation.

Speaking of foamy macrophages part deaux deaux…
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an adult with bilateral upper-lid yellow lesions?
Xanthelsasma

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:
No other issues whatsoever? Adult-onset xanthogranuloma
Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma
The ‘xanthelasma’ are ulcerated? Necrobiotic xanthogranuloma

Speaking of foamy macrophages part deaux deaux…
When are JXG unilateral?

Retina
Necrobiotic xanthogranuloma
1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux... What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions? Xanthelasma

When The presence of 'foamy macrophages'

Unilateral? The presence of Touton giant cells

Speaking of foamy macrophages part deaux deaux... What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and: No other issues whatsoever? Adult-onset xanthogranuloma

Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma

The ‘xanthelasma’ are ulcerated? Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present? Erdheim-Chester dz

Retina
Iris/Ciliary Body

Choroid

RPE

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

*In three words, what sort of condition is JXG?*

It is a... **nonneoplastic histiocytic proliferation**

**Speaking of foamy macrophages part deaux...**

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a
Adult with bilateral upper-lid yellow lesions?

**Xanthelsasma**

**When** The presence of *Touton* giant cells

**Unilateral?**

The presence of *'foamy macrophages'*

**Speaking of foamy macrophages part deaux deaux...**

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:

*No other issues whatsoever?* **Adult-onset xanthogranuloma**

*Recently diagnosed asthma?* **Adult-onset asthma with periocular xanthogranuloma**

*The ‘xanthelasma’ are ulcerated?* **Necrobiotic xanthogranuloma**

*Proptosis and/or terrible systemic symptoms are present?* **Erdheim-Chester dz**
Intraocular Tumors of Childhood

Erdheim-Chester disease
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a...nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux deaux...

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an adult with bilateral upper-lid yellow lesions? Xanthelsasma

Collectively, these conditions are known as the...

Adult-onset xanthogranuloma
Adult-onset asthma with periocular xanthogranuloma
Necrobiotic xanthogranuloma
Erdheim-Chester dz

Intraocular Tumors of Childhood

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG? It is a... nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux deaux...
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a
Adult with bilateral upper-lid yellow lesions?
Xanthelsasma

When The presence of Touton giant cells
Unilateral?

Adult-onset xanthogranuloma
Adult-onset asthma with periocular xanthogranuloma

Collectively, these conditions are known as the...
Adult xanthogranulomas
Intraocular Tumors of Childhood

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- The presence of 'foamy macrophages'...

In three words, what sort of condition are the AXGs?

Collectively, these conditions are known as the...

Adult xanthogranulomas

- Adult-onset xanthogranuloma
- Adult-onset asthma with periocular xanthogranuloma
- Necrobiotic xanthogranuloma
- Proptosis and/or highly suggestive systemic symptoms are present? Erdheim-Chester dz

Retina

- 1) Juvenile xanthogranuloma
- 2) Medulloepithelioma: Benign but locally aggressive neoplasia of nonpigmented epithelium of CB.
- 3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light.
- 4) Brushfield spots: Strong association with Down syndrome; 15% of non-Down pop.
- 5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.
- 6) Iris cysts: Can be pupillary, stromal, secondary.

- 1) Juvenile xanthogranuloma
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What dz comes to mind if instead of a toddler, an adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:
- No other issues whatsoever? Adult-onset xanthogranuloma
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Collectively, these conditions are known as the... Adult xanthogranulomas.

'Nonneoplastic histiocytic proliferations'... Why does that sound so familiar?

In three words, what sort of condition are the AXGs? They are... nonneoplastic histiocytic proliferations.

The presence of... Touton giant cells... lateral?

The presence of... 'foamy macrophages'...
Intraocular Tumors of Childhood

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Collectively, these conditions are known as the…
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In three words, what sort of condition is JXG?
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How does JXG usually present? (Hint: It’s not ophthalmic)
As orangish skin papules

At what age does it present?
The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?
Unilateral

In what three ways are the iris nodules clinically significant?
--They are prone to spontaneous bleeding, with subsequent
hyphema and secondary glaucoma
--They are in the DDx as a ‘masquerade syndrome’ in pediatrics
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?
It is self-limited, usually resolving by age 5 years

What are the two hallmarks of JXG histology?
The presence of Touton giant cells
The presence of ‘foamy macrophages’

Speaking of foamy macrophages part deaux deaux…
What dz comes to mind if, instead of a toddler
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‘Nonneoplastic histiocytic proliferations’…Why does that sound so familiar?
Because that’s the phrase we used to describe/define JXG

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The majority before age 1 year, and almost all by 2 years.

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In what three ways are the iris nodules clinically significant?

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Iris/Ciliary Body

Choroid

RPE

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This implies that JXG is somehow related to the AXGs. Is that the case?

Indeed it is.

What is the nature of this relationship?

They (JXG and the AXGs) together comprise the non-Langerhans cell histiocytoses.
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Intraocular Tumors of Childhood

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In three words, what sort of condition is JXG? It is a nonneoplastic histiocytic proliferation.

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Choroid

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The presence of Touton giant cells.
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Speaking of foamy macrophages part deaux deaux…

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In three words, what sort of condition are the AXGs? They are nonneoplastic histiocytic proliferations.

Collectively, these conditions are known as the... Adult xanthogranulomas

Juvenile xanthogranuloma

What if the adult has what could only be described as crazy bad xanthelasma (papillary yellow plaques)?

No other issues whatsoever? Adult-onset xanthogranuloma

Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated? Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present? Erdheim-Chester dz

Non-Langerhans cell histiocytoses...Why does that sound so familiar? Because that's the phrase we used to describe/define JXG.

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Juvenile xanthogranuloma

No question—summary slide, advance when ready
In three words, what sort of condition is JXG?
Nonneoplastic histiocytic proliferation

How does JXG usually present?
As orangish skin papules

At what age does it present?
The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?
Unilateral

In what three ways are the iris nodules clinically significant?
-- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
-- They are in the DDx as a 'masquerade syndrome' in peds uveitis
-- If enough nodules are present, heterochromia iridis will result

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Collectively, these conditions are known as the...
Adult xanthogranulomas
Juvenile xanthogranuloma

Do the AXGs have Touton giant cells like their juvenile cousin?
Indeed they do
In three words, what sort of condition is JXG?
It is a nonneoplastic histiocytic proliferation.

How does JXG usually present? (Hint: It's not ophthalmic)
As orangish skin papules.

At what age does it present?
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What is the natural history of JXG?
It is self-limited, usually resolving by age 5 years.

Collectively, these conditions are known as the...

<table>
<thead>
<tr>
<th>Adult xanthogranulomas</th>
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<td>Proptosis and/or terrible systemic symptoms are present?</td>
<td>Erdheim-Chester dz</td>
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In three words, what sort of condition are the AXGs?
They are nonneoplastic histiocytic proliferations.

What if the adult has what could only be described as crazy bad xanthelasma (pi)?
Adult-onset xanthogranuloma
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They (JXG and the AXGs) together comprise the non-Langerhans cell histiocytoses.
**Intraocular Tumors of Childhood**

**That there is something called a non-Langerhans cell histiocytosis implies that Langerhans histiocytosis is a thing. Is there?**

*non-Langerhans cell histiocytes*

**What is the nature of this relationship?**
They (JXG and the AXGs) together comprise the non-Langerhans cell histiocytes.

**What if the adult has what could only be described as crazy bad xanthelasma (pi)?**
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In three words, what sort of condition are the AXGs?
They are nonneoplastic histiocytic proliferations.

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Do the AXGs have Touton giant cells like their juvenile cousin? Indeed they do.

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What are the two hallmarks of JXG histology?
The presence of Touton giant cells
The presence of 'foamy macrophages'
In three words, what sort of condition is JXG?

It is a nonneoplastic histiocytic proliferation.

How does JXG usually present? (Hint: It's not ophthalmic)

As orangish skin papules.

At what age does it present?

The majority before age 1 year, and almost all by 2 years.

When JXG iris nodules are present, are they uni-, or bilateral?

Unilateral.

In what three ways are the iris nodules clinically significant?

1. They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma.
2. They are in the DDx as a 'masquerade syndrome' in peds uveitis.
3. If enough nodules are present, heterochromia iridis will result.

What is the natural history of JXG?

It is self-limited, usually resolving by age 5 years.

Collectively, these conditions are known as the...

Juvenile xanthogranuloma.

Iris/Ciliary Body

Choroid

RPE

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.


3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light.

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Speaking of foamy macrophages part deaux deaux…

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No other issues whatsoever?

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Recently diagnosed asthma?

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Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**
   - Nonneoplastic histiocytic proliferation
   - <2 years old
   - +/- skin papules
   - Iris nodules bleed → hyphema → increased IOP → glaucoma
   - Self-limited; regresses by age 5
   - Treat inflammation and IOP
   - Path: Touton giant cells

2) **Medulloepithelioma**
   - Benign but locally aggressive neoplasia of nonpigmented epithelium of CB
   - Presents: Iris mass before age 10 years
   - Can bleed → hyphema → increased IOP → glaucoma
   - Locally invasive → death
   - Tx: Enucleate

3) **Lisch nodules**
   - Strong association with NF1
   - Lighter on dark irides; darker on light

4) **Brushfield spots**
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In three words, what sort of condition is JXG?

It is a…

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How does JXG usually present? (Hint: It's not ophthalmic)

As orangish skin papules

At what age does it present?

The majority before age 1 year, and almost all by 2

When JXG iris nodules are present, are they uni-, or bilateral?

Unilateral

In what three ways are the iris nodules clinically significant?

--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
--They are in the DDx as a 'masquerade syndrome' in peds uveitis
--If enough nodules are present, heterochromia iridis will result

What is the natural history of JXG?

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Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- **What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?**
  - Imaging → soft tissue mass + lytic lesions?
  - **Langerhans-cell histiocytosis** (and there it is!)

