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

1) ?

2) ?

Iris/Ciliary Body

3) ?

4) ?

5) ?

6) ?

Choroid

RPE

Retina

Six tumors of the iris/ciliary body
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma
2) Medulloepithelioma
3) Lisch nodules
4) Brushfield spots
5) Iris mammillations
6) Iris cysts

Six tumors of the iris/ciliary body

Choroid
RPE
Retina
1) **Juvenile xanthogranuloma**

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

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**Iris/Ciliary Body**

1. **Juvenile xanthogranuloma**: Nonneoplastic histiocytic proliferation. Present 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.


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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In three words, what sort of condition is JXG?  
It is a…
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**

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

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

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Iris/Ciliary Body

Choroid

RPE

Retina
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)*

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1) **Juvenile xanthogranuloma**

- Nonneoplastic histiocytic proliferation
- Present before 2 years old
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Self-limited; regresses by age 5
- 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)**
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)*
As orangish skin papules

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

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

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

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

JXG: Skin papules. The orangish color is classic
**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

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**Iris/Ciliary Body**

**Choroid**

**RPE**

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Intraocular Tumors of Childhood

Iris/Ciliary Body

Choroid

RPE

Retina
Iris/Ciliary Body

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**Intraocular Tumors of Childhood**

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Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

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- 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

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Unilateral

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Iris/Ciliary Body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

JXG: Iris lesion
Intraocular Tumors of Childhood

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*In what three ways are the iris nodules clinically significant?*

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Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**

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Iris/Ciliary Body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

JXG: Spontaneous hyphema
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*What is the natural history of JXG?*

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

1) **Juvenile xanthogranuloma**

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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

Choroid

RPE

Retina
Intraocular Tumors of Childhood

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Iris/Ciliary Body

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RPE

Retina
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
The presence of *‘foamy macrophages’*

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Intraocular Tumors of Childhood

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The presence of **eponym**

*Unilateral?*

*When*

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Iris/Ciliary Body

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When Unilateral

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Choroid

RPE

Retina
Intraocular Tumors of Childhood

Touton giant cells

Foamy macrophages

JXG
Intraocular Tumors of Childhood

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

*RPE*

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*Retina*

*Choroid*

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**Intraocular Tumors of Childhood**

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*Retina*
Intraocular Tumors of Childhood

Iris/Ciliary Body

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*This histology—‘foamy macrophages’--is often described with other, equivalent terms. What are they?*  
Foamy = ‘lipid filled’  
Macrophages = ‘histiocytes’

*What is the natural history of JXG?*  
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Choroid

RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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*What are the two hallmarks of JXG histology?*

This histology—‘foamy macrophages’—is often described with other, equivalent terms. What are they?
Foamy = ‘lipid filled’
Macrophages = ‘histiocytes’

*If enough nodules are present, heterochromia iridis will result*

*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?**
It is a...\textit{nonneoplastic histiocytic proliferation}

**How does JXG usually present? (Hint: It's not ophthalmic)**
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**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?**
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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? First clue--more forthcoming
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma

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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? Clue #2
Intraocular Tumors of Childhood

Iris/Ciliary Body

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*Unilateral*

*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?
Associated with chronic diarrhea? Need another?

Choroid

Retina
1) **Juvenile xanthogranuloma**

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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?  
Associated with chronic diarrhea?  
And CNS symptoms--seizures, dementia, coma?  

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Last chance--answer is next!
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'

**When is the presence of JXG unilateral or bilateral?**
Unilateral

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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?*
*Associated with chronic diarrhea?*
*And **CNS symptoms**--seizures, dementia, coma?*

**Whipple’s disease**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

*What are the two hallmarks of JXG histology?*  
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*When unilateral?*  
Unilateral

*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?  
Associated with chronic diarrhea?  
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**Whipple’s disease**

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

Retina

Choroid

Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**

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*When Unilateral?*

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Associated with chronic diarrhea?
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**Whipple’s disease**

**Broadly speaking, what sort of condition is Whipple’s?**
It is infectious
1) Juvenile xanthogranuloma

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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?*  
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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?*  
The bacterium Tropheryma whipplei
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**

- **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 old

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

- **When unilateral?**

- **Speaking of ‘foamy macrophages’…**

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

- **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?**

- **Associated with chronic migratory arthritis?**
  - Yes

- **And CNS symptoms—seizures, dementia, coma?**

- **Choroid**

- **Retina**
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**

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

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

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---

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.

