Q

Aniridia: T/F

- Nystgamus is commonly associated

A

Aniridia: T/F

- Nystgamus is commonly associated True

Q

Aniridia: T/F

- Nystgamus is commonly associated True

Is this a sensory or a motor nystagmus?

A

Aniridia: T/F

- Nystgamus is commonly associated True

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

Q

Aniridia: T/F

- Nystgamus 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?

A

Aniridia: T/F

- Nystgamus 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?
Pendular

Q

Aniridia: T/F

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

A

Aniridia: T/F

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

Q

Aniridia: T/F

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

Where are limbal stem cells found?

A

Aniridia: T/F

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

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

Q

Aniridia: T/F

- Nystgamus 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

Aniridia: T/F

- Nystgamus 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

Aniridia: T/F

- Nystgamus 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.

A

Aniridia: T/F

- Nystgamus 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

Q

Aniridia: T/F

- Nystgamus 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?

A

Aniridia: T/F

- Nystgamus 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

Q

Aniridia: T/F

- Nystgamus 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 th
The basal layer

So, the stem cells are deep. Why?

Q/A

Aniridia: T/F

- Nystgamus 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 th
The basal layer

So, the stem cells are deep. Why?
By being deep…

A

Aniridia: T/F

- Nystgamus 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
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**

Q

Aniridia: T/F

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

How common is limbal stem cell deficiency in aniridia?

Q/A

Aniridia: T/F

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

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

A

Aniridia: T/F

- Nystgamus 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

Aniridia: T/F

- Nystgamus 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?

