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) ?
3) ?
4) ?
5) ?
6) ?

Six tumors of the iris/ciliary body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

Six tumors of the iris/ciliary body

Choroid

RPE

Retina
1) **Juvenile xanthogranuloma**

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

- It is a nonneoplastic histiocytic proliferation.
- It usually presents as orangish skin papules.
- It typically presents in the majority before age 1 year, and almost all by age 2.
- When iris nodules are present, they are unilateral.
- In the clinical setting, they are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma.
- They are in the differential diagnosis (DDx) as a 'masquerade syndrome' in pediatric uveitis.
- If enough nodules are present, heterochromia iridis will result.
- It is self-limited, usually resolving by age 5 years.
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

Iris/Ciliary Body

Choroid

RPE

Retina
1) Juvenile xanthogranuloma

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

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

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

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
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
Intraocular Tumors of Childhood

JXG: Skin papules. The orangish color is classic
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

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

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

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

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**Intraocular Tumors of Childhood**

Choroid

RPE

Retina
Iris/Ciliary Body

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

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

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

*When JXG iris nodules are present, are they uni-, or bilateral?*
In three words, what sort of condition is JXG?
It is a… **nonneoplastic histiocytic proliferation**

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

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

*When JXG iris nodules are present, are they uni-, or bilateral?*
Unilateral
Intraocular Tumors of Childhood

JXG: Iris lesion
1) **Juvenile xanthogranuloma**

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

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

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

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

*In what three ways are the iris nodules clinically significant?*
--
--
--
Intraocular Tumors of Childhood

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*In three words, what sort of condition is JXG?*
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*When JXG iris nodules are present, are they uni-, or bilateral?*
Unilateral

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

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**Iris/Ciliary Body**

**Choroid**

**RPE**

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

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

It is a… **nonneoplastic histiocytic proliferation**

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

As orangish skin papules

*At what age does it present?*

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

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

Unilateral

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

--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

JXG: Spontaneous hyphema
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**

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

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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Retina

Choroid

RPE
Intraocular Tumors of Childhood

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   As orangish skin papules

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

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

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

   *Should JXG nodules be removed surgically?*

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

Iris/Ciliary Body

1) **Juvenile xanthogranuloma**

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   *When JXG iris nodules are present, are they uni-, or bilateral?*
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   *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
   --Heterochromia iridis will result

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

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

Choroid

RPE

Retina
Iris/Ciliary Body

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*When are the JXG iris nodules clinically significant?*
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
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--If enough nodules are present, heterochromia iridis will result

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

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

Intraocular Tumors of Childhood

Choroid

RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

*In what three ways are the iris nodules clinically significant?*
--They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
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Retina

Choroid

RPE
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

*When unilateral?*
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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Choroid

RPE

Retina
Intraocular Tumors of Childhood

Touton giant cells

Foamy macrophages

JXG
**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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

*When unilateral?*

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Iris/Ciliary Body

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

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

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

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

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

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

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

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

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

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

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

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

When unilateral?

Unilateral

Speaking of ‘foamy macrophages’...
What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis? First clue--more forthcoming
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

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

In what three ways are the iris nodules clinically significant?
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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

‘foamy macrophages’

Speaking of ‘foamy macrophages’…
What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?
And a hx of chronic migratory arthritis?

Clue #2
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 manner of presentation in the eye is:*
Unilateral

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

*Speaking of ‘foamy macrophages’…*
*What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?*
*And a hx of chronic migratory arthritis?*
*Associated with chronic diarrhea?* — Need another?

Choroid

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 2

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

*When unilateral?*
The presence of ‘foamy macrophages’

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

Retina

Choroid
1) **Juvenile xanthogranuloma**

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

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

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

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

*When are JXG iris nodules unilateral?*  
Unilateral

**Speaking of ‘foamy macrophages’…**

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

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

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

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

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

**When?**
The presence of ‘foamy macrophages’

**Unilateral?**

**Speaking of ‘foamy macrophages’…**
What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?
And a hx of chronic migratory arthritis?
Associated with chronic diarrhea?
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*
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 years

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

1) Juvenile xanthogranuloma

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

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

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

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

When Unilateral

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

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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

*When unilateral?*

*Speaking of ‘foamy macrophages’…*
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

*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 years*

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

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

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

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

### Choroid

**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 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

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 (remember, they have GI issues)