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Can be pupillary, stromal, secondary.

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---
Whipple’s disease: Duodenal biopsy, high mag. The image shows the characteristic feature of foamy macrophages in the lamina propria.
Intraocular Tumors of Childhood

Iris/Ciliary Body

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*What are the two hallmarks of JXG histology?*
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*When unilateral?*
‘foamy macrophages’

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

*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 middle-aged white guy with… a hx of chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms—seizures, dementia, coma?*
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---What other finding will a duodenal biopsy reveal?---The presence of bacteria within macrophages located in intestinal villi---stain

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

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Intraocular Tumors of Childhood

Small-intestine biopsy stained with periodic acid-Schiff. Note the numerous macrophages in the lamina propria (arrows).
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 young child with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions?*

Pic forthcoming

*When? Unilateral*

*The presence of Touton giant cells*

*The presence of ‘foamy macrophages’*

*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

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

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

Condition?
Intraocular Tumors of Childhood

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*Speaking of foamy macrophages part deaux…*
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**Adult with bilateral upper-lid yellow lesions?**

**Xanthelasma**

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Intraocular Tumors of Childhood

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**Xanthelsasma**

The presence of Touton giant cells
'foamy macrophages'

When unilateral? When bilateral?

Are xanthelasmas a harbinger of elevated serum lipids?

The presence of Touton giant cells

What is the natural history of JXG?
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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.

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

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Can be pupillary, stromal, secondary.
Intraocular Tumors of Childhood

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

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

*Pic forthcoming*

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2) Medulloepithelioma

Iris/Ciliary Body

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

Choroid

RPE

Retina

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

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2) **Medulloepithelioma**

**Iris/Ciliary Body**

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

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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?*

**Choroid**

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Intraocular Tumors of Childhood

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*How does it present?*

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*How does it present?*
As an iris mass

Very rare

Benign, but very aggressive locally

Enucleation is usually required
Intraocular Tumors of Childhood

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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:

- Sectoral cataract

Is it common, or rare? Very rare

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

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Iris/Ciliary Body
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  - Very rare

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

Retina
Intraocular Tumors of Childhood

Medulloepithelioma/diktyoma
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

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.
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2) **Medulloepithelioma**

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Iris/Ciliary Body

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Choroid

RPE

Retina

Intraocular Tumors of Childhood

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### Intraocular Tumors of Childhood

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**RPE**

**Retina**

How aggressive is ‘very aggressive’? Aggressive enough to result in death
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Enucleation is usually required
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3) **Lisch nodules**

4) **Brushfield spots**

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

**NF1**

Is it associated with NF2? 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
Intraocular Tumors of Childhood

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In this context, what does NF1 stand for?

Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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**In this context, what does NF1 stand for?**

Neurofibromatosis type 1

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**Intraocular Tumors of Childhood**

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**Iris/Ciliary Body**

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**Retina**
Intraocular Tumors of Childhood

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In this context, what does NF1 stand for?

Neurofibromatosis type 1

What is the eponymous name for NF1?

NF1

Retina
Intraocular Tumors of Childhood

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NF1

In this context, what does NF1 stand for?
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What is the eponymous name for NF1?
von Recklinghausen’s disease

Retina
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Retina

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*In a word, what sort of condition is it?*
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### Iris/Ciliary Body

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A phakomatosis

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Intraocular Tumors of Childhood

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

Phakomatoses are known also as what sort of syndrome? Neurocutaneous syndromes

Lisch nodules are most strongly associated with what congenital condition? NF1
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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In general terms, how do phakomatoses present?
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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**Are Lisch nodules associated with NF2?**
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What is the prevalence of Lisch nodules in NF1?

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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Are Lisch nodules clinically significant?

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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

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**Intraocular Tumors of Childhood**

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

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

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

**Melanocytes and neuroglial cells**

**Melanocytic lesions**

- ?
- ?
- ?
- ?

**Neuroglial lesions**

- ?
- ?
- ?
- ?

**Give four classic examples of each:**

(YMMV of course)

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

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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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Retina
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

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

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 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

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

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Intraocular Tumors of Childhood

Retina

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

- Melanocytes
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Lisch nodules are most strongly associated with what congenital condition?

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What is the prevalence of Lisch nodules in NF1?

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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)?

- Melanocytic lesions are of no clinical significance beyond establishing the diagnosis,
- Neuroglial lesions are associated with significant ocular and/or systemic morbidity

Are Lisch nodules clinically significant?