- **What is the natural history of JXG?**
  - It is self-limited, usually resolving by age 5 years

- **In what three ways are the iris nodules clinically significant?**
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- **What are the two hallmarks of JXG histology?**
  - The presence of Touton giant cells
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- **Speaking of foamy macrophages part three …**

  - **What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?**

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- **When is the presence of JXG nodules uni- or bilateral?**
  - Unilateral

- **Iris/Ciliary Body**

- **Choroid**

- **RPE**

- **Retina**
Intraocular Tumors of Childhood

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- Nonneoplastic histiocytic proliferation
- <2 years old
- +/- skin papules
- Iris nodules bleed \(\rightarrow\) hyphema \(\rightarrow\) increased IOP \(\rightarrow\) glaucoma
- Self-limited; regresses by age 5
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- Presents: Iris mass before age 10 years
- Can bleed \(\rightarrow\) hyphema \(\rightarrow\) increased IOP \(\rightarrow\) glaucoma
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What is the natural history of JXG?

- It is self-limited, usually resolving by age 5 years

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For more on the Langerhans (and non-Langerhans) histiocytoses, see slide-set K20
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma
   - Nonneoplastic histiocytic proliferation.
   - <2 years old.
   - +/- skin papules.
   - Iris nodules bleed → hyphema → increased IOP → glaucoma.
   - Self-limited; regresses by age 5.
   - Treat inflammation and IOP.
   - Path: Touton giant cells.

2) Medulloepithelioma
   - Benign but locally aggressive neoplasia of nonpigmented epithelium of CB.
   - Presents: Iris mass before age 10 years.
   - Can bleed → hyphema → increased IOP → glaucoma.
   - Locally invasive → death.
   - Tx: Enucleate.

3) Lisch nodules
   - Strong association with NF1.
   - Lighter on dark irides; darker on light.

4) Brushfield spots
   - Strong association with Down syndrome; 15% of non-Down pop.

5) Iris mammillations
   - Tiny, numerous.
   - Same color as iris.
   - Weak association with NF1, Nevus of Ota.

6) Iris cysts
   - Can be pupillary, stromal, secondary.

In three words, what sort of condition is JXG?
- It is a…
- nonneoplastic histiocytic proliferation

How does JXG usually present? (Hint: It's not ophthalmic)
- As orangish skin papules

At what age does it present?
- The majority before age 1 year, and almost all by 2 years.

When JXG iris nodules are present, are they uni-, or bilateral?
- Unilateral

In what three ways are the iris nodules clinically significant?
- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma.
- They are in the DDx as a 'masquerade syndrome' in peds uveitis.
- If enough nodules are present, heterochromia iridis will result.

What is the natural history of JXG?
- It is self-limited, usually resolving by age 5 years.

What are the two hallmarks of JXG histology?
- The presence of Touton giant cells.
- The presence of 'foamy macrophages'.

Speaking of foamy macrophages part three …
- What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?
- Imaging → soft tissue mass + lytic lesions?
- Langerhans-cell histiocytosis (and there it is!)
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- Nonneoplastic histiocytic proliferation
- <2 years old
- +/- skin papules
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Self-limited; regresses by age 5
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In three words, what sort of condition is JXG?

It is a… nonneoplastic histiocytic proliferation

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The presence of Touton giant cells
The presence of 'foamy macrophages'

Speaking of foamy macrophages part three …

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?

Imaging → soft tissue mass + lytic lesions?

Langerhans-cell histiocytosis (and there it is!)

Speaking of foamy macrophages part whatever …

What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?

No family hx for anything like this? → Clue #2

Speaking of foamy macrophages part whatever …

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a 8 y.o. with a superotemporal orbital mass?

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Langerhans-cell histiocytosis (and there it is!)

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What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?

No family hx for anything like this? → Clue #2
1) Juvenile xanthogranuloma

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What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?
Imaging → soft tissue mass + lytic lesions?
Langerhans-cell histiocytosis (and there it is!)

What are the two hallmarks of JXG histology?
The presence of Touton giant cells
The presence of 'foamy macrophages'

Speaking of foamy macrophages part whatever …
What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?
No family hx for anything like this?
DFE → Exudative RD?

Clue #3
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?

Imaging \(\rightarrow\) soft tissue mass + lytic lesions?

**Langerhans-cell histiocytosis** (and there it is!)

When?

- The presence of Touton giant cells
- The presence of 'foamy macrophages'
- Unilateral?

Speaking of foamy macrophages part whatever …

What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?

No family hx for anything like this?

**DFE \(\rightarrow\) Exudative RD?**

**DFE also \(\rightarrow\) Retinal vascular microaneurysms, telangiectasias, dilatation?**
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?
Imaging → soft tissue mass + lytic lesions?
**Langerhans-cell histiocytosis** (and there it is!)
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- Nonneoplastic histiocytic proliferation
- Occurs in <2 years old
- +/- skin papules
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Self-limited; regresses by age 5
- Treat inflammation and IOP
- Path: Touton giant cells

2) Medulloepithelioma

- Benign but locally aggressive neoplasia of nonpigmented epithelium of CB
- Presents: Iris mass before age 10 years
- Can bleed → hyphema → increased IOP → glaucoma
- Locally invasive → death
- Tx: Enucleate

3) Lisch nodules

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- Tiny, numerous
- Same color as iris
- Weak association with NF1, Nevus of Ota

6) Iris cysts

- Can be pupillary, stromal, secondary

In three words, what sort of condition is JXG?
It is a...

nonneoplastic histiocytic proliferation

How does JXG usually present? (Hint: It's not ophthalmic)
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The majority before age 1 year, and almost all by 2

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Coats dz

Where are the foamy macrophages found in Coats dz?
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?

Imaging → soft tissue mass + lytic lesions?

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Unilateral?

Speaking of foamy macrophages part whatever …

What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?

No family hx for anything like this?

DFE → Exudative RD?

DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?

Coats dz

Where are the foamy macrophages found in Coats dz?

In the subretinal exudate
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a 8 y.o. with with a superotemporal orbital mass?
Imaging → soft tissue mass + lytic lesions?
Langerhans-cell histiocytosis (and there it is!)

Speaking of foamy macrophages part whatever …
What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?
No family hx for anything like this?
DFE → Exudative RD?
DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?
Coats dz

Where are the foamy macrophages found in Coats dz?
In the subretinal exudate (also present: substance crystals)
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

**Speaking of foamy macrophages part three ...**

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a 8 y.o. with a superotemporal orbital mass?

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- The presence of 'foamy macrophages'

Unilateral?

Speaking of foamy macrophages part whatever ...

What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?

No family hx for anything like this?

**DFE** \(\rightarrow\) Exudative RD?

DFE also \(\rightarrow\) Retinal vascular microaneurysms, telangiectasias, dilatation?

**Coats dz**

Where are the foamy macrophages found in Coats dz?

In the subretinal exudate (also present: cholesterol crystals)
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed→hyphema→increased IOP→glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma**

**Iris/Ciliary Body**

*What is the other name by which medulloepithelioma is known?*

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**RPE**

**Retina**

**Choroid**

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**RPE**

**Retina**

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**Retina**

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**Retina**
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) Medulloepithelioma

What is the other name by which medulloepithelioma is known? Diktyoma

Iris/Ciliary Body

Choroid

RPE

Retina
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma**

**Iris/Ciliary Body**

*What is the other name by which medulloepithelioma is known?* Diktyoma

*Which specific component of the iris/CB is involved in medulloepithelioma?*
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed→hyphema→increased IOP→glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) Medulloepithelioma

Iris/Ciliary Body

What is the other name by which medulloepithelioma is known?
Diktyoma

Which specific component of the iris/CB is involved in medulloepithelioma?
The nonpigmented epithelium of the ciliary body
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma**

**Iris/Ciliary Body**

- What is the other name by which medulloepithelioma is known? Diktyoma

- Which specific component of the iris/CB is involved in medulloepithelioma? **The nonpigmented epithelium of the ciliary body**

3) **Lisch nodules:** Strong association with NF1. Lighter on dark irides; darker on light

4) **Brushfield spots:** Strong association with Down syndrome; 15% of non-Down pop.

5) **Iris mammillations:** Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

6) **Iris cysts:** Can be pupillary, stromal, secondary

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**Iris/Ciliary Body**

- **What extremely important function does the nonpigmented epi of the CB perform?**
  - It is responsible for the creation of aqueous humor

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**Choroid**

- **RPE**

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**Retina**
1) 

**Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma**

**Iris/Ciliary Body**

*What is the other name by which medulloepithelioma is known?*
Diktyoma

*Which specific component of the iris/CB is involved in medulloepithelioma?*

**The nonpigmented epithelium of the ciliary body**

*What extremely important function does the nonpigmented epi of the CB perform?*
It is responsible for the creation of aqueous humor
Intraocular Tumors of Childhood

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2) **Medulloepithelioma**

Iris/Ciliary Body

**What is the other name by which medulloepithelioma is known?**
Diktyoma

**Which specific component of the iris/CB is involved in medulloepithelioma?**
The nonpigmented epithelium of the ciliary body

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Iris/Ciliary Body

RPE

Retina
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2) **Medulloepithelioma**

**Iris/Ciliary Body**

- **What is the other name by which medulloepithelioma is known?** Diktyoma
- **Which specific component of the iris/CB is involved in medulloepithelioma?** The nonpigmented epithelium of the ciliary body
- **How does it present?**
  - Glaucoma
  - Hyphema
  - Sectoral cataract

**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed \rightarrow hyphema \rightarrow increased IOP \rightarrow glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma**

**Iris/Ciliary Body**

*What is the other name by which medulloepithelioma is known?*
Diktyoma

*Which specific component of the iris/CB is involved in medulloepithelioma?*
The nonpigmented epithelium of the ciliary body

*How does it present?*
As an iris mass

**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

Medulloepithelioma/diktyoma
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2) **Medulloepithelioma**

**Iris/Ciliary Body**

*What is the other name by which medulloepithelioma is known?*  
Diktyoma

*Which specific component of the iris/CB is involved in medulloepithelioma?*  
The nonpigmented epithelium of the ciliary body

*How does it present?*  
As an iris mass along with one or more of the following:

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**Choroid**

**Retina**

**RPE**
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2) Medulloepithelioma

Iris/Ciliary Body

What is the other name by which medulloepithelioma is known? Diktyoma

Which specific component of the iris/CB is involved in medulloepithelioma? The nonpigmented epithelium of the ciliary body

How does it present? As an iris mass along with one or more of the following:
-- Glaucoma
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-- Sectoral cataract

Retina
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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*Which specific component of the iris/CB is involved in medulloepithelioma?*
The nonpigmented epithelium of the ciliary body

*How does it present?*
As an iris mass along with one or more of the following:
--Glaucoma
--Hyphema
--Sectoral cataract

*Is it common, or rare?*
Very rare

*Is it benign, or malignant?*
It is benign, but very aggressive locally

*How is it managed?*
Enucleation is usually required
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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**Iris/Ciliary Body**

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*Which specific component of the iris/CB is involved in medulloepithelioma?*  
The nonpigmented epithelium of the ciliary body

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**Choroid**

*Retina**

**RPE**
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It can have features of both. Either way, it is very locally aggressive.
Intraocular Tumors of Childhood

Medulloepithelioma/diktyoma
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma**

   *What is the other name by which medulloepithelioma is known?*
   Diktyoma

   *Which specific component of the iris/CB is involved in medulloepithelioma?*
   The nonpigmented epithelium of the ciliary body

   *How does it present?*
   As an iris mass along with one or more of the following:
   -- Glaucoma
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   -- Sectoral cataract

   *Is it common, or rare?*
   Very rare

   *Is it a benign, or malignant lesion?*
   It can have features of both. Either way, it is very locally aggressive.