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

Choroid

Retina

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

Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma

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

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

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

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

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

Whipple’s disease

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

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

Iris/Ciliary Body

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*When is JXG unilateral?*
Unilateral

*‘foamy macrophages’*

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

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*When unilateral or bilateral?*
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Broadly speaking, what sort of condition is Whipple’s?
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What infection agent is responsible for Whipple’s?
The bacterium *Tropheryma whipplei*

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*
**Iris/Ciliary Body**

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**Retina**

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Small-intestine biopsy stained with periodic acid-Schiff. Note the numerous macrophages in the lamina propria (arrows).
Intraocular Tumors of Childhood

1) **Juvenile xanthogranuloma**

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

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

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

When
- Unilateral

Unilateral

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

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

Condition?
1) **Juvenile xanthogranuloma**

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

**Xanthelsasma**

The presence of **Touton giant cells**

When Unilateral?

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**Xanthelsasma**

Are xanthelasmas a harbinger of elevated serum lipids?

The presence of **Touton giant cells**  
When unilateral?  
The presence of "foamy macrophages"  
Unilateral?

Choroid

**RPE**

What is the natural history of JXG?  
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Retina

1. **Juvenile xanthogranuloma**

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


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

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

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

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

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Xanthelasmas

The presence of Touton giant cells

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Choroid

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

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

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

*What is the natural history of JXG?*  
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**Intraocular Tumors of Childhood**

- **Iris/Ciliary Body**
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2) Medulloepithelioma

What is the other name by which medulloepithelioma is known?

Iris/Ciliary Body

Choroid

RPE

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

Iris/Ciliary Body

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

Very rare

Benign, but very aggressive locally

Enucleation is usually required

Choroid

RPE

Retina
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Iris/Ciliary Body

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- **What is the other name by which medulloepithelioma is known?** Diktyoma
- **Which specific component of the iris/CB is involved in medulloepithelioma?** The nonpigmented epithelium of the ciliary body
- **What extremely important function does the nonpigmented epi of the CB perform?**

**Choroid**

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It is responsible for the creation of aqueous humor

**Choroid**

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### Iris/Ciliary Body

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- RPE

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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 is it managed?
Enucleation is usually required
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### Choroid

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Intraocular Tumors of Childhood

Medulloepithelioma/diktyoma
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Very rare

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It is benign, but very aggressive locally

*How is it managed?*

Enucleation is usually required
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- **Is a tendency to metastasize one of its ‘malignant features’?** No, this lesion rarely metastasizes; it does its damage locally
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Is it common, or rare?
Very rare

Retina

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

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

2) Medulloepithelioma

Iris/Ciliary Body

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

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

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

Is it common, or rare?
Very rare

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

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

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Very rare

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How is it managed?

Choroid

RPE

Retina
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Is it common, or rare?
Very rare

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How is it managed?
Enucleation is usually required
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3) Lisch nodules
4) Brushfield spots

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

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Intraocular Tumors of Childhood

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

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Iris/Ciliary Body

Retina
Intraocular Tumors of Childhood

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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
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**Intraocular Tumors of Childhood**

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**Iris/Ciliary Body**

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

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

Retina
Intraocular Tumors of Childhood

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**Neurocutaneous** syndromes

In general terms, how do phakomatoses present?

Retina
Intraocular Tumors of Childhood

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

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

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

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Iris/Ciliary Body

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Are Lisch nodules associated with NF2?
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Intraocular Tumors of Childhood

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*NF1*

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What is the prevalence of Lisch nodules in NF1?

Retina
Intraocular Tumors of Childhood

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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.

Retina
Intraocular Tumors of Childhood

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**Are Lisch nodules clinically significant?**

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Intraocular Tumors of Childhood

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

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

- Lisch nodules
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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; 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

---

**Iris/Ciliary Body**

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

**Melanocytes and neuroglial cells**

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Intraocular Tumors of Childhood

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Melanocytic lesions

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--?
--?
--?

Neuroglial lesions

--?
--?
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--?

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

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NF1

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

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

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

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Intraocular Tumors of Childhood
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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)?

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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Retina
Intraocular Tumors of Childhood

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Lisch nodules are most strongly associated with what congenital condition?