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

Give four classic examples of each:

- 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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**Iris/Ciliary Body**

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**Choroid**

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**Intraocular Tumors of Childhood**

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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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**Melanocytic lesions**

- Café au lait spots
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- Choroidal lesions

**Neuroglial lesions**

- Nodular neurofibromas
- Plexiform neurofibromas
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- Prominent corneal nerves

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**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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**Melanocytic lesions**

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- NF1

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**Retina**

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

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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?**

*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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Intraocular Tumors of Childhood

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4) **Brushfield spots**

5) **Iris mammillations**

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

- Down syndrome
- At least 90%
- 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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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%
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Retina

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**Iris/Ciliary Body**

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

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**What is the clinical significance of Brushfield spots?**
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**Retina**

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Mammillations? Aren’t those a CNS thingamajig?

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Retina
Intraocular Tumors of Childhood

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

Intraocular Tumors of Childhood
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Intraocular Tumors of Childhood

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**With what phakomatosis are they associated?**
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NF1 (albeit not nearly as strongly as Lisch nodules)
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Mammillations? Aren’t those a CNS thingamajig?
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OK, then what are iris mammillations?
Tiny pigmented iris nodules

‘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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**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.

‘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)
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

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?
Intraocular Tumors of Childhood

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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 three words
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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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/Ciliary Body

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)
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)

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Choroid

RPE

Retina
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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Choroid

Five tumors of the choroid

1) ?
2) ?
3) ?
4) ?
5) ?
**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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**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**

**RPE**

**Retina**
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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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?
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**

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.

Retina
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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**Choroid**

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

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

Iris/Ciliary Body

Choroid

Five tumors of the choroid

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

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

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**Nevus:** Common. Benign

2) Melanocytoma

3) Osteoma

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma
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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Iris/Ciliary Body

Melanocytoma is a variant of what common choroidal finding?

Choroid

1) Nevus: Common. Benign
2) Melanocytoma
3) Osteoma
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma

RPE

Retina
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**Iris/Ciliary Body**

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

It is a particular sort of **choroidal nevus**
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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6) **Iris cysts:** Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

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**Iris/Ciliary Body**

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

*It is a particular sort of choroidal nevus*

*From what structure does it commonly arise?*

**Choroid**

1) **Nevus:** Common. Benign

2) **Melanocytoma**

3) **Osteoma**

4) **Isolated/focal choroidal hemangioma**

5) **Diffuse choroidal hemangioma**

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**RPE**

**Retina**
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

**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

**Choroid**

1) Nevus: Common. Benign

2) **Melanocytoma**

3) Osteoma

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

**RPE**

**Retina**

Melanocytoma is a variant of what common choroidal finding?

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The optic disc
Intraocular Tumors of Childhood

Melanocytoma
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

1. **Nevus:** Common. Benign

2. **Melanocytoma**

3. **Osteoma**

4. **Isolated/focal choroidal hemangioma**

5. **Diffuse choroidal hemangioma**

**Choroid**

1. **Nevus:** Benign. Can be isolated/focal or diffuse

2. **Melanocytoma**

3. **Osteoma**

4. **Isolated/focal choroidal hemangioma**

5. **Diffuse choroidal hemangioma**

**RPE**

**Retina**

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**Melanocytoma**

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

- 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
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

- **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

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**Choroid**

1) **Nevus:** Common. Benign
2) **Melanocytoma**
3) **Osteoma**
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**RPE**

**Retina**
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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?
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**

**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
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

**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? *Yes*

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**Choroid**

1) **Nevus**: Common. Benign
2) **Melanocytoma**
3) **Osteoma**
4) **Isolated/focal choroidal hemangioma**
5) **Diffuse choroidal hemangioma**

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**RPE**

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**Retina**
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

1) **Nevus:** Common. Benign

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

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
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Is there a racial predilection?
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Choroid

1) Nevus: Common. Benign
2) Melanocytoma
3) Osteoma
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma

RPE

Retina
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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**Choroid**

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Does melanocytoma have the potential to undergo malignant transformation?
Intraocular Tumors of Childhood

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**Melanocytoma**

2) **Melanocytoma**

3) **Osteoma**

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**Choroid**

**RPE**

**Retina**
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Approximately what percent of cases will transform?
Intraocular Tumors of Childhood