--
--
--

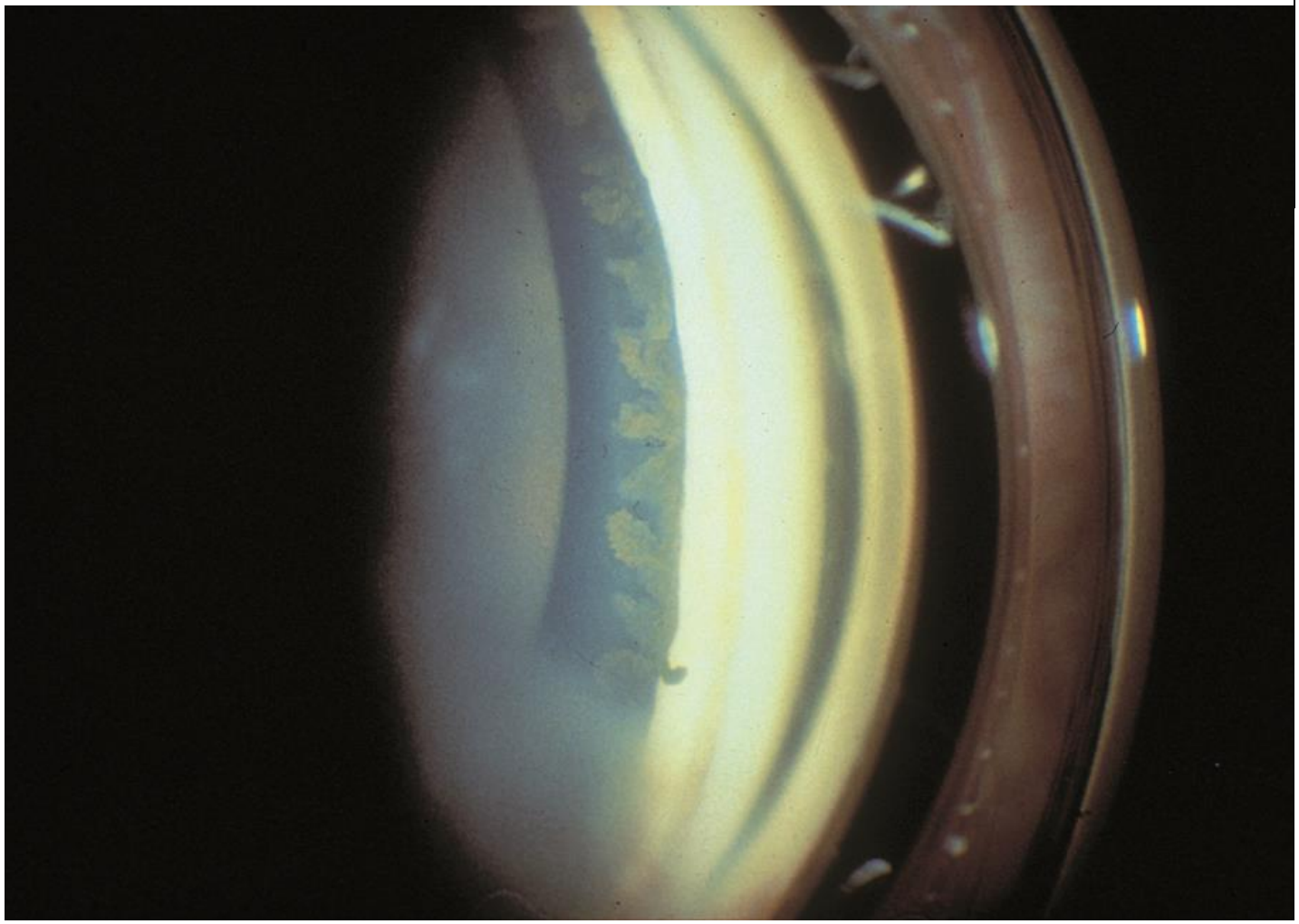
A

Aniridia: T/F

- Nystgamus 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

Q

Aniridia: T/F

- Nystgamus 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?

A

Aniridia: T/F

- Nystgamus 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?
It is called **'aniridia-associated keratopathy'**

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates

A/Q

Aniridia: T/F

- Nystgamus 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 you got a 50-50 shot…