NF1

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

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

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

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

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

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

- **Melanocytes**
- **neuroglial cells**

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**Melanocytic lesions**
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**Neuroglial lesions**
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**Give four classic examples of each: (YMMV of course)**

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Melanocytic lesions

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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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(No question—proceed when ready)
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Iris/Ciliary Body

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

Are Lisch nodules dark, or light?
Yes. 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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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? Wolfflin nodules
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**Iris/Ciliary Body**

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Intraocular Tumors of Childhood

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*Mammillations? Aren’t those a CNS thingamajig?*
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**Iris/Ciliary Body**

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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
Iris mammilations
**Intraocular Tumors of Childhood**

Iris/Ciliary Body

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Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

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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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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?
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_With what phakomatosis are they associated?_ 
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_In addition to NF1, iris mammillations have another important association. What is it?_
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Iris/Ciliary Body

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

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

4) Brushfield spots: Strong association with Down syndrome

5) Iris mammillations

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
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**With what phakomatosis are they associated?**
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*In addition to NF1, iris mammillations have another important association. What is it?*
Oculodermal melanocytosis, aka **nevus of Ota**
Oculodermal melanocytosis (nevus of Ota)
Intraocular Tumors of Childhood

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

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Iris/Ciliary Body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

Five tumors of the choroid

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

RPE

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**

Five tumors of the choroid

Retina

RPE
Intraocular Tumors of Childhood

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Choroid

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

But not 6) ?

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

Iris/Ciliary Body

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Choroid

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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.
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

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

Iris/Ciliary Body

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Choroid

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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

Iris/Ciliary Body

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Choroid

1) **Nevus:** Common. Benign
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Note the factoids, then proceed
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

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**Choroid**

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2) **Melanocytoma**

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**RPE**

**Retina**
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

It is a particular sort of **choroidal nevus**

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**Choroid**

1) **Nevus**: Common. Benign

2) **Melanocytoma**

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**Retina**

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**RPE**

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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

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

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**Iris/Ciliary Body**

**Melanocytoma** is a variant of what common choroidal finding?
It is a particular sort of choroidal nevus
From what structure does it commonly arise?
The optic disc

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**Choroid**

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

**Retina**
Melanocytoma
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

It is a particular sort of choroidal nevus

**From what structure does it commonly arise?**

The optic disc

**Does it have a unilateral/bilateral predilection?**

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**Choroid**

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**RPE**

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**Retina**
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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2) **Melanocytoma**: It is a particular sort of choroidal nevus

3) **Osteoma**

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**Choroid**

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

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

3) **Does it have a unilateral/bilateral predilection?**
   
   **Yes, it is virtually always**
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**Iris/Ciliary Body**

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

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

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

**Intraocular Tumors of Childhood**

1) Nevus: Common. Benign
2) Melanocytoma
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**Choroid**

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**RPE**

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

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**Choroid**

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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?*

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**RPE**

**Retina**
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2) **Melanocytoma**: 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

3) **Osteoma**

4) **Isolated/focal choroidal hemangioma**

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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

   Is there a racial predilection?

   No

   Does it have the potential to undergo malignant transformation?

   Yes
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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2) **Melanocytoma**

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**Choroid**

1) **Neovascularization**

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

It is a particular sort of chorial nevus

**From what structure does it commonly arise?**

The optic disc

**Does it have a unilateral/bilateral predilection?**

Yes, it is virtually always unilateral

**Is there a racial predilection?**

No

**Does it affect visual acuity?**

Yes, in a minority of cases. But in almost all cases, it does affect visual fields.
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?
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...
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6) **Iris cysts**: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

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**Iris/Ciliary Body**

1) **Nevus**: Common. Benign

2) **Melanocytoma**

3) **Osteoma**

4) **Isolated/focal choroidal hemangioma**

5) **Diffuse 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, 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.
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**Iris/Ciliary Body**

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Intraocular Tumors of Childhood

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

- **Bone**
- **Very rare**
- **Benign**
- **Yes, it is more common in females**
- **Teens**
- **If significant vision loss occurs, what osteoma complication is usually the culprit?**
  - **Choroidal neovascular membrane**

---

**Intraocular Tumors of Childhood**

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Osteoma: Bone

Choroidal osteoma: Bone

Isolated/focal choroidal hemangioma

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

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Is it common or rare?

Very rare

Is it benign or malignant?