   *Is a tendency to metastasize one of its ‘malignant features’?*
   No, this lesion rarely metastasizes; it does its damage locally
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) Medulloepithelioma

What is the other name by which medulloepithelioma is known? Diktyoma

Which specific component of the iris/CB is involved in medulloepithelioma? The nonpigmented epithelium of the ciliary body

How does it present? As an iris mass along with one or more of the following:
-- Glaucoma
-- Hyphema
-- Sectoral cataract

Is it common, or rare? Very rare

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2) **Medulloepithelioma**

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**Iris/Ciliary Body**

- **What is the other name by which medulloepithelioma is known?** Diktyoma
- **Which specific component of the iris/CB is involved in medulloepithelioma?** The nonpigmented epithelium of the ciliary body
- **How does it present?** As an iris mass along with one or more of the following: -- Glaucoma -- Hyphema -- Sectoral cataract
- **Is it common, or rare?** Very rare
- **Is it a benign, or malignant lesion?** It can have features of both. Either way, it is **very locally aggressive**.

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**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

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2) Medulloepithelioma

Iris/Ciliary Body

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-- Glaucoma
-- Hyphema
-- Sectoral cataract

Is it common, or rare?
Very rare

Is it a benign, or malignant lesion?
It can have features of both. Either way, it is very locally aggressive.

How aggressive is ‘very aggressive’?
Aggressive enough to result in death
Intraocular Tumors of Childhood

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2) Medulloepithelioma

**Iris/Ciliary Body**

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The nonpigmented epithelium of the ciliary body

*How does it present?*
As an iris mass along with one or more of the following:
--Glaucoma
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*Is it common, or rare?*
Very rare

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It can have features of both. Either way, it is very locally aggressive.

*How is it managed?*

**Choroid**

**RPE**

**Retina**
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2) **Medulloepithelioma**

**Iris/Ciliary Body**

*What is the other name by which medulloepithelioma is known?*

Diktyoma

*Which specific component of the iris/CB is involved in medulloepithelioma?*

The nonpigmented epithelium of the ciliary body

*How does it present?*

As an iris mass along with one or more of the following:

-- Glaucoma
-- Hyphema
-- Sectoral cataract

*Is it common, or rare?*

Very rare

*Is it a benign, or malignant lesion?*

It can have features of both. Either way, it is very locally aggressive.

*How is it managed?*

Enucleation is usually required
1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed→hyphema→increased IOP→glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
2) **Medulloepithelioma** (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed→hyphema→increased IOP→glaucoma. Locally invasive→death. Tx: Enucleate
3) **Lisch nodules**
4) **Brushfield spots**

*Lisch nodules are most strongly associated with what congenital condition?*
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NF1
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In this context, what does NF1 stand for?

Neurofibromatosis type 1

In a word, what sort of condition is it?

A phakomatosis
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**Retina**

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Intraocular Tumors of Childhood

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**Intraocular Tumors of Childhood**

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**In general terms, how do phakomatoses present?**
With multiple lesions in two or more organ systems, including the skin and CNS
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5) **Iris mammillations:** Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) **Iris cysts:** Can be pupillary, stromal, secondary.

**Retina**

**Most NF1 lesions are associated with one of two cell types. What are they?**

**Lisch nodules are most strongly associated with what congenital condition?**

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**Most NF1 lesions are associated with one of two cell types. What are they?**

Melanocytes and neuroglial cells

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Neuroglial lesions
--?
--?
--?
--?

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--?
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**Most NF1 lesions are associated with one of two cell types. What are they?**

**Melanocytes** and **neuroglial cells**

**Melanocytic lesions**
- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

**Neuroglial lesions**
- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

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Give four classic examples of each: (YMMV of course)

Neuroglial lesions
--Nodular neurofibromas
--Plexiform neurofibromas
--Optic glioma
--Prominent corneal nerves

Lisch nodules are most strongly associated with what congenital condition? NF1

In what fundamental way do these lesions differ (other than the cell type of origin, duh)?

The melanocytic lesions are of no clinical significance beyond establishing the diagnosis, whereas the neuroglial lesions are associated with significant ocular and/or systemic morbidity.

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Intraocular Tumors of Childhood

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- Yes, but the relationship is far weaker—Lisch nodules occur in NF2, but so sporadically that they are not expected

What is the prevalence of Lisch nodules in NF1?

- The rule-of-thumb is that Lisch nodule prevalence equals the age of the patient times 10. Thus, 50% of 5 year olds will have them, 60% of 6 year olds, etc. At age 10 and beyond, essentially 100% of NF1 patients have Lisch nodules.

Are Lisch nodules clinically significant?

- No; their only significance is as a diagnostic marker for NF1

Most NF1 lesions are associated with one of two cell types. What are they?

- Melanocytes
- Neuroglial cells

In what fundamental way do these lesions differ (other than the cell type of origin, duh)?

- The melanocytic lesions are of no clinical significance beyond establishing the diagnosis, whereas the neuroglial lesions are associated with significant ocular and/or systemic morbidity

Give four classic examples of each: (YMMV of course)

**Melanocytic lesions**

- Café au lait spots
- Axillary/inguinal freckles
- Lisch nodules
- Choroidal lesions

**Neuroglial lesions**

- Nodular neurofibromas
- Plexiform neurofibromas
- Optic glioma
- Prominent corneal nerves

Are Lisch nodules clinically significant?

- No; their only significance is as a diagnostic marker for NF1
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) Medulloepithelioma (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate

3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light

4) Brushfield spots: Strong association with Down syndrome; 15% of non-Down pop.

5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

6) Iris cysts: Can be pupillary, stromal, secondary

Retina

Most NF1 lesions are associated with one of two cell types. What are they?

- Melanocytes and neuroglial cells

Lisch nodules are most strongly associated with what congenital condition?

- NF1

Are Lisch nodules associated with NF2?

- Yes, but the relationship is far weaker—Lisch nodules occur in NF2, but so sporadically that they are not expected

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(No question—proceed when ready)
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3) **Lisch nodules**

4) **Brushfield spots**

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**Iris/Ciliary Body**

**Lisch nodules are most strongly associated with what congenital condition?**

**NF1**

**Are Lisch nodules dark, or light?**

- It depends. Lisch nodules are lighter than the rest of the iris when the iris in question is dark, but darker than the rest when the iris is light.

**What is the prevalence of Lisch nodules in NF1?**

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No; their only significance is as a diagnostic marker for NF1

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**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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3) **Lisch nodules**
4) **Brushfield spots**

**Retina**

*Lisch nodules are most strongly associated with what congenital condition?*

NF1

Are Lisch nodules dark, or light?

It depends. Lisch nodules are **lighter** than the rest of the iris when the iris in question is dark, but **darker** than the rest when the iris is light.

What is the prevalence of Lisch nodules in NF1?

The rule-of-thumb is that Lisch nodule prevalence equals the age of the patient times 10. Thus, 50% of 5 year olds will have them, 60% of 6 year olds, etc. At age 10 and beyond, essentially 100% of NF1 patients have Lisch nodules.

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**Iris/Ciliary Body**

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4) Brushfield spots
5) Iris mammillations

**Brushfield spots are most strongly associated with what congenital condition?**
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3) **Lisch nodules**: Strong association with NF1. Lighter on dark irides; darker on light

4) **Brushfield spots**: Strong association with Down syndrome

5) **Iris mammillations**
Intraocular Tumors of Childhood

Brushfield spots
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3) **Lisch nodules:** Strong association with NF1. Lighter on dark irides; darker on light
4) **Brushfield spots**
5) **Iris mammillations**

**Iris/Ciliary Body**

*Brushfield spots are most strongly associated with what congenital condition?* Down syndrome

*What is the prevalence of Brushfield spots in the Down population?* At least 90%

*What is the clinical significance of Brushfield spots?* They have none

*When a clinically identical iris finding occurs in a non-Down individual, what are the lesions called?* Wolfflin nodules
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3) **Lisch nodules:** Strong association with NF1. Lighter on dark irides; darker on light irides

4) **Brushfield spots**

5) **Iris mammillations**

**Iris/Ciliary Body**

**Brushfield spots are most strongly associated with what congenital condition?**

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**What is the prevalence of Brushfield spots in the Down population?**

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5) **Iris mammillations**

**Iris/Ciliary Body**

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**Iris/Ciliary Body**

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Intraocular Tumors of Childhood

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Mammillations? Aren’t those a CNS thingamajig?

Iris/Ciliary Body

Retina

Choroid
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You’re thinking of the **mammillary bodies**, paired structures that are part of the limbic system
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**OK, then what are iris mammillations?**
Tiny pigmented iris nodules which, when present, are found in vast numbers diffusely scattered across the iris surface
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**Are they unilateral, or bilateral?**
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**With what phakomatosis are they associated?**
NF1 (albeit not nearly as strongly as Lisch nodules)
Intraocular Tumors of Childhood

Iris/Ciliary Body

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‘Tiny pigmented iris nodules associated with NF1’--given this, how on earth are you supposed to differentiate between Lisch nodules and mammillations?

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Tiny pigmented iris nodules which, when present, are found in vast numbers diffusely scattered across the iris surface.

