Before you begin: This is a big topic, and big topics beget big slide-sets. There’s a natural break near the halfway mark (slide 213); I placed a \textit{break time!} slide at that point to mark it.
Intraocular Tumors of Childhood

Four intraocular locations (i.e., structures/tissues)
Intraocular Tumors of Childhood

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

1) ?

2) ?

Iris/Ciliary Body

3) ?

4) ?

5) ?

6) ?

Six tumors of the iris/ciliary body

Choroid

RPE

Retina
Intraocular Tumors of Childhood

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

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

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

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

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

6) **Iris cysts**

**Six tumors of the iris/ciliary body**
1) **Juvenile xanthogranuloma**

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

It is a...
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

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Iris/Ciliary Body

- **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.


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

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

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

---

In three words, what sort of condition is JXG?

It is a... **nonneoplastic histiocytic proliferation**
1) **Juvenile xanthogranuloma**

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

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

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Intraocular Tumors of Childhood

**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- In three words, what sort of condition is JXG?
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- How does JXG usually present? (Hint: It’s not ophthalmic)
  As orangish skin papules

- 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
Intraocular Tumors of Childhood

JXG: Skin papules. The orangish color is classic
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Iris/Ciliary Body

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Intraocular Tumors of Childhood

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#### Iris/Ciliary Body

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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Intraocular Tumors of Childhood

JXG: Iris lesion
Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

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Choroid

RPE

Retina
JXG: Spontaneous hyphema
Intraocular Tumors of Childhood

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

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

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

Should JXG nodules be removed surgically? Only if the glaucoma is uncontrollable.
1) **Juvenile xanthogranuloma**

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

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2) Medulloepithelioma

3) Lisch nodules

4) Brushfield spots

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Intraocular Tumors of Childhood

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   What are the two hallmarks of JXG histology?
   The presence of **Touton giant cells**
   The presence of...
   (we’ll come back to this one)

   What is the natural history of JXG?
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What are the two hallmarks of JXG histology?
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**What is a giant cell?**
It is an aggregate of histiocytes. (The formal term for the aggregate is syncytium). Put simply, it is a bunch of histiocytes that have glommed together.
# Intraocular Tumors of Childhood

## Iris/Ciliary Body

1) **Juvenile xanthogranuloma**

- **In three words, what sort of condition is JXG?** Nonneoplastic histiocytic proliferation

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

- **At what age does it present?**
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What is a giant cell?
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What is the histologic hallmark of a giant cell?
It is multinucleated—the myriad nuclei of the involved histiocytes are all visible within it
Intraocular Tumors of Childhood

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RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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It is multinucleated—the myriad nuclei of the involved histiocytes are all visible within it.

Retina

C

RPE
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.

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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

JXG: Touton giant cells
Iris/Ciliary Body

1) **Juvenile xanthogranuloma**

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Histocytes are also known as...?
Macrophages

Histiocytes derive from what type of cell?
Monocytes

What does derive mean here? How does a monocyte become a histiocyte?
Monocytes are found within the vasculature. When a monocyte leaves the circulation and settles down within tissue, it becomes (ie, is re-classified) as a histiocyte.
1) **Juvenile xanthogranuloma**

- **In three words, what sort of condition is JXG?**
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  - The presence of... *giant cells*

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

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- The presence of histiocytes

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*What is the natural history of JXG?*
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- Benign but locally aggressive neoplasia of nonpigmented epithelium of CB
- Presents: Iris mass before age 10 years
- Can bleed → hyphema → increased IOP → glaucoma
- Locally invasive → death
- Tx: Enucleate

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

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Unilateral.

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

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

Choroid

RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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 Touton giant cells
   - The presence of ‘foamy macrophages’

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

Choroid

RPE

Retina
Intraocular Tumors of Childhood

Iris/Ciliary Body

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  - Foamy = ‘lipid filled’
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The presence of Touton giant cells.
The presence of 'foamy macrophages'.

This histology—'foamy macrophages'—is often described with other, equivalent terms. What are they?
Foamy = 'lipid filled'
Macrophages = ? (You mos def know this one)

If enough nodules are present, 'heterochromia iridis' will result.

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

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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

1) **Juvenile xanthogranuloma**

Retina

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RPE
Intraocular Tumors of Childhood

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## Intraocular Tumors of Childhood

### Iris/Ciliary Body

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   *What are the two hallmarks of JXG histology?*
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   *When are JXG iris nodules typically...*
   *...uni-, or bilateral?*
   Unilateral

   *In what three ways are the iris nodules clinically significant?*
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   *What are the two hallmarks of JXG histology?*
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   The presence of **‘lipid filled’ macrophages**

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

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### Retina

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Intraocular Tumors of Childhood

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

Speaking of ‘foamy macrophages’...
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis? First clue—more forthcoming
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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Speaking of 'foamy macrophages'...
What disease comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?
And a hx of chronic migratory arthritis?

Retina
Intraocular Tumors of Childhood

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What disease comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?
And a hx of chronic migratory arthritis?
Associated with chronic diarrhea?

Need another?

Choroid

Retina
**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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

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

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

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

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

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

**Choroid**

**Retina**
Intraocular Tumors of Childhood

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  Associated with chronic diarrhea?
  And CNS symptoms--seizures, dementia, coma?
  Whipple’s disease

Retina
Intraocular Tumors of Childhood

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

Whipple’s disease

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

It is infectious.

The bacterium Tropheryma whipplei.
1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG?
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It is self-limited, usually resolving by age 5 years

Speaking of ‘foamy macrophages’…

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

Whipple’s disease

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

The bacterium Tropheryma whipplei

What infection agent is responsible for Whipple’s?

The presence of... ‘foamy macrophages’
1) Juvenile xanthogranuloma

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

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

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

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

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

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

Speaking of ‘foamy macrophages’...
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a middle-aged white guy with bilateral panuveitis?
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Whipple’s disease

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

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

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

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

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

Speaking of ‘foamy macrophages’...

What dz comes to mind if, instead of a young child with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules, the pt in question was a middle-aged white guy with iris nodules...

Whipple’s disease

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

What infection agent is responsible for Whipple’s?
The bacterium Tropheryma whippelii

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

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

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

2) Medulloepithelioma

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

3) Lisch nodules

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

4) Brushfield spots

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

5) Iris mammillations

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

6) Iris cysts

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

1) Juvenile xanthogranuloma

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

2) Medulloepithelioma

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

3) Lisch nodules

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

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- Can be pupillary, stromal, secondary.

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

Speaking of ‘foamy macrophages’...

What disease comes to mind if, instead of a young child with iris nodules, the patient 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.

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

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

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

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 nodule usually presents 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'

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

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

Whipple’s disease

 Speaking of 'foamy macrophages'...

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

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

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What infection agent is responsible for Whipple’s?
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Retina

1) Juvenile xanthogranuloma

1) Juvenile xanthogranuloma

2) Medulloepithelioma

3) Lisch nodules

4) Brushfield spots

5) Iris mammillations

6) Iris cysts

Intraocular Tumors of Childhood

Iris/Ciliary Body

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

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

What infection agent is responsible for Whipple’s?
The bacterium Tropheryma whipplei

Retina

1) Juvenile xanthogranuloma

1) Juvenile xanthogranuloma

2) Medulloepithelioma

3) Lisch nodules

4) Brushfield spots

5) Iris mammillations

6) Iris cysts

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)
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The presence of 'foamy macrophages'

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What other finding will a duodenal biopsy reveal?
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Whipple’s disease

 Speaking of 'foamy macrophages'...

