Nystagmus is commonly associated

Aniridia: T/F
- Nystagmus is commonly associated (True)

**Aniridia: T/F**
Q

- Nystagmus is commonly associated: True

Is this a sensory or a motor nystagmus?

Aniridia: T/F
Nystagmus is commonly associated: True

Is this a sensory or a motor nystagmus?
Sensory (more on this shortly)

Aniridia: T/F
Q

- Nystagmus is commonly associated  **True**

*Is this a sensory or a motor nystagmus?*
Sensory (more on this shortly)

*Is it a jerk, or a pendular nystagmus?*
Nystagmus is commonly associated \textbf{True}

\textit{Is this a sensory or a motor nystagmus?}
Sensory (more on this shortly)

\textit{Is it a jerk, or a pendular nystagmus?}
Pendular

\textbf{Aniridia: T/F}
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency
• Nystagmus is commonly associated: True
• Aniridia is associated with limbal stem cell deficiency: True
Nystagmus is commonly associated  True
Aniridia is associated with **limbal stem cell deficiency** True

Where are limbal stem cells found?
Nystagmus is commonly associated: True

Aniridia is associated with **limbal stem cell deficiency**: True

Where are limbal stem cells found?
Um, at the limbus?
Nystagmus is commonly associated  True
Aniridia is associated with limbal stem cell deficiency  True

Where are limbal stem cells found?
Um, at the limbus?
Yes, but where at the limbus--in what eponymous structures?
A

- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency  True

Where are limbal stem cells found?
Um, at the limbus?

Yes, but where at the limbus--in what eponymous structures?
The palisades of Vogt
Q

- Nystagmus is commonly associated True
- Aniridia is associated with **limbal stem cell deficiency** True

**Where are limbal stem cells found?**

Um, at the limbus?

Yes, but where at the limbus--in what eponymous structures?

The **palisades of Vogt**

**Briefly describe these palisades.**
Where are limbal stem cells found?
Um, at the limbus?

Yes, but where at the limbus--in what eponymous structures?
The **palisades of Vogt**

**Briefly describe these palisades.**
They are a series of pigmented ridges around the limbus
Nystagmus is commonly associated: True

Aniridia is associated with **limbal stem cell deficiency** True

---

**Q**

**Aniridia: T/F**

- Nystagmus is commonly associated: True
- Aniridia is associated with **limbal stem cell deficiency** True

---

**Where are limbal stem cells found?**

Um, at the limbus?

Yes, but where at the limbus--in what eponymous structures?

The **palisades of Vogt**

**Briefly describe these palisades.**

They are a series of pigmented ridges around the limbus

**At which level of the epithelium are the stem cells found?**
Nystagmus is commonly associated \textit{True}.

Aniridia is associated with \textit{limbal stem cell deficiency} \textit{True}.

\textit{Where are limbal stem cells found? Um, at the limbus?}

\textit{Yes, but where \textit{at the limbus}--in what eponymous structures?}

\textit{The \textit{palisades of Vogt}}

\textit{Briefly describe these palisades.}

\textit{They are a series of pigmented ridges around the limbus}

\textit{At which level of the epithelium are the stem cells found?}

\textit{The basal layer}
Nystagmus is commonly associated: True
Aniridia is associated with limbal stem cell deficiency: True

Where are limbal stem cells found?
Um, at the limbus?
Yes, but where at the limbus--in what eponymous structures?
The palisades of Vogt

Briefly describe these palisades.
They are a series of pigmented ridges around the limbus

At which level of the epithelium are the stem cells found?
The basal layer

So, the stem cells are deep. Why?
Nystagmus is commonly associated: True
Aniridia is associated with limbal stem cell deficiency: True

Where are limbal stem cells found?
Um, at the limbus?
Yes, but where at the limbus--in what eponymous structures?
The palisades of Vogt

Briefly describe these palisades.
They are a series of pigmented ridges around the limbus.

