Intraocular Tumors of Childhood

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

Four intraocular locations (i.e., structures/tissues)

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

Iris/Ciliary Body

1) ?

2) ?

Six tumors of the iris/ciliary body

1) ?

2) ?

3) ?

4) ?

5) ?

6) ?

Choroid

RPE

Retina
**Intraocular Tumors of Childhood**

1) **Juvenile xanthogranuloma**

2) **Medulloepithelioma**

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

**Six tumors of the iris/ciliary body**

**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) **Juvenile xanthogranuloma**

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

It is a… nonneoplastic histiocytic proliferation

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

As orangish skin papules

At what age does it present?

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

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

Unilateral

In what three ways are the iris nodules clinically significant?

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

What is the natural history of JXG?

It is self-limited, usually resolving by age 5 years
1) **Juvenile xanthogranuloma**

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**Iris/Ciliary Body**

1. Juvenile xanthogranuloma
2. Medulloepithelioma
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JXG: Skin papules. The orangish color is classic
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Iris/Ciliary Body

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JXG: Iris lesion
Iris/Ciliary Body

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Intraocular Tumors of Childhood
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JXG: Spontaneous hyphema
Intraocular Tumors of Childhood

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

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

*When unilateral?*

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At what age does it present?  
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What are the two hallmarks of JXG histology?  
- The presence of **Touton giant cells**  
- The presence of ‘foamy macrophages’

When is the lesion Unilateral?

In what three ways are the iris nodules bleeding clinically significant?  
- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma  
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What is the natural history of JXG?  
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Intraocular Tumors of Childhood

Touton giant cells

Foamy macrophages

JXG
Intraocular Tumors of Childhood

Iris/Ciliary Body

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The presence of Touton giant cells

*When unilateral?*
Unilateral

*‘foamy macrophages’*

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

**Retina**

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

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

Choroid

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

*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?*
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.

6) **Iris cysts**

Can be pupillary, stromal, secondary.
1) **Juvenile xanthogranuloma**

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As orangish skin papules

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

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

**In what three ways are the iris nodules clinically significant?**
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**What is the natural history of JXG?**
It is self-limited, usually resolving by age 5 years

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

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*
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) **Juvenile xanthogranuloma**

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

When unilateral? Bilateral?

_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
Iris/Ciliary Body

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*At what age does it present?*  
Unilateral

*When bilateral?*  
When enough nodules are present, heterochromia iridis will result

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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

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The presence of ‘foamy macrophages’

*When unilateral?*  
Lateral

*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?
Intraocular Tumors of Childhood

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How can JXG induce hyphema and glaucoma?
- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
- They are in the DDx as a ‘masquerade syndrome’ in pediatrics 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

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

Iris/Ciliary Body

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Whipple’s disease

Retina
Intraocular Tumors of Childhood

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**Whipple’s disease**

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

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*When Unilateral? Are JXG iris nodules unilateral?*
Unilateral

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

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

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

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

Iris/Ciliary Body

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When unilateral? Unilateral

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

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What is the histology of JXG?
The presence of 'foamy macrophages'

Speaking of ‘foamy macrophages’...

What disease 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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And CNS symptoms--seizures, dementia, coma?

Whipple’s disease

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

What infection agent is responsible for Whipple’s?

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

Iris/Ciliary Body

1) Juvenile xanthogranuloma

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

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

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

*The majority bilateral?*
Unilateral

*What are the two hallmarks of JXG histology?*
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

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

*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 chronic migratory arthritis? Associated with chronic diarrhea? And CNS symptoms—seizures, dementia, coma? Whipple’s disease

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Intraocular Tumors of Childhood

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The presence of *Touton* giant cells

*When are JXG nodules unilateral or bilateral?*
Unilateral

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

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

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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

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

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How does JXG typically present clinically? They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma.

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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).
Intraocular Tumors of Childhood

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_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 an Adult with bilateral upper-lid yellow lesions?*  
--- Pic forthcoming

*In what three ways are the iris nodules clinically significant?*
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It is self-limited, usually resolving by age 5 years

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.
Intraocular Tumors of Childhood

Condition?
Intraocular Tumors of Childhood

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Xanthelsasma

**In what three ways are the iris nodules clinically significant?**  
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**What is the natural history of JXG?**  
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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.

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Intraocular Tumors of Childhood

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   - *Are xanthelasmas a harbinger of elevated serum lipids?*
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Xanthelasma

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

*When are xanthelasma unilatateral?*  
The presence of Touton giant cells.