When foamy macrophages are found in a biopsy performed on a Whipple’s pt, from what site was the biopsy collected?
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What other finding will a duodenal biopsy reveal?
The presence of acid-fast bacteria within macrophages located in intestinal villi

Speaking of 'foamy macrophages'...

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

Whipple’s disease

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

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'

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

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

Broadly speaking, what sort of condition is Whipple's?
It is an infection

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.

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

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

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

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

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

  Whipple’s disease.

Whipple’s disease

- Broadly speaking, what sort of condition is Whipple’s?
  It is an infection.

- What infection agent is responsible for Whipple’s?
  The bacterium Tropheryma whippelii.

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

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

- In three words, what sort of condition is JXG?
Small-intestine biopsy stained with periodic acid-Schiff. Note the numerous macrophages in the lamina propria (arrows).
Intraocular Tumors of Childhood

Iris/Ciliary Body

1) Juvenile xanthogranuloma

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

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

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

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'.

For more on Whipple’s dz, see slide-set U24

Choroid

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, a hx of chronic migratory arthritis, associated with chronic diarrhea, and CNS symptoms—seizures, dementia, coma?
Whipple’s disease.

Retina

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

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

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

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

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

Speaking of foamy macrophages part deaux…
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions?
(Pic forthcoming—give the dx after seeing it)

The presence of Touton giant cells 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?
If is self-limited, usually resolving by age 5 years.
Intraocular Tumors of Childhood

*Condition?*
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

In three words, what sort of condition is JXG?
Nonneoplastic histiocytic proliferation

Speaking of foamy macrophages part deaux…

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

Xanthelasma

The presence of ‘Touton giant cells’
The presence of ‘foamy macrophages’
The presence of ‘lipid-laden cells’

When
The presence in the choroid is unilateral?
Unilateral

In what three ways are the iris nodules clinically significant?

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

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

When
The presence of nodules is bilateral?
Bilateral

Intraocular Tumors of Childhood

RPE

Retina

Choroid
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

Are xanthelasmas a harbinger of elevated serum lipids?

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

Intraocular Tumors of Childhood

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

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

Are xanthelasmas a harbinger of elevated serum lipids?

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

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

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

Intraocular Tumors of Childhood

Choroid

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

RPE

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

Retina

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

1) Juvenile xanthogranuloma

+/- 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.

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

Speaking of foamy macrophages part deaux...

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

1) Juvenile xanthogranuloma

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

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

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

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

- Intraocular Tumors of Childhood

- The presence of ‘Touton giant cells’ unilateral/lateral?
  - Unilateral

- ‘foamy macrophages’

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

- Choroid

- RPE

- Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- In three words, what sort of condition is JXG? Nonneoplastic histiocytic proliferation.
- Speaking of foamy macrophages part deaux...
- What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was a 
Adult with bilateral upper-lid yellow lesions?
- Xanthelasmas
- The presence of 'Touton giant cells' is characteristic of JXG.
- Are xanthelasmas a harbinger of elevated serum lipids?
  - Yes, and when they are, they usually are a sign of lipid derangement.
- Can they be congenital?
  - Yes, and when they are, they usually are a sign of lipid derangement.

2) Medulloepithelioma

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

3) Lisch nodules

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

4) Brushfield spots

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

5) Iris mammillations

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

6) Iris cysts

- Can be pupillary, stromal, secondary.

In three words, what sort of condition is JXG?

- It is a...nonneoplastic histiocytic proliferation.

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

- As orangish skin papules.

At what age does it present?

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

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

- Unilateral.

In what three ways are the iris nodules clinically significant?

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

What is the natural history of JXG?

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

What are the two hallmarks of JXG histology?

- The presence of Touton giant cells.
- The presence of 'foamy macrophages'.
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

The presence of 'Touton giant cells'
When...? The presence...? Unilateral?

Speaking of foamy macrophages part deaux deaux…
What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide)

(No question yet—advance to the pic)
Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux deaux...

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

Xanthelsasma

Speaking of foamy macrophages part deaux deaux...

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

No other issues whatsoever?

Give the diagnosis
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux deaux...

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

Xanthelsasma

Speaking of foamy macrophages part deaux deaux...

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

Retina


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

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

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

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

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

nonneoplastic histiocytic proliferation

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

As orangish skin papules

At what age does it present?

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

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

Unilateral

In what three ways are the iris nodules clinically significant?

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

What is the natural history of JXG?

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

What are the two hallmarks of JXG histology?

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

Speaking of foamy macrophages part deaux deaux...

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

Speaking of foamy macrophages part deaux deaux...

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and: No other issues whatsoever? Adult-onset xanthogranuloma
1) Juvenile xanthogranuloma

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

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

The presence of 'Touton' giant cells
When: The presence of 'foamy macrophages'
Unilateral?

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

**Ditto**

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

The presence of ‘Touton giant cells’? Unilateral?

When: The presence of ‘foamy macrophages’?

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

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

Xanthelsasma

The presence of Touton giant cells
When The presence of 'foamy macrophages'
Unilateral? Bilateral?

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

Retina
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

When The presence of 'Touton giant cells'
Unilateral?

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

Retina

1) Juvenile xanthogranuloma

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; 15% of non-Down pop.

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

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

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

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

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

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

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

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

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

Speaking of foamy macrophages part deaux deaux...

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

When The presence of 'Touton giant cells'
Unilateral?

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

Necrobiotic xanthogranuloma
1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux deaux...

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

**Xanthelsasma**

When The presence of 'Touton giant cell' unilateral? Unilateral

The presence of 'foamy macrophages'

Speaking of foamy macrophages part deaux deaux...

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

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

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

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

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

Last one
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

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

The presence of 'foamy macrophages'
When? Unilateral?

Speaking of foamy macrophages part deaux deaux…
What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:
No other issues whatsoever? Adult-onset xanthogranuloma
Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma
The ‘xanthelasma’ are ulcerated? Necrobiotic xanthogranuloma
Proptosis and/or terrible systemic symptoms are present? Erdheim-Chester dz

Retina
Intraocular Tumors of Childhood

Erdheim-Chester disease
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux deaux...

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

Collectively, these conditions are known as the... Adrenalphobia

Speaking of foamy macrophages part deaux deaux...

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

Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated? Necrobiotic xanthogranuloma

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

--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'.

Intraocular Tumors of Childhood

Retina
Iris/Ciliary Body

Choroid

RPE

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux...

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

Xanthelsasma

Collectively, these conditions are known as the...

Adult xanthogranulomas

Speaking of foamy macrophages part deaux deaux...

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

No other issues whatsoever?

Adult-onset xanthogranuloma

Recently diagnosed asthma?

Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated?

Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present?

Erdheim-Chester dz

Retina

1) Juvenile xanthogranuloma

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


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

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

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

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

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

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

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

The presence of 'foamy macrophages'

Speaking of foamy macrophages part deaux deaux...

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

No other issues whatsoever?

Adult-onset xanthogranuloma

Recently diagnosed asthma?

Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated?

Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present?

Erdheim-Chester dz

Speaking of foamy macrophages part deaux deaux...

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

Xanthelsasma

Collectively, these conditions are known as the...

Adult xanthogranulomas

Speaking of foamy macrophages part deaux deaux...

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

No other issues whatsoever?

Adult-onset xanthogranuloma

Recently diagnosed asthma?

Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated?

Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present?

Erdheim-Chester dz

Speaking of foamy macrophages part deaux deaux...

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

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux…

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

**Xanthelasma**

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

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

- No other issues whatsoever?
- Recently diagnosed asthma?
- The ‘xanthelasma’ are ulcerated?
- Proptosis and/or terrible systemic symptoms are present?