At which level of the epithelium are the stem cells found?
The basal layer

So, the stem cells are deep. Why?
By being deep...
Nystagmus is commonly associated True

Aniridia is associated with **limbal stem cell deficiency** True

Where are limbal stem cells found?
Um, at the limbus?

Yes, but where at the limbus--in what eponymous structures?
The **palisades of Vogt**

Briefly describe these palisades.
They are a series of **pigmented ridges** around the limbus

At which level of the epithelium are the stem cells found?
The **basal layer**

So, the stem cells are deep. Why?
By being deep...The overlying pigment absorbs UV light, thereby protecting the stem cells from **mutagenesis**
Nystagmus is commonly associated

Aniridia is associated with limbal stem cell deficiency

How common is limbal stem cell deficiency in aniridia?
Q/A

- Nystagmus is commonly associated  True
- Aniridia is associated with **limbal stem cell deficiency**  True

---

**Aniridia: T/F**

- How common is limbal stem cell deficiency in aniridia?
- Very--about % of aniridia pts suffer with it
A

- Nystagmus is commonly associated True
- Aniridia is associated with **limbal stem cell deficiency** True

How common is limbal stem cell deficiency in aniridia?
Very--about 90% of aniridia pts suffer with it
Q

- Nystagmus is commonly associated  True
- Aniridia is associated with **limbal stem cell deficiency**  True

How common is limbal stem cell deficiency in aniridia?  
Very--about 90% of aniridia pts suffer with it

How does limbal stem cell deficiency manifest in aniridic pts?

- --
- --
- --
Nystagmus is commonly associated: True

Aniridia is associated with limbal stem cell deficiency: True

How common is limbal stem cell deficiency in aniridia?
Very--about 90% of aniridia pts suffer with it

How does limbal stem cell deficiency manifest in aniridic pts?
In all the ways one would expect…
--corneal erosions/ulcers
--vascular pannus
--conjunctivilization of the cornea
Phakic patient with aniridia. Note the corneal pannus obscuring the view of the angle inferiorly
Nystagmus is commonly associated  True

Aniridia is associated with limbal stem cell deficiency  True

How common is limbal stem cell deficiency in aniridia?
Very--about 90% of aniridia pts suffer with it

How does limbal stem cell deficiency manifest in aniridic pts?
In all the ways one would expect…
--corneal erosions/ulcers
--vascular pannus
--conjunctivilization of the cornea

What is aniridia-associated keratopathy called?
- Nystagmus is commonly associated: True
- Aniridia is associated with **limbal stem cell deficiency**: True

**How common is limbal stem cell deficiency in aniridia?**
Very--about 90% of aniridia pts suffer with it

**How does limbal stem cell deficiency manifest in aniradic pts?**
In all the ways one would expect…
--corneal erosions/ulcers
--vascular pannus
--conjunctivilization of the cornea

**What is aniridia-associated keratopathy called?**
It is called ‘**aniridia-associated keratopathy**’
Nystagmus is commonly associated  True
Aniridia is associated with limbal stem cell deficiency  True
Presents unilaterally and bilaterally in roughly equal rates
A/Q

- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral

Aniridia: T/F
Nystagmus is commonly associated: True
Aniridia is associated with limbal stem cell deficiency: True
Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
Bilateral congenital aniridia
Q

- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about 1/2 of cases, a rudimentary iris root is present

Aniridia: T/F
Nystagmus is commonly associated: True

Aniridia is associated with limbal stem cell deficiency: True

Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral

The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
Aniridia: Note the iris roots
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia
- Nystagmus is commonly associated  **True**
- Aniridia is associated with limbal stem cell deficiency  **True**
- Presents unilaterally and bilaterally in roughly equal rates  **False; it is almost always bilateral**
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  **False; it’s a misnomer because a rudimentary iris root is **always** present**
- Aniridia is strongly associated with foveal and optic nerve hypoplasia  **True**
Aniridia: Foveal/optic nerve hypoplasia
Nystagmus is commonly associated  True
Aniridia is associated with limbal stem cell deficiency True
Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
The term 'aniridia' is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present
Aniridia is strongly associated with foveal and optic nerve hypoplasia  True