‘Tiny pigmented iris nodules associated with NF1’--given this, how on earth are you supposed to differentiate between Lisch nodules and mammillations? By appearance. Iris mammillations are always the same color as the rest of the iris. In contrast and as stated previously, Lisch nodules are lighter when the iris is dark, but darker when the iris is light.

**With what phakomatosis are they associated?**
**NF1** (albeit not nearly as strongly as Lisch nodules)
Intraocular Tumors of Childhood

Iris/Ciliary Body

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With what phakomatosis are they associated?
NF1 (albeit not nearly as strongly as Lisch nodules)

In addition to NF1, iris mammillations have another important association. What is it?
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With what phakomatosis are they associated? 
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In addition to NF1, **iris mammillations have another important association.**

What is it? 
Oculodermal melanocytosis, aka **nevus of Ota**
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5) **Iris mammillations**

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**Mammillations? Aren’t those a CNS thingamajig?**
You’re thinking of the mammillary bodies, paired structures that are part of the limbic system.

**OK, then what are iris mammillations?**
Tiny pigmented iris nodules which, when present, are found in vast numbers diffusely scattered across the iris surface.

**Are they unilateral, or bilateral?**
Usually unilateral, but bilaterality occurs frequently enough that it can’t be used to rule them out.

**With what phakomatosis are they associated?**
NF1 (albeit not nearly as strongly as Lisch nodules).

*In addition to NF1, iris mammillations have another important association. What is it?*
Oculodermal melanocytosis, aka nevus of Ota
Intraocular Tumors of Childhood

Oculodermal melanocytosis (nevus of Ota)
Iris/Ciliary Body

1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.

2) **Medulloepithelioma** (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Locally invasive $\rightarrow$ death. Tx: Enucleate.

3) **Lisch nodules**: Strong association with NF1. Lighter on dark irides; darker on light.

4) **Brushfield spots**: Strong association with Down syndrome.

5) **Iris mammillations**: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

6) **Iris cysts**: Can be pupillary, stromal, secondary (see the Iris issues is kids slide-set).
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**Iris/Ciliary Body**

**Choroid**

Five tumors of the choroid

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**RPE**

**Retina**
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**Iris/Ciliary Body**

**Choroid**

- 1) Nevus
- 2) Melanocytoma
- 3) Osteoma
- 4) Isolated/focal choroidal hemangioma
- 5) Diffuse choroidal hemangioma

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**Five tumors of the choroid**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

1) Nevus
2) Melanocytoma
3) Osteoma
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma

But not 6)

What common sort of choroidal tumor—common in adults—is absent from this list?

Retina
Iris/Ciliary Body

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Choroid

1) **Nevus**
2) **Melanocytoma**
3) **Osteoma**
4) **Isolated/focal choroidal hemangioma**
5) **Diffuse choroidal hemangioma**
6) **Metastases**

**Five tumors of the choroid**

What common sort of choroidal tumor—common in adults—is absent from this list?

Choroidal tumors arising as **metastases** from a nonocular primary. In adults, metastasis of solid tumors to the uveal tract is common. It almost never happens in children.
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Five tumors of the choroid

But not 6) Metastases

What common sort of choroidal tumor—common in adults—is absent from this list? Choroidal tumors arising as metastases from a nonocular primary. In adults, metastasis of solid tumors to the uveal tract is common. It almost never happens in children.

If a child does suffer an ophthalmic metastasis, where does it tend to occur?
Intraocular Tumors of Childhood

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**Five tumors of the choroid**

But not 6) **Metastases**

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If a child does suffer an ophthalmic metastasis, where does it tend to occur? The **orbit**

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**Iris/Ciliary Body**

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**Choroid**
Intraocular Tumors of Childhood

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Melanocytoma is a variant of what common choroidal finding?

- It is a particular sort of choroidal nevus
- From what structure does it commonly arise? The optic disc
- Is it unilateral, or bilateral? It is virtually always unilateral
- Is there a racial predilection? No
- Does it affect visual acuity? Only in a minority of cases. But in almost all cases, it does affect visual fields.
- Does melanocytoma have the potential to undergo malignant transformation? Yes
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

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RPE

Retina

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From what structure does it commonly arise?
Intraocular Tumors of Childhood

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From what structure does it commonly arise?

The optic disc

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Retina

RPE

Choroid

Iris/Ciliary Body

Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

Melanocytoma

[Images of eye scans showing melanocytoma]
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**Iris/Ciliary Body**

**Choroid**

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**Iris/Ciliary Body**

**Choroid**

**RPE**

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**Melanocytoma** is a variant of what common choroidal finding?

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**Iris/Ciliary Body**

- Nevus: Common. Benign
- Melanocytoma
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**Choroid**

- Melanocytoma is a variant of what common choroidal finding?
  - **It is a particular sort of choroidal nevus**

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**Retina**

**RPE**

**Choroid**

**Iris/Ciliary Body**

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Yes, it is virtually always unilateral

**Is there a racial predilection?**

No

**Does it affect visual acuity?**

Only in a minority of cases. But in almost all cases, it does affect visual fields.

**Does melanocytoma have the potential to undergo malignant transformation?**

Yes
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**Iris/Ciliary Body**

**Choroid**

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**From what structure does it commonly arise?**
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   No

   **Does it affect visual acuity?**
   Only in a minority of cases. But in almost all cases, it does affect visual fields.

   **Does melanocytoma have the potential to undergo malignant transformation?**

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Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
2) Medulloepithelioma (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate
3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light
4) Brushfield spots: Strong association with Down syndrome
5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota
6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

1) Nevus: Common. Benign
2) Melanocytoma: From what structure does it commonly arise? The optic disc
   Does it have a unilateral/bilateral predilection? Yes, it is virtually always unilateral
   Is there a racial predilection? No
   Does it affect visual acuity? Only in a minority of cases. But in almost all cases, it does affect visual fields.
   Does melanocytoma have the potential to undergo malignant transformation? Yes

Melanocytoma is a variant of what common choroidal finding?
It is a particular sort of choroidal nevus
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4) Isolated/focal choroidal hemangioma
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**Melanocytoma is a variant of what common choroidal finding?**
It is a particular sort of choroidal nevus

**From what structure does it commonly arise?**
The optic disc

**Does it have a unilateral/bilateral predilection?**
Yes, it is virtually always unilateral

**Is there a racial predilection?**
No

**Does it affect visual acuity?**
Only in a minority of cases. But in almost all cases, it does affect visual fields.

**Does melanocytoma have the potential to undergo malignant transformation?**
Approximately what percent of cases will transform?

Yes
1-2
Iris/Ciliary Body

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Choroid

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   - From what structure does it commonly arise? The optic disc

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3) **Osteoma**

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Retina

**Melanocytoma is a variant of what common choroidal finding?**

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Nevus: Common. Benign

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Osteoma

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

In a word, what is a choroidal osteoma composed of?

Bone

Very rare

Benign

Yes, it is more common in females

Teens

Choroidal neovascular membrane
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Intraocular Tumors of Childhood

Osteoma
Intraocular Tumors of Childhood

Osteoma: FP, and $b$-scan demonstrating

buzzword describing a $b$-scan finding illustrated above
Intraocular Tumors of Childhood

Osteoma: FP, and b-scan demonstrating ‘shadowing’
Osteoma: Another example
Osteomas (same pt, different cuts).
Note how bright the lesions are
**In a word, what is a choroidal osteoma composed of?**

Bone

---

**Is it common or rare?**

Very rare

---

**Benign or malignant?**

Benign

---

**Does it have a gender predilection?**

Yes, it is more common in females

---

**Is it more typically found in pre-teens, or teens?**

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---

**If significant vision loss occurs, what osteoma complication is usually the culprit?**

Choroidal neovascular membrane

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**Intraocular Tumors of Childhood**

- Neovascularization. <2 years old.
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**RPE**

**Retina**
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---

**Choroid**

In a word, what is a choroidal osteoma composed of?

- Bone

**Is it common or rare?**

- Very rare

**Is it benign or malignant?**

- Benign

**Is there a gender predilection?**

- Yes, it is more common in females

**Is it more typically found in pre-teens, or teens?**

- Teens

**If significant vision loss occurs, what osteoma complication is usually the culprit?**

- Choroidal neovascular membrane
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Intraocular Tumors of Childhood

Choroid

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RPE

Retina
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Choroid

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Is it benign or malignant? Benign.

Does there a gender predilection? Yes, it is more common in M v F.

Choroid

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**In a word, what is a choroidal osteoma composed of?**

*Bone*

**Is it common or rare?**

*Very rare*

**Is it benign or malignant?**

*Benign*

**Does there a gender predilection?**

*Yes, it is more common in females*

**Intraocular Tumors of Childhood**

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Retina

RPE

Iris/Ciliary Body

Choroid

Bone

Retina
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Intraocular Tumors of Childhood

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**Is it more typically found in pre-teens, or teens?**
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**Choroid**

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**RPE**

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### Intraocular Tumors of Childhood

#### Iris/Ciliary Body

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#### Choroid

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### In a word, what is a choroidal osteoma composed of?

- **Bone**

### Is it common or rare?

- Very rare

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### Does there a gender predilection?

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### If significant vision loss occurs, what osteoma complication is usually the culprit?

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Choroidal neovascular membrane
Osteoma with CNVM in a 13 y.o. female
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**If significant vision loss occurs, what osteoma complication is usually the culprit?**

Choroidal neovascular membrane

---

**True osteomas are indeed rare; however, secondary osteoma-like lesions can be found in eyes with what sorts of history?**

Eyes that have suffered severe chronic inflammation (especially if they become phthisical)

**Choroid**

3) **Osteoma**

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**RPE**

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3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

By what other name is this lesion known?