**Adult xanthogranulomas**

Adult-onset xanthogranuloma

Adult-onset asthma with periocular xanthogranuloma

Necrobiotic xanthogranuloma

Erdheim-Chester dz

Collectively, these conditions are known as the...

**Adult xanthogranulomas**

In three words, what sort of condition is JXG?

It is a 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.

2) Medulloepithelioma

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

3) Lisch nodules

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

4) Brushfield spots

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

5) Iris mammillations

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

6) Iris cysts

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

1) Juvenile xanthogranuloma

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

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

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

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

Adult-onset xanthogranuloma
Adult-onset asthma with periocular xanthogranuloma
Necrobiotic xanthogranuloma
Erdheim-Chester dz...
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux...

What dz comes to mind if, instead of a toddler with JXG, the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:

- No other issues whatsoever?
- Recently diagnosed asthma?
- The 'xanthelasma' are ulcerated?
- Proptosis and/or terrible systemic symptoms are present?

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

Xanthelsasma

‘Nonneoplastic histiocytic proliferations’...Why does that sound so familiar?

The presence of 'Touton giant cells'

‘foamy macrophages’

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

They are...nonneoplastic histiocytic proliferations.

When the presence of 'Touton giant cells' noted? Bilateral?

Where the presence of 'foamy macrophages' noted? Unilateral?

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

Adult-onset xanthogranuloma

Adult-onset asthma with periocular xanthogranuloma

Necrobiotic xanthogranuloma

Erdheim-Chester dz

Path: Touton giant cells.

Patients usually present with: Iris nodules bleed...hyphema...increased IOP...glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP.

1) Juvenile xanthogranuloma

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

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

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

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

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

1) Juvenile xanthogranuloma

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

Speaking of foamy macrophages part deaux…

What dz comes to mind if, instead of a toddler with JXG or any other dz, it were the adult who had what could only be described as crazy-bad xanthelasma (pic next slide), and:

- No other issues whatsoever?
- Recently diagnosed asthma?
- The ‘xanthelasma’ are ulcerated?
- Proptosis and/or terrible systemic symptoms are present?

Nonneoplastic histiocytic proliferations’…Why does that sound so familiar? Because that’s the phrase we used to describe/define JXG

Collectively, these conditions are known as the… **Adult xanthogranulomas**

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

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

‘Nonneoplastic histiocytic proliferations’…Why does that sound so familiar? Because that’s the phrase we used to describe/define JXG

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

How does JXG usually present? As orangish skin papules.

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

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

In what three ways are the iris nodules clinically significant? 1. They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma. 2. They are in the DDX as a 'masquerade syndrome' in pediatric uveitis. 3. If enough nodules are present, heterochromia iridis will result.

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

Intraocular Tumors of Childhood

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


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

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.

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

Speaking of foamy macrophages part deaux deaux…

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and: No other issues whatsoever? Adult-onset xanthogranuloma. Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma. The 'xanthelasma' are ulcerated? Necrobiotic xanthogranuloma. Proptosis and/or terrible systemic symptoms are present? Erdheim-Chester dz.

Collectively, these conditions are known as the... Adult xanthogranulomas.
In three words, what sort of condition is JXG? It is a nonneoplastic histiocytic proliferation.

How does JXG usually present? As orangish skin papules.

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

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

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

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

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

Speaking of foamy macrophages part deaux deaux…

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and:
- No other issues whatsoever?
- Recently diagnosed asthma?
- Adult-onset asthma with periocular xanthogranuloma?
- The 'xanthelasma' are ulcerated?
- Proptosis and/or terrible systemic symptoms are present?
- Erdheim-Chester dz

Collectively, these conditions are known as the adult xanthogranulomas.
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 pediatric 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'.

Collectively, these conditions are known as the Adult xanthogranulomas.

Because that's the phrase we used to describe/define JXG.

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

What if the adult has what could only be described as crazy-bad xanthelasma (pic next slide), and: No other issues whatsoever? Adult-onset xanthogranuloma. Recently diagnosed asthma? Adult-onset asthma with periocular xanthogranuloma. The 'xanthelasma' are ulcerated? Necrobiotic xanthogranuloma. Proptosis and/or terrible systemic symptoms are present? Erdheim-Chester dz.

Speaking of foamy macrophages part deaux deaux… What if, instead of a toddler with iris nodules, the pt in question was a Adult with bilateral upper-lid yellow lesions? Xanthelasma.
In three words, what sort of condition is JXG?
It is a nonneoplastic histiocytic proliferation.

How does JXG usually present?
As orangish skin papules.

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

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

In what three ways are the iris nodules clinically significant?
-- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma.
-- They are in the differential diagnosis (DDx) as a 'masquerade syndrome' in pediatric 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.

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 neurofibromatosis type 1. Lighter on dark irides; darker on light.

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

5) Iris mammillations: Tiny, numerous. Same color as iris. Weak association with neurofibromatosis type 1, Nevus of Ota.

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

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

Speaking of foamy macrophages part deaux deaux...

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

Recently diagnosed asthma?
Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated?
Necrobiotic xanthogranuloma

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

Speaking of foamy macrophages part deaux deaux...

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

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

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

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

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

Recently diagnosed asthma?
Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated?
Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present?
Erdheim-Chester disease
In three words, what sort of condition is JXG?
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How does JXG usually present?
As orangish skin papules.

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

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

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

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

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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

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

2. **Medulloepithelioma:** 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.

---

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

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Speaking of foamy macrophages part deaux deaux…

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

- No other issues whatsoever?
- Recently diagnosed asthma?
- Adult-onset xanthogranuloma
- The 'xanthelasma' are ulcerated?
- Proptosis and/or terrible systemic symptoms are present?

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

**Adult-onset xanthogranuloma**
**Adult-onset asthma with periocular xanthogranuloma**
**Necrobiotic xanthogranuloma**
**Erdheim-Chester dz**
In three words, what sort of condition is JXG?

Nonneoplastic histiocytic proliferation

How does JXG usually present?

As orangish skin papules

At what age does it present?

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

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

Unilateral

In what three ways are the iris nodules clinically significant?

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

What is the natural history of JXG?

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

Collectively, these conditions are known as the...

Juvenile xanthogranuloma

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; 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.

What are the two hallmarks of JXG histology?

The presence of Touton giant cells

The presence of 'foamy macrophages'

Speaking of foamy macrophages part deaux deaux…

What if the adult has what could only be described as crazy-bad xanthelasma (pi?

No other issues whatsoever?

Adult-onset xanthogranuloma

Recently diagnosed asthma?

Adult-onset asthma with periocular xanthogranuloma

The 'xanthelasma' are ulcerated?: Necrobiotic xanthogranuloma

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

Collectively, these conditions are known as the...

Adult xanthogranulomas

Non-Langerhans cell histiocytoses

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

They are nonneoplastic histiocytic proliferations

What if the adult has what could only be described as crazy bad xanthelasma (pi?

Non-Langerhans cell histiocytoses

Adult-onset xanthogranuloma

Adult-onset asthma with periocular xanthogranuloma

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

JXG

No question—summary slide, advance when ready
Intraocular Tumors of Childhood

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

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

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

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

Do the AXGs have Touton giant cells like their juvenile cousin?
Intraocular Tumors of Childhood

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

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

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

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

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

Nonneoplastic histiocytic proliferations...Why does that sound so familiar?
Because that's the phrase we used to describe/define JXG.