The foveal and optic nerve hypoplasia lead to poor central acuity (usually no better than 20/100, and often significantly worse). In turn, the poor visual acuity produces the congenital sensory nystagmus that is associated with aniridia.
Nystagmus is commonly associated True

Aniridia is associated with limbal stem cell deficiency True

Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral

The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present False; it’s a misnomer because a rudimentary iris root is always present

Aniridia is strongly associated with foveal and optic nerve hypoplasia True

Is there a correlation between the extent of aniridia and the severity of the foveal hypoplasia?
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True

Is there a correlation between the extent of aniridia and the severity of the foveal hypoplasia? No
Q

- Nystagmus is commonly associated \textbf{True}
- Aniridia is associated with limbal stem cell deficiency \textbf{True}
- Presents unilaterally and bilaterally in roughly equal rates \textbf{False; it is almost always bilateral}
- The term ‘aniridia’ is a misnomer because, in about \(\frac{1}{2}\) of cases, a rudimentary iris root is present \textbf{False; it’s a misnomer because a rudimentary iris root is \textit{always} present}
- Aniridia is strongly associated with foveal and optic nerve hypoplasia \textbf{True}
- Patients complain of (and infants suffer from) photophobia
Aniridia: T/F

- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia True
- Patients complain of (and infants suffer from) photophobia  True
Nystagmus is commonly associated  True
Aniridia is associated with limbal stem cell deficiency True
Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present False; it’s a misnomer because a rudimentary iris root is always present
Aniridia is strongly associated with foveal and optic nerve hypoplasia True
Patients complain of (and infants suffer from) photophobia True
Familial cases are at risk for Wilms tumor
- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency  True
- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia  True
- Patients complain of (and infants suffer from) photophobia  True
- Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

**Aniridia: T/F**
Aniridia: T/F

- Nystagmus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present True
- Aniridia is strongly associated with foveal and optic nerve hypoplasia True
- Patients complain of (and infants suffer from) photophobia True
- Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

What sort of neoplasms is a Wilms tumor?

- A nephroblastoma

As childhood intra-abdominal malignancies go, is Wilms tumor common?

- Yes--it's the most common intra-abdominal malignancy of childhood

Does Wilms tumor have a favorable prognosis?

- Yes; at present, >90% case are curable if caught early
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
- Patients complain of (and infants suffer from) photophobia: True
- Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

Aniridia: T/F

What sort of neoplasm is a Wilms tumor?

A nephroblastoma

Wilms tumor
Wilm’s tumor
Q

- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris can be seen  False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia True
- Patients complain of (and infants suffer from) photophobia  True
- Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

What sort of neoplasm is a Wilms tumor?
A nephroblastoma

As childhood intra-abdominal malignancies go, is Wilms tumor common?
Yes--it’s the most common intra-abdominal malignancy of childhood

Does Wilms tumor have a favorable prognosis?
Yes; at present, >90% case are curable if caught early
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about 1/2 of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
- Patients complain of (and infants suffer from) photophobia: True
- Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

**Aniridia: T/F**

*What sort of neoplasm is a Wilms tumor?*
A nephroblastoma

*As childhood intra-abdominal malignancies go, is Wilms tumor common?*
Yes--it’s the most common intra-abdominal malignancy of childhood

**Wilms tumor**
Nystagmus is commonly associated True
Aniridia is associated with limbal stem cell deficiency True
Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral
The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present False; it’s a misnomer because a rudimentary iris root is always present
Aniridia is strongly associated with foveal and optic nerve hypoplasia True
Patients complain of (and infants suffer from) photophobia True
Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

What sort of neoplasm is a Wilms tumor? A nephroblastoma
As childhood intra-abdominal malignancies go, is Wilms tumor common? Yes--it’s the most common intra-abdominal malignancy of childhood
Does Wilms tumor have a favorable prognosis? Yes; at present, >90% case are curable if caught early
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
- Patients complain of (and infants suffer from) photophobia: True
- Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