A

Aniridia: T/F

- Nystgamus 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

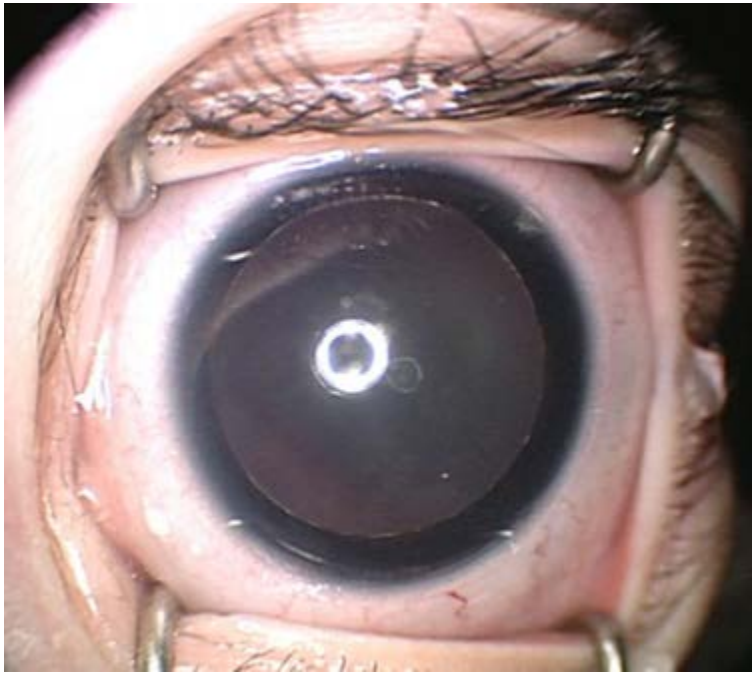
Aniridia: T/F

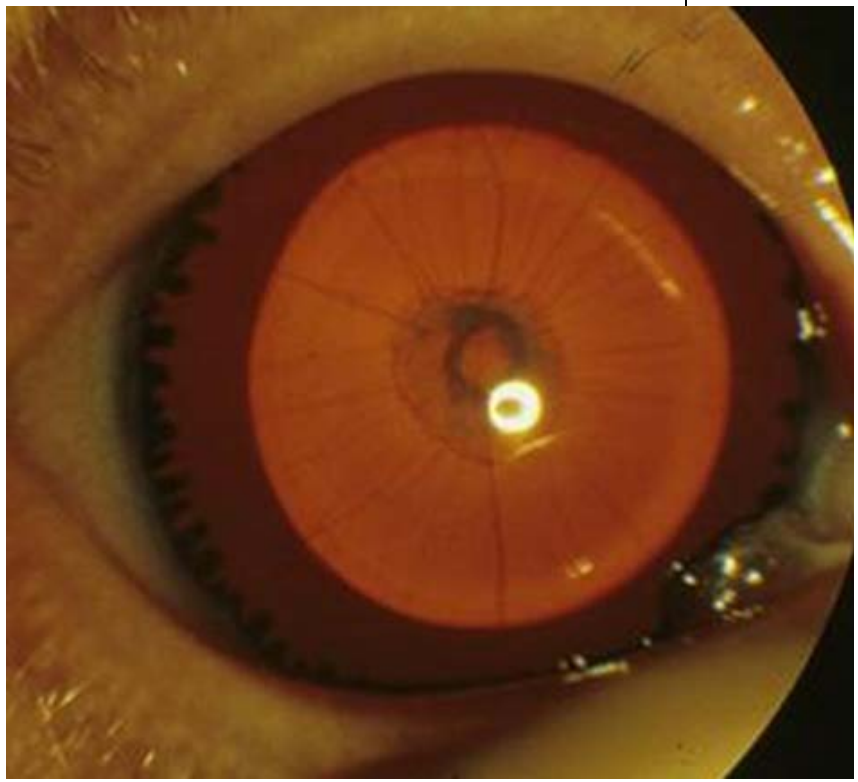
- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is almost always bilateral
- The term ‘aniridia’ is a misnomer because, in about ½ of cases, a rudimentary iris root is present

A

Aniridia: T/F

- Nystgamus 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

Q

Aniridia: T/F

- Nystgamus 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

A

Aniridia: T/F

- Nystgamus 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

Aniridia: T/F

- **Nystgamus is commonly associated True**
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is
- rudimentary iris root is present False; it's a misnomer because a rudimentary iris root is *always* present
- Aniridia is strongly associated with *<u>foveal and optic nerve hypoplasia</u>* 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.

Q

Aniridia: T/F

- Nystgamus 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?

A

Aniridia: T/F

- Nystgamus 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

Aniridia: T/F

- Nystgamus 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

A

Aniridia: T/F

- Nystgamus 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

Q

Aniridia: T/F

- Nystgamus 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

A

Aniridia: T/F

- Nystgamus 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

Q

Aniridia: T/F

- Nystgamus 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 ⅓ of cases, a rudime… a rudime…
- Aniridia… lasia 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

Aniridia: T/F

- Nystgamus 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 ⅓ of cases, a rudime… a rudime…
- Aniridia… lasia 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

Wilm’s tumor

Q

Aniridia: T/F

- Nystgamus 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 rudime… a rudime…
- Aniridia… lasia 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?

A

Aniridia: T/F

- Nystgamus 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 ⅓ of cases, a rudime… a rudime…
- Aniridia… lasia 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

Q

Aniridia: T/F

- Nystgamus 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 ⅓ of cases, a rudime… a rudime…
- Aniridia… lasia 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?

A

Aniridia: T/F

- Nystgamus 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 ⅓ of cases, a rudime… a rudime…
- Aniridia… lasia 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

Q

Aniridia: T/F

- Nystgamus 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 associat
  True
- Patients complain of (and i
- 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*?*
--**W**ilms tumor
--**A**niridia
--**G**
--**R**

A

Aniridia: T/F

- Nystgamus 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 associat
  True
- Patients complain of (and i
- 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*?*
--**W**ilms tumor
--**A**niridia
--**G**enitourinary abnormalities
--**R**etardation