What if the adult has what could only be described as crazy-bad xanthelasma (papular xanthoma)?
Adult-onset xanthogranuloma
Adult-onset asthma with periocular xanthogranuloma
Proptosis and/or terrible systemic symptoms are present? Erdheim-Chester dz

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

What is the nature of this relationship?
They (JXG and the AXGs) together comprise the non-Langerhans cell histiocytoses.
In three words, what sort of condition is JXG? It is a nonneoplastic histiocytic proliferation.

How does JXG usually present? As orangish skin papules. At what age does it present? The majority before age 1 year, and almost all by 2 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.

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

Intraocular Tumors of Childhood

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

It is a... nonneoplastic histiocytic proliferation

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

As orangish skin papules

At what age does it present?

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

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

Unilateral

In what three ways are the iris nodules clinically significant?

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

What is the natural history of JXG?

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

Collectively, these conditions are known as the... 

Juvenile xanthogranuloma

Iris/Ciliary Body

Choroid

RPE

Intraocular Tumors of Childhood

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

2) Medulloepithelioma: Benign but locally aggressive neoplasia of nonpigmented epithelium of CB. Presents: Iris mass before age 10 years. Can bleed \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.

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What are the two hallmarks of JXG histology?

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Adult-onset xanthogranuloma

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The 'xanthelasma' are ulcerated?

Necrobiotic xanthogranuloma

Proptosis and/or terrible systemic symptoms are present?

Erdheim-Chester dz

Collectively, these conditions are known as the... 

Adult xanthogranulomas

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

Non-Langerhans cell histiocytes
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three…

What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass? (Clue 1 of 2)

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?
If self-limited, usually resolving by age 5 years

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- Nonneoplastic histiocytic proliferation
- Presents: Iris mass before age 10 years
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Self-limited; regresses by age 5
- Treat inflammation and IOP
- Path: Touton giant cells

2) Medulloepithelioma

- Benign but locally aggressive neoplasia of nonpigmented epithelium of CB
- Presents: Iris mass before age 10 years
- Iris nodules bleed → hyphema → increased IOP → glaucoma
- Locally invasive → death
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6) Iris cysts

- Can be pupillary, stromal, secondary

In three words, what sort of condition is JXG?

- It is a…

- nonneoplastic histiocytic proliferation

How does JXG usually present?

- 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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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’
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …
What dz comes to mind if, instead of a toddler with iris nodules, the pt in question was an 8 y.o. with a superotemporal orbital mass?
Imaging → soft tissue mass + lytic lesions?  

Clue 1 of 2

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

In what three ways are the iris nodules clinically significant?
-- They are prone to spontaneous bleeding, with subsequent hyphema and secondary glaucoma
-- They are in the DDx as a ‘masquerade syndrome’ in peds uveitis
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What are the two hallmarks of JXG histology?
The presence of Touton giant cells
The presence of ‘foamy macrophages’

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

Clue 1 of 2

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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Clue 1 of 2

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Clue 1 of 2

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Clue 1 of 2

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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-- 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’
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

2) Medulloepithelioma

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

3) Lisch nodules

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

4) Brushfield spots

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

5) Iris mammillations

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

6) Iris cysts

- Can be pupillary, stromal, secondary

In three words, what sort of condition is JXG?

It is a…

nonneoplastic histiocytic proliferation

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

As orangish skin papules

At what age does it present?

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

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

Unilateral

In what three ways are the iris nodules clinically significant?

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What is the natural history of JXG?

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

Speaking of foamy macrophages part three …

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

Imaging → soft tissue mass + lytic lesions?

Langerhans-cell histiocytosis (and there it is!)
Iris/Ciliary Body

Choroid

RPE

Retina

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …

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

Imaging → soft tissue mass + lytic lesions?

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

Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …

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

Imaging → soft tissue mass + lytic lesions?

For more on the Langerhans (and non-Langerhans) histiocytoses, see slide-set K20
1) **Juvenile xanthogranuloma**

*Speaking of foamy macrophages part three …*

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

*Imaging → soft tissue mass + lytic lesions?*  
**Langerhans-cell histiocytosis** (and there it is!)

- **Histology:***
  - The presence of **Touton giant cells**
  - The presence of 'foamy macrophages'

**Clue #1**

- **Imaging:***
  - Soft tissue mass + lytic lesions?
  - Langerhans-cell histiocytosis

---

**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.

2) **Medulloepithelioma:**

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

3) **Lisch nodules:**

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

4) **Brushfield spots:**

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

5) **Iris mammillations:**

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

6) **Iris cysts:**

- Can be pupillary, stromal, secondary.

In three words, what sort of condition is JXG?

*It is a…*

**nonneoplastic histiocytic proliferation**

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

*As orangish skin papules*
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

2) Medulloepithelioma

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

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Can be pupillary, stromal, secondary.

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

nonneoplastic histiocytic proliferation

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

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

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

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

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

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

Speaking of foamy macrophages part whatever …
What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?
No family hx for anything like this? Clue #2
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.

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

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

- Nonneoplastic histiocytic proliferation

How does JXG usually present?

As orangish skin papules

At what age does it present?

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

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

Unilateral

In what three ways are the iris nodules clinically significant?

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

What is the natural history of JXG?

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

What are the two hallmarks of JXG histology?

The presence of Touton giant cells

The presence of ‘foamy macrophages’

Speaking of foamy macrophages part three …

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

Langerhans-cell histiocytosis (and there it is!)

Speaking of foamy macrophages part whatever …

What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria? No family hx for anything like this?

DFE → Exudative RD? ← Clue #3
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

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

2) Medulloepithelioma

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

Speaking of foamy macrophages part three ...

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

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

Speaking of foamy macrophages part whatever ...

What dz comes to mind if the pt in question was a 6 y.o. boy with unilateral leukocoria?
No family hx for anything like this?

DFE → Exudative RD?
DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?

Clue last
Ira/Ciliary Body

Choroid

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …

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

Imaging → soft tissue mass ± lytic lesions?

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What are the two hallmarks of JXG histology?

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’foamy macrophages’

Speaking of foamy macrophages part whatever …

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

No family hx for anything like this?

DFE → Exudative RD?

DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?

Coats dz

Retina
Intraocular Tumors of Childhood

Exudates; MAs; telangiectasias
Exudative RD (note also the vasc abnormalities)

Coats disease
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

- Nonneoplastic histiocytic proliferation
- Present in <2 years old
- +/- skin papules
- Iris nodules bleed → hyphema → increased IOP → glaucoma
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In three words, what sort of condition is JXG?

- It is a nonneoplastic histiocytic proliferation

How does JXG usually present?

- As orangish skin papules

At what age does it present?

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

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

What is the natural history of JXG?

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

What are the two hallmarks of JXG histology?

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

Speaking of foamy macrophages part three …

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

- Imaging → soft tissue mass + lytic lesions
- Langerhans-cell histiocytosis (and there it is!)

Speaking of foamy macrophages part whatever …

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

- No family hx for anything like this?
- DFE → Exudative RD?
- DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?
- Coats dz

Where are the foamy macrophages found in Coats dz?
1) Juvenile xanthogranuloma

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

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- 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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**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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**Unilateral**

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

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

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

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

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

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

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

**Speaking of foamy macrophages part whatever ...**

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

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

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

**Coats dz**

**Where are the foamy macrophages found in Coats dz?**

**In the subretinal exudate**
Intraocular Tumors of Childhood

1) Juvenile xanthogranuloma

Speaking of foamy macrophages part three …

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

**Imaging → soft tissue mass + lytic lesions?**

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

---

What are the two hallmarks of JXG histology?