**Aniridia: T/F**

- What sort of neoplasm is a Wilms tumor?
  A nephroblastoma

- As childhood intra-abdominal malignancies go, is Wilms tumor common?
  Yes--it’s the most common intra-abdominal malignancy of childhood

- Does Wilms tumor have a favorable prognosis?
  Yes; at present, >90% case are curable if caught early
Nystagmus is commonly associated True
Aniridia is associated with limbal stem cell deficiency True
Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral
The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present False; it’s a misnomer because a rudimentary iris root is always present
Aniridia is strongly associated with foveal and optic nerve hypoplasia True
Patients complain of (and infants suffer from) photophobia True
Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

What are the other components of the WAGR complex?
-- Wilms tumor
-- Aniridia
-- G
-- R

WAGR complex
A

- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is *always* present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia  True
- Patients complain of (and infants suffer from) photophobia  True
- Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex

What are the other components of the WAGR complex?
- Wilms tumor
- Aniridia
- Genitourinary abnormalities
- Retardation

WAGR complex
Nystagmus is commonly associated **True**
Aniridia is associated with limbal stem cell deficiency **True**
Presents unilaterally and bilaterally in roughly equal rates **False; it is almost always bilateral**
The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present **False; it’s a misnomer because a rudimentary iris root is *always* present**
Aniridia is strongly associated with foveal and optic nerve hypoplasia **True**
Patients complain of (and infants suffer from) photophobia **True**
Familial cases are at risk for Wilms tumor **False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex**
Aniridia is associated with glaucoma
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
- Patients complain of (and infants suffer from) photophobia: True
- Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma: True
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
- Patients complain of (and infants suffer from) photophobia: True
- Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma: True
- Aniridia is associated with early-onset cataracts
Nystagmus is commonly associated: True
Aniridia is associated with limbal stem cell deficiency: True
Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral
The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present
Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
Patients complain of (and infants suffer from) photophobia: True
Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
Aniridia is associated with glaucoma: True
Aniridia is associated with early-onset cataracts: True
Bilateral childhood cataracts in aniridia
Nystagmus is commonly associated. True

Aniridia is associated with limbal stem cell deficiency. True

Presents unilaterally and bilaterally in roughly equal rates. False; it is almost always bilateral.

The term 'aniridia' is a misnomer because, in about ½ of cases, a rudimentary iris root is present. False; it’s a misnomer because a rudimentary iris root is always present.

Aniridia is strongly associated with foveal and optic nerve hypoplasia. True

Patients complain of (and infants suffer from) photophobia. True

Familial cases are at risk for Wilms tumor. False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex.

Aniridia is associated with glaucoma. True

Aniridia is associated with early-onset cataracts. True

Don’t think of aniridia as an iris condition! The BCSC characterizes it as a panophthalmic disorder.
Nystagmus is commonly associated  True
Aniridia is associated with limbal stem cell deficiency  True
Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present
Aniridia is strongly associated with foveal and optic nerve hypoplasia  True
Patients complain of (and infants suffer from) photophobia  True
Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
Aniridia is associated with glaucoma  True
Aniridia is associated with early-onset cataracts  True

How ‘early’ do aniridia pts get their ‘early onset’ cataracts?
• Nystagmus is commonly associated  True  
• Aniridia is associated with limbal stem cell deficiency True  
• Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral  
• The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present  
• Aniridia is strongly associated with foveal and optic nerve hypoplasia True  
• Patients complain of (and infants suffer from) photophobia  True  
• Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex  
• Aniridia is associated with glaucoma True  
• Aniridia is associated with early-onset cataracts True  

How ‘early’ do aniridia pts get their ‘early onset’ cataracts?  
Childhood to young adulthood  