Circumscribed choroidal hemangioma

Is it common, or rare? Rare

Is it associated with a systemic condition, ie, is it syndromic? No

How does it present? As a reddish-orange mass in the macula

What is its characteristic pattern on a -scan ultrasonography? It is one of 'high internal reflectivity'
Intraocular Tumors of Childhood

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By what other name is this lesion known? **Circumscribed choroidal hemangioma**
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2) **Medulloepithelioma (aka diktyoma):** Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate

3) **Lisch nodules:** Strong association with NF1. Lighter on dark irides; darker on light

4) **Brushfield spots:** Strong association with Down syndrome

5) **Iris mammillations:** Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

6) **Iris cysts:** Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

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1) **Nevus:** Common. Benign

2) **Melanocytoma:** Usually juxtapapillary. Malignant transformation extremely rare

3) **Osteoma:** Benign bony tumor, most common in teen years, females. Risk of CNVM

4) **Isolated/focal choroidal hemangioma**

5) **Diffuse choroidal hemangioma**

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**By what other name is this lesion known?**

**Is it common, or rare?**

**Is it associated with a systemic condition, ie, is it syndromic?**

**How does it present?**

**What is its characteristic pattern on a -scan ultrasonography?**

It is one of 'high internal reflectivity'
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By what other name is this lesion known? Circumscribed choroidal hemangioma

Is it common, or rare? Rare

Retina

Iris/Ciliary Body

Choroid

RPE
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By what other name is this lesion known? **Circumscribed choroidal hemangioma**

Is it common, or rare? Rare

Is it associated with a systemic condition, ie, is it syndromic? No

How does it present? As a reddish-orange mass in the macula

What is its characteristic pattern on an ultrasound? It is one of 'high internal reflectivity'
**Intraocular Tumors of Childhood**

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**By what other name is this lesion known?**

**Circumscribed choroidal hemangioma**

**Is it common, or rare?**

Rare

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**Intraocular Tumors of Childhood**

**Choroid**

*By what other name is this lesion known?*
**Circumscribed choroidal hemangioma**

*Is it common, or rare?*
Rare

*Is it associated with a systemic condition, ie, is it syndromic?*
No

*How does it present?*
As a reddish-orange mass in the macula

*What is its characteristic pattern on a -scan ultrasonography?*
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As a reddish-orange mass in the macula

Iris/Ciliary Body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

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Iris/Ciliary Body

Choroid

RPE

Retina

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Intraocular Tumors of Childhood

Circumscribed choroidal hemangioma: High internal reflectivity on a-scan
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**Isolated/focal choroidal hemangioma**

**Diffuse choroidal hemangioma**

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**By what other name is this lesion known?** CIRCUMSCRIBED choroidal hemangioma

**Is it common, or rare?** Rare

**Is it associated with a systemic condition, ie, is it syndromic?** No

**How does it present?** As a reddish-orange mass in the macula

**What is its characteristic pattern on a-scan ultrasonography?**

"High internal reflectivity"--what other choroidal lesion’s a-scan is described the same way?

It is one of high internal reflectivity

3) Osteoma. Benign bony tumor, most common in teen years, females. Risk of CNVM

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**Iris/Ciliary Body**

**Choroid**

**Retina**

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**By what other name is this lesion known?**

Circumscribed choroidal hemangioma

**Is it common, or rare?**

Rare

**Is it associated with a systemic condition, ie, is it syndromic?**

No

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'High internal reflectivity'--what other choroidal lesion’s a-scan is described the same way?

Choroidal nevus

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It is one of high internal reflectivity

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5) **Diffuse choroidal hemangioma**

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With what condition is the diffuse choroidal hemangioma associated?

Sturge-Weber syndrome (SWS)

Diffuse choroidal hemangioma is present in what percent of SWS?

About 50%

What does the fundus look like in an eye with a diffuse choroidal hemangioma?

The coloration is a very red—much more so than an unaffected fundus

What food-related term is used to describe the fundus appearance in SWS?

'Tomato catsup fundus'

Can the choroidal hemangioma be present bilaterally?

Yes, but it’s uncommon

Does the choroidal hemangioma have malignant potential?

No
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In a word, what sort of condition is SWS?
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A phakomatosis
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Diffuse choroidal hemangioma: Very rare. Characteristic a- and b-scan pattern. Associated with Sturge-Weber syndrome (SWS), very red fundus appearance, 'tomato catsup fundus'. Bilateral but uncommon. No malignant potential. Phakomatosis. Encephalotrigeminal angiomatosis. Also known as encephalofacial angiomatosis or cerebrofacial angiomatosis.
Intracocular Tumors of Childhood

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In a word, what sort of condition is SWS? A phakomatosis

What is the noneponymous name for SWS? Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

What is the hallmark skin finding in SWS? The port-wine stain

In one word, what sort of lesion is the port-wine stain? An angioma

When does it present? At birth

What is the typical pattern of distribution? It comports to the distribution of one or more divisions of CN5

Does it always present in this manner? No. Some cases will cross the midline of the face

All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS? No
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Intraocular Tumors of Childhood

With what condition is the diffuse choroidal hemangioma associated? Sturge-Weber syndrome (SWS)

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Nevus:

- Common. Benign.

Melanocytoma:

- Usually juxtapapillary. Malignant transformation extremely rare.

Osteoma:

- Benign bony tumor, most common in teen years, females. Risk of CNVM.

Isolated/focal choroidal hemangioma:

- Very rare. Characteristic a-scan pattern.

Diffuse choroidal hemangioma:

- Associated with Sturge-Weber syndrome (SWS).
- Present in about 50% of SWS.
- The coloration is a very red—much more so than an unaffected fundus.
- 'Tomato catsup fundus.'
- Can be present bilaterally but uncommonly.
- No malignant potential.

Sturge-Weber syndrome (SWS):

- A phakomatoses.
- Non-epithelial somatic mutation phakomatoses.
- Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis).

What is the hallmark skin finding in SWS?
The port-wine stain.

In one word, what sort of lesion is the port-wine stain? An angioma.

With what condition is the diffuse choroidal hemangioma associated? Sturge-Weber syndrome (SWS).
Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/− skin papules. Iris nodules bleed ⇒ hyphema ⇒ increased IOP ⇒ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

Medulloepithelioma (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed ⇒ hyphema ⇒ increased IOP ⇒ glaucoma. Locally invasive ⇒ death. Tx: Enucleate

Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light

Brushfield spots: Strong association with Down syndrome

Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues is kids slide-set)

Nevus: Common. Benign

Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

Diffuse choroidal hemangioma is present in what percent of SWS? About 50%

What does the fundus look like in an eye with a diffuse choroidal hemangioma? The coloration is a very red—much more so than an unaffected fundus

What food-related term is used to describe the fundus appearance in SWS? 'Tomato catsup fundus'

Can the choroidal hemangioma be present bilaterally? Yes, but it's uncommon

Does the choroidal hemangioma have malignant potential? No

In a word, what sort of condition is SWS? A phakomatosis

What is the noneponymous name for SWS? Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

What is the hallmark skin finding in SWS? The port-wine stain

In one word, what sort of lesion is the port-wine stain? An angioma

When does it present?
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. 
   <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. 
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6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

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1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

5) Diffuse choroidal hemangioma: Sturge-Weber syndrome (SWS)

   In a word, what sort of condition is SWS? 
   A phakomatosis

   What is the noneponymous name for SWS? 
   Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

   What is the hallmark skin finding in SWS? 
   The port-wine stain

   In one word, what sort of lesion is the port-wine stain? 
   An angioma

   When does it present? 
   At birth
Intraocular Tumors of Childhood

Sturge-Weber: Port-wine stain
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed \(\rightarrow\) hyphema \(\rightarrow\) increased IOP \(\rightarrow\) glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.

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6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set).

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3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM.

4) Isolated/focal choroidal hemangioma: Very rare. Characteristic \(\alpha\)-scan pattern.


  - In a word, what sort of condition is SWS? A phakomatosis.
  - What is the noneponymous name for SWS? Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis).

  - What is the hallmark skin finding in SWS? The port-wine stain.
  - In one word, what sort of lesion is the port-wine stain? An angioma.
  - When does it present? At birth.
  - What is the typical pattern of distribution? It comports to the distribution of one or more divisions of CN5. Does it always present in this manner? No. Some cases will cross the midline of the face. All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS? No.
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3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) Isolated/focal choroidal hemangioma: Very rare. Characteristic \textit{a}-scan pattern

5) Diffuse choroidal hemangioma

   With what condition is the diffuse choroidal hemangioma associated? \textbf{Sturge-Weber syndrome (SWS)}

   \textbf{In a word, what sort of condition is SWS?} A phakomatosi

   \textbf{What is the noneponymous name for SWS?} Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

   \textbf{What is the hallmark skin finding in SWS?} The \textbf{port-wine stain}

   \textbf{In one word, what sort of lesion is the port-wine stain?} An angioma

   \textbf{When does it present?} At birth

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4) Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

5) Diffuse choroidal hemangioma: With what condition is the diffuse choroidal hemangioma associated? Sturge-Weber syndrome (SWS). Diffuse choroidal hemangioma is present in what percent of SWS? About 50%. What does the fundus look like in an eye with a diffuse choroidal hemangioma? The coloration is a very red—much more so than an unaffected fundus. What food-related term is used to describe the fundus appearance in SWS? 'Tomato catsup fundus'. Can the choroidal hemangioma be present bilaterally? Yes, but it's uncommon. Does the choroidal hemangioma have malignant potential? No. In a word, what sort of condition is SWS? A phakomatosis. What is the noneponymous name for SWS? Encephalotrigeminal angiomatosis (you might also see encephalotrigeminal or cerebrofacial angiomatosis). What is the hallmark skin finding in SWS? The port-wine stain. In one word, what sort of lesion is the port-wine stain? An angioma. When does it present? At birth. Does it always present in this manner? No. Some cases will cross the midline of the face. All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS? No.
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Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern.

Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). About 50% present. The coloration is a very red—much more so than an unaffected fundus. 'Tomato catsup fundus.' Yes, but it's uncommon. No malignant potential. A phakomatosis. Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis).

What is the hallmark skin finding in SWS? The port-wine stain.

In one word, what sort of lesion is the port-wine stain? An angioma.

When does it present? At birth.

What is the typical pattern of distribution? It comports to the distribution of one or more divisions of CN5.

Does it always present in this manner? No, some cases will cross the midline of the face.
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2) Medulloepithelioma (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate

3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light

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6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

In a word, what sort of condition is SWS?
A phakomatosis

What is the noneponymous name for SWS?
Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

What is the hallmark skin finding in SWS?
The port-wine stain

In one word, what sort of lesion is the port-wine stain?
An angioma

When does it present?
At birth

What is the typical pattern of distribution?
It comports to the distribution of one or more divisions of CN5

Does it always present in this manner?
No, some cases will cross the midline of the face

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**Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-scan pattern

**Diffuse choroidal hemangioma:** Associated with Sturge-Weber syndrome (SWS). About 50% of cases. The coloration is a very red—much more so than an unaffected fundus. 'Tomato catsup fundus'. Yes, but it's uncommon. No

**Sturge-Weber syndrome (SWS):**

**In a word, what sort of condition is SWS?**
A phakomatosis

**What is the noneponymous name for SWS?**
Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

**What is the hallmark skin finding in SWS?**
The **port-wine stain**

**In one word, what sort of lesion is the port-wine stain?**
An angioma

**When does it present?**
At birth

**What is the typical pattern of distribution?**
It comports to the distribution of one or more divisions of CN5

**Does it always present in this manner?**
No, some cases will cross the midline of the face

**All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS?**
No
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/− skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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1) **Nevus:** Common. Benign

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4) **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-CTA scan pattern

5) **Diffuse choroidal hemangioma**

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**With what condition is the diffuse choroidal hemangioma associated?**

Sturge-Weber syndrome (SWS)

**Diffuse choroidal hemangioma is present in what percent of SWS?**

About 50%

What does the fundus look like in an eye with a diffuse choroidal hemangioma?

The coloration is a very red—much more so than an unaffected fundus

What food-related term is used to describe the fundus appearance in SWS?

'Tomato catsup fundus'

Can the choroidal hemangioma be present bilaterally?

Yes, but it’s uncommon

Does the choroidal hemangioma have malignant potential?

No
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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4) **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-scan pattern

5) **Diffuse choroidal hemangioma**

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**With what condition is the diffuse choroidal hemangioma associated?**
Sturge-Weber syndrome (SWS)

**Diffuse choroidal hemangioma is present in what percent of SWS?**
About half
Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

Isolated/focal choroidal hemangioma: Very rare. Characteristic a- and b-scan pattern

5) Diffuse choroidal hemangioma

With what condition is the diffuse choroidal hemangioma associated? Sturge-Weber syndrome (SWS)

Diffuse choroidal hemangioma is present in what percent of SWS? About half

What does the fundus look like in an eye with a diffuse choroidal hemangioma? The coloration is a very red--much more so than an unaffected fundus

What food-related term is used to describe the fundus appearance in SWS? 'Tomato catsup fundus'

Can the choroidal hemangioma be present bilaterally? Yes, but it's uncommon

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6) **Iris cysts:** Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set).

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**Iris/Ciliary Body**

**Choroid**

**Intraocular Tumors of Childhood**

With what condition is the diffuse choroidal hemangioma associated? Sturge-Weber syndrome (SWS)

Diffuse choroidal hemangioma is present in what percent of SWS? About half

What does the fundus look like in an eye with a diffuse choroidal hemangioma? The coloration is a very red, much more so than an unaffected fundus.

5) **Diffuse choroidal hemangioma**

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**RPE**

**Retina**
Intraocular Tumors of Childhood

Sturge-Weber: Tomato catsup fundus OD
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2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

5) Diffuse choroidal hemangioma

**With what condition is the diffuse choroidal hemangioma associated?** Sturge-Weber syndrome (SWS)

**Diffuse choroidal hemangioma is present in what percent of SWS?** About half

**What does the fundus look like in an eye with a diffuse choroidal hemangioma?** The coloration is a very red, much more so than an unaffected fundus

**What food-related term is used to describe the fundus appearance in SWS?** 'Tomato catsup fundus'

**Can the choroidal hemangioma be present bilaterally?** Yes, but it's uncommon

**Does the choroidal hemangioma have malignant potential?** No

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**Iris/Ciliary Body**

**Choroid**

**Retina**

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**Intraocular Tumors of Childhood**

*With what condition is the diffuse choroidal hemangioma associated?* Sturge-Weber syndrome (SWS)

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5) **Diffuse choroidal hemangioma**

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**Retina**

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RPE
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What food-related term is used to describe the fundus appearance in SWS? ‘Tomato catsup fundus’

Can the choroidal hemangioma be present bilaterally? Yes, but it's uncommon

Does the choroidal hemangioma have malignant potential? No

With what condition is the diffuse choroidal hemangioma associated? Sturge-Weber syndrome (SWS)

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**Isolated/focal choroidal hemangioma:** Very rare. Characteristic a -scan pattern

**Diffuse choroidal hemangioma:**

- **With what condition is the diffuse choroidal hemangioma associated?**
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5) **Diffuse choroidal hemangioma**

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RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

1) ?

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**Iris/Ciliary Body**

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**RPE**

1) **Congenital hypertrophy of the RPE (CHRPE)**

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**Retina**
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**Iris/Ciliary Body**

**What is the clinical appearance of CHRPE?**

**Choroid**

- 1) **Congenital hypertrophy of the RPE (CHRPE)**

**Retina**

**RPE**

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Flat, mainly black lesion(s) ranging in size from a 1 mm up to ~10

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1) **Congenital hypertrophy of the RPE (CHRPE)**

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**RPE**

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**Retina**

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**Iris/Ciliary Body**

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**Iris/Ciliary Body**

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**Iris/Ciliary Body**
Intraocular Tumors of Childhood

CHRPE
Intraocular Tumors of Childhood

Iris/Ciliary Body

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**RPE**

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**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Flat, mainly black lesion(s) ranging in size from a 1 mm up to ~10

**Is it common, or rare?**
Common

Choroid

1) **Congenital hypertrophy of the RPE (CHRPE)**

Retina

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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Retina

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What is the clinical appearance of CHRPE?
Flat, mainly black lesion(s) ranging in size from a 1 mm up to ~10

Is it common, or rare?
Common

Is it a hamartoma or a choristoma?

RPE

Choroid

Is it common, or rare?
Common

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Intraocular Tumors of Childhood

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RPE

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Intraocular Tumors of Childhood

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RPE

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**Iris/Ciliary Body**

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**Retina**

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**What is the clinical appearance of CHRPE?**
Flat, mainly black lesion(s) ranging in size from a 1 mm up to ~10

**Is it common, or rare?**
Common

**Is it a hamartoma or a choristoma?**
It is neither

**Choroid**

- Very rare
- Found in Sturge-Weber syndrome
- Unilateral
- Characteristic a-scan pattern
- Risk of CNVM

**RPE**

- 1) **Congenital hypertrophy of the RPE (CHRPE)**

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Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

1) **Congenital hypertrophy of the RPE (CHRPE)**

**CHRPE is characterized according to its presentation. In what two ways does it present?**

--Solitary CHRPE
--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

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CHRPE is characterized according to its presentation. In what two ways does it present?

- **CHRPE**

- **or**

CHRPE: Large lesion(s) surrounded by a few smaller ones
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Intraocular Tumors of Childhood

Solitary

Grouped

CHRPE
Intraocular Tumors of Childhood

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RPE

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CHRPE is characterized according to its presentation. In what two ways does it present?

--Solitary CHRPE

--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

What descriptive name is used with regard to the appearance of Multifocal/Grouped CHRPE?
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

1) Congenital hypertrophy of the RPE (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?
--Solitary CHRPE
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What descriptive name is used with regard to the appearance of Multifocal/Grouped CHRPE? ‘Bear tracks’
Intraocular Tumors of Childhood

CHRPE: Bear tracks
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**Iris/Ciliary Body**

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and non-eponymous) of this syndrome?

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**CHRPE**

1) **Congenital hypertrophy of the RPE** (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?

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--Solitary  CHRPE

--Multifocal or **Grouped**  CHRPE: Large lesion(s) surrounded by a few smaller ones
1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

2) **Medulloepithelioma** (aka *diktyoma*): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate

3) **Lisch nodules**: Strong association with NF1. Lighter on dark irides; darker on light

4) **Brushfield spots**: Strong association with Down syndrome

5) **Iris mammillations**: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

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**A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?**

Familial adenomatous polyposis, aka **Gardner syndrome**

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**RPE**

1) **Congenital hypertrophy of the RPE (CHRPE)**

CHRPE is characterized according to its presentation. In what two ways does it present?

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**--Solitary** CHRPE

**--Multifocal** or **Grouped** CHRPE: Large lesion(s) surrounded by a few smaller ones
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

A CHRPE-like lesion
(both eponymous and non-)
- Familial adenomatous polyposis, aka Gardner syndrome
- CHRPE is characterized according to its presentation. In what two ways does it present?
  -- Solitary CHRPE
  -- Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

Take careful note of the modifier ‘like’ here, because while CHRPE and the lesions associated with Gardner syndrome are ophthalmoscopically similar, they are not the same!

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?
-- Solitary CHRPE
-- Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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5) **Iris mammillations**: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

6) **Iris cysts**: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

7) **Nevus**: Common. Benign

8) **Melanocytoma**: Usually juxtapapillary. Malignant transformation extremely rare

9) **Osteoma**: Benign bony tumor, most common in teen years, females. Risk of CNVM

10) **Isolated/focal choroidal hemangioma**: Very rare. Characteristic + scan pattern

11) **Diffuse choroidal hemangioma**: Unilateral. Found in Sturge-Weber syndrome

CHRPE is characterized according to its presentation. In what two ways does it present?

--- Solitary CHRPE

--- Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?

Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

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**Iris/Ciliary Body**

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**A CHRPE-like lesion is associated with a potentially fatal inherited syndrome.** What is the name (both eponymous and noneponymous) of this syndrome?

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What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

--If it is bilateral (regular CHRPE is almost always unilateral)

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**RPE**

1) **Congenital hypertrophy of the RPE (CHRPE)**

**CHRPE is characterized according to its presentation. In what two ways does it present?**

--Solitary CHRPE

--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

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**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?

Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

-- If it is bilateral (regular CHRPE is almost always unilateral )

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RPE

1) **Congenital hypertrophy of the RPE** (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?

-- **Solitary** CHRPE

-- **Multifocal** or **Grouped** CHRPE: Large lesion(s) surrounded by a few smaller ones

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CHRPE-like lesions of Gardner syndrome: Bilateral presentation
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?
Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?
--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are distributed pattern

RPE

1) **Congenital hypertrophy of the RPE (CHRPE)**

CHRPE is characterized according to its presentation. In what two ways does it present?
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6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues is kids slide-set)

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RPE

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CHRPE is characterized according to its presentation. In what two ways does it present?