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Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

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Retina
Intraocular Tumors of Childhood

Osteoma
Osteoma: FP, and b-scan demonstrating finding illustrated above.
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
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### Iris/Ciliary Body

### Choroid

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### RPE

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Intraocular Tumors of Childhood

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Osteoma: Bone

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

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Choroidal neovascular membrane

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Osteoma with CNVM in a 13 y.o. female
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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)
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Intraocular Tumors of Childhood

RPE
Retina

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

4) Isolated/focal choroidal hemangioma

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By what other name is this lesion known?

Circumscribed choroidal hemangioma

Is it common, or rare?

Rare

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

No

How does it present?

As a reddish-orange mass in the macula

What is its characteristic pattern on a -scan ultrasonography?

It is one of 'high internal reflectivity'
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5) Diffuse choroidal hemangioma

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Circumscribed choroidal hemangioma

Is it common, or rare?

Rare

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

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

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

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

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

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

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

Diffuse choroidal hemangioma

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

Is it common, or rare? Rare

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

4) Isolated/focal choroidal hemangioma

5) Diffuse choroidal hemangioma
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

RPE

Retina
Intraocular Tumors of Childhood

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

Isolated/focal choroidal hemangioma

Diffuse choroidal hemangioma

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By what other name is this lesion known?

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Rare

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

No

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Retina

Iris/Ciliary Body

Choroid

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Intraocular Tumors of Childhood

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

Is it common, or rare?
Rare

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

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

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

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**Isolated/focal choroidal hemangioma**

**Diffuse choroidal hemangioma**
Intraocular Tumors of Childhood

Circumscribed 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**

5. **Diffuse choroidal hemangioma**

**Retina**

By what other name is this lesion known?

**Circumscribed choroidal hemangioma**

Is it common, or rare?

Rare

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

No

How does it present?

As a reddish-orange mass in the macula

What is its characteristic pattern on a-scan ultrasonography?

It is one of 'high internal reflectivity'
Intraocular Tumors of Childhood

Iris/Ciliary Body

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7) Nevus: Common. Benign

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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’

Choroid

RPE

Retina
Intraocular Tumors of Childhood

Circumscribed choroidal hemangioma: High internal reflectivity on a-scan
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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'

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

Choroidal nevus

It is one of high internal reflectivity

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

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

Is it common, or rare? Rare

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

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

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

It is one of: high internal reflectivity

3) Osteoma: benign bony tumor, most common in teen years, females. Risk of CNVM
4) Isolated/focal choroidal hemangioma
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Intracocular Tumors of Childhood

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With what condition is the diffuse choroidal hemangioma associated?

Sturge-Weber syndrome (SWS)

Diffuse choroidal hemangioma is present in what percent of SWS?

About 50%

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

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

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

'Tomato catsup fundus'

Can the choroidal hemangioma be present bilaterally?

Yes, but it's uncommon

Does the choroidal hemangioma have malignant potential?

No

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

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5) Diffuse choroidal hemangioma

- Sturge-Weber syndrome (SWS)
- About 50% of SWS patients
- The coloration is a very red—much more so than an unaffected fundus
- 'Tomato catsup fundus'
- Yes, but it's uncommon
- No

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


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

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

Diffuse choroidal hemangioma

1) Sturge-Weber syndrome (SWS)

In a word, what sort of condition is SWS?

Retina

RPE
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5) Diffuse choroidal hemangioma

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

Sturge-Weber syndrome (SWS)

Intraocular Tumors of Childhood

With what condition is the diffuse choroidal hemangioma associated?

Sturge-Weber syndrome (SWS)

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Sturge-Weber syndrome (SWS)

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What is the noneponymous name for SWS?

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Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Present in about 50% of patients with SWS. The coloration is a very red—much more so than an unaffected fundus. Fundus appearance in SWS is described as 'Tomato catsup fundus.' It is uncommon for SWS to be bilateral. The choroidal hemangioma does not have malignant potential.

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

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

**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?**

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**Intraocular Tumors of Childhood**

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

In a word, what condition is the diffuse choroidal hemangioma associated with? **Sturge-Weber syndrome (SWS)**

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

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Intraocular Tumors of Childhood

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

Nevus

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 what percent of SWS? About 50%

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

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

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

Does the choroidal hemangioma have malignant potential? No

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

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

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

Intraocular Tumors of Childhood

Sturge-Weber syndrome (SWS)
Intraocular Tumors of Childhood

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1) **Nevus**: Common. Benign

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

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**When does it present?**
At birth

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**Intraocular Tumors of Childhood**

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**Sturge-Weber syndrome (SWS)**

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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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**What does the fundus look like in an eye with a diffuse choroidal hemangioma?**
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Yes, but it's uncommon

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At birth
Intraocular Tumors of Childhood

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

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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

2. **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?*
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   *What food-related term is used to describe the fundus appearance in SWS?*
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   *Can the choroidal hemangioma be present bilaterally?*
   Yes, but it's uncommon

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   No

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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 is present in about 50% of Sturge-Weber syndrome (SWS). The coloration is a very red—much more so than an unaffected fundus. 'Tomato catsup fundus'. Yes, but it’s uncommon. No. A phakomatosis

What is the non-prenomous name for SWS?