Q

Aniridia: T/F

- Nystgamus 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

A

Aniridia: T/F

- Nystgamus 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

Q

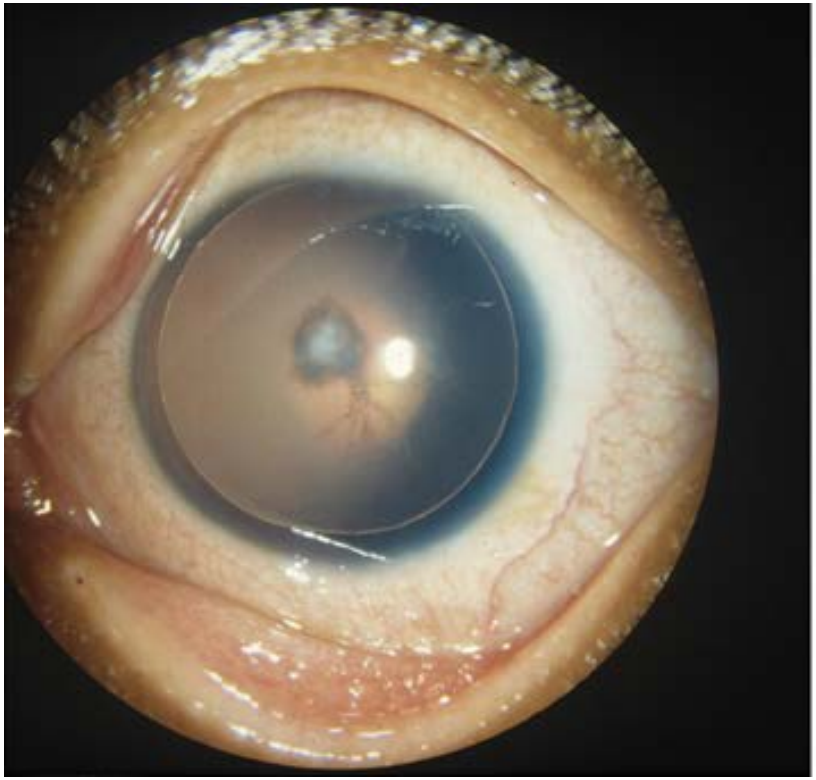
Aniridia: T/F

- Nystgamus 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

A

Aniridia: T/F

- Nystgamus 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

Aniridia: T/F

- **Nystgamus** 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 is a misnomer because, in about ½ of cases, a rudi 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 is a ***panophthalmic disorder***

Q

Aniridia: T/F

- Nystgamus 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 co… …True
- Familial ca… …**radic** cases deve…
- Aniridia is a…

How ‘early’ do aniridia pts get their ‘early onset’ cataracts?

- **Aniridia is associated with early-onset cataracts True**

A

Aniridia: T/F

- Nystgamus 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 co… …True
- Familial ca… …**radic** cases deve…
- Aniridia is a…
- **Aniridia is associated with early-onset cataracts True**

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

Q

Aniridia: T/F

- Nystgamus 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 co… …True
- Familial ca… …**radic** cases deve…
- Aniridia is a…
- **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

Aniridia: T/F

- Nystgamus 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 co… True
- Familial ca… **radic** cases deve…
- Aniridia is a…
- **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 % of aniridia cases

A

Aniridia: T/F

- Nystgamus 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 co… …True
- Familial ca… …**radic** cases deve…
- Aniridia is a…
- **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

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

Is glaucoma common in aniridia?

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A/Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

Is glaucoma common in aniridia?
Yes. Estimates vary, but it's a safe bet that at least % of aniridics develop glaucoma

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

Is glaucoma common in aniridia?
Yes. Estimates vary, but it's a safe bet that at least 50% of aniridics develop glaucoma

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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?

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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?

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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?

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 stemmin

How does this occlusion process proceed?

Is glaucoma in aniridia
Angle closure

What is the mechanism
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.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 stemmin

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.

Is glaucoma in aniridia
Angle closure

What is the mechanism
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.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 stemmin

Is glaucoma in aniridia
Angle closure

What is the mechanism
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.

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? That is, in such cases, when will the glaucoma declare itself?

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 stemmin

Is glaucoma in aniridia
Angle closure

What is the mechanism
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.

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? That is, in such cases, when will the glaucoma declare itself?
No earlier than the second decade, and often much later

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 gla *variety?*
Angl

In what ways is the angle abnormal?

Wha
That idia the angle is structurally
norm dimentary iris root. In contrast,
the angle in sporadic aniridia tends to be congenitally abnormal.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 gla *variety?*
Angl

In what ways is the angle abnormal?
The TM is poorly developed. Further, corneal endothelium may grow across the angle producing PAS.

