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In this context, what is a transcription factor?
A protein that regulates the transcription process for a specific gene.
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--the **translation** of the RNA code into a protein.

*Transcription* | In this context, what is a transcription factor?
A protein that regulates the transcription process for a specific gene

*Translation* | Do transcription factors play an important role in the genetic process?
Indeed they do. In fact, about 10% of all genes in humans code for transcription factors!
With respect to genetics, to what does the term Central Dogma refer?

It refers to the two steps involved in transforming genetic information into protein:

--- The first step is the **transcription** of DNA code into RNA code; followed by

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Does transcription-factor dysfunction play a role in ophthalmic dz?
Indeed it does—a number of important ophthalmic conditions can be traced to transcription-factor mutations
With respect to genetics, to what does the term **Central Dogma** refer?

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The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?

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With respect to genetics, to what does the term **Central Dogma** refer?

It refers to the two steps involved in transforming genetic information into protein: --the transcription process into RNA; followed by --the translation process into a protein.

*Transcription*  

**In this context, what is a transcription factor?**

A protein that regulates the transcription process for a specific gene.

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*The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?*

--PAX2  
--PAX3  
--PAX6
In the present context, what is the origin of the word PAX? Where does it come from?

The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?

-- PAX2
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In the present context, what is the origin of the word PAX? Where does it come from? It is a portmanteau of the term ‘PAired (homeo)boX’.

The Fundamentals book refers to PAX genes both as ‘paired homeobox’ and ‘paired box’ genes.

The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?

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Generally speaking, what are PAX genes involved in?

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Generally speaking, what are PAX genes involved in?
Morphogenesis

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--PAX3?
--PAX6?
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Generally speaking, what are PAX genes involved in? Morphogenesis

Of these three PAX genes, which is most important to the development of the eye? PAX6. The Fundamentals book refers to it as “the master switch for eye development.” The Peds book says, “The PAX6 gene is the master control gene for eye morphogenesis.”

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--PAX2
--PAX3
--PAX6!

Next let’s take a closer look at PAX6
There are four ocular abnormalities attributed to the PAX6 gene. What are they?

The mnemonic is...
There are four ocular abnormalities attributed to the PAX6 gene. What are they?

The mnemonic is... PAX6
There are four ocular abnormalities attributed to the PAX6 gene. What are they?
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- Peters anomaly
- Anirida
- PX
- 6
There are four ocular abnormalities attributed to the PAX6 gene. What are they?

- Peters anomaly
- Anirida
- Congenital cataract
- Aniridic-like syndrome (PAX6)
There are four ocular abnormalities attributed to the PAX6 gene. What are they?

- Peters anomaly
- Anirida
- Congenital cataract
- Foveal hypoplasia

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

Peters anomaly
Anirida
Congenital cataract
Foveal hypoplasia

Endeavor to remember all of these. But if you have to pick just one to remember, make it…
There are four ocular abnormalities attributed to the PAX6 gene. What are they?

- Anirida
- Congenital cataract
- Foveal hypoplasia
- Peters anomaly

Endeavor to remember all of these. But if you have to pick just one to remember, make it... anirida.
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Peters anomaly
Anirida
Congenital cataract
Foveal hypoplasia

Endeavor to remember all of these. But if you have to pick just one to remember, make it... **aniridia.*** *Almost all cases of aniridia are 2ndry to mutations involving PAX6.*
There are four ocular abnormalities attributed to the PAX6 gene. What are they?

- Peters anomaly
- Anirida
- Congenital cataract
- Foveal hypoplasia

One final takeaway point regarding anirida...

Endeavor to remember all of these. But if you have to pick just one to remember, make it... **anirida**. *Almost all cases of anirida are secondary to mutations involving PAX6.*
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

The takeaway point: Don't think of aniridia as an iris condition!
The BCSC characterizes it is a panophthalmic disorder Because all are tied to PAX6, you know foveal hypoplasia, ON hypoplasia and cataracts are associated with aniridia.

One final takeaway point regarding aniridia…

Because all are tied to PAX6, you know foveal hypoplasia, ON hypoplasia and cataracts are associated with aniridia.

foveal and optic nerve hypoplasia

cataracts
Q

PAX Ophthalmicana

...But you need to know the three other eye findings also associated with aniridia:

One final takeaway point regarding aniridia...

(Hints forthcoming)
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.
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…But you need to know the three other eye findings also associated with aniridia:

foveal and optic nerve hypoplasia
Angle-related condition
cataracts

One final takeaway point regarding aniridia… (Hints forthcoming)
Nystagmus

limbal stem cell deficiency

Aniridia is associated with limbal stem cell deficiency

Presents unilaterally and bilaterally in roughly equal rates

The term 'aniridia' is a misnomer because, in about ½ of cases, a rudimentary iris root is present

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Nystagmus

limbal stem cell deficiency

foveal and optic nerve hypoplasia

glaucoma

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--PAX2
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--PAX6

Next, we’ll do PAX3
With what eponymous syndrome is PAX3 associated?
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**Waardenburg Syndrome**
Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?

--
--
--
Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?
--Heterochromia iridis
--Synophrys
--Dystopia canthorum

PAX Ophthalmicana
Waardenburg syndrome: Heterochromia iridis, dystopia canthorum, and mild synophrys
(What the heck is synophrys?)
Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?
--Heterochromia iridis
--Synophrys
--Dystopia canthorum

What the heck is synophrys?

The presence of a white forelock (ie, an isolated streak of white hair in the forehead region)

The formal medical term for a unibrow
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What the heck is synophrys?
The formal medical term for a unibrow
Congenital lid abnormalities: Matching

Waardenburg syndrome: Synophrys
Congenital lid abnormalities: Matching

Waardenburg syndrome: Heterochromia iridis, dystopia canthorum, and mild synophrys

*(What the heck is dystopia canthorum?)*
What 3 ophthalmic findings are classic for Waardenburg syndrome?

- Heterochromia iridis
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What the heck is dystopia canthorum?
What 3 ophthalmic findings are classic for Waardenburg syndrome?
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What the heck is dystopia canthorum?
Lateral displacement of the canthi (ie, telecanthus) PLUS laterally displaced lacrimal puncta
Waardenburg Syndrome

What 3 ophthalmic findings are classic for Waardenburg syndrome?
-- Heterochromia iridis
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What non-ophthalmic finding is classic for Waardenburg syndrome?
The presence of a white forelock (ie, an isolated streak of white hair in the forehead region)

What the heck is dystopia canthorum?
Lateral displacement of the canthi (ie, telecanthus) PLUS laterally displaced lacrimal puncta

How on earth are you supposed to recognize that the puncta are too lateral?

Draw an imaginary vertical line from the upper to the lower puncta. If this line crosses the cornea, the puncta are displaced. (Next time you examine a pt at the slit-lamp, take note of whether such a line crosses their cornea [it won’t].)
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Dystopia canthorum. Note the telecanthus, and laterally displaced lacrimal puncta.
Waardenburg Syndrome

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-- Heterochromia iridis
--Synophrys
--Dystopia canthorum

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--Dystopia canthorum

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The presence of a white forelock (ie, an isolated streak of **white hair** in the forehead region)
Congenital lid abnormalities: Matching

Waardenburg syndrome: White forelock
Congenital lid abnormalities: Matching

Note that Waardenburg syndrome has forms that do not involve heterochromia.
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The Fundamentals book lists three transcription-factor (genes) that re especially important for the eye, ranked as follows:

- PAX2
- PAX3
- PAX6

Last and most definitely least…PAX2 mutations present with hypoplasia of the optic nerve, and non-eye hypoplasia
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The Fundamentals book lists three transcription-factor (genes) that are especially important for the eye—what are they?

--PAX2
--PAX3
--PAX6

Last and most definitely least…PAX2 mutations present with colobomas of the optic nerve, and renal hypoplasia