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

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

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

An angioma

When does it present?

At birth

What is the typical pattern of distribution?

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

Does it always present in this manner?
Intraocular Tumors of Childhood

What condition is the diffuse choroidal hemangioma associated with?

Sturge-Weber syndrome (SWS)

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

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

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When does it present?
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Does it always present in this manner?
No, some cases will cross the midline of the face
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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Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Present in about 50% of SWS. The coloration is a very red—much more so than an unaffected fundus. This appearance is referred to as 'tomato catsup fundus'. The choroidal hemangioma can be present bilaterally, but it's uncommon. It does not have malignant potential. It is associated with Sturge-Weber syndrome (SWS), also known as encephalotrigeminal angiomatosis, encephalofacial angiomatosis, or cerebrofacial angiomatosis. The hallmark skin finding in SWS is the port-wine stain, which is an angioma that presents at birth. It comports to the distribution of one or more divisions of CN5. It does not always present in this manner; some cases will cross the midline of the face. All infants with SWS have a port-wine stain. Not all infants with a port-wine stain have SWS.
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Diffuse choroidal hemangioma: Sturge-Weber syndrome (SWS)

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

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

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

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

5) Diffuse choroidal hemangioma

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

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

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

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

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

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Intraocular Tumors of Childhood

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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**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

To summarize, the diffuse choroidal hemangioma is associated with Sturge-Weber syndrome (SWS). It is present in about half of the cases of SWS. The fundus appearance in an eye with a diffuse choroidal hemangioma is characterized by a very red coloration, much more so than an unaffected fundus.
Sturge-Weber: Tomato catsup fundus OD
Intracocular Tumors of Childhood

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

Does the choroidal hemangioma have malignant potential? No

Iris/Ciliary Body

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**Intracocular Tumors of Childhood**

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

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

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

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

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Diffuse choroidal hemangioma

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Can the choroidal hemangioma be present bilaterally?

Yes, but it’s uncommon

RPE

Retina
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**Intraocular Tumors of Childhood**

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

Sturge-Weber syndrome (SWS)

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

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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5) **Diffuse choroidal hemangioma**

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


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

4) Brushfield spots: Strong association with Down syndrome.

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

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

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Iris/Ciliary Body

Retina

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Yes, but it’s uncommon

Does the choroidal hemangioma have malignant potential?
No

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Stargardt disease
- Autosomal recessive choroidal hemangioma. Very rare. Characteristics a Scan pattern

5) Diffuse choroidal hemangioma
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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

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**Intraocular Tumors of Childhood**

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- No

---

**Diffuse choroidal hemangioma**

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
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Choroid

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RPE

1) ?

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Iris/Ciliary Body

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RPE

1) Congenital hypertrophy of the RPE (CHRPE)

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**Intraocular Tumors of Childhood**

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**Iris/Ciliary Body**

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

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**Retina**

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

- Flat, mainly black lesion(s) ranging in size from a 1 mm up to ~10
- Common
- It is neither a hamartoma or a choristoma

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**Choroid**

- On extremely rare males. Risk of CNVM a diagnostic a-scan pattern
- Diffuse choroidal hemangioma: Unilateral. Found in Sturge-Weber syndrome

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**RPE**

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

Retina

Choroid

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

RPE

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

CHRPE
## Intraocular Tumors of Childhood

### Iris/Ciliary Body

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### Choroid

**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

### Retina

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

### RPE

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Common

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

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**Retina**
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

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

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

Iris/Ciliary Body

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Choroid

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

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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RPE

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

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

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**Choroid**

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

- 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

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

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

CHRPE: Bear tracks
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

RPE

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

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

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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. The name of this syndrome is:

Familial adenomatous polyposis, aka Gardner syndrome

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

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

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

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

**CHRPE** is characterized according to its presentation. In what two ways does it present?
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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome?
Familial adenomatous polyposis, aka **Gardner syndrome**