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3) Lisch nodules: Stark contrast to JXG and Medulloepithelioma; FDG-PET F-18 uptake.
4) Brushfield spots: Hyperpigmented macules in the iris. Can be associated with cataracts, Down syndrome, and other syndromes.
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Approximately what percent of cases will transform?
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Nevus: Common. Benign

Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

Osteoma: Bone. Very rare. Benign. Yes, it is more common in females. Teens. If significant vision loss occurs, choroidal neovascular membrane is usually the culprit

Choroidal osteoma

Choroid

Osteoma

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

RPE

Retina
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Diffuse choroidal hemangioma

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

Intraocular Tumors of Childhood

Retina

RPE

Choroid

Iris/Ciliary Body

Intraocular Tumors of Childhood

Retina

RPE

Choroid

Iris/Ciliary Body
Intraocular Tumors of Childhood

Osteoma
Intraocular Tumors of Childhood

Osteoma: FP, and b-scan demonstrating

buzzword describing a b-scan finding illustrated above
Osteoma: FP, and \( b \)-scan demonstrating ‘shadowing’
Intraocular Tumors of Childhood

Osteoma: Another example
Intraocular Tumors of Childhood

Osteomas (same pt, different cuts).
Note how bright the lesions are
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In a word, what is a choroidal osteoma composed of?
Bone

*Is it common or rare?*
Very rare

---

**Intraocular Tumors of Childhood**

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**Choroid**

3) **Osteoma**

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

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**RPE**

**Retina**
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

Intraocular Tumors of Childhood

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**Intraocular Tumors of Childhood**

**Bone**

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Yes, it is more common in females

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Teens

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

Choroidal neovascular membrane

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**Choroidal Osteoma**

- Bone
- Common
- Benign
- Extremely rare
- More typically found in teens
- More likely to result in choroidal neovascular membrane

**Isolated/focal choroidal hemangioma**

**Diffuse choroidal hemangioma**
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**Intraocular Tumors of Childhood**

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---

**Iris/Ciliary Body**

**Choroid**

3) Osteoma

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

---

**Retina**

**RPE**
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Intraocular Tumors of Childhood

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Retina

RPE

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Intraocular Tumors of Childhood

Osteoma with CNVM in a 13 y.o. female
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Very rare

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Teens

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)

Intraocular Tumors of Childhood

Retina

Choroid

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Intraocular Tumors of Childhood

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Retina

RPE

Choroid

3) Osteoma
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma
Intraocular Tumors of Childhood

By what other name is this lesion known?

Iris/Ciliary Body

Choroid

RPE

Retina

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Iris/Ciliary Body

Choroid

RPE

Retina

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

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

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

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

Iris/Ciliary Body

Choroid

Retina

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 -scan ultrasonography? It is one of 'high internal reflectivity'

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**Iris/Ciliary Body**

**Choroid**

By what other name is this lesion known?

**Circumscribed choroidal hemangioma**

Is it common, or rare?

Rare

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**Iris/Ciliary Body**

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**Retina**

**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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## Intraocular Tumors of Childhood

### Iris/Ciliary Body

<table>
<thead>
<tr>
<th>Question</th>
<th>Answer</th>
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</thead>
<tbody>
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**Choroid**

- **Circumscribed 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

- **Isolated/focal choroidal hemangioma**

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---

**RPE**

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Rare

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

'High internal reflectivity' is also described for:
- Circumscribed choroidal hemangioma
- Choroidal nevus
- Isolated/focal choroidal hemangioma
- Diffuse choroidal hemangioma

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**RPE**

**Retina**
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**Choroid**

By what other name is this lesion known?

**Circumscribed choroidal hemangioma**

Is it common, or rare?

Rare

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No

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

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

Sturge-Weber syndrome (SWS)

---

With what condition is the diffuse choroidal hemangioma associated?

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

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

Intraocular Tumors of Childhood

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

RPE

Retina

Intraocular Tumors of Childhood

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Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Presents in about 50% of patients. The fundus appearance is described as 'tomato catsup fundus.' Can be bilateral. Does not have malignant potential.