Aniridia is associated with early-onset cataracts True
- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present  False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia True
- Patients complain of (and infants suffer from) photophobia  True
- Familial cases are at risk for Wilms tumor  False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma True
- Aniridia is associated with early-onset cataracts True

**How ‘early’ do aniridia pts get their ‘early onset’ cataracts?**
Childhood to young adulthood

**How common are early-onset cataracts in aniridia?**
A/Q

- Nystagmus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present False; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia True
- Patients complain of (and infants suffer from) photophobia True
- Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma True
- Aniridia is associated with early-onset cataracts True

How ‘early’ do aniridia pts get their ‘early onset’ cataracts? Childhood to young adulthood

How common are early-onset cataracts in aniridia? They occur in at least 50% of aniridia cases
- Nystagmus is commonly associated: True
- Aniridia is associated with limbal stem cell deficiency: True
- Presents unilaterally and bilaterally in roughly equal rates: False; it is almost always bilateral.
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present: False; it’s a misnomer because a rudimentary iris root is always present.
- Aniridia is strongly associated with foveal and optic nerve hypoplasia: True
- Patients complain of (and infants suffer from) photophobia: True
- Familial cases are at risk for Wilms tumor: False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex.
- Aniridia is associated with glaucoma: True
- Aniridia is associated with early-onset cataracts: True

**Aniridia: T/F**

- How ‘early’ do aniridia pts get their ‘early onset’ cataracts? Childhood to young adulthood
- How common are early-onset cataracts in aniridia? They occur in at least 50% of aniridia cases

**Aniridia is associated with early-onset cataracts** True
Aniridia: T/F

- Nystagmus is commonly associated  True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates  False; it is almost always bilateral
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Is glaucoma common in aniridia?

Yes. Estimates vary, but it’s a safe bet that at least 50% of aniridics develop glaucoma.

Is glaucoma a significant factor in the visual prognosis of aniridic pts?

Yes. Glaucomatous optic neuropathy is a significant cause of blindness in aniridics, as are complications stemming from glaucoma surgery.

Is glaucoma in aniridia of the open-angle or angle-closure variety?

Angle closure

What is the mechanism of angle closure?

That depends on the nature of the aniridia. In familial aniridia the angle is structurally normal at birth, but becomes occluded over time by the rudimentary iris root. In contrast, the angle in sporadic aniridia tends to be congenitally abnormal.
A/Q

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Aniridia: T/F
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Is glaucoma in aniridia of the open-angle or angle-closure variety?

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**Aniridia: T/F**

- **Is glaucoma common in aniridia?**
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- **Is glaucoma a significant factor in the visual prognosis of aniridic pts?**
  Yes. Glaucomatous optic neuropathy is a significant cause of blindness in aniridics, as are complications stemming from glaucoma surgery.

- **Is glaucoma in aniridia of the open-angle or angle-closure variety?**
  Angle closure

- **How does this occlusion process proceed?**
  Gradually. Over time, the iris stump rotates anteriorly, resulting in contact with the TM. Eventually, synechiae form, and the angle will occlude.

- **How much time are we talking about here?**
  No earlier than the second decade, and often much later.

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In what ways is the angle abnormal?  The TM is poorly developed. Further, corneal endothelium may grow across the angle producing PAS. In such cases, when will the glaucoma declare itself?  Early--these cases present as congenital glaucoma

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Is glaucoma open-angle or angle-closure variety?
Angle-closure

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Defects involving what gene are the cause of aniridia?

The PAX6 gene
- Nystagmus is commonly associated. True
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Defects involving what gene are the cause of aniridia?
The PAX6 gene

What other ocular abnormalities are associated with defects of the PAX6 gene?
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\textbf{Defects involving what gene are the cause of aniridia?}

The PAX6 gene

\textbf{What other ocular abnormalities are associated with defects of the PAX6 gene?}

I'm glad you asked…
What four ocular abnormalities are attributed to the PAX6 gene?