Wha
That idia the angle is structurally
norm dimentary iris root. In contrast,
the angle in sporadic aniridia tends to be congenitally abnormal.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 gla *variety?*
Angl

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?

Wha
That idia the angle is structurally
norm dimentary iris root. In contrast,
the angle in sporadic aniridia tends to be congenitally abnormal.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is

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 gla *variety?*
Angl

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

Wha
That idia the angle is structurally
norm udimentary iris root. In contrast,
the angle in sporadic aniridia tends to be congenitally abnormal.

- **Aniridia is associated with glaucoma True**
- Aniridia is associated with early-onset cataracts True

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is a
- T
  r
  r
- 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

Defects involving what gene are the cause of aniridia?

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is a
- T
  r
  r
- 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

Defects involving what gene are the cause of aniridia?
The PAX6 gene

Q

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is a
- T
  r
  r
- 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

Defects involving what gene are the cause of aniridia?
The PAX6 gene

What other ocular abnormalities are associated with defects of the PAX6 gene?

A

Aniridia: T/F

- Nystgamus is commonly associated True
- Aniridia is associated with limbal stem cell deficiency True
- Presents unilaterally and bilaterally in roughly equal rates False; it is a
- T
  r
  r
- 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

Defects involving what gene are the cause of aniridia?
The PAX6 gene

What other ocular abnormalities are associated with defects of the PAX6 gene?
I'm glad you asked…

Q

Aniridia: T/F

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…

P

A nirida

X

6

Start with the 'P' and work down

A

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

P eters anomaly

A nirida

X

6

A

Aniridia: T/F

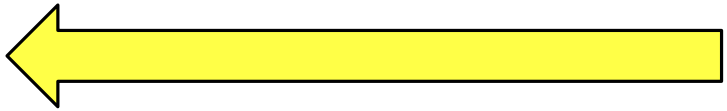
What four ocular abnormalities are attributed to the PAX6 gene?

P eters anomaly

A nirida

Congenital catara***X***

6



A

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

P eters anomaly

A nirida

Congenital catara***X***

foveal ***6*** ypoplasia

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

Q

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

Peters anomaly

What is Peters anomaly*?*

A

Aniridia: T/F

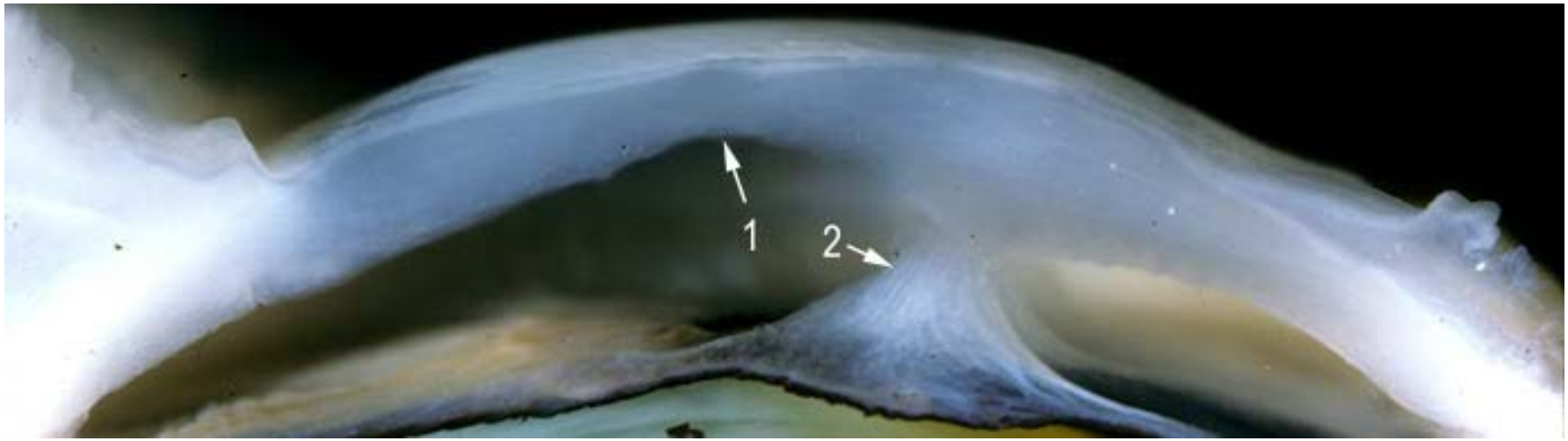
What four ocular abnormalities are attributed to the PAX6 gene?