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

---

Speaking of foamy macrophages part whatever …

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

**No family hx for anything like this?**

**DFE → Exudative RD?**

**DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?**

**Coats dz**

*Where are the foamy macrophages found in Coats dz?*

In the subretinal exudate (*also present: cholesterol crystals*)

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Retina

Choroid
1) Juvenile xanthogranuloma

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**DFE → Exudative RD?**

**DFE also → Retinal vascular microaneurysms, telangiectasias, dilatation?**

**Coats dz**

Where are the foamy macrophages found in Coats dz?

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

2) Medulloepithelioma

What is the other name by which medulloepithelioma is known?

Iris/Ciliary Body
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2) **Medulloepithelioma**

**Iris/Ciliary Body**

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

**Retina**

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**RPE**
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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?

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

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

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*How does it present?*

Choroid

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*How does it present?*
As an iris mass

**Choroid**

**RPE**

**Retina**
Intraocular Tumors of Childhood

Medulloepithelioma/diktyoma
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As an iris mass along with one or more of the following:
--?
--?
--?

**Choroid**

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Is it common, or rare?
Very rare

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

How is it managed?
Enucleation is usually required
Intraocular Tumors of Childhood

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Is a tendency to metastasize one of its ‘malignant features’?

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*Is a tendency to metastasize one of its ‘malignant features’?*
- No, this lesion rarely metastasizes; it does its damage locally

*Intraocular Tumors of Childhood*
Intraocular Tumors of Childhood

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

3) **Lisch nodules**

4) **Brushfield spots**

---

**Lisch nodules are most strongly associated with what congenital condition?**
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Iris/Ciliary Body

Retina

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NF1
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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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NF1

In this context, what does NF1 stand for?

**Retina**

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**Intraocular Tumors of Childhood**

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

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

Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

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

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

Iris/Ciliary Body

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**Iris/Ciliary Body**

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**In this context, what does NF1 stand for?**
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**In a word, what sort of condition is it?**

A phakomatosis

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A phakomatosis

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

Retina
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3) Lisch nodules

4) Brushfield spots

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

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

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

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**Iris/Ciliary Body**

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Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

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

Intraocular Tumors of Childhood

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

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**Intraocular Tumors of Childhood**

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

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

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

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

Melanocytic lesions are of no clinical significance beyond establishing the diagnosis, whereas the neuroglial lesions are associated with significant ocular and/or systemic morbidity.
Intraocular Tumors of Childhood

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

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

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

**NF1**

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

Yes

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

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5) **Iris mammillations**

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

**Down syndrome**

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

At least 90%

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

They have none

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

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Intraocular Tumors of Childhood

Brushfield spots
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**

5) **Iris mammillations**

**Iris/Ciliary Body**

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

- Down syndrome

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

- At least 90%

*What is the clinical significance of Brushfield spots?*

- They have none

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

- Wolfflin nodules
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**Retina**
Intraocular Tumors of Childhood

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**Mammillations? Aren’t those a CNS thingamajig?**

You’re thinking of the **two words**, paired structures that are part of the limbic system
Intraocular Tumors of Childhood

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Choroid

Retina
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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

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**Iris/Ciliary Body**

**Retina**
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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Oculodermal melanocytosis, aka **nevus of Ota**
### Intraocular Tumors of Childhood

#### Iris/Ciliary Body

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Intraocular Tumors of Childhood

Oculodermal melanocytosis (nevus of Ota).
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Note the presence of pigmentation in addition to that of the eye structure.
Intraocular Tumors of Childhood

Oculodermal melanocytosis (nevus of Ota).
Note the presence of periocular pigmentation in addition to that of the episclera.
Intraocular Tumors of Childhood

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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 is kids slide-set)
(This is a good point in the set to take a break)
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**Iris/Ciliary Body**

**Choroid**

Five tumors of the choroid

Retina

RPE
Intraocular Tumors of Childhood

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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
Intraocular Tumors of Childhood

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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 is kids slide-set)

Choroid

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

Five tumors of the choroid

But not 6)

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

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

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**Iris/Ciliary Body**

**Choroid**

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

But not 6) **Metastases**

---

**Retina**

**Five tumors of the choroid**

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

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

Iris/Ciliary Body

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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2) Melanocytoma
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4) Isolated/focal choroidal hemangioma
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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.

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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6) Metastases

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

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

Retina
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
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**Iris/Ciliary Body**

- Nevus: Common. Benign
- **Melanocytoma**
- Osteoma
- Iris mammillations
- Iris cysts

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**Choroid**

- Neovascular glioma
- Segmental choroidal hemangioma
- Diffuse choroidal hemangioma
- Branch retinal artery occlusion
- Macular hole
- Central retinal vein occlusion
- Macular edema

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

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

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2) **Melanocytoma**

3) **Osteoma**

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5) **Diffuse choroidal hemangioma**

Melanocytoma is a variant of what common choroidal finding? It is a particular sort of choroidal nevus
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5) Diffuse choroidal hemangioma

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

From what structure does it commonly arise? The optic disc

Is it unilateral, or bilateral? It is virtually always unilateral

Is there a racial predilection? No

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

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

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It is a particular sort of choroidal nevus

From what structure does it commonly arise?
The optic disc

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RPE

Retina

Iris/Ciliary Body

Choroid
Intraocular Tumors of Childhood

Melanocytoma
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Intraocular Tumors of Childhood

1) **Nevis:** Common. Benign

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

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

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

The optic disc

**Does it have a unilateral/bilateral predilection?**
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**Iris/Ciliary Body**

**Choroid**

**RPE**

**Retina**

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

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

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

The optic disc

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

Yes, it is virtually always [unilateral]
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**Choroid**

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**Retina**

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**RPE**

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**Iris/Ciliary Body**

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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.

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

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**RPE**

**Retina**

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**Iris/Ciliary Body**

---

**Choroid**

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

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

The optic disc

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

Yes, it is virtually always unilateral

**Is there a racial predilection?**

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**Melanocytoma**

---

1) **Nevus:** Common. Benign

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**Intraocular Tumors of Childhood**

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**Iris/Ciliary Body**

**Choroid**

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3) **Osteoma**
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**RPE**

**Retina**

**Intraocular Tumors of Childhood**

Retina
Intraocular Tumors of Childhood

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

From what structure does it commonly arise?
The optic disc

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

Is there a racial predilection?
No

Does it affect visual acuity?
Intraocular Tumors of Childhood

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From what structure does it commonly arise?
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Does it affect visual acuity?
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**Melanocytoma is a variant of what common choroidal finding?**

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

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**Iris/Ciliary Body**

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

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

Yes

---

**Choroid**

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**RPE**

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**Retina**
Intraocular Tumors of Childhood

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   *It is a particular sort of choroidal nevus*

   *From what structure does it commonly arise?*
   
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**Iris/Ciliary Body**

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**Iris/Ciliary Body**

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**Does it affect visual acuity?**

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

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

Approximately what percent of cases will transform?

**Choroid**

1) **Nevus**: Common. Benign

2) **Melanocytoma**

3) **Osteoma**

4) **Isolated/focal choroidal hemangioma**

5) **Diffuse choroidal hemangioma**

**Retina**

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

---

**Melanocytoma**

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

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

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

The optic disc

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

Yes, it is virtually always unilateral

**Is there a racial predilection?**

No

**Does it affect visual acuity?**

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

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

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1-2
In a word, what is a choroidal osteoma composed of?