There are four main abnormalities, and the term PAX6 acts as its own mnemonic. Start with the ‘P’ and make your way down…

Aniridia: T/F

Start with the ‘P’ and work down
What four ocular abnormalities are attributed to the PAX6 gene?
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Aniridia
Peters anomaly
Congenital cataract

Aniridia: T/F
What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Anirida
- Congenital cataract
- Foveal hypoplasia

If you use your imagination, the 6 looks like a lower-case h…
What four ocular abnormalities are attributed to the PAX6 gene?

Peters anomaly

What is Peters anomaly?
What four ocular abnormalities are attributed to the PAX6 gene?

**Peters anomaly**

*What is Peters anomaly?*
A condition characterized by defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.
1. Defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium

2. Adhesions extending from the iris to the posterior corneal defect

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Peters anomaly

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How does it present?
What four ocular abnormalities are attributed to the PAX6 gene?

Peter's anomaly

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How does it present?
As a corneal opacity at birth (it’s the P in the infamous mnemonic for congenital cloudy cornea).
What four ocular abnormalities are attributed to the PAX6 gene?

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A condition characterized by defect of the posterior central cornea, including the absence of Descemet’s and subjacent endothelium. Adhesions extending from the iris to the posterior corneal defect are often present.

How does it present?
As a corneal opacity at birth (it’s the P in the infamous STUMPED mnemonic for congenital cloudy cornea). The opacity ranges in severity from a faint haze to an opaque, elevated and vascularized mess.
Peters anomaly: Hazy cornea
What four ocular abnormalities are attributed to the PAX6 gene?

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Note: There are two S’s and two E’s.
What are four ocular abnormalities attributed to the PAX6 gene?

What is the STUMPED mnemonic for a cloudy cornea in an infant?

- Sclerocornea; Stromal dystrophy (CHSD)
- Trauma (e.g., forceps injury)
- Ulcer
- Mucopolysaccharidosis
- Peters anomaly
- Endothelial dystrophy (CHED); Elevated IOP (congenital glaucoma)
- Dermoid of the cornea

Note: There are two S’s and two E’s.

How does it present?
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What four ocular abnormalities are attributed to the PAX6 gene?

Of the four, which is most strongly associated with PAX6 mutations?

- Aniridia?
- Peters anomaly?
- Congenital cataract?
- Foveal hypoplasia?
What four ocular abnormalities are attributed to the PAX6 gene?

Of the four, which is most strongly associated with PAX6 mutations? **Aniridia.** As the *Fundamentals* book puts it, “PAX6 mutations are the basis of virtually all cases of aniridia.” [emphasis added]
What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Aniridia
- Congenital cataract
- Foveal hypoplasia

If you use your imagination, the 6 looks like a lower-case h…
What four ocular abnormalities are attributed to the PAX6 gene?

- Anirida
- Congenital cataracts
- Foveal hypoplasia
- Peters anomaly

What sort of gene is PAX6 anyway? A homeobox gene.

If you use your imagination, the 6 looks like a lower-case h...
What sort of gene is PAX6 anyway?
A homeobox gene

What is a homeobox gene?

What four ocular abnormalities are attributed to the PAX6 gene?

- Peters anomaly
- Anirida
- Congenital cataract
- Foveal hypoplasia

If you use your imagination, the 6 looks like a lower-case h…
What sort of gene is PAX6 anyway?
A homeobox gene

What is a homeobox gene?
One that regulates morphogenesis

What four ocular abnormalities are attributed to the PAX6 gene?

If you use your imagination, the 6 looks like a lower-case h…
What four ocular abnormalities are attributed to the **PAX6** gene?

- Nasal Peters anomaly
- Aniridia
- Congenital cataract
- Foveal hypoplasia

If you use your imagination, the 6 looks like a lower-case h…

As the BCSC *Peds* book puts it, “The **PAX6** gene is the master control gene for eye morphogenesis.”