What characteristics of a CHRPE-like presentation increase the likelihood that it is a component of Gardner syndrome?
-- If it is bi-v unilateral (regular CHRPE is almost always bi-v unilateral)
Intraocular Tumors of Childhood

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

**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)

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

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

--**Solitary** CHRPE

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

Iris/Ciliary Body

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

**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’ distribution pattern)

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**RPE**

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

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

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

Iris/Ciliary Body

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

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’)

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

CHRPE-like lesions of Gardner syndrome: Scattered distribution
Intraocular Tumors of Childhood

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

RPE
1) Congenital hypertrophy of the RPE (CHRPE)

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

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

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

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

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

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

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**Intraocular Tumors of Childhood**

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### Iris/Ciliary Body

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

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

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

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

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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

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

--If the shape of the lesions is pisciform

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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

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed→hyphema→increased IOP→glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells.
3) Lisch nodules: Strong association with NF1. Lighter on dark irides; darker on light
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

What does pisciform mean?
'Fish-shaped'

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

RPE

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

Iris/Ciliary Body

1) **Juvenile xanthogranuloma** (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed \( \rightarrow \) hyphema \( \rightarrow \) increased IOP \( \rightarrow \) glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
2) **Medulloepithelioma** (aka diktyoma): Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed \( \rightarrow \) hyphema \( \rightarrow \) increased IOP \( \rightarrow \) glaucoma. Locally invasive \( \rightarrow \) death. Tx: Enucleate
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A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. What is the name (both eponymous and noneponymous) of this syndrome? Familial adenomatous polyposis, aka **Gardner syndrome**

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

What does pisciform mean? It means ‘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
--**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

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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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’)

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

Iris/Ciliary Body

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Retina

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Choroid

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RPE
Intraocular Tumors of Childhood

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

--If it is bilateral (regular CHRPE is almost always unilateral)
--If the lesions are 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 vs hyperpigmented
--They point towards the optic nerve head

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**Iris/Ciliary Body**

**Retina**

**Choroid**

**Iris/Ciliary Body**

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

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

--If it is bilateral (regular CHRPE is almost always unilateral)
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**The tails of these fish-shaped lesions have two telltale (tell-tail?) characteristics--what are they?**

--They are hypopigmented vs hyperpigmented
--They point towards the optic nerve head
Intraocular Tumors of Childhood

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

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

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

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
CHRPE-like lesions of Gardner syndrome: Hypopigmented tail pointing toward ONH
**Intraocular Tumors of Childhood**

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

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

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

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

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

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

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

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

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

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

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

- **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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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 characteristic 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

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

RPE
Intraocular Tumors of Childhood

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
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 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

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

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)

Retina

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

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

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
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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

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

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

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

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

Intraocular Tumors of Childhood

**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

1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota
6) Iris cysts: Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

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

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

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

What is the treatment of choice? Prophylactic colectomy

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

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

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

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- If it is bilateral (regular CHRPE is almost always unilateral)
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- Benign tumors of the skin
- Benign tumors of bone
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Retina

1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

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


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**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?**
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**Retina**

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

- Muir-Torre syndrome
- Peutz Jeghers syndrome

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**Intraocular Tumors of Childhood**

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Intraocular Tumors of Childhood

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

RPE

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Iris/Ciliary Body

RPE

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**Intraocular Tumors of Childhood**

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

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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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**RPE**

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What is family history of? Congenital hypertrophy of the RPE
-- If it is **bilateral** (regular CHRPE is almost always unilateral)
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2) **Melanocytoma:** Usually juxtapapillary. Malignant transformation extremely rare

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

4) **Isolated/focal choroidal hemangioma:** Very rare. Characteristic

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

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?

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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

Iris/Ciliary Body

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What sorts of sebaceous lesions? --Basal-cell carcinomas with sebaceous differentiation

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What sorts of sebaceous lesions?

--Sebaceous-cell carcinomas
--Sebaceous-cell adenomas
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When 'colon cancer + ophthalmic issue' is mentioned, two syndromes should come to mind. One is Gardner syndrome. What is the other? **Muir-Torre syndrome** and **Hereditary Non-Polyposis Colorectal Cancer**

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

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**Iris/Ciliary Body**

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**RPE**

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**Retina**

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When "colon cancer + ophthalmic issue" is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? Muir-Torre syndrome and **Peutz-Jeghers syndrome**

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How is Peutz-Jeghers pronounced? Pyoots je·grz
Intraocular Tumors of Childhood

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When "colon cancer + ophthalmic issue" is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? **Peutz-Jeghers syndrome**

--- If it's **Muir-Torre syndrome** (regarding skin and eyes)

--- If it's **Peutz-Jeghers syndrome**

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

--- If the shape of the lesions is pisciform

How is Peutz-Jeghers pronounced? **Pyoots jeh-grz**
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 mamillations:** Tiny, numerous. Same color as iris. Weak association with NF1, Nevus of Ota

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**Iris/Ciliary Body**

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

When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? Muir-Torre syndrome and **Peutz-Jeghers syndrome**

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines of (but not necessarily limited to) the eyelids

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**Retina**

---

**Ciliary Body**

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**RPE**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Familial adenomatous polyposis, aka **Gardner syndrome**

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

Muir-Torre syndrome and **Peutz-Jeghers syndrome**

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?
Simple lentigines of (but not necessarily limited to) the eyelids
Intraocular Tumors of Childhood

Iris/Ciliary Body

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When "colon cancer + ophthalmic issue" is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? Muir-Torre syndrome and Peutz-Jeghers syndrome

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines

What are simple lentigines?

Flat melanocytic lesions histologically similar to ephelides

By what variant of the term 'simple lentigines' are they also known? 'Lentigo simplex'

Does lentigo simplex have malignant potential? No
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**

---

**When “colon cancer + ophthalmic issue” is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other?**

Muir-Torre syndrome and **Peutz-Jeghers syndrome**

---

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?

**Simple lentigines**

---

What are simple lentigines?

Flat melanocytic lesions histologically similar to ephelides

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**Intraocular Tumors of Childhood**

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**Iris/Ciliary Body**

---

**Retina**

---

**Choroid**

---

**RPE**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? Muir-Torre syndrome and Peutz-Jeghers syndrome

What is the main ophthalmic issue of Peutz-Jeghers syndrome? Simple lentigines. What are simple lentigines? Flat melanocytic lesions histologically similar to ephelides.
Intraocular Tumors of Childhood

Iris/Ciliary Body

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What are simple lentigines? Flat melanocytic lesions histologically similar to ephelides

What are ephelides (singular, ephelis)? Freckles
Intraocular Tumors of Childhood

Peutz-Jeghers syndrome: Eyelid simple lentigines
Intraocular Tumors of Childhood

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Muir-Torre syndrome and Peutz-Jeghers syndrome

What are simple lentigines?
Flat melanocytic lesions histologically similar to ephelides

By what variant of the term ‘simple lentigines’ are they also known?

' Lentigo simplex'
Intraocular Tumors of Childhood

Iris/Ciliary Body

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What are **simple lentigines?** Flat melanocytic lesions histologically similar to ephelides

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**Simple lentigines**

By what variant of the term ‘simple lentigines’ are they also known? ‘Lentigo simplex’
Intraocular Tumors of Childhood

Iris/Ciliary Body

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**Iris/Ciliary Body**

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What are simple lentigines? Flat melanocytic lesions histologically similar to ephelides

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Intraocular Tumors of Childhood

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Muir-Torre syndrome and **Peutz-Jeghers syndrome**

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What are simple lentigines? Flat melanocytic lesions histologically similar to ephelides

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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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

---

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

Muir-Torre syndrome and **Peutz-Jeghers syndrome**

---

What are simple lentigines?

- Flat melanocytic lesions histologically similar to ephelides
- By what variant of the term 'simple lentigines' are they also known? 'Lentigo simplex'

---

I coulda sworn lentigo simplex had malignant potential. You sure about this?

Yes, I’m sure. You’re thinking of a pre-malignant melanocytic lesion of the skin.

---

**Does lentigo simplex have malignant potential?**

No
Intraocular Tumors of Childhood

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- Muir-Torre syndrome
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What are simple lentigines?
Flat melanocytic lesions histologically similar to ephelides

By what variant of the term 'simple lentigines' are they also known? 'Lentigo simplex'

Does lentigo simplex have malignant potential?
No

I coulda sworn lentigo simplex had malignant potential. You sure about this?
Yes, I'm sure. You're thinking of lentigo maligna, a pre-malignant melanocytic lesion of the skin.
Intraocular Tumors of Childhood

Iris/Ciliary Body

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When ‘colon cancer + ophthalmic issue’ is mentioned, three syndromes should come to mind. One is Gardner syndrome. What is the other? Muir-Torre syndrome and Peutz-Jeghers syndrome

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines of (but not necessarily limited to) the eyelids

Are lentigo simplex eyelid lesions the classic harbinger of Peutz-Jehgers syndrome? Yes

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

Retina

1) Congenital hypertrophy of the RPE (CHRPE): Large lesion(s) surrounded by a few smaller ones

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

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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 main ophthalmic manifestation of Peutz-Jeghers syndrome? **Simple lentigines** of (but not necessarily limited to) the eyelids

Are lentigo simplex eyelid lesions the classic harbinger of Peutz-Jeghers syndrome? No, pigmented lesions of the **perioral** region are the classic/most common finding

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
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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What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? **Simple lentigines** of (but not necessarily limited to) the eyelids

Are lentigo simplex eyelid lesions the classic harbinger of Peutz-Jeghers syndrome? No, pigmented lesions of the perioral region are the classic/most common finding
Intraocular Tumors of Childhood

Characteristic circumoral pigmentation in a patient with Peutz-Jeghers syndrome
Intraocular Tumors of Childhood

Speaking of: Did you notice the pigmented lip lesions in this pic?
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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**

**Gardner syndrome**

- SOLITARY CHRPE
- MULTIFOCAL or GROUPED CHRPE: Large lesion(s) surrounded by a few smaller ones

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

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**Peutz-Jeghers syndrome**

**What is the main ophthalmic manifestation of Peutz-Jeghers syndrome?**

Simple lentigines of (but not necessarily limited to) the eyelids

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**Does Peutz-Jeghers present with multiple adenomatous polyps of the colon a la Gardner syndrome?**

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Intraocular Tumors of Childhood

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

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

What is the main ophthalmic manifestation of Peutz-Jeghers syndrome? Simple lentigines of (but not necessarily limited to) the eyelids

Does Peutz-Jeghers present with multiple adenomatous polyps of the colon a la Gardner syndrome? Yes

--Congenital 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 $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells
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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

Iris/Ciliary Body

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

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

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

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Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

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**Retina**

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

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**Intraocular Tumors of Childhood**

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Iris/Ciliary Body

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- Combined hamartoma of the retina and RPE

- 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?

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

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RPE
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3) Combined hamartoma of the retina and RPE

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

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

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Retina

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

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RPE

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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

If this image is a question, it might be asking about the clinical characteristics or diagnosis of various intraocular tumors. The text provides a detailed overview of different conditions, their symptoms, and treatments, with a focus on juvenile xanthogranuloma, medulloepithelioma, and other associated conditions such as NF1 and Down syndrome. The diagram highlights the relationship between the RPE and retina, with notes on various hamartomas and choristomas, as well as other tumors and conditions. The text is a comprehensive resource for understanding the various intraocular tumors of childhood and their clinical implications.
Intraocular Tumors of Childhood

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

What is the reverse of that?
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RPE

Retina

1) Combined hamartoma of the retina and RPE

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

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### Intraocular Tumors of Childhood

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

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

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RPE cells (duh) and retinal glial cells
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**How does it present clinically?**

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**RPE**

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**Iris/Ciliary Body**

**Choroid**

**RPE**

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**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

**How does it present clinically?**
As a variably pigmented, slightly elevated retinal mass of the retina

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RPE cells (duh) and retinal glial cells

How does it present clinically?
As a variably pigmented, slightly elevated retinal mass of the peripapillary retina

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

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Intraocular Tumors of Childhood

Combined hamartoma of retina and RPE
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How does it present clinically?
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With what more sinister dz entity is it often confused?

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

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

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

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

4) Brushfield spots: Strong association with Down syndrome

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

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

1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

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


1) 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?**

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

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**RPE**

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

**Retina**

1) Combined hamartoma of the retina and RPE

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

Retina

RPE

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

With what more sinister dz entity is it often confused?

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

How can one avoid making such a disastrous mistake?

By taking pains to carefully determine the anatomic location of the tumor in question--choroidal melanomas originate behind Bruch’s membrane, whereas combined hamartomas of the retina and RPE are located wholly in front of it

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1) Combined hamartoma of the retina and RPE

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

Choroid

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

1) 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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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) **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