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What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

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- If the lesions are scattered throughout multiple sectors of the eyes (ie, not ‘grouped’)

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Retina

1) Congenital hypertrophy of the RPE (CHRPE)
CHRPE-like lesions of Gardner syndrome: Scattered distribution
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.

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5) **Iris mammillations:** Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota.

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**Iris/Ciliary Body**

- **Nevus:** Common. Benign.
- **Melanocytoma:** Usually juxtapapillary. Malignant transformation extremely rare.
- **Osteoma:** Benign bony tumor, most common in teen years, females. Risk of CNVM.
- **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-Scan pattern.
- **Diffuse choroidal hemangioma:** Unilateral. Found in Sturge-Weber syndrome.

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**RPE**

1) **Congenital hypertrophy of the RPE (CHRPE):**

CHRPE is characterized according to its presentation. In what two ways does it present?

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- **Solitary**
- **Multifocal** or **Grouped**

CHRPE: Large lesion(s) surrounded by a few smaller ones.
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

1) Congenital hypertrophy of the RPE (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?
--Solitary CHRPE
--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka Gardner syndrome

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?
--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are scattered throughout multiple sectors of the eyes (ie, not ‘grouped’)
--If the shape of the lesions is pisciform
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What does pisciform mean?
\'Fish-shaped\'

1) Congenital hypertrophy of the RPE (CHRPE)
Iris/Ciliary Body

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**What does pisciform mean?** It means ‘fish-shaped’

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1) **Congenital hypertrophy of the RPE (CHRPE)**

CHRPE is characterized according to its presentation. In what two ways does it present?

-- **Solitary** CHRPE
-- **Multifocal** or **Grouped** CHRPE: Large lesion(s) surrounded by a few smaller ones
CHRPE-like lesions of Gardner syndrome: Pisciform shape
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed \( \rightarrow \) hyphema \( \rightarrow \) increased IOP \( \rightarrow \) glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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**Iris/Ciliary Body**

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?

Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

--If it is bilateral (regular CHRPE is almost always unilateral)

--If the lesions are scattered throughout multiple sectors of the eyes (ie, not ‘grouped’)

--If the shape of the lesions is pisciform

The tails of these fish-shaped lesions have two telltale (tell-tail?) characteristics--what are they?

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**Retina**

1) **Nevus**: Common. Benign

2) **Melanocytoma**: Usually juxtapapillary. Malignant transformation extremely rare

3) **Osteoma**: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) **Isolated/focal choroidal hemangioma**: Very rare. Characteristic a-scan pattern

5) **Diffuse choroidal hemangioma**: Unilateral. Found in Sturge-Weber syndrome

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**Intraocular Tumors of Childhood**

1) **Congenital hypertrophy of the RPE (CHRPE)**: Retina

Choroid

Iris/Ciliary Body

CHRPE is characterized according to its presentation. In what two ways does it present?

--Solitary

--Multifocal or Grouped

CHRPE: Large lesion(s) surrounded by a few smaller ones

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?

Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

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The tails of these fish-shaped lesions have two telltale (tell-tail?) characteristics--what are they?
--They are hypopigmented vs hyperpigmented
--They point towards the optic nerve head (hypo- vs hyperpigmented location in eye)

1) **Nevus:** Common. Benign
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4) **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-scan pattern
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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

**CHRPE** is characterized according to its presentation. In what two ways does it present?
--Solitary
--Multifocal or Grouped: CHRPE: Large lesion(s) surrounded by a few smaller ones

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka **Gardner syndrome**

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**Retina**
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**Iris/Ciliary Body**

A **CHRPE-like lesion** is associated with a potentially fatal inherited syndrome. **What is the name (both eponymous and noneponymous) of this syndrome?**

Familial adenomatous polyposis, aka **Gardner syndrome**

**What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?**

-- If it is bilateral (regular CHRPE is almost always unilateral)

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-- If the shape of the lesions is pisciform

**The tails of these fish-shaped lesions have two telltale (tell-tail?) characteristics—what are they?**

-- They are hypopigmented

-- They point towards the optic nerve head

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**Retina**
Intraocular Tumors of Childhood

CHRPE-like lesions of Gardner syndrome: Hypopigmented tail pointing toward ONH
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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**Iris/Ciliary Body**

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of Gardner syndrome increase the likelihood that it is a component of Gardner syndrome?

--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are scattered throughout multiple sectors of the eyes (ie, not 'grouped')
--If the shape of the lesions is pisciform

What is the most clinically important (and ominous) component to Gardner syndrome?

Pts develop thousands of colonic polyps, a significant number of which are malignant

Other than the colonic and RPE lesions, what are the findings in Gardner syndrome?

--Benign tumors of the skin
--Benign tumors of bone
--Dental anomalies

**Retina**

CHRPE is characterized according to its presentation. In what two ways does it present?

--**Solitary** CHRPE
--**Multifocal** or **Grouped** CHRPE: Large lesion(s) surrounded by a few smaller ones
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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka Gardner syndrome

What characteristics of Gardner syndrome increase the likelihood that it is a component of Gardner syndrome?
--If it is bilateral (regular CHRPE is almost always unilateral)
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What is the most clinically important (and ominous) component to Gardner syndrome? Pts develop thousands of colonic polyps, a significant number of which are malignant

CHRPE is characterized according to its presentation. In what two ways does it present?
--Solitary CHRPE
--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones
Gardner syndrome: Colonic polyps
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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6) **Iris cysts:** Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

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**A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?**

Familial adenomatous polyposis, aka **Gardner syndrome**

**What is the most clinically important (and ominous) component to Gardner syndrome?**

Pts develop thousands of **colonic polyps, a significant number of which are malignant**

**What proportion of untreated Gardner syndrome pts will develop colon cancer?**

All of them

By what age will this occur?

Age 40, maybe a little later
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed→hyphema→increased IOP→glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
2) Medulloepithelioma (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed→hyphema→increased IOP→glaucoma. Locally invasive→death. Tx: Enucleate
3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light
4) Brushfield spots: Strong association with Down syndrome
5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

Nevus: Common. Benign
Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare
Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM
Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern
Diffuse choroidal hemangioma: Unilateral. Found in Sturge-Weber syndrome

CHRPE is characterized according to its presentation. In what two ways does it present?
--Solitary CHRPE
--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka Gardner syndrome

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?
--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are scattered throughout multiple sectors of the eyes (ie, not 'grouped')
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What is the treatment of choice? **Prophylactic colectomy**

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Other than the colonic and RPE lesions, what are the findings in Gardner syndrome?

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**When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What are the other two?**

- Muir-Torre syndrome
- Peutz Jeghers syndrome

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

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Retina

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Another is Muir-Torre syndrome:

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When ‘colon cancer + ophthalmic issue’ is mentioned, two syndromes should come to mind. One is Gardner syndrome. What is the other?

One is Gardner syndrome. The other is **Muir-Torre syndrome**

What is the main ophthalmic manifestation of Muir-Torre syndrome?

Multiple sebaceous lesions of (but not necessarily limited to) the eyelids

Does Muir-Torre present with multiple adenomatous polyps of the colon a la Gardner syndrome?

No; Muir-Torre is an example of a disease spectrum called Hereditary Non-Polyposis Colorectal Cancer

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Multiple skin-colored to yellow–pink papules (arrows) on the face of a 64-year-old woman with a history of colon and cervical cancer. A skin biopsy confirmed a diagnosis of sebaceous adenoma resulting from Muir–Torre syndrome.
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**Iris/Ciliary Body**

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**A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?**

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**What sorts of sebaceous lesions?**

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**What sorts of sebaceous lesions?**

--Sebaceous-cell carcinomas
--Sebaceous-cell adenomas
--Basal-cell carcinomas with sebaceous differentiation

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Intraocular Tumors of Childhood

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2) Medulloepithelioma (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate
3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light
4) Brushfield spots: Strong association with Down syndrome
5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

1) Nevus: Common. Benign
2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare
3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM
4) Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka Gardner syndrome

What is the main ophthalmic manifestation of Muir-Torre syndrome?
Multiple sebaceous lesions of (but not necessarily limited to) the eyelids.

Does Muir-Torre present with multiple adenomatous polyps of the colon a la Gardner syndrome?
No; Muir-Torre is an example of a disease spectrum called Hereditary Non-Polyposis Colorectal Cancer
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka **Gardner syndrome**

- If it is bilateral (regular CHRPE is almost always unilateral)
- If the lesions are scattered throughout multiple sectors of the eyes (ie, not ‘grouped’)
- If the shape of the lesions is pisciform

When ‘colon cancer + ophthalmic issue’ is mentioned, two syndromes should come to mind. One is Gardner syndrome. What is the other? **Muir-Torre syndrome**

- Multiple sebaceous lesions of (but not necessarily limited to) the eyelids

Does Muir-Torre present with multiple adenomatous polyps of the colon a la Gardner syndrome? No; Muir-Torre is an example of a disease spectrum called Hereditary **Non-Polyposis Colorectal Cancer**

RPE

1) **Congenital hypertrophy of the RPE (CHRPE)**

CHRPE is characterized according to its presentation. In what two ways does it present?

- **Solitary** CHRPE
- **Multifocal** or **Grouped** CHRPE: Large lesion(s) surrounded by a few smaller ones

Retina

- **Congenital hypertrophy of the RPE (CHRPE)**
- **Melanocytoma**: Usually juxtapapillary. Malignant transformation extremely rare

- **Osteoma**: Benign bony tumor, most common in teen years, females. Risk of CNVM

- **Isolated/focal choroidal hemangioma**: Very rare. Characteristic a-scan pattern

- **Diffuse choroidal hemangioma**: Unilateral. Found in Sturge-Weber syndrome
Intraocular Tumors of Childhood

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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?

Familial adenomatous polyposis, aka **Gardner syndrome**

When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other?

Muir-Torre syndrome and **Peutz-Jeghers syndrome**

How is Peutz-Jeghers pronounced?