*‘foamy macrophages’*

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

---

1. **Juvenile xanthogranuloma**: Nonneoplastic histiocytic proliferation. Typically presents in children under 2 years old. Clinical manifestations include skin papules and iris nodules, which can lead to hyphema and increased intraocular pressure (IOP) resulting in glaucoma. This condition is usually self-limited, with regression by age 5. Treatment options include inflammation and IOP management. Pathologically, Touton giant cells are characteristic.

2. **Medulloepithelioma**: A benign but locally aggressive neoplasm of nonpigmented epithelium of the choroid. It typically presents as an iris mass before 10 years of age and can result in hyphema and increased IOP leading to glaucoma. This condition is locally invasive and can be fatal. Treatment involves enucleation.

3. **Lisch nodules**: Associated with Neurofibromatosis Type 1 (NF1). They are typically lighter in dark irides and darker in light irides.

4. **Brushfield spots**: Associated with Down syndrome. They occur in 15% of the non-Down population.

5. **Iris mammillations**: Small, numerous, and of the same color as the iris. They have a weak association with NF1 and Nevoid of Ota.

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

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**Intraocular Tumors of Childhood**

**1) Juvenile xanthogranuloma**

*In three words, what sort of condition is JXG?*
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**Adult with bilateral upper-lid yellow lesions?**

**Xanthelasma**

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

**Can they be congenital?**

**Pic forthcoming**

---

**Choroid**

The presence of Touton giant cells...

The presence of 'foamy macrophages'

**Unilateral?**

---

**RPE**

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

---

**Retina**

**1)**  **Juvenile xanthogranuloma**

*1) Intraocular Tumors of Childhood*...
Iris/Ciliary Body

1) **Juvenile xanthogranuloma**

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

**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?**  

Xanthelasma

When unilateral? The presence of Touton giant cells.  
When bilateral? The presence of *'foamy macrophages'*

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

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

RPE

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

Intraocular Tumors of Childhood

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.

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

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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

What is the other name by which medulloepithelioma is known?

Iris/Ciliary Body

What is the other name by which medulloepithelioma is known?

Choroid

RPE

Retina
Intraocular Tumors of Childhood

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Diktyoma
Intraocular Tumors of Childhood

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2) Medulloepithelioma

Iris/Ciliary Body

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Which specific component of the iris/CB is involved in medulloepithelioma?
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| Iris/Ciliary Body | Choroid | RPE | Retina |
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It is responsible for the creation of aqueous humor
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2) **Medulloepithelioma**

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<thead>
<tr>
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The nonpigmented epithelium of the ciliary body

*How does it present?*

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

- Glaucoma
- Hyphema
- Sectoral cataract

*Is it common, or rare?*

Very rare

*Is it benign, or malignant?*

It is benign, but very aggressive locally

*How is it managed?*

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Intraocular Tumors of Childhood

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Retina
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Very rare

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

*RPE*

How aggressive is ‘very aggressive’?
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**

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**Iris/Ciliary Body**

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

Diktyoma

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

The nonpigmented epithelium of the ciliary body

*How does it present?*

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

-- Glaucoma
-- Hyphema
-- Sectoral cataract

*Is it common, or rare?*

Very rare

*Is it benign, or malignant?*

It is benign, but **very aggressive locally**

*How aggressive is ‘very aggressive’?*

Aggressive enough to result in death
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**Is it benign, or malignant?**

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**How is it managed?**

Enucleation is usually required
Intraocular Tumors of Childhood

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*How does it present?*

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**Choroid**

*Is it common, or rare?*

Very rare

*Is it benign, or malignant?*

It is benign, but very aggressive locally

*How is it managed?*

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

3) **Lisch nodules**

4) **Brushfield spots**

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

**NF1**
**Intraocular Tumors of Childhood**

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3) **Lisch nodules**
   - Lisch nodules are most strongly associated with what congenital condition? 
     - NF1

4) **Brushfield spots**

5) **Iris cysts**
   - Iris/Ciliary Body
   - Iris/Ciliary Body
   - Choroid
   - Retina

6) Iris/Ciliary Body

**Retina**
Intraocular Tumors of Childhood

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In this context, what does NF1 stand for?

Intraocular Tumors of Childhood

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5) **Iris cysts**

6) **Iris/Ciliary Body**

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**Retina**

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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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

**NF1**

*In this context, what does NF1 stand for?* Neurofibromatosis type 1
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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**

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

Neurofibromatosis type 1

*What is the eponymous name for NF1?*

von Recklinghausen’s disease

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

A phakomatosis
Intraocular Tumors of Childhood

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3) **Lisch nodules**

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5) **Iris cysts** can be pupillary, stromal, secondary

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**Retina**

__Lisch nodules are most strongly associated with what congenital condition?__

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Iris/Ciliary Body

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**Iris/Ciliary Body**

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A phakomatosis

**Phakomatoses are known also as what sort of syndrome?**

A neurocutaneous syndrome

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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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