**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**

**Choroidal neovascular membrane**
Intraocular Tumors of Childhood

Osteoma
Intraocular Tumors of Childhood

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

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

Osteomas (same pt, different cuts). Note how bright the lesions are.
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Choroidal neovascular membrane

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RPE

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Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

Choroid

RPE

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

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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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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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Isolated/focal choroidal hemangioma: Circular, well-demarcated lesion often in the macula. Risk of CNVM.

Diffuse choroidal hemangioma: Can be diffuse across the choroid, may involve the RPE. Can cause serous retinal detachment. Self-limited. Rare.

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

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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).


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

4) Isolated/focal choroidal hemangioma (aka circumscribed choroidal hemangioma): By what other name is this lesion known?
   Circumscribed choroidal hemangioma

   Is it common, or rare?
   Rare

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

   No

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

   What is its characteristic pattern on an -scan ultrasonography?
   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

   5) Diffuse choroidal hemangioma
Intraocular Tumors of Childhood

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

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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Iris/Ciliary Body

Choroid

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

Is it common, or rare? Rare

Is it associated with a systemic condition, ie, is it syndromic? No
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**Iris/Ciliary Body**

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- Isolated/focal choroidal hemangioma
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**Choroid**

- Circumscribed choroidal hemangioma

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

**Circumscribed choroidal hemangioma**

**Is it common, or rare?**

Rare

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

No

**How does it present?**

As a reddish-orange mass in the macula

**Retina**

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

4) **Isolated/focal choroidal hemangioma**

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Circumscribed choroidal hemangioma
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**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’
Circumscribed choroidal hemangioma: High internal reflectivity on a-scan
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5) Diffuse choroidal hemangioma

Choroid

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

Is it common, or rare?
Rare

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

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

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

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

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

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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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Sturge-Weber syndrome (SWS)

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

Sturge-Weber syndrome (SWS)
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**In a word, what sort of condition is SWS?**

A phakomatosis

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With what condition is the diffuse choroidal hemangioma associated?

**Sturge-Weber syndrome (SWS)**

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**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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**What is the noneponymous name for SWS?**

Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)
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Sturge-Weber syndrome (SWS)

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

What is the nonponymous 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

Intracocular Tumors of Childhood

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

Sturge-Weber syndrome (SWS) is associated with about 50% of cases. The coloration is a very red—much more so than an unaffected fundus. The coloration is a very red—much more so than an unaffected fundus. '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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Isolated/focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

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

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

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

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Intraocular Tumors of Childhood

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

11) Diffuse choroidal hemangioma: Intraocular Tumors of Childhood

- What condition is the diffuse choroidal hemangioma associated with? Sturge-Weber syndrome (SWS)
- About 50% of people with SWS have a diffuse choroidal hemangioma.
- 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 angioma (you might also see encephalofacial or cerebrofacial angioma)
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6) **Iris cysts:** Can be pupillary, stromal, secondary (see the Iris issues in kids slide-set)

**Intracocular Tumors of Childhood**

- **Nevus:** Common. Benign
- **Melanocytoma:** Usually juxtapapillary. Malignant transformation extremely rare
- **Osteoma:** Benign bony tumor, most common in teen years, females. Risk of CNVM
- **Isolated/focal choroidal hemangioma:** Very rare. Characteristic a- scan pattern
- **Diffuse choroidal hemangioma:** Present in what condition? Sturge-Weber syndrome (SWS)
  - Present in approximately what percent of SWS? About 50%
  - What does the fundus look like in an eye with a diffuse choroidal hemangioma? The coloration is a very red—much more so than an unaffected fundus
  - What food-related term is used to describe the fundus appearance in SWS? 'Tomato catsup fundus'
  - Can the choroidal hemangioma be present bilaterally? Yes, but it's uncommon
  - Does the choroidal hemangioma have malignant potential? No
  - In a word, what sort of condition is SWS? A phakomatosis
  - What is the noneponymous name for SWS? Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)
  - What is the hallmark skin finding in SWS? The port-wine stain
  - In one word, what sort of lesion is the port-wine stain? An angioma
  - When does it present? At birth

**Sturge-Weber syndrome (SWS)**
Intraocular Tumors of Childhood

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

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

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

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

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

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

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**Intracocular Tumors of Childhood**

**Iris/Ciliary Body**

Choroid

Intraocular Tumors of Childhood

With what condition is the diffuse choroidal hemangioma associated?

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

**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?**

- Very rare. Characteristic abnormal *a*-scan pattern

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

- 'Tomato catsup fundus'

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

- Yes, but it's uncommon

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

- No

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

- A phakomatosis

**What is the noneponymous name for SWS?**

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

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

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

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**Fundus appearance in SWS**

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

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**Fundus appearance in SWS**

- **Tomato catsup fundus**

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**Fundus appearance in SWS**

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

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

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

Brushfield spots: Strong association with Down syndrome

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

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

Nevus: Common. Benign

Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

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

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

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

Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Diffuse choroidal hemangioma is present in about 50% of SWS cases. The coloration is a very red—much more so than an unaffected fundus. This appearance is known as 'tomato catsup fundus'. Choroidal hemangioma can be present bilaterally but is uncommon. It does not have malignant potential. Sturge-Weber syndrome (SWS) is a phakomatosis. Its noneponymous name is encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis). The hallmark skin finding in SWS is the port-wine stain. In one word, what sort of lesion is the port-wine stain? An angioma. It presents at birth and its typical pattern of distribution comports to the distribution of one or more divisions of CN5. Does it always present in this manner? No.
**Intracocular 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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**Nevus:** Common. Benign

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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**

- **Associated with:** Sturge-Weber syndrome (SWS)
- **Prevalence:** About 50%
- **Fundus appearance:** The coloration is very red—much more so than an unaffected fundus
- **Synonym:** ‘Tomato catsup fundus’
- **Bilaterality:** Yes, but uncommon
- **Malignancy:** No

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

- **Non-eponym name:** Encephalotrigeminal angiomatosis (you might also see encephalofacial or cerebrofacial angiomatosis)

**Port-wine stain:** The hallmark skin finding in SWS

- **Type of lesion:** An angioma
- **Presentation:** At birth
- **Distribution pattern:** It comports to the distribution of one or more divisions of CN5
- **Presentation:** No, some cases will cross the midline of the face
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/− skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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

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

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

Diffuse choroidal hemangioma: Sturge-Weber syndrome (SWS)

Diffuse choroidal hemangioma is present in what percent of SWS?

Sturge-Weber syndrome (SWS)

With what condition is the diffuse choroidal hemangioma associated?