Pyoots jeh-grz

RPE

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Intraocular Tumors of Childhood

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**RPE**

1) **Congenital hypertrophy of the RPE (CHRPE)**

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are scattered throughout multiple sectors of the eyes (ie, not “grouped”)
--If the shape of the lesions is pisciform

When ‘colon cancer + ophtalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? **Muir-Torre syndrome** and **Peutz-Jeghers syndrome**

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**Retina**

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**Iris/Ciliary Body**

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka **Gardner syndrome**

When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? Muir-Torre syndrome and Peutz-Jeghers syndrome.

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines of (but not necessarily limited to) the eyelids

Does Peutz-Jeghers present with multiple adenomatous polyps of the colon a la Gardner syndrome? Yes

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**Retina**

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**Congenital hypertrophy of the RPE (CHRPE):**

- In what two ways does it present? --Solitary --Multifocal or Grouped

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name of this syndrome? **Gardner syndrome**

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Muir-Torre syndrome and Peutz-Jeghers syndrome

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?

Simple lentigines of (but not necessarily limited to) the eyelids

When a CHRPE-like lesion is present?

--Congenital CHRPE

--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines of (but not necessarily limited to) the eyelids

Does Peutz-Jeghers present with multiple adenomatous polyps of the colon a la Gardner syndrome? Yes

What are simple lentigines? Flat melanocytic lesions histologically similar to ephelides. By what variant of the term ‘simple lentigines’ are they also known? ‘Lentigo simplex’

Does lentigo simplex have malignant potential? No

RPE
Iris/Ciliary Body

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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. **What is the name (both eponymous and noneponymous) of this syndrome?**

**Familial adenomatous polyposis, aka Gardner syndrome**

When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. **What is the other?**

- **Muir-Torre syndrome** and Peutz-Jeghers syndrome

**What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?**

- Simple lentigines

**What are simple lentigines?**

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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Muir-Torre syndrome and Peutz-Jeghers syndrome are the others.

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**What are simple lentigines?**
Flat melanocytic lesions histologically similar to ephelides

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What are ephelides (singular, ephelis)?

Intraocular Tumors of Childhood

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Iris/Ciliary Body

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Familial adenomatous polyposis, aka Gardner syndrome

What are simple lentigines?
Flat melanocytic lesions histologically similar to ephelides

What are ephelides (singular, ephelis)? Freckles
Intraocular Tumors of Childhood

Peutz-Jeghers syndrome: Eyelid simple lentigines
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Intraocular Tumors of Childhood

Iris/Ciliary Body

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Muir-Torre syndrome and Peutz-Jeghers syndrome

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?

Simple lentigines of (but not necessarily limited to) the eyelids

Does Peutz-Jehgers present with multiple adenomatous polyps of the colon a la Gardner syndrome?

Yes

What are simple lentigines?

Flat melanocytic lesions histologically similar to ephelides

By what variant of the term ‘simple lentigines’ are they also known?

‘Lentigo simplex’
1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed→hyphema→increased IOP→glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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**Iris/Ciliary Body**

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**Retina**

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**Simple lentigines**
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**Iris/Ciliary Body**

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. **What is the name (both eponymous and noneponymous) of this syndrome?**

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When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other?

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**What are simple lentigines?**

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**Does lentigo simplex have malignant potential?**

No
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Intraocular Tumors of Childhood

Iris/Ciliary Body

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What are simple lentigines? Flat melanocytic lesions histologically similar to ephelides

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Iris/Ciliary Body

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When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other?

- Muir-Torre syndrome and
- Peutz-Jeghers syndrome

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?

- Simple lentigines

Simple lentigines: Flat melanocytic lesions histologically similar to ephelides

By what variant of the term ‘simple lentigines’ are they also known?

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Does lentigo simplex have malignant potential?

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I coulda sworn lentigo simplex had malignant potential. You sure about this?

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Intraocular Tumors of Childhood

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Familial adenomatous polyposis, aka **Gardner syndrome**

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When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other?

Muir-Torre syndrome and **Peutz-Jeghers syndrome**

What are simple lentigines?

Flat melanocytic lesions histologically similar to ephelides

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‘Lentigo simplex’

**Does lentigo simplex have malignant potential?**

No

I coulda sworn lentigo simplex had malignant potential. You sure about this?

Yes, I’m sure. You’re thinking of ‘Lentigo maligna’, a pre-malignant melanocytic lesion of the skin.
**Intraocular Tumors of Childhood**

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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2) **Medulloepithelioma** (aka *diktyoma*): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed → hyphema → increased IOP → glaucoma. Locally invasive → death. Tx: Enucleate

3) **Lisch nodules**: Strong association with NF1. Lighter on dark irides; darker on light

4) **Brushfield spots**: Strong association with Down syndrome

5) **Iris mammillations**: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka *Gardner syndrome*

When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? *Muir-Torre syndrome* and *Peutz-Jeghers syndrome*

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? **Simple lentigines** of (but not necessarily limited to) the eyelids

Are lentigo simplex eyelid lesions the classic harbinger of Peutz-Jeghers syndrome? Yes

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1) **Nevus**: Common. Benign

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3) **Osteoma**: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) **Isolated/focal choroidal hemangioma**: Very rare. Characteristic α-scan pattern

5) **Diffuse choroidal hemangioma**: Unilateral. Found in Sturge-Weber syndrome

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RPE
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

CHRPE is characterized according to its presentation. In what two ways does it present?

--Solitary CHRPE

--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka Gardner syndrome

When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other?

Muir-Torre syndrome and Peutz-Jeghers syndrome

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?

Simple lentigines of (but not necessarily limited to) the eyelids

Are lentigo simplex eyelid lesions the classic harbinger of Peutz-Jeghers syndrome?

No, pigmented lesions of the perioral region are the classic/most common finding

- Retinal

- Ciliary

- Uveal

- Conjunctival

- Scleral

- Conjunctival, episcleral

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**Are a CHRPE-like lesion and the Retina**

--Solitary CHRPE

--Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones
Intraocular Tumors of Childhood

Characteristic circumoral pigmentation in a patient with Peutz-Jeghers syndrome
Intraocular Tumors of Childhood

Speaking of: Did you notice the pigmented lip lesions in this pic?
Iris/Ciliary Body

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What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines of (but not necessarily limited to) the eyelids

Does Peutz-Jehgers present with multiple adenomatous polyps of the colon a la Gardner syndrome? Yes
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**Retina**

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**Congenital hypertrophy of the RPE (CHRPE):**

- In what two ways does it present?
  - Solitary
  - Multifocal or Grouped

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? **Gardner syndrome**

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Intraocular Tumors of Childhood

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Choroid

1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

3) Osteoma: Benign bony tumor, most common in teen years, females. Risk of CNVM

4) Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern


RPE

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Retina

1) ?
Intraocular Tumors of Childhood

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**Retina**

1) **Retinoblastoma** (see the slide-set dedicated to it)
Intraocular Tumors of Childhood

**Iris/Ciliary Body**

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**RPE**

1. **Congenital hypertrophy of the RPE (CHRPE)**: Common. Deeply pigmented. A CHRPE-like finding is associated with Gardner syndrome

**Retina**

1. **Combined hamartoma of the retina and RPE**

**1) Retinoblastoma** (see the slide-set dedicated to it)
Intraocular Tumors of Childhood

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What is a hamartoma?

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1) **Retinoblastoma:** (see the slide-set dedicated to it)

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**What is a hamartoma?**

A tumor composed of histologically normal cells found in their normal location

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Intraocular Tumors of Childhood

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**Retina**

1) **Combined hamartoma of the retina and RPE**

   - **RPE**
   - **Retina**

   - 1) **Congenital hypertrophy of the RPE (CHRPE):** Common. Deeply pigmented. A CHRPE-like finding is associated with Gardner syndrome

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**What is a hamartoma?**

A tumor composed of histologically abnormal cells found in their normal location
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Retinoblastoma (see the slide-set dedicated to it)

What is a hamartoma? A tumor composed of histologically normal cells found in an abnormal location?

What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?

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Intraocular Tumors of Childhood

1) Congenital hypertrophy of the RPE (CHRPE): Common. Deeply pigmented. A CHRPE-like finding is associated with Gardner syndrome

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**Intraocular Tumors of Childhood**

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**What is a hamartoma?**

A tumor composed of histologically abnormal cells found in their normal location

**What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?**

A choristoma

---

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Iris/Ciliary Body

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2) Combined hamartoma of the retina and RPE

3) Retinoblastoma (see the slide-set dedicated to it)

What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location? A choristoma

What is a hamartoma? A tumor composed of histologically normal cells found in an abnormal location

That a lesion is a hamartoma (or choristoma) indicates what about its onset?

That a lesion is a hamartoma (or choristoma) indicates what about its status vis-a-vis malignancy?

That it is congenital

That it is benign
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**Intraocular Tumors of Childhood**

**What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?**

A choristoma

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**What is a hamartoma?**

A tumor composed of histologically normal cells found in their normal location

---

*That a lesion is a hamartoma (or choristoma) indicates what about its onset?*

That it is congenital

---

**RPE**

1) Congenital hypertrophy of the RPE (CHRPE): Common. Deeply pigmented. A CHRPE-like finding is associated with Gardner syndrome

**Retina**

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Retinal Tumors

1) Combined hamartoma of the retina and RPE

RPE

Retina

1) Retinoblastoma (see the slide-set dedicated to it)

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What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?
A choristoma

What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

That a lesion is a hamartoma (or choristoma) indicates what about its onset?
That it is congenital

That a lesion is a hamartoma (or choristoma) indicates what about its status vis a vis malignancy?
That it is benign
Intraocular Tumors of Childhood

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RPE

Retina

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**Iris/Ciliary Body**

**Choroid**

**Intraocular Tumors of Childhood**

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Combined hamartoma of retina and RPE
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**With what more sinister dz entity is it often confused?**

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Choroidal melanoma--eyes have been enucleated because of this misdiagnosis

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Iris/Ciliary Body

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**RPE**

<table>
<thead>
<tr>
<th>Retina</th>
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**Choroidal melanoma**

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**How can one avoid making such a disastrous mistake?**
By taking pains to carefully determine the anatomic location of the tumor in question—choroidal melanomas originate behind Bruch’s membrane, whereas combined hamartomas of the retina and RPE are located wholly in front of it

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1) Combined hamartoma of the retina and RPE: Benign, congenital retinal lesion

1) Retinoblastoma (see the slide-set dedicated to it)

No question—summary/review slide