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



Peters anomaly

Q

Aniridia: T/F

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.

How does it present?

Q/A

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

***P*eters 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.

How does it present?
As a corneal opacity at birth (it's the ***P*** in the infamous [redacted] *mnemonic for congenital cloudy cornea*).

A

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

***P*eters 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.

How does it present?
As a corneal opacity at birth (it's the ***P*** in the infamous *STUMPED mnemonic for congenital cloudy cornea*).

A

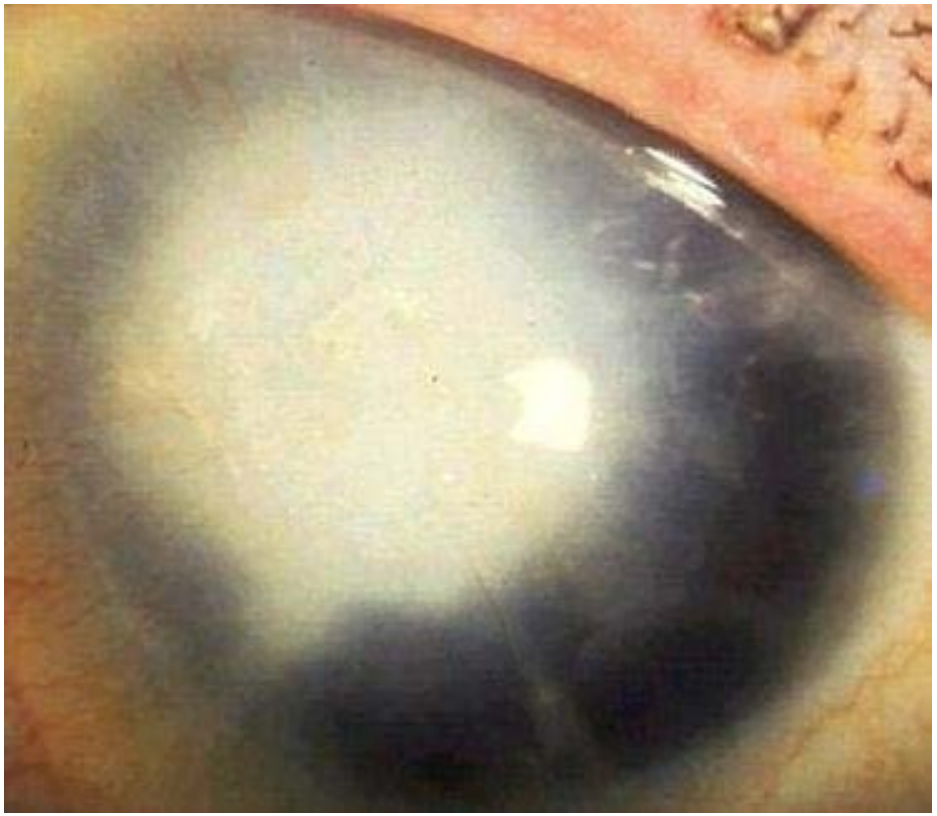
Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

***P*eters 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.

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

Q

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

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

S
T
U
M
Peters anomaly
E
D

Note: There are two S's and two E's

iris to the posterior cor

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.

A

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

What is the STUMPED mnemonic for a cloudy cornea in an infant?
Sclerocornea; **S**tromal dystrophy (CHSD)
Trauma (eg, forcep injury)
Ulcer
Mucopolysaccharidosis
Peters anomaly
Endothelial dystrophy (CHED); **E**levated IOP (congenital glaucoma)
Dermoid of the cornea

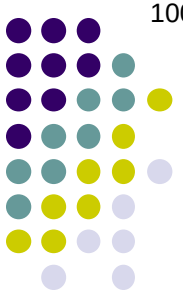
Note: There are two S's and two E's

iris to the posterior cor

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.