One that **regulates morphogenesis**

*What sort of gene is PAX6 anyway? A homeobox gene*
Nystagmus is commonly associated True
Aniridia is associated with limbal stem cell deficiency True

- Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral
- The term 'aniridia' is a misnomer because, in about ½ of cases, a rudimentary iris root is present False; it's a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia True

- Patients complain of (and infants suffer from) photophobia True
- Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma True
- Aniridia is associated with early-onset cataracts True

Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia?

True

- Patients complain of (and infants suffer from) photophobia True
- Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
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- Aniridia is associated with early-onset cataracts True
Why is **sporadic** aniridia associated with Wilms tumor, but not **familial** aniridia?

The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. *Inherited* genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, *sporadic* cases of aniridia are usually the result of the wholesale deletion of a chunk of genetic material in the PAX6 ‘neighborhood.’ And since WT1 is its next-door neighbor, it is often affected as well by these deletions.

Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

**True**

- Patients complain of (and infants suffer from) photophobia  True
- Familial cases are at risk for Wilms tumor  False; 1/3 of **sporadic** cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma True
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Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.
Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia? The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, sporadic cases of aniridia are usually the result of the wholesale deletion of a chunk of genetic material in the PAX6 ‘neighborhood.’ And since WT1 is its next-door neighbor, it is often affected as well. Because WT1 encodes a transcription factor, this situation eliminates all expression of its and nearby genes.

Does this ‘neighborhood deletion effect’ account for the other manifestations of the WAGR complex?

True

- Patients complain of (and infants suffer from) photophobia.
- Familial cases are at risk for Wilms tumor. False, 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex.
- Aniridia is associated with glaucoma True
- Aniridia is associated with early-onset cataracts True

What are the other components of the WAGR complex?

- Wilms tumor
- Aniridia
- Genitourinary abnormalities
- Retardation
Aniridia: T/F

Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia?
The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, sporadic cases of aniridia are usually the result of the wholesale deletion of a chunk of genetic material in the PAX6 ‘neighborhood.’ And since WT1 is its next-door neighbor, it is often affected as well. Because sporadic cases are associated with the deletion of WT1, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

Does this ‘neighborhood deletion effect’ account for the other manifestations of the WAGR complex?
Indeed it does.

What are the other components of the WAGR complex?
--Wilms tumor
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WAGR complex
Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia? The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, sporadic cases of aniridia are usually the result of the **wholesale deletion of a chunk of genetic material in the PAX6 ‘neighborhood.’** And since WT1 is its next-door neighbor, it is often affected as well.

Because sporadic aniridia is associated with Wilms tumor, it must be screened for it. Does this ‘neighborhood deletion effect’ account for the other manifestations of the WAGR complex? Indeed it does.

By what general term are such contiguous gene-deletion syndromes known?

What are the other components of the WAGR complex?
--Wilms tumor
--Aniridia
--Genitourinary abnormalities
--Retardation

WAGR complex
Aniridia: T/F

- Nystagmus is commonly associated. True
- Aniridia is associated with limbal stem cell deficiency. True
- Presents unilaterally and bilaterally in roughly equal rates. False; it is almost always bilateral.
- The term 'aniridia' is a misnomer because, in about ½ of cases, a rudimentary iris root is present. False; it's a misnomer because a rudimentary iris root is always present.
- Aniridia is strongly associated with foveal and optic nerve hypoplasia. True
- Patients complain of (and infants suffer from) photophobia. True
- Familial cases are at risk for Wilms tumor. False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex.
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Does this ‘neighborhood deletion effect’ account for the other manifestations of the WAGR complex? Indeed it does.

By what general term are such contiguous gene-deletion syndromes known? They are called ‘contiguous gene-deletion syndromes’

What are the other components of the WAGR complex?--Wilms tumor--Aniridia--Genitourinary abnormalities--Retardation

WAGR complex
Why is **sporadic** aniridia associated with Wilms tumor, but not **familial** aniridia? The PAX6 gene and the **Wilms tumor gene (called **WT1**)** are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself; thus, these familial cases are not associated with a risk for Wilms tumor. In contrast, sporadic cases of aniridia are usually the result of a **whole deletion** of a chunk of genetic material in the PAX6 'neighborhood.' Because **WT1** is next-door neighbor to PAX6, it is often affected in these deletions. Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?