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

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5) Diffuse choroidal hemangioma: In a word, what sort of condition is the diffuse choroidal hemangioma associated with? Sturge-Weber syndrome (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)
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Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

What is the hallmark skin finding in SWS?
The port-wine stain
Intraocular Tumors of Childhood

Sturge-Weber: Port-wine stain
Intraocular Tumors of Childhood

**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 in 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:** Sturge-Weber syndrome (SWS)
In a word, what sort of condition is SWS?
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Sturge-Weber syndrome (SWS)
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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. Fundus appearance resembles a “tomato catsup fundus.” Can be present bilaterally. No malignant potential. In a word, what condition is SWS? A phakomatosis

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What is the hallmark skin finding in SWS?
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In one word, what sort of lesion is the port-wine stain?
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When does it present?
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In one word, what sort of lesion is the port-wine stain? An angioma

When does it present? At birth

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

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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
Sturge-Weber syndrome (SWS)

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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?

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

The coloration is a very red--much more so than an unaffected fundus
'Tomato catsup fundus'

Yes, but it's uncommon

No

A phakomatosis

Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

The port-wine stain

An angioma

At birth

What is the typical pattern of distribution?
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.


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

Brushfield spots: Strong association with Down syndrome.

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

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Nevus: Common. Benign.

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

Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Present in about 50% of cases. The fundus look is a very red--much more so than an unaffected fundus. This is referred to as 'tomato catsup fundus'. The choroidal hemangioma can be present bilaterally, but it's uncommon. It does not have malignant potential.


Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis).

Port-wine stain: The hallmark skin finding in SWS. An angioma.

At birth.

The coloration is a very red--much more so than an unaffected fundus. This is referred to as 'tomato catsup fundus'. It comports to the distribution of one or more divisions of CN5.
**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?**
Intracocular Tumors of Childhood

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

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

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

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   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?
Intraocular Tumors of Childhood

With what condition is the diffuse choroidal hemangioma associated?

Sturge-Weber syndrome (SWS)

In a word, what sort of condition is SWS?
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At birth

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**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?*

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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

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

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

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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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**Iris/Ciliary Body
**

**Choroid
**

**RPE
**

**Retina
**

---

**Intracocular 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

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**5) Diffuse choroidal hemangioma**
**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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**Intracocular 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

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

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**Iris/Ciliary Body**

**Choroid**

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**Retina**
Intraocular Tumors of Childhood

Sturge-Weber: Tomato catsup fundus OD
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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?*
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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'

---

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**
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1) 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

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Iris/Ciliary Body

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

---

5) **Diffuse choroidal hemangioma**
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**

**Intraocular Tumors of Childhood**

**Retina**

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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

**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?**

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**Can the choroidal hemangioma be present bilaterally?**

Yes, but it’s uncommon

---

**Diffuse choroidal hemangioma**

- Associated with Sturge-Weber syndrome (SWS) and some cases of isolated choroidal hemangioma
- Very rare
- Characteristic a-scan pattern
- Associated with SWS: About half of patients
- Fundus appearance: Very red, often described as ‘tomato catsup fundus’
- Can be bilateral
- No malignant potential

- Associated with isolated choroidal hemangioma: Very rare
- Associated with Sturge-Weber syndrome (SWS) and isolated choroidal hemangioma
- Very rare
- Characteristic a-scan pattern
- Associated with SWS: About half of patients
- Fundus appearance: Very red, often described as ‘tomato catsup fundus’
- Can be bilateral
- No malignant potential
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)

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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

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

5) **Diffuse choroidal hemangioma**
**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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1. **Nevus:** Common. Benign
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**RPE**

1. ?

**Retina**
# Intraocular Tumors of Childhood

**Iris/Ciliary Body**

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**RPE**

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

**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Retina

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

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

- Flat, mainly black lesion(s) ranging in size from a 1 mm up to ~10
- Common
- It is neither a hamartoma nor a choristoma
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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Choroid

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RPE

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Retina
Intraocular Tumors of Childhood

CHRPE
Intraocular Tumors of Childhood

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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?

RPE
1) Congenital hypertrophy of the RPE (CHRPE)

Retina
Intraocular Tumors of Childhood

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Is it common, or rare?
Common

Ch

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Retina
Iris/Ciliary Body

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Retina

**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?**

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Intraocular Tumors of Childhood
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**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

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

**RPE**

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Intraocular Tumors of Childhood

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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
Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

Solitary

Grouped

CHRPE
Intraocular Tumors of Childhood

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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)**

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**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

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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
Intraocular Tumors of Childhood

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?**

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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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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Intraocular Tumors of Childhood

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A CHRPE-like lesion is:
(both eponymous and non-eponymous)
Familial adenomatous polyposis, aka Gardner syndrome

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

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!
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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?**

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### RPE

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Intraocular Tumors of Childhood