Sturge-Weber syndrome (SWS)

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

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

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

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1) **Nevus:** Common. Benign

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

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

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

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

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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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- and b-scan pattern

5) Diffuse choroidal hemangioma

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

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

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

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

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

Does the choroidal hemangioma have malignant potential? No
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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**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

**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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**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

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

2) **Diffuse choroidal hemangioma**

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**RPE**

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

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

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

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

Nevus: Common. Benign

Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

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

Diffuse choroidal hemangioma: Associated with Sturge-Weber syndrome (SWS). Present in about half of SWS cases. The fundus appearance is characterized by a very red coloration, much more so than an unaffected fundus. The term 'tomato catsup fundus' is used to describe this appearance in SWS. Can be present bilaterally but uncommonly so. The choroidal hemangioma itself has no malignant potential.
1) **Juvenile xanthogranuloma (JXG):** Nonneoplastic histiocytic proliferation. <2 years old. +/- skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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

5) **Diffuse choroidal hemangioma**

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

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

**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’
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Iris/Ciliary Body

Choroid

RPE

Retina

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

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

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

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

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

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5) Diffuse choroidal hemangioma: Very rare. Characteristic a-scan pattern

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Iris/Ciliary Body

Choroid

RPE

Retina
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**Iris/Ciliary Body**

**Choroid**

**RPE**

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

Does the choroidal hemangioma have malignant potential? No

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

Sturge-Weber syndrome (SWS): Diffuse choroidal hemangioma is present in what percent of SWS? About half

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Does the choroidal hemangioma have malignant potential? No

Sturge-Weber syndrome (SWS): Diffuse choroidal hemangioma is present in what percent of SWS? About half

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

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

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

Does the choroidal hemangioma have malignant potential? No
1) Juvenile xanthogranuloma (JXG): Nonneoplastic histiocytic proliferation. <2 years old. +/− skin papules. Iris nodules bleed → hyphema → increased IOP → glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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

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

4) Brushfield spots: Strong association with Down syndrome

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

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

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1) Nevus: Common. Benign

2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

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

5) Diffuse choroidal hemangioma

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

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

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

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

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

Does the choroidal hemangioma have malignant potential?
No

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Iris/Ciliary Body

Choroid

RPE

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

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**Iris/Ciliary Body**

**Retina**

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

Sturge-Weber syndrome (SWS)

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

About half

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

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

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

‘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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1) Isolated focal choroidal hemangioma: Very rare. Characteristic a-scan pattern

2) Isolated juxtapapillary choroidal hemangioma: Very rare. Characteristic a-scan pattern

3) Isolated neovascular choroidal hemangioma: Very rare. Characteristic a-scan pattern

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

5) **Diffuse choroidal hemangioma**

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For more on SWS, see slide-set P10
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**Iris/Ciliary Body**

**Choroid**

1) **Nevus:** Common. Benign

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

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

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**RPE**

1) ?
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

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

Retina
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**Iris/Ciliary Body**

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

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

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

- Common

- It is neither a hamartoma or a choristoma

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**RPE**

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

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**Retina**
Intraocular Tumors of Childhood

Iris/Ciliary Body

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

Choroid

1) Congenital hypertrophy of the RPE (CHRPE)

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

Retina
Intraocular Tumors of Childhood

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

*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

1) Congenital hypertrophy of the RPE (CHRPE)

RPE

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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

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**RPE**

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

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

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Retina

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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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**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

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

Retina
Iris/Ciliary Body

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

1) **Nevus**: 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)**

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
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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**RPE**

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

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

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CHRPE

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**on**

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?

--Solitary  CHRPE

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

Solitary

Grouped

CHRPE
**Iris/Ciliary Body**

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**RPE**

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

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

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

**Iris/Ciliary Body**

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**RPE**

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

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

--- Solitary CHRPE

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

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

‘Bear tracks’
Intraocular Tumors of Childhood

CHRPE: Bear tracks
Intraocular Tumors of Childhood

**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?
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Retina
Iris/Ciliary Body

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

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

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Iris/Ciliary Body

A **CHRPE-like lesion**

(both eponymous and non-eponymous)

Familial adenomatous polyposis, aka Gardner syndrome

A CHRPE-like lesion is associated with a potentially fatal inherited syndrome. Name: **Familial adenomatous polyposis**, aka Gardner syndrome

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

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

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

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**RPE**

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

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

-- **Solitary** CHRPE

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

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**Retina**

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Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

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

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

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

Iris/Ciliary Body

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

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

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

4) Brushfield spots: Strong association with Down syndrome

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

Nevus: Common. Benign

Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare

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

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

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

RPE

1) Congenital hypertrophy of the RPE (CHRPE)

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

--Solitary CHRPE

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

Iris/Ciliary Body

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

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

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**CHRPE is characterized according to its presentation. In what two ways does it present?**
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CHRPE-like lesions of Gardner syndrome: Bilateral presentation
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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Intraocular Tumors of Childhood
Intraocular Tumors of Childhood

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RPE

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Intraocular Tumors of Childhood

CHRPE-like lesions of Gardner syndrome: Scattered distribution
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**Iris/Ciliary Body**

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

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

-- If it is bilateral (regular CHRPE is almost always unilateral)
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-- If the shape of the lesions is pisciform

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**Retina**

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Intraocular Tumors of Childhood

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**What does pisciform mean?**

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

RPE

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---Solitary CHRPE
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Intraocular Tumors of Childhood

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

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

--?
--?

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**Retina**

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**Intraocular Tumors of Childhood**

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

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

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Intraocular Tumors of Childhood

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**Intraocular Tumors of Childhood**

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**Intraocular Tumors of Childhood**

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Intraocular Tumors of Childhood

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Iris/Ciliary Body

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

**Retina**

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

Retina/Choroid/Bruch Membrane

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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

--They are hypopigmented

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**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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**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 location in eye
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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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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: Large lesion(s) surrounded by a few smaller ones

--Multifocal or Grouped

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

--Solitary
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Familial adenomatous polyposis, aka Gardner syndrome

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

--Multifocal or Grouped
Intraocular Tumors of Childhood

CHRPE-like lesions of Gardner syndrome: Hypopigmented tail pointing toward ONH
Iris/Ciliary Body

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

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

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

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

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

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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

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

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**Iris/Ciliary Body**

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

- 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
Gardner syndrome: Colonic polyps
Intraocular Tumors of Childhood

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

What proportion of untreated Gardner syndrome pts will develop colon cancer? All of them. By what age will this occur? Age 40, maybe a little later.
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10) **Isolated/focal choroidal hemangioma**: Very rare. Characteristic a-scan pattern

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

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**Retina**

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

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

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

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

**All** of them

By what age will this occur?

Age 40, maybe a little later
Intraocular Tumors of Childhood

Iris/Ciliary Body

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**By what age will this occur?**

40, maybe a little later

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**Iris/Ciliary Body**

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**All** of them

**What is the treatment of choice?**

**Prophylactic colectomy**

**By what age will this occur?**

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

Retina

1) Nevoid (aka congenital): Common. Benign
2) Melanocytoma: Usually juxtapapillary. Malignant transformation extremely rare
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Other than the colonic and RPE lesions, what are the findings in Gardner syndrome?
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CHRPE is characterized according to its presentation. In what two ways does it present?
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Intraocular Tumors of Childhood

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**ACHRPE**

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Speaking of eye dentistry: When a pt has teephus issues, several conditions should spring immediately to mind. One is Gardners; what are the other three?

--Gardner syndrome
--?
--?
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**Retina**

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--Axenfeld-Reiger

--Incontinentia pigmenti

Dental anomalies

--Benign tumors of bone

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Retina

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

What is the eponym for the dental abnormalities in congenital syphilis?

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

RPE

Intraocular Tumors of Childhood

1) Congenital hypertrophy of the RPE (CHRPE): Retina, Choroid, Iris/Ciliary Body

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Familial adenomatous polyposis, aka Gardner syndrome

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

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Speaking of eye dentistry: When a pt has teephus issues, several conditions should spring immediately to mind. One is Gardner’s; what are the other three?
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Hutchinson teeth

What is the classic description re the shape of Hutchinson teeth?
‘Peg-shaped’ (For more on congenital syphilis, see slide-set K4)
Intraocular Tumors of Childhood

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Intraocular Tumors of Childhood

Congenital syphilis: Hutchinson teeth
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**Iris/Ciliary Body**

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**Speaking of eye dentistry:** What should spring immediately to mind when a pt has teephus issues?

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---Iris cysts
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---Iris nodules
---Iris spots
---Osteoma
---Retinal vessels
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---Retinal pigment epithelium (RPE)

---RPE: Intraocular Tumors of Childhood (slide-set)

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

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---In three words, what sort of condition is A-R?