In general terms, what sort of gene is the Wilms tumor gene?

A tumor suppressor gene

What prominent ocular tumor of childhood results from inactivation of a tumor suppressor gene?

Retinoblastoma

**Aniridia: T/F**

- Patients complain of (and infants suffer from) photophobia  **True**
- Familial cases are at risk for Wilms tumor  **False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex**
- Aniridia is associated with glaucoma  **True**
- Aniridia is associated with early-onset cataracts  **True**
Aniridia: T/F

Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia? True
The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, and thus do not affect the viability of the nearby WT1. In contrast, sporadic cases of aniridia are usually the result of the wholesale deletion of a chunk of genetic material in the PAX6 'neighborhood.' And since WT1 is its next-door neighbor, it is often affected as well by these deletions. Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor? True

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Why is sporadic aniridia associated with Wilms tumor, but not familial aniridia? The PAX6 gene and the Wilms tumor gene (called WT1) are adjacent to one another on chromosome 11p. Inherited genetic abnormalities leading to familial aniridia are located within the PAX6 gene itself, whereas in sporadic cases these abnormalities result in the wholesale deletion of a chunk of genetic material that includes both PAX6 and WT1. Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor? Via periodic renal ultrasound

True

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If a child tests positive for the Wilms tumor defect, how should they be screened for Wilms tumor?
Via periodic renal ultrasound

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Because of the PAX6/WT1 spatial relationship, all infants presenting with sporadic aniridia must undergo genetic screening for the Wilms tumor defect.

If a child tests positive for the Wilms tumor defect, how may be screened for Wilms tumor?

Via periodic renal ultrasound

How often, and for how long?
Every 3 months until age 7 years

True

- Patients complain of (and infants suffer from) photophobia True
- Familial cases are at risk for Wilms tumor False; 1/3 of sporadic cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma True
- Aniridia is associated with early-onset cataracts True
Nystagmus is commonly associated **True**

**Aniridia is associated with limbal stem cell deficiency** **True**

Presents unilaterally and bilaterally in roughly equal rates **False; it is almost always bilateral**

The term 'aniridia' is a misnomer because, in about ½ of cases, a rudimentary iris root is present **False; it's a misnomer because a rudimentary iris root is always present**

Aniridia is strongly associated with foveal and optic nerve hypoplasia **True**

Patients complain of (and infants suffer from) photophobia **True**

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Aniridia is associated with glaucoma **True**

Aniridia is associated with early-onset cataracts **True**
- Nystagmus is commonly associated \textbf{True}
- \textbf{Aniridia is associated with limbal stem cell deficiency \textbf{True}}
- Presents unilaterally and bilaterally in roughly equal rates \textbf{False}; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about \(\frac{1}{2}\) of cases, a rudimentary iris root is present \textbf{False}; it’s a misnomer because a rudimentary iris root is always present
- Aniridia is strongly associated with foveal and optic nerve hypoplasia \textbf{True}
- Patients complain of (and infants suffer from) photophobia \textbf{True}
- Familial cases are at risk for Wilms tumor \textbf{False}; 1/3 of \textit{sporadic} cases develop Wilms tumor as part of the WAGR complex
- Aniridia is associated with glaucoma \textbf{True}
- Aniridia is associated with early-onset cataracts \textbf{True}

<table>
<thead>
<tr>
<th>Associated condition</th>
<th>% of aniridia pts with the associated condition</th>
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<tbody>
<tr>
<td>\textit{Limbal stem-cell deficiency}</td>
<td>90</td>
</tr>
<tr>
<td>\textit{Glaucoma}</td>
<td>\textbf{To recap:} 50+</td>
</tr>
<tr>
<td>\textit{Early-onset cataracts}</td>
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