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**RPE**

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

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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 bi- v unilateral (regular CHRPE is almost always bi- v unilateral)
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Intraocular Tumors of Childhood

CHRPE-like lesions of Gardner syndrome: Bilateral presentation
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 (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

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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 adenomatosus 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 distribution pattern

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

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--If the lesions are scattered throughout multiple sectors of the eyes (ie, not ‘grouped’)}
CHRPE-like lesions of Gardner syndrome: Scattered distribution
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What does pisciform mean?
'Fish-shaped'

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CHRPE-like lesions of Gardner syndrome: Pisciform shape
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**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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**Iris/Ciliary Body**

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

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

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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 location in eye

Retina

1) Congenital hypertrophy of the 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

2) Neuropigmentary changes (aka Brushfield spots) are associated with what syndrome?
Down syndrome

3) A choroidal mass (aka isolated/focal choroidal hemangioma) is characterized by what characteristic a-scan pattern?

4) Diffuse choroidal hemangioma is found in what syndrome?
Sturge-Weber syndrome
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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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CHRPE-like lesions of Gardner syndrome: Hypopigmented tail pointing toward ONH
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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

Other than the colonic and RPE lesions, what are the findings in Gardner syndrome?
--Benign tumors of skin
--Benign tumors of bone
--Dental anomalies

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

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Gardner syndrome: Colonic polyps
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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

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

**What is the treatment of choice?**

Prophylactic colectomy

**By what age will this occur?**

40, maybe a little later

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

--- **Solitary** CHRPE
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Intraocular Tumors of Childhood

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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?**

- **Muir-Torre syndrome**

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### Retina

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

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RPE

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**Isolated/focal choroidal hemangioma**

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**Intraocular Tumors of Childhood**

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Muir-Torre syndrome

How is Muir-Torre pronounced? mure (rhymes with ‘pure’) tore-AY

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Retina

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What is the main ophthalmic manifestation of Muir-Torre syndrome? Multiple sebaceous lesions of (but not necessarily limited to) the eyelids

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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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1) **Congenital hypertrophy of the RPE (CHRPE)**
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.
**Intraocular Tumors of Childhood**

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What sorts of sebaceous lesions?

------Basal-cell carcinomas with sebaceous differentiation

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--Sebaceous-cell adenomas
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**Intraocular Tumors of Childhood**

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Intraocular Tumors of Childhood

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RPE

Intraocular Tumors of Childhood

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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) ?
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

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Iris/Ciliary Body

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**What is a hamartoma?**
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What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location?

**What is a choristoma?**
A normal cell found in an abnormal location...

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That a lesion is a hamartoma (or choristoma) indicates what about its onset?
That it is congenital

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**So, what combination of hamartomatous cells of the retina and RPE are involved in a combined hamartoma of the retina and RPE?**

RPE cells (duh) and retinal glial cells

**How does it present clinically?**

As a variably pigmented, slightly elevated retinal mass of the peripapillary retina

**With what more sinister dz entity is it often confused?**

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**Retina**

1) Combined hamartoma of the retina and RPE

1) Retinoblastoma (see the slide-set dedicated to it)
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: 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 \( \alpha \)-scan pattern


1) Retinoblastoma (see the slide-set dedicated to it)

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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

2) **Combined hamartoma of the retina and RPE**

3) **Retinoblastoma** (see the slide-set dedicated to it)

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Choroidal melanoma--eyes have been enucleated because of this misdiagnosis
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Iris/Ciliary Body

Choroid

RPE

Retina

What is a hamartoma?
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So, what combination of hamartomatous cells of the retina and RPE are involved in a combined hamartoma of the retina and RPE?

How can one avoid making such a disastrous mistake?

With what more sinister dz entity is it often confused?

**Choroidal melanoma**

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Iris/Ciliary Body

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RPE

Retina

1) Combined hamartoma of the retina and RPE

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

With what more sinister dz entity is it often confused?
Choroidal melanoma—eyes have been enucleated because of this misdiagnosis

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1) Retinoblastoma (see the slide-set dedicated to it)
Combined hamartoma of retina and RPE. Note the entire lesion is above Bruchs
Intraocular Tumors of Childhood

Iris/Ciliary Body

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4) **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-scan pattern
5) **Diffuse choroidal hemangioma:** Unilateral. Found in Sturge-Weber syndrome

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:** Benign, congenital retinal lesion

No question—summary/review slide