---An anterior-segment dysgenesis (for more on A-R, see slide-set FELT7)

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(For more on congenital syphilis, see slide-set K4)

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RPE Intraocular Tumors of Childhood

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

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

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**Intraocular Tumors of Childhood**

**Iris/Ciliary Body**

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

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(For more on congenital syphilis, see slide-set K4)

**In three words, what sort of condition is A-R?**

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(For more on A-R, see slide-set FELT7)

**What is the eponymous name of IP?**

**Bloch-Sulzberger syndrome**

**In one word, what sort of condition is IP?**

A phakomatosis

(For more on IP, see slide-set P10)
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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 are the other two?

- Muir-Torre syndrome
- Peutz Jeghers syndrome

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Intraocular Tumors of Childhood

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RPE

1) Congenital hypertrophy of the RPE (CHRPE)

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

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

Iris/Ciliary Body

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

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

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

RPE

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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
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---

Retina

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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**How is Muir-Torre pronounced?** Mure (rhymes with ‘pure’) tore-ay

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Intraocular Tumors of Childhood

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**Iris/Ciliary Body**

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

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

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**RPE**

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

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

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

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Intraocular Tumors of Childhood

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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

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

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

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

When 'colon cancer + ophthalmic issue' is mentioned, two syndromes should come to mind. One is Gardner syndrome. What is the other?
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Does Muir-Torre present with multiple adenomatous polyps of the colon a la Gardner syndrome?
No; Muir-Torre is an example of a disease spectrum called Hereditary Non-Polyposis Colorectal Cancer
Multiple skin-colored to yellow–pink papules (arrows) on the face of a 64-year-old woman with a history of colon and cervical cancer. A skin biopsy confirmed a diagnosis of sebaceous adenoma resulting from Muir–Torre syndrome.
Intraocular Tumors of Childhood

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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
- One is Gardner syndrome. Multiple extraintestinal manifestations of (both eponymous and noneponymous) syndrome?
- If it is bilateral (regular CHRPE is almost always unilateral)
- If the lesions are scattered throughout multiple sectors of the eye (not “grouped”)
- If the shape of the lesions is pisciform

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

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 à la Gardner syndrome?
- No; Muir-Torre is an example of a disease spectrum called Hereditary Non-Polyposis Colorectal Cancer

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

--Sebaceous-cell carcinomas
--Sebaceous-cell adenomas
--Basal-cell carcinomas with sebaceous differentiation
Intraocular Tumors of Childhood

Iris/Ciliary Body

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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

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?
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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

How is Peutz-Jeghers pronounced? Pyoots jeh-grz

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

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

Familial adenomatous polyposis, aka **Gardner syndrome**

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

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How is Peutz-Jeghers pronounced?

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What is the main ophthalmic manifestation of Peutz-Jeghers 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 are both associated with MSI + tumors. One of the cancers that can develop in Peutz-Jeghers is present?

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

Yes
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**
- **Peutz-Jeghers 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 are the other two.

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

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

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1) **Retina**

2) **Choroid**

- **Congenital hypertrophy of the RPE (CHRPE):**
  - In what two ways does it present?
  - **Solitary** CHRPE: Single large lesion or patch
  - **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?
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’
Does lentigo simplex have malignant potential?
No
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**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

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

Yes

What are simple lentigines?

Flat melanocytic lesions histologically similar to ephelides
Intraocular Tumors of Childhood

Iris/Ciliary Body

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Flat melanocytic lesions histologically similar to ephelides

What are ephelides (singular, ephelis)?
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?

**Gardner syndrome**

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

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**Simple lentigines**

**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
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 irises

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**

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

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 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

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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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

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

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

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**Does lentigo simplex have malignant potential?** No
**Intraocular Tumors of Childhood**

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**Retina**

++ Retinal Holes

++ Multifocal choroiditis

++ Cystoid Macular Edema

++ Retinal Detachment

++ Macular telangiectasis

++ Retinal angiomatosus vasoclonus

++ Retinal telangiectasis

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**Choroid**

++ Behcet's disease

++ Cystoid macular edema

++ Pneuma chorioidis

++ CNVM

++ Neovascular RPE

++ Eales disease

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**Ocular Blood Vessels**

++ Venous dilatation

++ Venous tortuosity

++ Retinal vasculopathy

---

**Eyelid**

++ Lid margin deformities

++ Ptosis

++ Eyelid nevi

++ Canaliculus obstruction

---

**Ciliary Body**

++ Iris transillumination defect

++ Iris transillumination defect + secondary cataract

++ Iris plicata

++ Iris mammillations

++ Glaucoma

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**Iris/Ciliary Body**

++ Iris transillumination defect

++ Iris transillumination defect + secondary cataract

++ Iris plicata

++ Lisch nodules

++ Brushfield spots

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**CONGENITAL HYPERTROPHY OF THE RPE (CHRPE)**

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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 are simple lentigines? Flat melanocytic lesions histologically similar to ephelides

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-- Lentigo simplex

-- Lentigo maligna

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

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Does lentigo simplex have malignant potential? No

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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.
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**Iris/Ciliary Body**

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

**Peutz-Jeghers syndrome**

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

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

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Intraocular Tumors of Childhood

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**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 are simple lentigines?

Flat melanocytic lesions histologically similar to ephelides

Simple lentigines are also known as ‘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.

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Can also be a term for ‘Lentigo maligna melanoma’

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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**
Iris/Ciliary Body

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

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

What is the name of the 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

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 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 (not ‘grouped’)
-- If the shape of the lesions is pisciform

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

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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

---

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

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

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

---

1) **Congenital hypertrophy of the RPE (CHRPE):** Retina, Choroid, Iris/Ciliary Body

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

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

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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

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

When ‘colon cancer + eye 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?

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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
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?
Intraocular Tumors of Childhood

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

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

Retina

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

**Additional Tumors**

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Choroid

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RPE

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

Retina

1) ?
Intraocular Tumors of Childhood

Iris/Ciliary Body

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RPE

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

Retina

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

Iris/Ciliary Body

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Intraocular Tumors of Childhood

**Iris/Ciliary Body**

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**RPE**

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

**Retina**

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

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

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

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

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

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

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

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

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

   - 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

### RPE

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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

RPE Retina

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**Retina**

**RPE**

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**Intraocular Tumors of Childhood**

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

A **choristoma**

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**What is a hamartoma?**

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

That it is congenital

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**Iris/Ciliary Body**

**Choroid**

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As a variably pigmented, slightly elevated retinal mass of the area retina
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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 $\rightarrow$ hyphema $\rightarrow$ increased IOP $\rightarrow$ glaucoma. Self-limited; regresses by age 5. Treat inflammation and IOP. Path: Touton giant cells

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

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

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

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

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

1) **Nevus:** Common. Benign

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

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

**Iris/Ciliary Body**

**Choroid**

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

As a hamartoma

**RPE**

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

**Retina**

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

**Choroidal melanoma**

- How 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 can one avoid making such a disastrous mistake?
  By taking pains to carefully determine the anatomic location of the tumor in question

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

**What is a hamartoma?**

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RPE cells (duh) and retinal glial cells

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; in contrast, combined hamartomas of the retina and RPE are located in front of it.

With what more sinister dz entity is it often confused?
Choroidal melanoma — eyes have been enucleated because of this misdiagnosis
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**Combined hamartoma of the retina and RPE**

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

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1) **Combined hamartoma of the retina and RPE**: Benign, congenital retinal lesion

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

No question—summary/review slide