Q

Aniridia: T/F



What four ocular abnormalities are attributed to the PAX6 gene?

P *eters anomaly?*

A *Nirida?*

Congenital catara ***X?***

foveal ***6*** *ypoplasia?*

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

A

Aniridia: T/F

What four ocular abnormalities are attributed to the PAX6 gene?

P eters anomaly

A nirida

Congenital catara***X***

foveal ***6*** ypoplasia

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]

Q

What sort of gene is PAX6 anyway?

*What four ocular abnormalities are attributed to the **PAX6 gene**?*

P eters anomaly

A nirida

Congenital catara **X**

foveal **6** poplasia

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

A

What sort of gene is PAX6 anyway?
A homeobox gene

What four ocular abnormalities are attributed to the ***PAX6 gene?***

P eters anomaly

A nirida

Congenital catara ***X***

foveal ***6*** poplasia

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

Q

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****?*

P eters anomaly

A nirida

Congenital catara ***X***

foveal ***6*** poplasia

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

A

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****?*

P eters anomaly

A nirida

Congenital catara ***X***

foveal ***6*** poplasia

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

What sort of gene is PAX6 anyway?
A homeobox gene

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

One that **regulates morphogenesis**

What four ocular abnormalities are attributed to the ***PAX6 gene?***

P eters anomaly

A niridia

Congenital catara***X***

foveal ***6*** poplasia

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

Q

Aniridia: T/F

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
- Aniridia is associated with glaucoma True
- Aniridia is associated with early-onset cataracts True

Q/A

Aniridia: T/F

Why is **sporadic** *aniridia associated with Wilms tumor, but not* **familial** *aniridia?*
The PAX6 gene and the Wilms tumor gene (called abb.(#)) 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 same . 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 same is its next-door neighbor, it is often affected as well by these deletions.
Because of the PAX6/ same 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
- Aniridia is associated with early-onset cataracts True

A

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 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
- Aniridia is associated with early-onset cataracts True

Q

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

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

Beca

unde

What are the other components of the WAGR complex*?*
--**W**ilms tumor
--**A**niridia
--**G**enitourinary abnormalities
--**R**etardation

True

- Patients complain of (and i
- 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

A

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

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*?*
--**W**ilms tumor
--**A**niridia
--**G**enitourinary abnormalities
--**R**etardation

True

- Patients complain of (and
- 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

Q

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

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*?*
--**W**ilms tumor
--**A**niridia
--**G**enitourinary abnormalities
--**R**etardation

True

- Patients complain of (and
- 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

A

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

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*?*
--**W**ilms tumor
--**A**niridia
--**G**enitourinary abnormalities
--**R**etardation

Beca st
unde

True

- Patients complain of (and i
- 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

Q

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 c
cases l in
the P ll by
these
Beca **nust**
undergo genetic screening for the Wilms tumor defect.

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

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

A

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 c
cases l in
the P ll by
these
Beca **must**
undergo genetic screening for the Wilms tumor defect.

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

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

Q

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 [illegible] cases [illegible] in the P[illegible] by these [illegible] Beca[illegible] **must** undergo genetic screening for the Wilms tumor defect.

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?*

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

A

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
cases
the P
these
Beca
undergo genetic screening for the Wilms tumor defect.

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

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

Q

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 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

- 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

A

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 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?
Via periodic renal ultrasound

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

Q

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 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 Wilm *How often, and for how long?* *y be screened for Wilms tumor?*
Via periodic renal ultrasound

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

A

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 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 Wilm… …y 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

Q

Aniridia: T/F

- Nystgamus 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 te… …es, a rudime… …se a rudime…
- Aniridi… …oplasia True
- Patien… …rue
- 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**

Associated condition	% of aniridia pts with the associated condition
Limbal stem-cell deficiency	*?*
Glaucoma	*?*
Early-onset cataracts	*?*

To recap:

A

Aniridia: T/F

- Nystgamus 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 te… …es, a rudime… …se a rudime…
- Aniridi… …oplasia True
- Patien… …rue
- 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**

Associated condition	% of aniridia pts with the associated condition
Limbal stem-cell deficiency	90
Glaucoma	50+
Early-onset cataracts	50+

To recap: