Q

On LHON

- LHON stands for

A

On LHON

- LHON stands for *Leber's hereditary optic neuropathy*

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- LHON is more likely to affect males vs females, despite the fact that transmission is organelle

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Where does LHON rank among inherited mitochondrial diseases in terms of incidence?

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Where does LHON rank among inherited mitochondrial diseases in terms of incidence?
It is #1--the most common

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What percentage of LHON cases are male, and what percentage are female?

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What percentage of LHON cases are male, and what percentage are female?
80-90 are male; 10-20 are female

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As female offspring inherit the same genotype, they would be expected to display the phenotype at rates equal to those of males

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- Presentation is typically unilateral; the fellow eye will almost [always vs never] become affected

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- Onset is typically in the #th - #th decades

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How old at onset were the youngest and oldest confirmed cases?
Youngest:
Oldest:

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How old at onset were the youngest and oldest confirmed cases?
Youngest: **1 year old**
Oldest: **80**

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  - Scotoma (usually location in VF)
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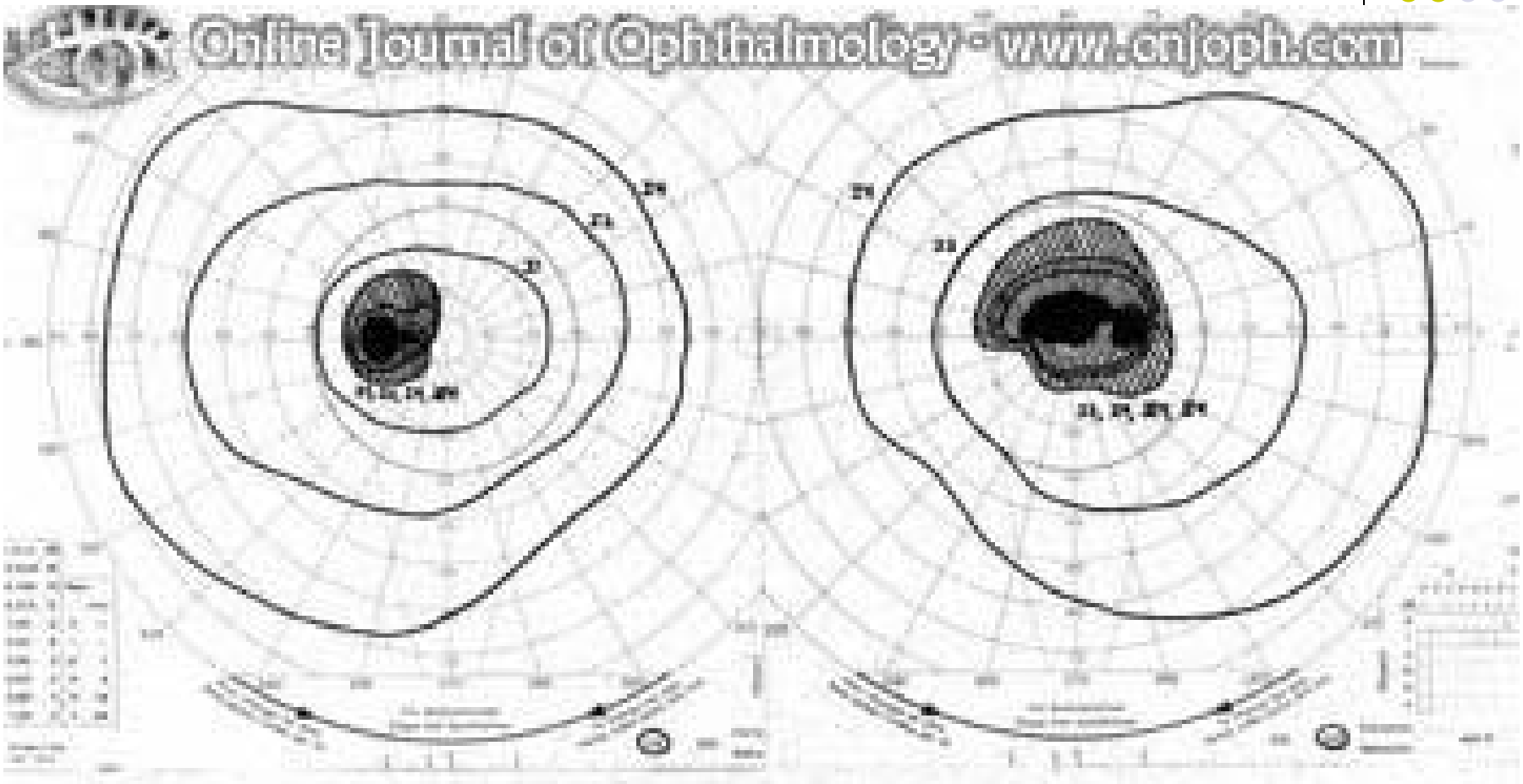

LHON: Central/cecocentral scotomata

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  - Dyschromatopsia (usually red - green vs blue - yellow)

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Color deficiency issues are a real pain. It is true that the majority of inherited defects are **red-green**, and the **vast** majority of **blue-yellow** defects are acquired. However, a significant proportion of acquired defects are **red-green**, not **blue-yellow**. Thus, if a patient has a **blue-yellow** defect, it is most assuredly acquired.
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*How can you tell if a **red-green** deficiency is acquired?*
1)
2)
3)
4)

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*How can you tell if a **red-green** deficiency is acquired?*
1) If it is in one eye only
2) If the patient is female (females can have inherited **red-green** defects, but it is highly unusual)
3) If it is sectoral (i.e., one portion of the visual field is desaturated compared to others)
4) The clinical setting; i.e., if the patient is complaining of decreased acuity, field loss, pain with movement, etc

Q

On LHON

- Classic DFE findings:
  - ONH…
  - ONH…
  -

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On LHON

- Classic DFE findings:
  - ONH…telangiectasias
  - ONH…pseudoedema
  -

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  - Retinal arteriolar…

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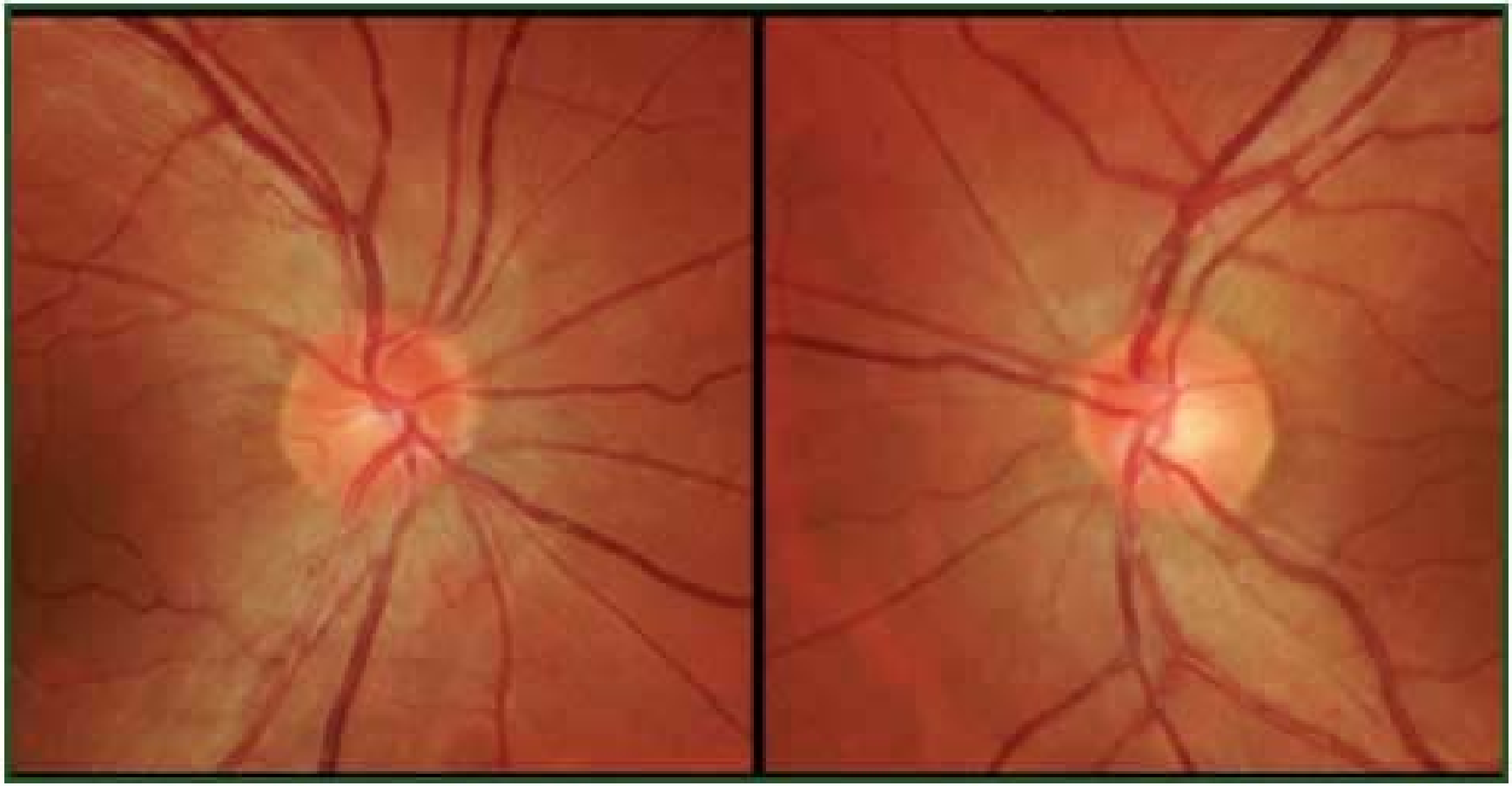
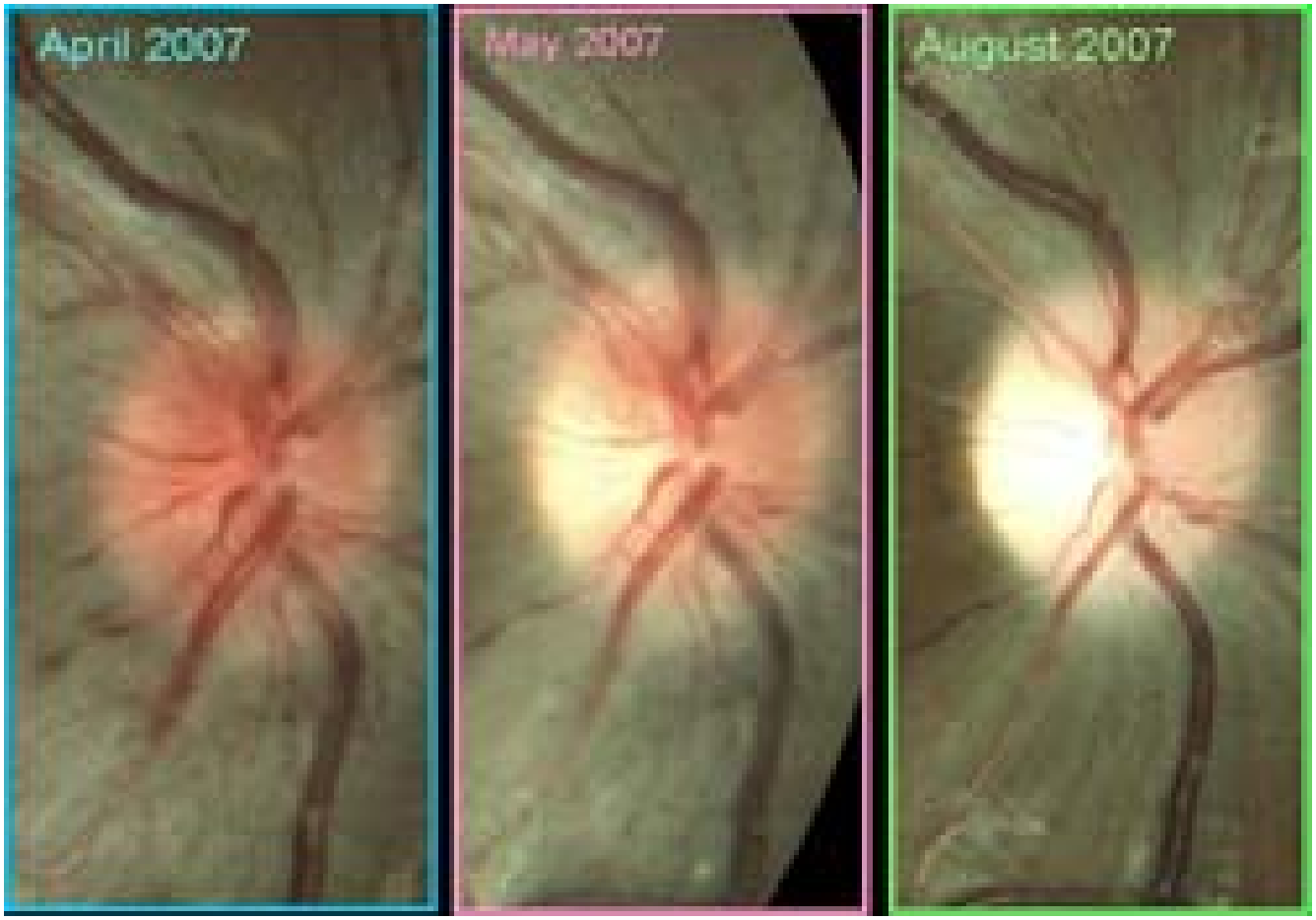
  - Retinal arteriolar…tortuosity

On LHON



6. A careful comparison of the discs revealed subtle findings—disc hyperemia, relative opacity of the retinal nerve fiber layer, and mild telangiectatic (corkscrew) vessels—that were more marked in the right eye.

On LHON



LHON: Progression of ONH atrophy

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- Classic DFE findings:
  - ONH…telangiectasias
  - ONH…pseudoedema
  - Retinal arteriolar…tortuosity
- Cardiac co-morbidity: eponym-eponym-eponym

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What is WPW?
An abnormality of cardiac **conduction**

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What is WPW?
An abnormality of cardiac **conduction**

What are the classic EKG findings in WPW?
--The PR interval is abnormally…*[long vs short]*
--
--

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What is WPW?
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What are the classic EKG findings in WPW?
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--The QRS complex is abnormally…*[wide vs narrow]*

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--The QRS complex onset is…*[classic descriptor]*

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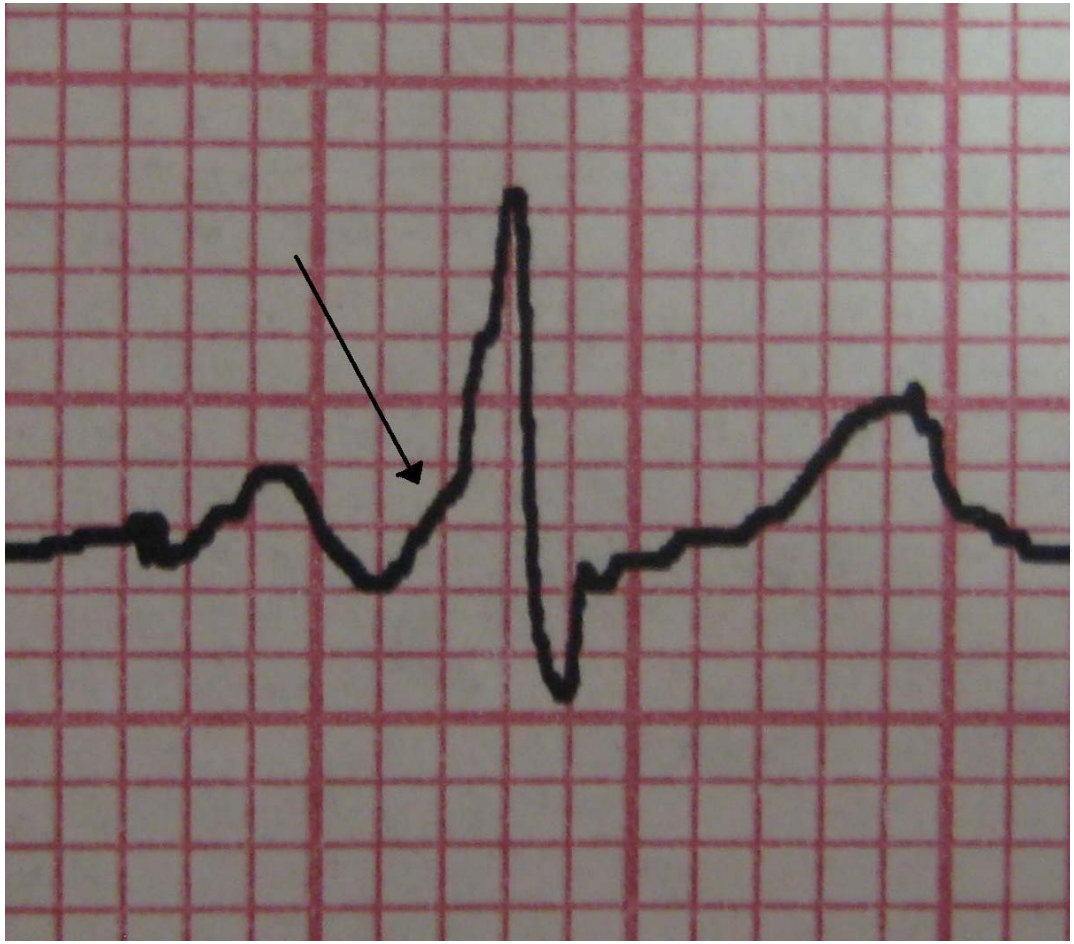
--The QRS complex onset is…'**slurred**'

On LHON



WPW: Slurred onset of the QRS complex

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WPW renders pts prone to what abnormal rhythm?

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WPW renders pts prone to what abnormal rhythm?
Supraventricular tachycardia (SVT)

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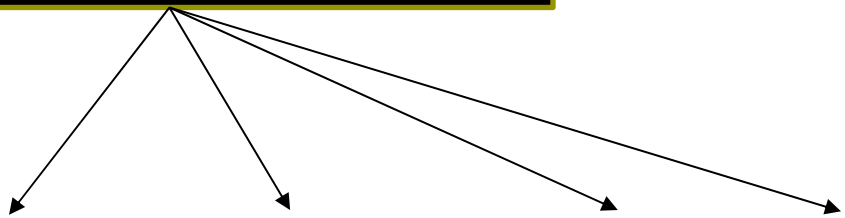

Speaking of cardiac conduction issues—when an eye dentist encounters those words, four conditions should come to mind (although admittedly, one of them probably needn't stay there for long).

No question yet—proceed when ready

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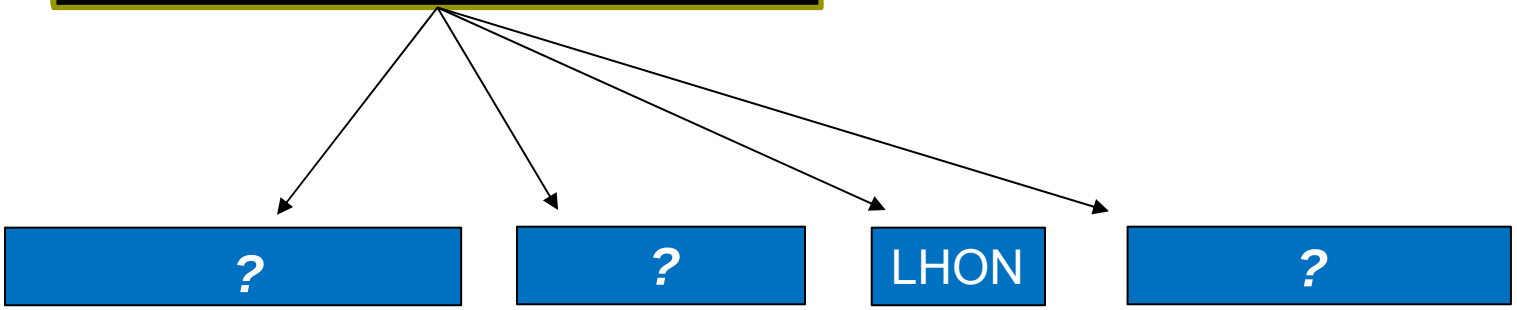

Speaking of cardiac conduction issues—when an eye dentist encounters those words, four conditions should come to mind (although admittedly, one of them probably needn't stay there for long). One is LHON. What are the other three?

A

On LHON

- Classic DFE findings:
  - ONH…telangiectasias
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  - Retinal arteriolar…tortuosity
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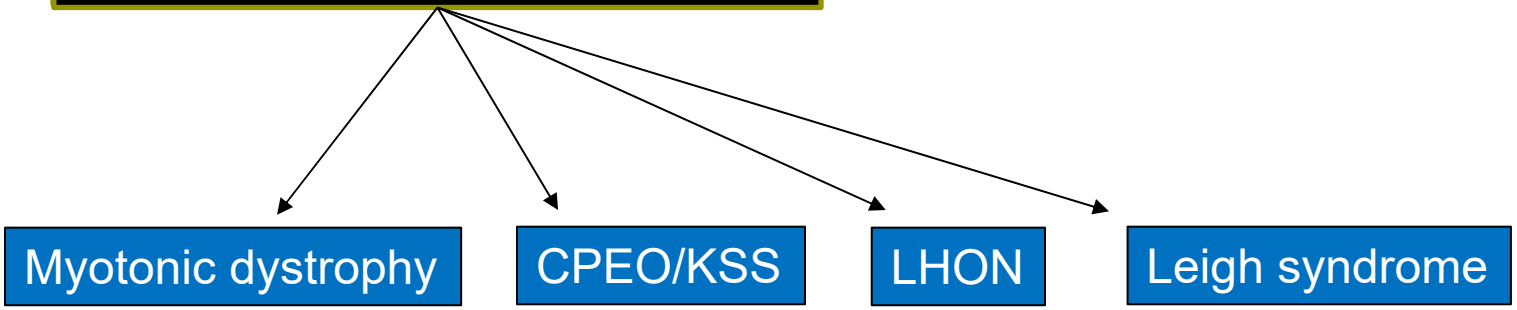

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Q

On LHON

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In a nutshell, what sort of condition is myotonic dystrophy?

Myotonic dystrophy

Speaking of cardia
four conditions sho
needn't stay there

A

On LHON

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Myotonic dystrophy

In a nutshell, what sort of condition is myotonic dystrophy?
An inherited AD progressive systemic condition that results in ophthalmoplegia

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Q/A

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What are its other ocular manifestations?
Bilateral symmetric ptosis--
Pigmentary retinopathy--
two-word description cataracts--

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What are its other ocular manifestations?
Bilateral symmetric ptosis--
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'Christmas tree' cataracts--

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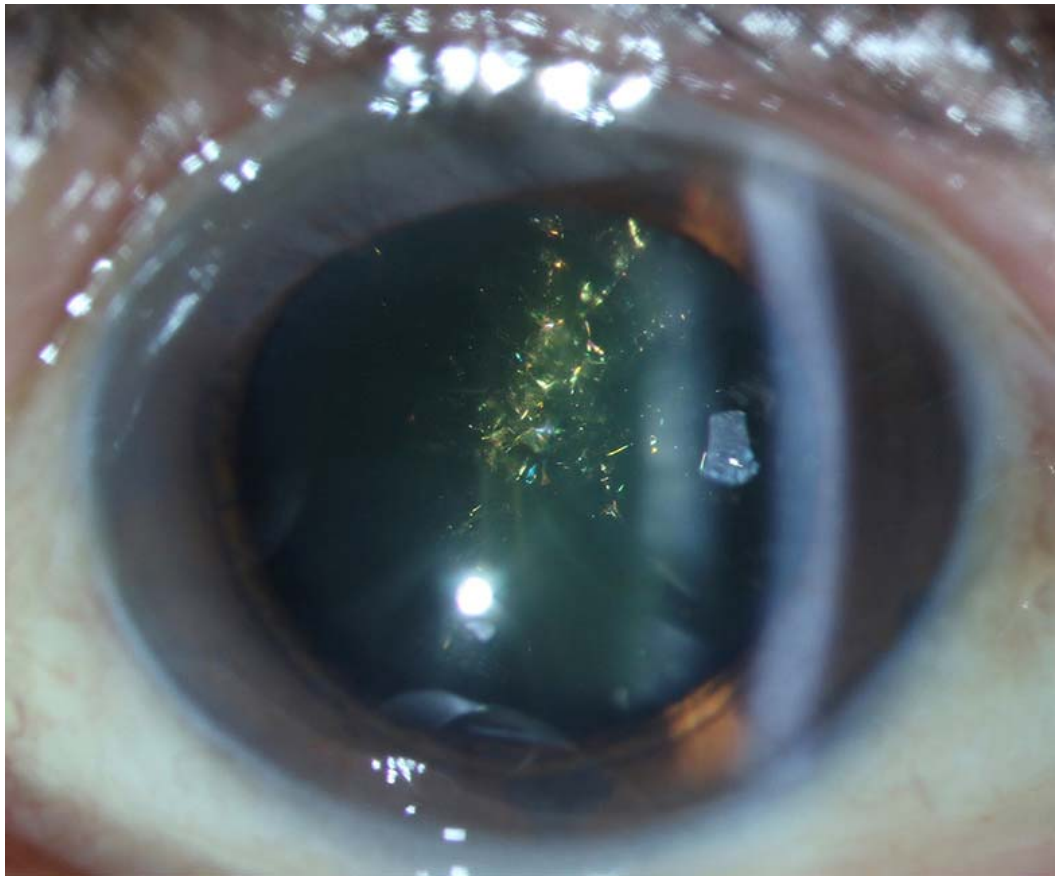
On LHON



Myotonic dystrophy: Christmas tree cataract

Q

On LHON

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Bilateral symmetric ptosis--
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--
--
--
--

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Q/A

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Cardiac conduction issues--
Myotonia--
Characteristic facies--
Frontal balding--
Low intelligence--

Speaking of cardia
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Bilateral symmetric ptosis--
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Cardiac conduction issues--
Myotonia--
Characteristic 'hatchet' facies--
Frontal balding--
Low intelligence--

Speaking of cardia
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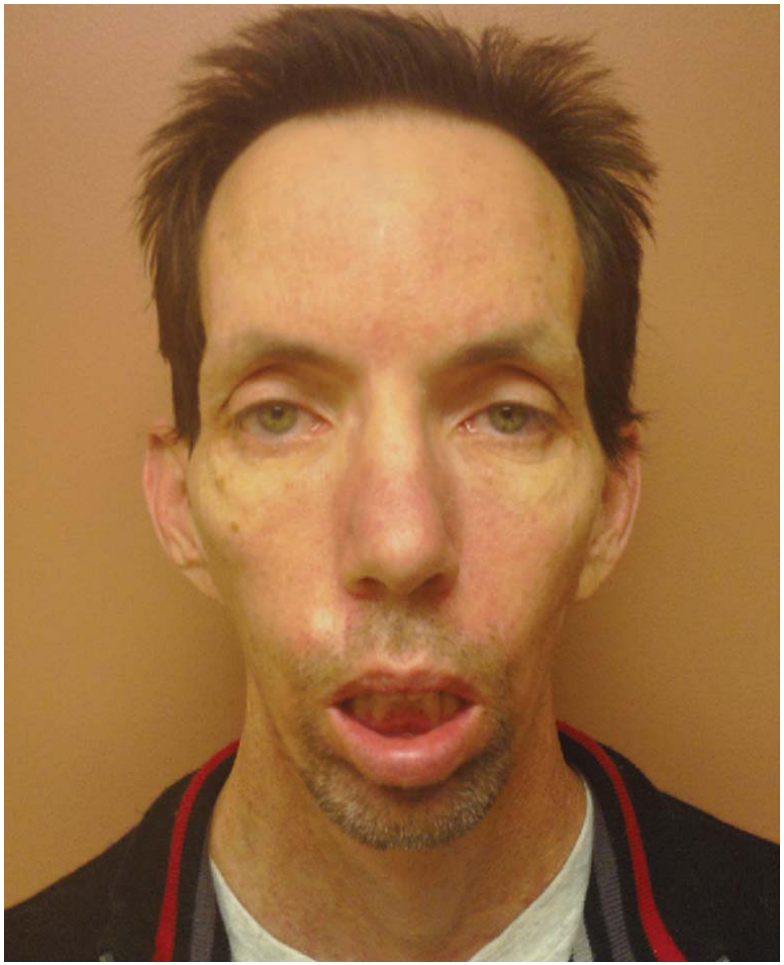
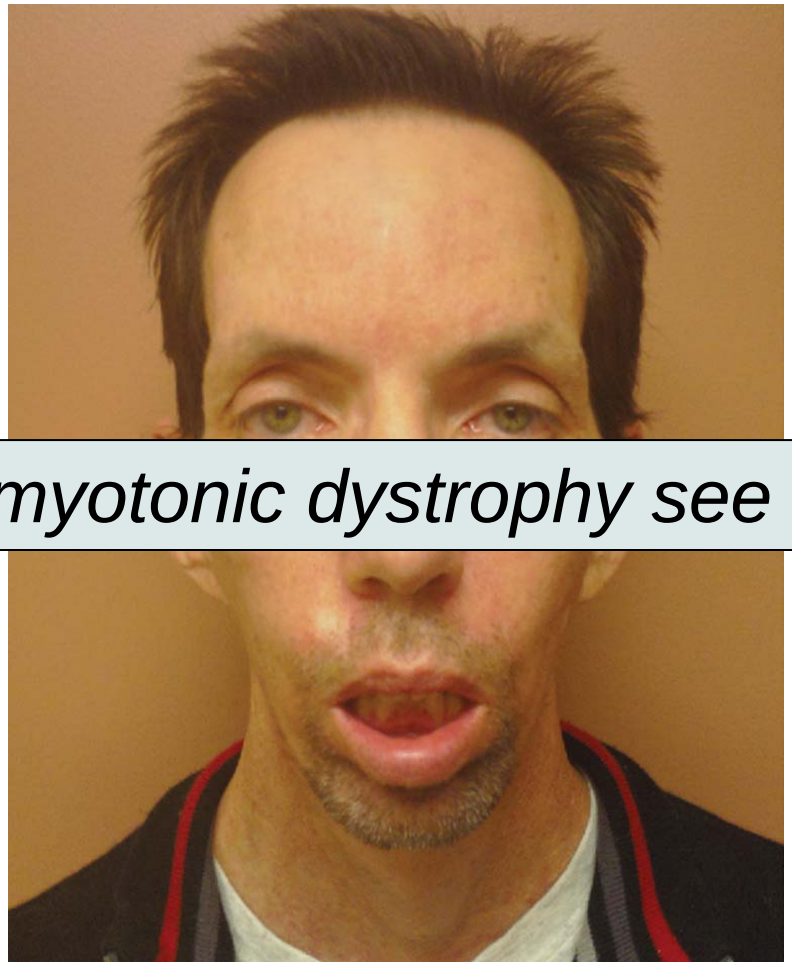
On LHON



Myotonic dystrophy: Hatchet face and frontal balding

On LHON



For more on myotonic dystrophy see slide-set O21

Myotonic dystrophy: Hatchet face and frontal balding

Q

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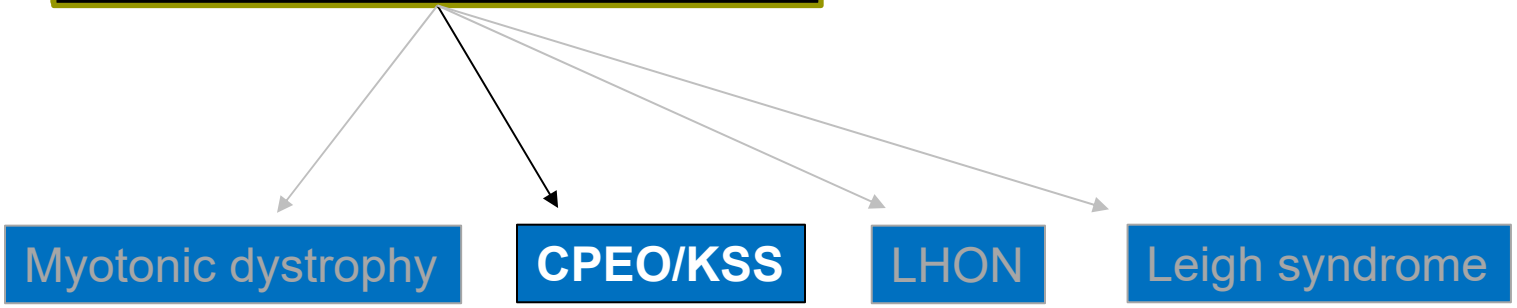

What do CPEO *and* KSS *stand for in this context?*
--CPEO:
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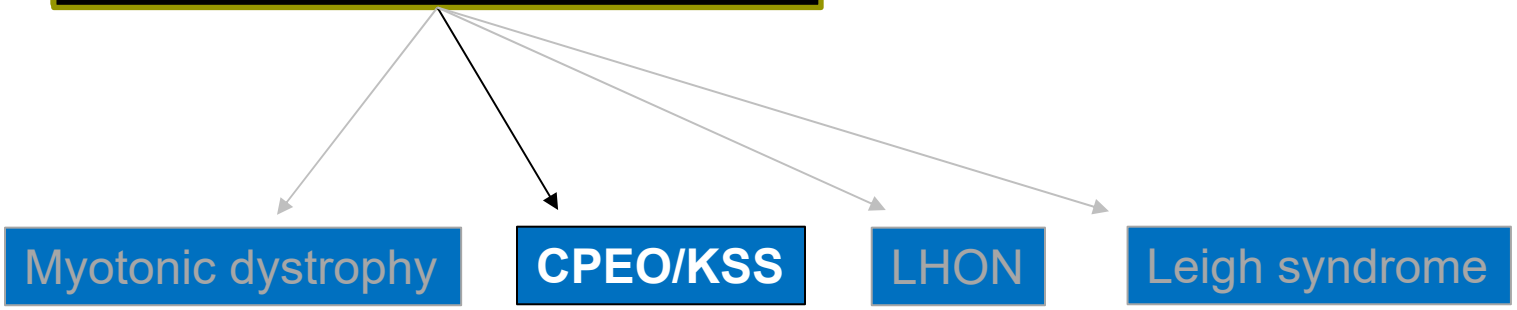

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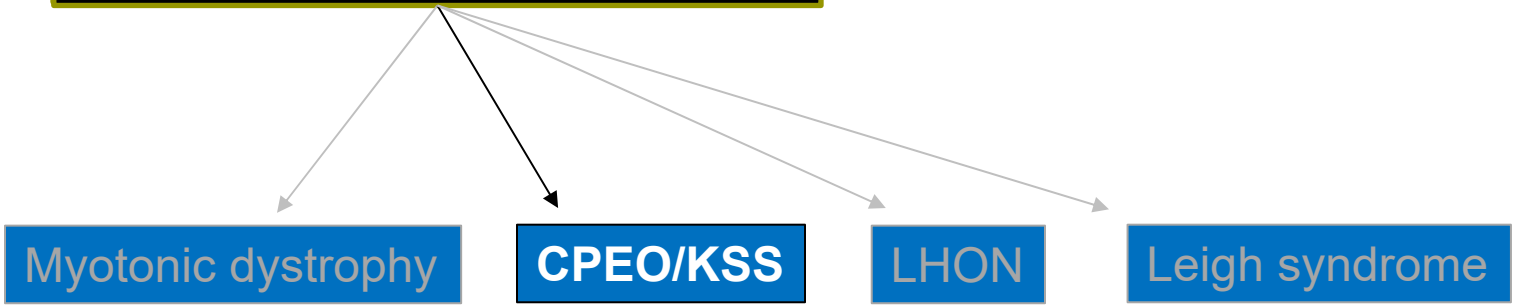

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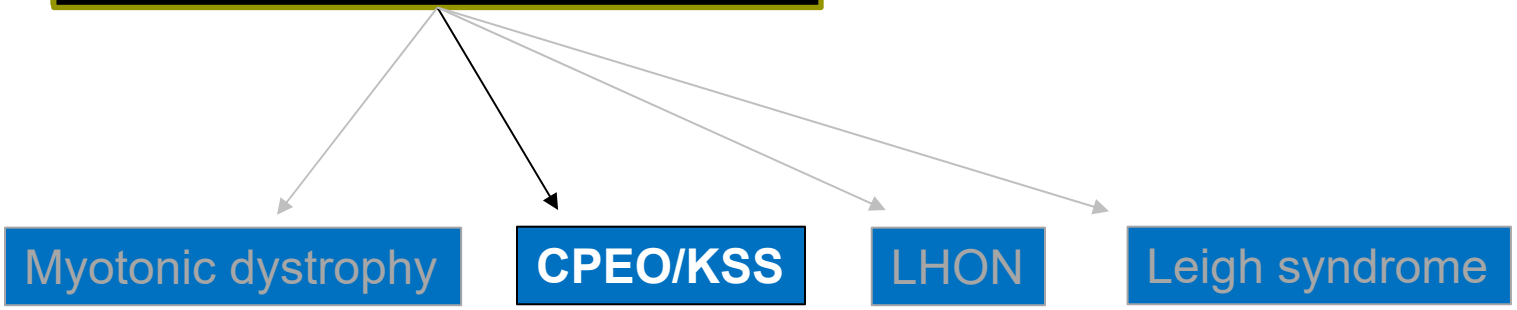

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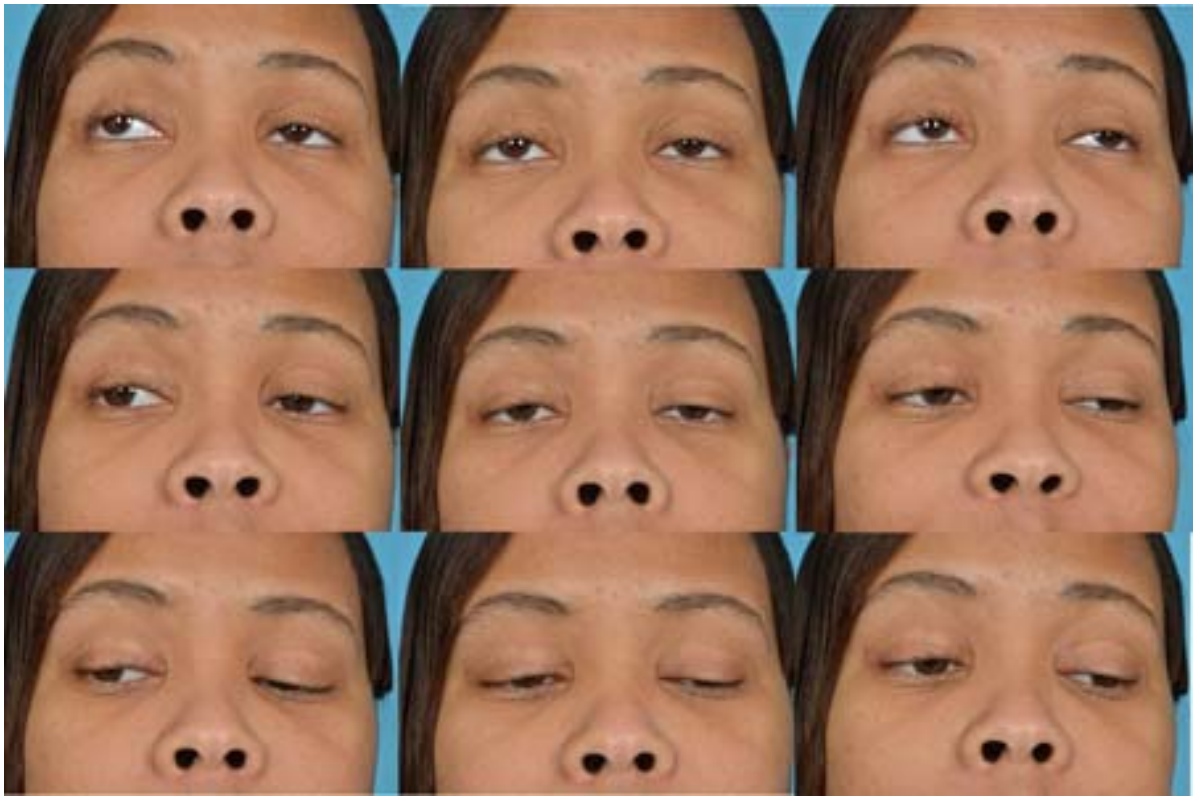
On LHON



CPEO: Symmetric ophthalmoplegia

Q

On LHON

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  - ONH…pseudoedema
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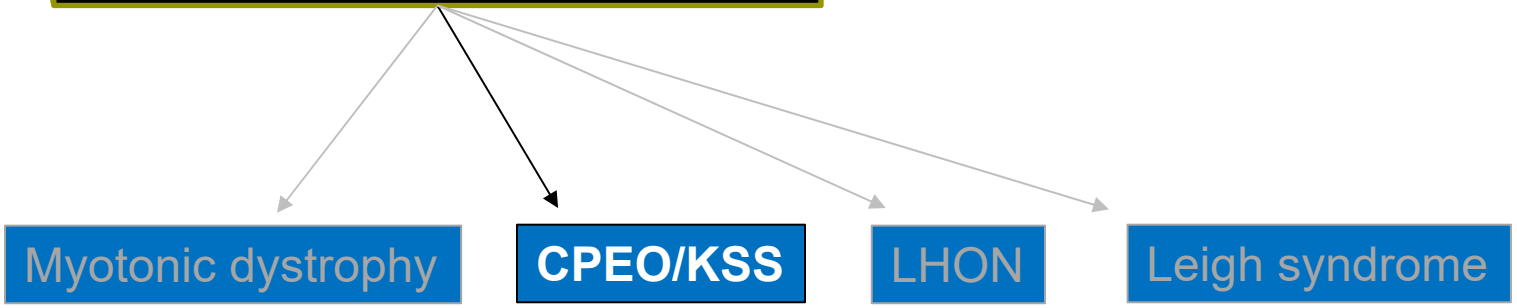
- **Cardiac co-morbidity**: Wolf-Parkinson-White



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Which muscles are affected first?

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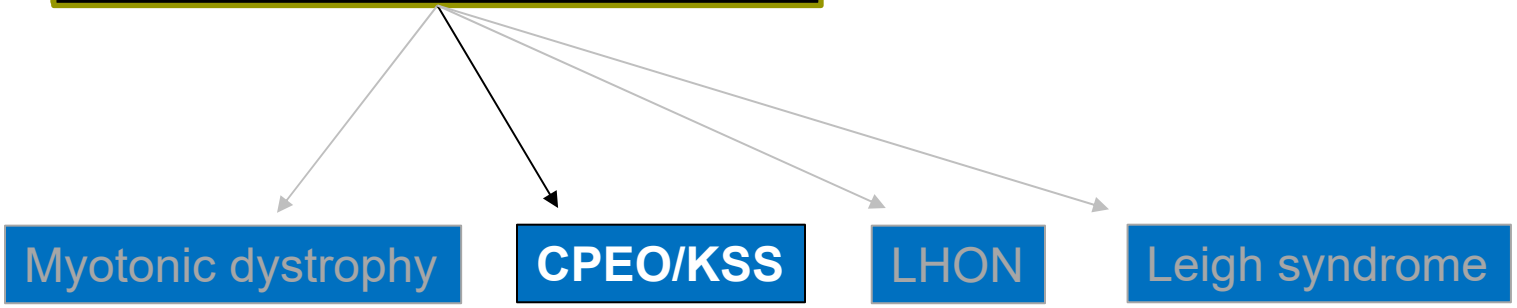

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Which muscles are affected first?
The levators, resulting in ptosis

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On LHON

CPEO: Progressive ptosis

Q

On LHON

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  - ONH…pseudoedema
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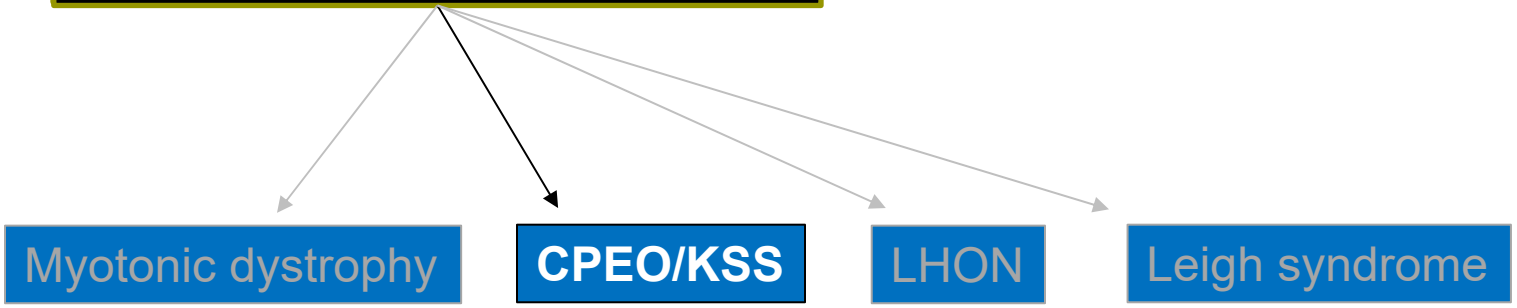

What do CPEO *and* KSS *stand for in this context?*
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KSS has a classic triad—what is it?

st encounters those words,
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On LHON

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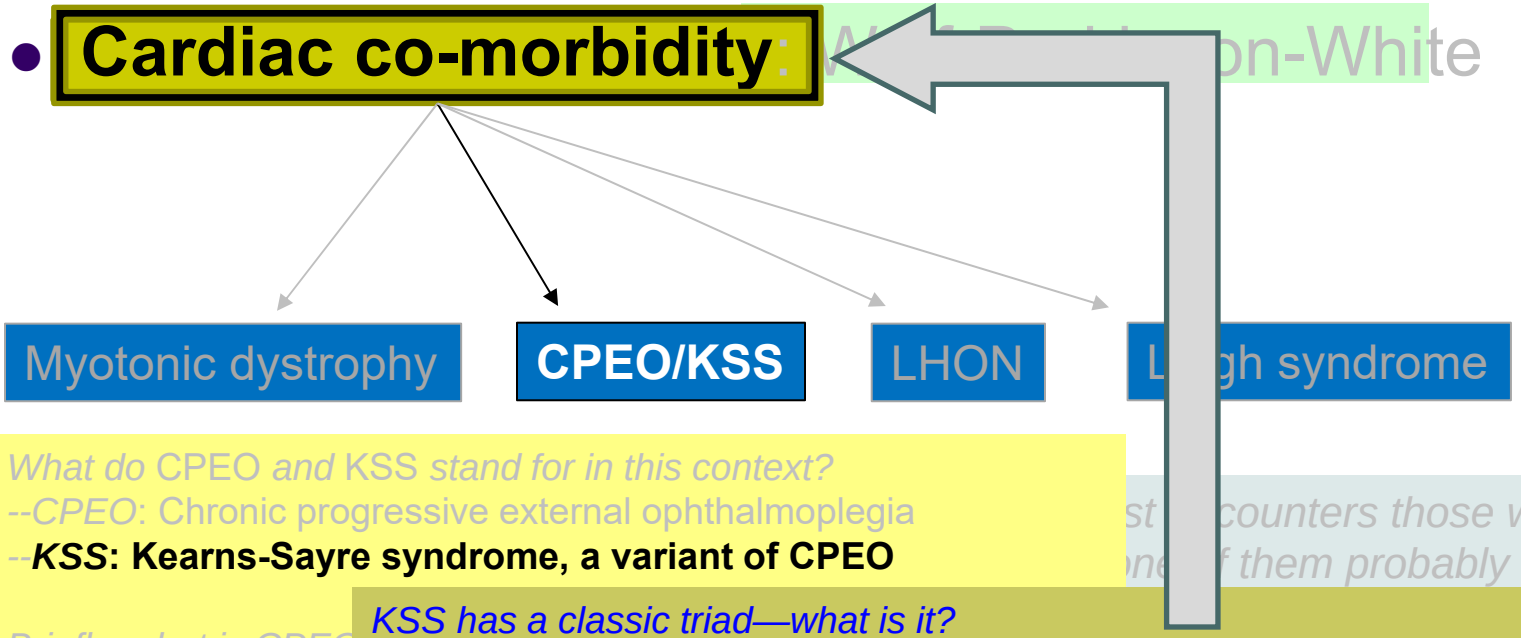

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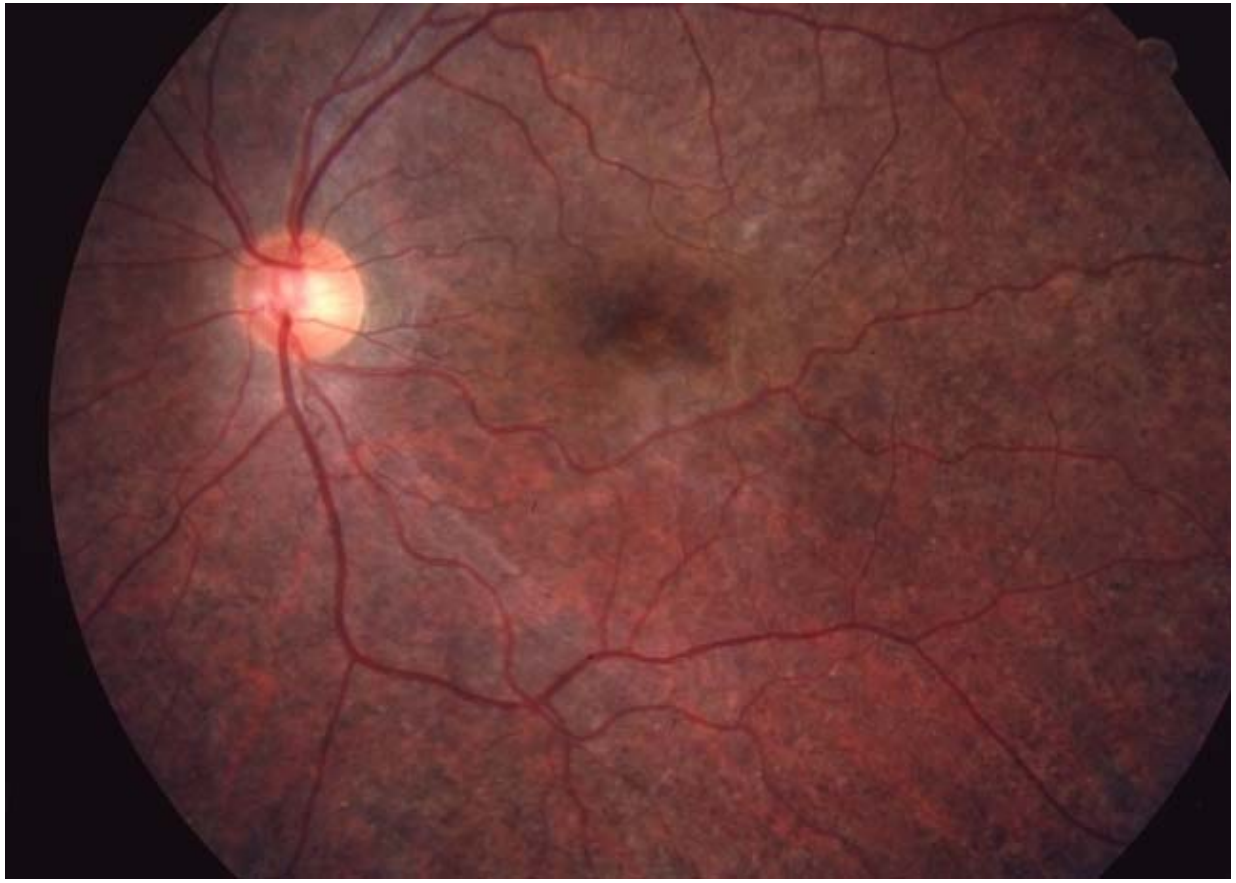
KSS has a classic triad—what is it?
CPEO + pigmentary retinopathy + **cardiac conduction abnormalities**

On LHON



CPEO: Pigmentary retinopathy

On LHON

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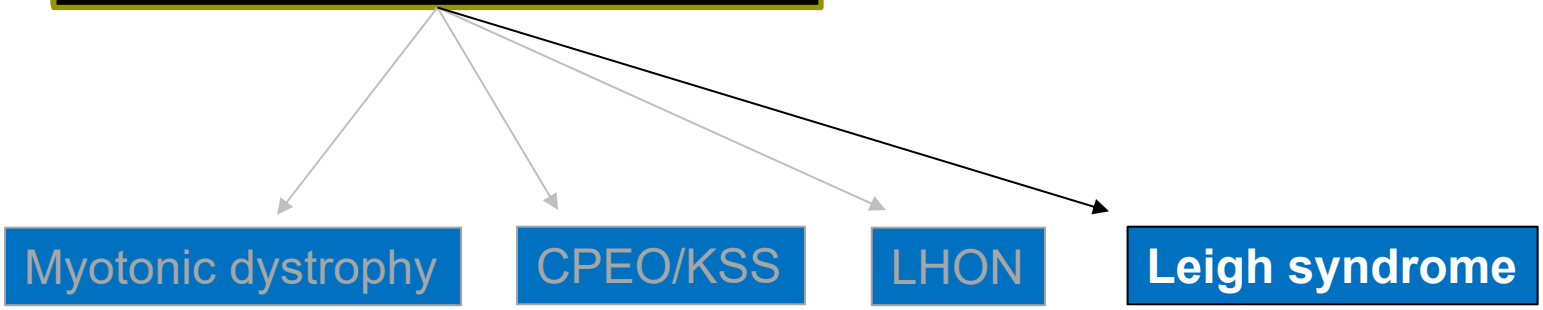
- **Cardiac co-morbidity**: Wolf-Parkinson-White



Speaking of cardiac conduction issues—when an eye dentist encounters those words, four conditions should come to mind (al… needn't stay there for long). One is LHO…

Leigh syndrome is the one you can probably forget. (It has a full entry in *Eyewiki*, but receives only one mention—in a Table—in the *BCSC*.)

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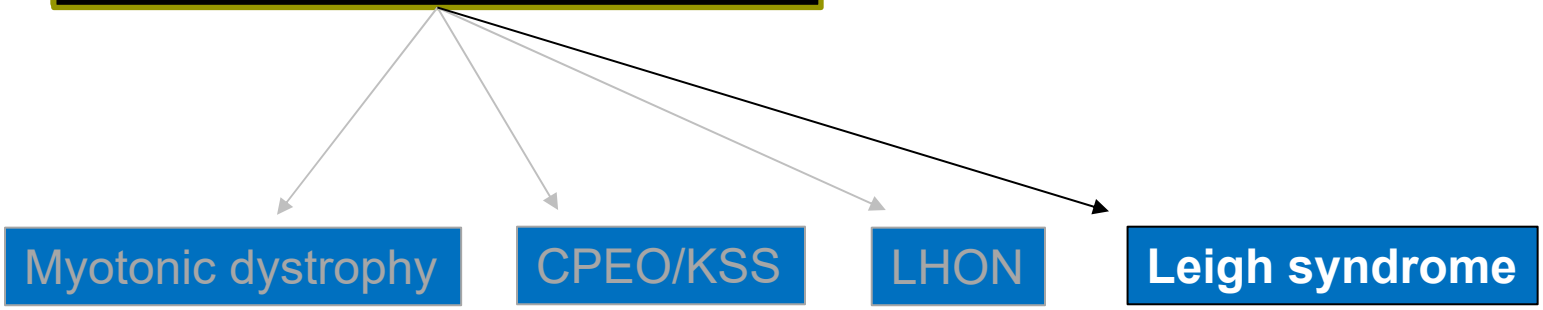

Speaking of cardiac conduction issues—when an eye dentist encounters those words, four conditions should come to mind (al needn't stay there for long). One is LHO

Leigh syndrome is the one you can probably forget. (It has a full entry in *Eyewiki*, but receives only one mention—in a Table—in the *BCSC*.) It is a mitochondrial condition that presents in childhood with cognitive and motor decline, ophthalmoplegia, and optic atrophy.

Q

On LHON

- Classic DFE findings:
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  - ONH…pseudoedema
  - Retinal arteriolar…tortuosity
- Cardiac co-morbidity: Wolf-Parkinson-White
- Diagnosis: Blood assay for abb + word

A

On LHON

- Classic DFE findings:
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  - ONH…pseudoedema
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- Cardiac co-morbidity: Wolf-Parkinson-White
- Diagnosis: Blood assay for mDNA mutation

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What are the genetic positions for the three most common mutations?

A

On LHON

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What are the genetic positions for the three most common mutations?
11778, 3460 and 14484

Q

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Which is associated with the poorest ultimate vision?

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Which carries the lowest likelihood of spontaneous visual recovery?

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- Cardiac co-morbidity: Wolf-Parkinson-White
- Diagnosis: Blood assay for mDNA mutation
- Treatment:

A

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- Cardiac co-morbidity: Wolf-Parkinson-White
- Diagnosis: Blood assay for mDNA mutation
- Treatment: None, unfortunately