In this context, what does NF1 stand for? Neurofibromatosis type 1

What is the eponymous name for NF1? von Recklinghausen’s disease

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

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

**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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

In general terms, how do phakomatoses present?
Intraocular Tumors of Childhood

Iris/Ciliary Body

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In this context, what does NF1 stand for? Neurofibromatosis type 1

What is the eponymous name for NF1? von Recklinghausen’s disease

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

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

In general terms, how do phakomatoses present? With multiple lesions in two or more organ systems, including the skin and CNS
Intraocular Tumors of Childhood

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Are Lisch nodules associated with NF2?
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**Lisch nodules are most strongly associated with what congenital condition?**

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**Are Lisch nodules associated with NF2?**

Yes, but the relationship is far weaker—Lisch nodules occur in NF2, but so sporadically that they are not expected
**Intraocular Tumors of Childhood**

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

**Iris/Ciliary Body**

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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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**Are Lisch nodules clinically significant?**
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*Are Lisch nodules clinically significant?*

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

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

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

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

Iris cysts: Can be pupillary, stromal, secondary

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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Melanocytic lesions --? --? --? --?

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

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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

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

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

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)?

Are Lisch nodules clinically significant? No; their only significance is as a diagnostic marker for NF1

Retina
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**Melanocytes and neuroglial cells**

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**Melanocytes**

--Café au lait spots

--Axillary/inguinal freckles

--Lisch nodules

--Choroidal lesions

**Neuroglial lesions**

--Nodular neurofibromas

--Plexiform neurofibromas

--Optic glioma

--Prominent corneal nerves

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

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.

Are Lisch nodules clinically significant?

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

Lisch nodules are most strongly associated with what congenital condition?

**NF1**

Are Lisch nodules associated with NF2?

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

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.

**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

Retina

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

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

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

Are Lisch nodules clinically significant? No; their only significance is as a diagnostic marker for NF1

In what fundamental way do these lesions differ (other than the cell type of origin, duh)? The melanocytic lesions are of no clinical significance beyond establishing the diagnosis, whereas the neuroglial lesions are associated with significant ocular and/or systemic morbidity
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Most NF1 lesions are associated with one of two cell types. What are they?
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Melanocytic lesions
- Lisch nodules

Neuroglial lesions
- Nodular neurofibromas
- Plexiform neurofibromas
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Give four classic examples of each: (YMMV of course) The melanocytic lesions are of no clinical significance beyond establishing the diagnosis

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Retina

(No question—proceed when ready)
Intraocular Tumors of Childhood

Iris/Ciliary Body

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3) Lisch nodules

4) Brushfield spots

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.

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Intraocular Tumors of Childhood

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3) **Lisch nodules**

4) **Brushfield spots**

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

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Retina
Intraocular Tumors of Childhood

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Retina
Intraocular Tumors of Childhood

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4) **Brushfield spots**

5) **Iris mammillations**

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**Iris/Ciliary Body**

**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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Down syndrome
Intraocular Tumors of Childhood

Brushfield spots
Intraocular Tumors of Childhood

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4) **Brushfield spots**

5) **Iris mammillations**

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

Down syndrome

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

At least 90%

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

They have none

When a clinically identical iris finding occurs in a non-Down individual, what are the lesions called?

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Brushfield spots are most strongly associated with what congenital condition?

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5) **Iris mammillations**

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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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

Down syndrome

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

At least 90%

*What is the clinical significance of Brushfield spots?*
Intraocular Tumors of Childhood

Iris/Ciliary Body

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5) **Iris mammillations**

**Retina**

- **Irish/Ciliary Body**

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

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

5) **Iris mammillations**

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**Iris/Ciliary Body**

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

Down syndrome

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*What is the clinical significance of Brushfield spots?*

They have none

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Wolfflin nodules
Intraocular Tumors of Childhood

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

5) **Iris mammillations**

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Mammillations? Aren’t those a CNS thingamajig?

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Choroid

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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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.
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

**OK, then what are iris mammillations?**
Intraocular Tumors of Childhood

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OK, then what are iris mammillations?
Tiny pigmented iris nodules which, when present, are found in vast numbers diffusely scattered across the iris surface
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**Are they unilateral, or bilateral?**
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**Iris/Ciliary Body**
Iris/Ciliary Body

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NF1 (albeit not nearly as strongly as Lisch nodules)
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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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

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

With what phakomatosis are they associated?

NF1 (albeit not nearly as strongly as Lisch nodules)
Intraocular Tumors of Childhood

Iris/Ciliary Body

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_Mammillations? Aren’t those a CNS thingamajig?_  
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_OK, then what are iris mammillations?_  
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_Are they unilateral, or bilateral?_  
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_With what phakomatosis are they associated?_  
NF1 (albeit not nearly as strongly as Lisch nodules)

_In addition to NF1, iris mammillations have another important association. What is it?_
Intraocular Tumors of Childhood

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

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

Iris/Ciliary Body

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Oculodermal melanocytosis (nevus of Ota)
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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

*Five tumors of the choroid*
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

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

RPE

Retina

*Five tumors of the choroid*
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

**Choroid**

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

**Retina**

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

Five tumors of the choroid

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

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

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

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

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

But not 6) 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

Five tumors of the choroid
Intraocular Tumors of Childhood

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

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1) **Nevus**: Common. Benign

2) **Melanocytoma**

3) **Osteoma**

4) **Isolated/focal choroidal hemangioma**

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Intraocular Tumors of Childhood

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

1) Nevus: Common. Benign
2) Melanocytoma
3) Osteoma
4) Isolated/focal choroidal hemangioma
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Intraocular Tumors of Childhood

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1) Nevus: Common. Benign. 2) Melanocytoma
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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?
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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
Intraocular Tumors of Childhood

Melanocytoma
Intraocular Tumors of Childhood

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

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 stromal or pupillary. Secondary (see the Iris issues in kids slide-set)

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**Iris/Ciliary Body**

1) **Nevus**: Common. Benign

2) **Melanocytoma**

3) **Osteoma**

4) **Isolated/focal choroidal hemangioma**

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**Choroid**

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**RPE**

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**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
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**Iris/Ciliary Body**

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**Choroid**

1) **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

Iris/Ciliary Body

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Choroid

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RPE

Retina

Melanocytoma is a variant of what common choroidal finding?
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From what structure does it commonly arise?
The optic disc

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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

**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?**

Yes
Intraocular Tumors of Childhood

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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**

**Choroid**

Melanocytoma is a variant of what common choroidal finding?

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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?

---

**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?
No

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

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**Iris/Ciliary Body**

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

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From what structure does it commonly arise?
The optic disc

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

Is there a racial predilection?
No

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

**Choroid**

1) **Nevus:** Common. Benign

2) **Melanocytoma**

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5) **Diffuse choroidal hemangioma**

**RPE**

**Retina**

Melanocytoma is a variant of what common choroidal finding?

It is a particular sort of choroidal nevus

From what structure does it commonly arise?
The optic disc

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

Is there a racial predilection?
No

Does it affect visual acuity?
Only in a minority of cases. But in almost all cases, it does affect visual fields.
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: Striking, darkly pigmented areas of iris before age 8 years. Same color as iris. Weak association with NF1. Nevus of Ota
4) Brushfield spots: Tiny, white, freely movable specks within the iris. 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

Iris/Ciliary Body

Choroid

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

Choroid

RPE

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Does melanocytoma have the potential to undergo malignant transformation?

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

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Yes
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Does melanocytoma have the potential to undergo malignant transformation?

Approximately what percent of cases will transform?

Melanocytoma

Nevus

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Melanocytoma
Iris/Ciliary Body

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3) Lisch nodules: Striking yellow nodule on irides in NF1

4) Brushfield spots: Lighter on dark irides; darker on light

5) Iris mammillations: Essential association with NF1, Nevus of Ota

6) Iris cysts: Can be very large

Choroid

1) Nevus: Common. Benign

2) Melanocytoma

3) Osteoma

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

Melanocytoma is a variant of what common choroidal finding?

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Is there a racial predilection?
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Does melanocytoma have the potential to undergo malignant transformation?

Approximately what percent of cases will transform?
1-2

RPE

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

- Bone
- Very rare
- Benign
- More common in females
- Teens
- Choroidal neovascular membrane

Choroid:
3) **Osteoma**
4) Isolated/focal choroidal hemangioma
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6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

---

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

Bone

---

**Choroid**

3) **Osteoma**

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

---

**Retina**

**RPE**

**Intraocular Tumors of Childhood**

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**Retina**

Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

Osteoma
Intraocular Tumors of Childhood

Osteoma: FP, and b-scan demonstrating ...
Intraocular Tumors of Childhood

Osteoma: FP, and b-scan demonstrating ‘shadowing’
Intraocular Tumors of Childhood

Osteoma: Another example
Osteomas (same pt, different cuts).
Note how bright the lesions are.
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 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: Bone

Very rare

Benign

Yes, it is more common in females

Teens

Choroidal neovascular membrane

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

Bone

Is it common or rare?

Is it benign or malignant?

Does there a gender predilection?

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

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

Choroidal neovascular membrane

Osteoma

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma
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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

1. Nevus
2. Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare
3. Osteoma
4. Isolated/focal choroidal hemangioma
5. Diffuse choroidal hemangioma

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Choroid

3) **Osteoma**
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma

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Retina

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RPE

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Iris/Ciliary Body

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Intraocular Tumors of Childhood

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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 in kids slide-set)

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

Is it common or rare?
Very rare

Is it benign or malignant?
Benign

Choroid

3) Osteoma
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma

RPE

Retina
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Osteoma

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

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

Bone

Is it common or rare?

Very rare

Is it benign or malignant?

Benign

Intraocular Tumors of Childhood

Choroid

Retina

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

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)

---

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

**Bone**

**Is it common or rare?**

Very rare

**Is it benign or malignant?**

Benign

**Does there a gender predilection?**

Yes, it is more common in females

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

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

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

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Osteoma

3) Osteoma

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

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

Bone

Is it common or rare?

Very rare

Is it benign or malignant?

Benign

Does there a gender predilection?

Yes, it is more common in

M v F

Intraocular Tumors of Childhood

Retina

Choroid

RPE

Iris/Cornea
In a word, what is a choroidal osteoma composed of?

Bone

Is it common or rare?
Very rare

Is it benign or malignant?
Benign

Does there a gender predilection?
Yes, it is more common in females

Intraocular Tumors of Childhood

Iris/Ciliary Body

Choroid

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RPE

Retina

1) Nevis
Common. Benign

2) Melanocytoma
Usually juxtapapillary. Malignant transformation extremely rare

3) Osteoma
Bone

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

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Yes, it is more common in females

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

Osteoma

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1) Nevus: Common. Benign

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3) Osteoma

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In a word, what is a choroidal osteoma composed of?
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Is it common or rare?
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Does there a gender predilection?
Yes, it is more common in females

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

Intraocular Tumors of Childhood

Retina

Choroid

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

Choroidal neovascular membrane

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

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Is it common or rare?

Very rare

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Does there a gender predilection?

Yes, it is more common in females

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

Choroidal neovascular membrane
Intraocular Tumors of Childhood

Osteoma with CNVM in a 13 y.o. female
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---

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

- Bone

**Is it common or rare?**

- Very rare

**Does 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

---

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

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

---

**Choroid**

- 3) **Osteoma**
- 4) Isolated/focal choroidal hemangioma
- 5) Diffuse choroidal hemangioma
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Osteoma

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

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

Bone

Is it common or rare?

Very rare

Does there a gender predilection?

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

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

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

Choroid

3) Osteoma

4) Isolated/focal choroidal hemangioma

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Retina

RPE
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Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma
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1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

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

Iris/Ciliary Body

Choroid

RPE

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 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)

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**Iris/Ciliary Body**

**Retina**

**Choroid**

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

4) **Isolated/focal choroidal hemangioma**

5) **Diffuse choroidal hemangioma**

---

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

**Circumscribed choroidal hemangioma**

**Is it common, or rare?**

**Rare**

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

**No**

**How does it present?**

As a reddish-orange mass in the macula

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

It is one of 'high internal reflectivity'
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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1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma

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

Is it common, or rare?
Rare

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Intraocular Tumors of Childhood
**Intraocular Tumors of Childhood**

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**Iris/Ciliary Body**

**Choroid**

**Retina**

---

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

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**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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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
Intraocular Tumors of Childhood

Circumscribed choroidal hemangioma
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Intraocular Tumors of Childhood

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

Is it common, or rare? Rare

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

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

What is its characteristic pattern on a-scan ultrasonography?

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### Intraocular Tumors of Childhood

#### Iris/Ciliary Body

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#### Choroid

**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

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

**Circumscribed choroidal hemangioma**

**Is it common, or rare?**

Rare

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

No

**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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Intraocular Tumors of Childhood

Circumscribed choroidal hemangioma: High internal reflectivity on a-scan
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

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- **By what other name is this lesion known?** Circumscribed choroidal hemangioma
- **Is it common, or rare?** Rare
- **Is it associated with a systemic condition, ie, is it syndromic?** No
- **How does it present?** As a reddish-orange mass in the macula
- **What is its characteristic pattern on a-scan ultrasonography?** It is one of 'high internal reflectivity'
- ‘High internal reflectivity’--what other choroidal lesion’s a-scan is described the same way?

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- **Isolated/focal choroidal hemangioma**
- **Osteoma**
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Nevus: Common. Benign

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Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

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

Is it common, or rare?
Rare

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

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

It is one of:
3) Osteoma
4) Isolated/focal choroidal hemangioma
5) Diffuse choroidal hemangioma

Retina

RPE

Choroid

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Intraocular Tumors of Childhood

### 192

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

**Sturge-Weber syndrome (SWS)**

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

About 50%

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

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

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

'Tomato catsup fundus'

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

Yes, but it’s uncommon

**Does the choroidal hemangioma have malignant potential?**

No

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**5) Diffuse choroidal hemangioma**
1) Juvenile xanthogranuloma (JXG): 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

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

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

5) Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Present in about 50% of SWS cases. The fundus appearance is a very red--much more so than an unaffected fundus. This is referred to as the 'Tomato catsup fundus'. It is uncommon for the choroidal hemangioma to be present bilaterally. It does not have malignant potential.
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?

A phakomatosis

What is the noneponymous name for SWS?

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

Intraocular Tumors of Childhood

With what condition is the diffuse choroidal hemangioma associated?

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

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**Intraretinal Tumors of Childhood**

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

Sturge-Weber syndrome (SWS)

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A phakomatosis

**Intraretinal Tumors of Childhood**

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RPE

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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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**Intraocular Tumors of Childhood**

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

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

**Choroid**

**RPE**

**Retina**

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

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

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

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

When does it present?
At birth

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

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

All infants with SWS have a port-wine stain. Do all infants with a port-wine stain have SWS?
No

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

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

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

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

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

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

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

**In a word, what sort of condition is SWS?**
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4) **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a-scan pattern
5) **Diffuse choroidal hemangioma**

**Sturge-Weber syndrome (SWS)**

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

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

Sturge-Weber: Port-wine stain
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Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

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

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

At birth

It comports to the distribution of one or more divisions of CN5

Does the choroidal hemangioma have malignant potential? No

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

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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: Present in Sturge-Weber syndrome (SWS). In about 50% of cases.

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

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5) Diffuse choroidal hemangioma: Present in what percent of SWS? About 50%

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

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

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

Does the choroidal hemangioma have malignant potential? No

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

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When does it present? At birth
Intraocular Tumors of Childhood

Sturge-Weber: Port-wine stain
In a word, what sort of condition is SWS? 
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When does it present? 
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What is the typical pattern of distribution?

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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?**
A phakomatosis

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**When does it present?**
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**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**

With what condition is the diffuse choroidal hemangioma associated?

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No, some cases will cross the midline of the face
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Diffuse choroidal hemangioma: 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?
In a word, what sort of condition is SWS?
A phakomatosis

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**Intracocular Tumors of Childhood**

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**Iris/Ciliary Body**

**Retina**

**RPE**

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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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5) **Diffuse choroidal hemangioma**
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**Diffuse choroidal hemangioma:**

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

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

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

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

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

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No

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Intracocular Tumors of Childhood

With what condition is the diffuse choroidal hemangioma associated?
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5) **Diffuse choroidal hemangioma**
Intraocular Tumors of Childhood

Sturge-Weber: Tomato catsup fundus OD
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**Intraocular Tumors of Childhood**

**Retina**

1) **Nevus:** Common. Benign

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5) **Diffuse choroidal hemangioma**

**Iris/Ciliary Body**

**RPE**

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

Sturge-Weber syndrome (SWS)

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About half

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Iris/Ciliary Body

Choroid

RPE

Retina

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Retina

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

Retina

RPE
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Sturge-Weber syndrome (SWS)

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

Retina

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

Iris/Ciliary Body

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Choroid

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

RPE

1) ?

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
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
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RPE

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

Iris/Ciliary Body

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RPE

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

Choroid

What is the clinical appearance of CHRPE?
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**Iris/Ciliary Body**

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

CHRPE
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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

Iris/Ciliary Body

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

Is it common, or rare?
Common

RPE

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

Retina
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### Iris/Ciliary Body

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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### Choroid

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

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### Retina
Iris/Ciliary Body

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Intraocular Tumors of Childhood

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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
Intraocular Tumors of Childhood

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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?

- **CHRPE**
- **or**

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

Iris/Ciliary Body

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

Solitary

Grouped

CHRPE
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

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

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

--- Solitary CHRPE

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

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

Iris/Ciliary Body

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RPE

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

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

--**Solitary** CHRPE

--**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?

‘Bear tracks’
Intraocular Tumors of Childhood

CHRPE: Bear tracks
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

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**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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**Retina**
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?
Familial adenomatous polyposis, aka Gardner syndrome

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?
--Solitary
--Multifocal or Grouped

Retina

1) Nevus
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**RPE**

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

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

---

**Solitary** CHRPE

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

---

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

Familial adenomatous polyposis, aka Gardner syndrome

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

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

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

If the shape of the lesions is pisciform

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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!
**Intraocular Tumors of Childhood**

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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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
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?
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)

RPE

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

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

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**RPE**

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

**CHRPE is characterized according to its presentation. In what two ways does it present?**
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CHRPE-like lesions of Gardner syndrome: Bilateral presentation
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**Iris/Ciliary Body**

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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 __distribution pattern__

---

**RPE**

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

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--**Solitary** CHRPE

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Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

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--If the lesions are scattered throughout multiple sectors of the eyes (ie, not ‘grouped’)
CHRPE-like lesions of Gardner syndrome: Scattered distribution
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

---

**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

---

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 $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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**A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?**

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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

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

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Intraocular Tumors of Childhood

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Iris/Ciliary Body

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

- What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?
  --If it is bilateral (regular CHRPE is almost always unilateral)
  --If the lesions are scattered throughout multiple sectors of the eye (i.e., not ‘grouped’)
  --If the shape of the lesions is pisciform

- What does pisciform mean?
  'Fish-shaped'

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

CHRPE is characterized according to its presentation. In what two ways does it present?
--Solitary CHRPE
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Intraocular Tumors of Childhood

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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

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RPE

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

CHRPE-like lesions of Gardner syndrome: Pisciform shape
**Intraocular Tumors of Childhood**

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

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

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

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

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

RPE

Retina

Choroid

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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**Iris/Ciliary Body**

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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

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

---They are hypopigmented
---They point towards the optic nerve head

---Multifocal or Grouped

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

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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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**Intraocular Tumors of Childhood**

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1) **Nevus**: Common. Benign

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

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

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

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

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

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

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**Solitary** CHRPE

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

Iris/Ciliary Body

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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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 characteristic of Gardner syndrome increases the likelihood that it is a component of Gardner syndrome?
--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are scattered throughout multiple sectors of the eyes (ie, not 'grouped')
--If the shape of the lesions is pisciform

What is the most clinically important (and ominous) component to Gardner syndrome?
Pts develop thousands of colonic polyps, a significant number of which are malignant

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

1) **Nevus:** Common. Benign
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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

Intraocular Tumors of Childhood

1) **Congenital hypertrophy of the RPE (CHRPE):** Retina
   Choroid
   Iris/Ciliary Body
   CHRPE is characterized according to its presentation. In what two ways does it present?
   --Solitary CHRPE
   --Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones

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

What is the most clinically important (and ominous) component to **Gardner syndrome**?
Pts develop thousands of **colonic polyps, a significant number of which are malignant**

What proportion of untreated Gardner syndrome pts will develop colon cancer?
All of them
By what age will this occur?
Age 40, maybe a little later
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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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?

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

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

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What is the 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**

---

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

--**Solitary** CHRPE

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

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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? 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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What is the most clinically important (and ominous) component to Gardner syndrome?
Pts develop thousands of colonic polyps, a significant number of which are malignant

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

By what age will this occur?
Age 40, maybe a little later
Intraocular Tumors of Childhood

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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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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?

40, maybe a little later

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**Retina**

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

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

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

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**Iris/Ciliary Body**

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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**

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

All of them

By what age will this occur?

40, maybe a little later

What is the treatment of choice?

Prophylactic colectomy

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

--Solitary CHRPE
--Multifocal or Grouped CHRPE

CHRPE: Large lesion(s) surrounded by a few smaller ones
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? Familial adenomatous polyposis, aka Gardner syndrome

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

By what age will this occur? 40, maybe a little later

What is the treatment of choice? Prophylactic colectomy

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

**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?**

---

**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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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 is the most clinically important (and ominous) component to Gardner syndrome?
Pts develop thousands of colonic polyps, a significant number of which are malignant

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

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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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

### 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 is the most clinically important (and ominous) component to Gardner syndrome? Pts develop thousands of colonic polyps, a significant number of which are malignant

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

**CHRPE is characterized according to its presentation. In what two ways does it present?**
---Solitary CHRPE
---Multifocal or Grouped CHRPE: Large lesion(s) surrounded by a few smaller ones
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What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?

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When ‘colon cancer + ophthalmic issue’ is mentioned, two syndromes should come to mind. **One is Gardner syndrome. What is the other?**

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**RPE**

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

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

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**Solitary** CHRPE

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**Retina**
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Muir-Torre syndrome

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

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What is the main ophthalmic manifestation of Muir-Torre syndrome?

When 'colon cancer + ophthalmic issue' is mentioned, two syndromes should come to mind. One is Gardner syndrome. What is the other?

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

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

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

-- If the shape of the lesions is pisciform

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What is the main ophthalmic manifestation of Muir-Torre syndrome?

Multiple sebaceous lesions of (but not necessarily limited to) the eyelids

Does Muir-Torre present with multiple adenomatous polyps of the colon a la Gardner syndrome?

No; Muir-Torre is an example of a disease spectrum called Hereditary Non-Polyposis Colorectal Cancer
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Multiple skin-colored to yellow–pink papules (arrows) on the face of a 64-year-old woman with a history of colon and cervical cancer. A skin biopsy confirmed a diagnosis of sebaceous adenoma resulting from Muir–Torre syndrome.
Intraocular Tumors of Childhood

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**What sorts of sebaceous lesions?**

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

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Muir-Torre syndrome

--- Sebaceous-cell carcinomas

--- Sebaceous-cell adenomas

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Choroid
1) **Nevus**: Common. Benign
2) **Melanocytoma**: Usually juxtapapillary. Malignant transformation extremely rare
3) **Osteoma**: Benign bony tumor, most common in teen years, females. Risk of CNVM
4) **Isolated/focal choroidal hemangioma**: Very rare. Characteristic a-scan pattern
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) ?
Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

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1) **Combined hamartoma of the retina and RPE

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

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

What is a hamartoma?

A hamartoma is a tumor composed of histologically abnormal cells found in their normal location. 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?

- Choroidal melanoma--eyes have been enucleated because of this misdiagnosis

1) **Congenital hypertrophy of the RPE (CHRPE):** Common. Deeply pigmented.

A CHRPE-like finding is associated with Gardner syndrome

- 1) Combined hamartoma of the retina and RPE

- 1) Retinoblastoma (see the slide-set dedicated to it)
Iris/Ciliary Body

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Choroid

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Intraocular Tumors of Childhood

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

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

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

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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

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

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

- **What is a hamartoma?**
  - A tumor composed of histologically abnormal cells found in their normal location

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**

1) **Retinoblastoma** (see the slide-set dedicated to it)
What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location? A choristoma

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

1) Retinoblastoma (see the slide-set dedicated to it)
Intraocular Tumors of Childhood


2) Combined hamartoma of the retina and RPE

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

What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location? A choristoma.

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

That a lesion is a hamartoma (or choristoma) indicates what about its status vis a vis malignancy? That it is benign.

What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location? A choristoma.
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.

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Nevus: Common. Benign.

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


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

Intraocular Tumors of Childhood

What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location? A choristoma.

What is a hamartoma? A tumor composed of histologically normal cells found in their abnormal location. What is the name of the reverse clinical entity, ie, one with normal cells found in an abnormal location? A choristoma.

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

Intraocular Tumors of Childhood

RPE

Retina

1) Combined hamartoma of the retina and RPE

1) Retinoblastoma (see the slide-set dedicated to it)
Intraocular Tumors of Childhood

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**What is a hamartoma?**

A tumor composed of histologically abnormal cells found in their normal location

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

A choristoma

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RPE

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[Diagram: RPE → Retina → Combined hamartoma of the retina and RPE]

1) Combined hamartoma of the retina and RPE

1) Retinoblastoma (see the slide-set dedicated to it)
Intraocular Tumors of Childhood

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What is a hamartoma?
A tumor composed of histologically normal cells found in their normal location

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A choristoma

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

What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

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

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Retinoblastoma (see the slide-set dedicated to it)

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?
RPE cells (duh) and retinal glial cells

RPE
Retina
1) Congenital hypertrophy of the RPE (CHRPE): Common. Deeply pigmented. A CHRPE-like finding is associated with Gardner syndrome
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10) Isolated/focal choroidal hemangioma: Very rare. Characteristic a- scan pattern


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

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**What is a hamartoma?**

A tumor composed of histologically abnormal cells found in their normal location

**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?**

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**RPE**

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

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**Retina**

1) Retinoblastoma (see the slide-set dedicated to it)
Intraocular Tumors of Childhood

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Retina

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Iris/Ciliary Body

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2) Combined hamartoma of the retina and RPE
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What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

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

RPE

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Intraocular Tumors of Childhood

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**What is a hamartoma?**

A tumor composed of histologically abnormal cells found in their normal location

**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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Iris/Ciliary Body

What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

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?
Choroidal melanoma--eyes have been enucleated because of this misdiagnosis

Retina

RPE

1) Combined hamartoma of the retina and RPE

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Intraocular Tumors of Childhood

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


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

---

What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

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

RPE

Retina

1) Combined hamartoma of the retina and RPE

RPE

Retina

1) Congenital hypertrophy of the RPE (CHRPE): Common. Deeply pigmented. A CHRPE-like finding is associated with Gardner syndrome

1) Combined hamartoma of the retina and RPE

RPE

Retina

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

What is a hamartoma?
A tumor composed of histologically abnormal cells found in their normal location

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?
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

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
Combined hamartoma of retina and RPE.
Note the entire lesion is above Bruchs
Iris/Ciliary Body

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Choroid

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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

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

_No question—summary/review slide_