LHON stands for Leber's hereditary optic neuropathy
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LHON is more likely to affect males, despite the fact that transmission is mitochondrial.
LHON stands for *Leber’s hereditary optic neuropathy*

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Where does LHON rank among inherited mitochondrial diseases in terms of incidence?
LHON stands for Leber's hereditary optic neuropathy.

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

On LHON

- LHON stands for *Leber’s hereditary optic neuropathy*
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*What percentage of LHON cases are male, and what percentage are female?*
On LHON

- LHON stands for *Leber’s hereditary optic neuropathy*
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**What percentage of LHON cases are male, and what percentage are female?**
80-90 are male; 10-20 are female
On LHON

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

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*How are mitochondrial disorders inherited; ie, what is the pattern?*
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What percentage of LHON cases are male, and what percentage are female?
80-90 are male; 10-20 are female.

How are mitochondrial disorders inherited; ie, what is the pattern?
Maternal; ie, women pass it along to all their biological offspring.
On LHON

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*Why are mitochondrial diseases inherited maternally?*
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Why are mitochondrial diseases inherited maternally?
Because all mitochondria derive from those present in the egg at the moment of conception (ie, none are contributed by the father via the sperm)
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Why are mitochondrial diseases inherited maternally?
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Given that LHON is a mitochondrial disorder, why is its strong male preponderance unusual?
LHON stands for *Leber’s hereditary optic neuropathy*

LHON is more likely to affect **males**, despite the fact that transmission is **mitochondrial**

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**Given that LHON is a mitochondrial disorder, why is its strong male preponderance unusual?**

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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OK then, so why don’t females develop LHON at the same rate as males?
LHON stands for *Leber’s hereditary optic neuropathy*. LHON is more likely to affect males, despite the fact that transmission is mitochondrial.

**Q/A**

- **What percentage of LHON cases are male, and what percentage are female?**
  
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- **OK then, so why don’t females develop LHON at the same rate as males?**
  
  This is not yet known, but estrogen seems to play a protective role.
A

**On LHON**

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Presentation is typically unilateral; the fellow eye will **almost always** become affected.
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What is the typical time interval between initial and fellow-eye presentation?
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*What is the typical time interval between initial and fellow-eye presentation?* 

Weeks to months to years.
LHON stands for Leber’s hereditary optic neuropathy.

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What is the typical time interval between initial and fellow-eye presentation?

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*Q*

What is the typical time interval between initial and fellow-eye presentation? Weeks to months

Are there cases in which the interval has been much longer—say, years?
On LHON

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**On LHON**

- What is the typical time interval between initial and fellow-eye presentation?
  - Weeks to months

- Are there cases in which the interval has been much longer--say, years?
  - Yes, intervals as long as 8 years have been reported
Q

- LHON stands for *Leber’s hereditary optic neuropathy*
- LHON is more likely to affect **males**, despite the fact that transmission is **mitochondrial**
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- Onset is typically in the **2nd - 4th** decades
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**Onset is typically in the 2nd-4th decades**

*What is the typically-cited age range of onset?*
LHON stands for *Leber’s hereditary optic neuropathy*

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**Onset is typically in the 2\(^{nd}\)-4\(^{th}\) decades**

*What is the typically-cited age range of onset?*

10-30
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LHON is more likely to affect males, despite the fact that transmission is mitochondrial.
Presentation is typically unilateral; the fellow eye will almost always become affected.
Onset is typically in the 2nd-4th decades.

What is the typically-cited age range of onset?
10-30

How old at onset were the youngest and oldest confirmed cases?
Youngest:
Oldest:
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**Onset is typically in the 2nd-4th decades**

- What is the typically-cited age range of onset?
  - 10-30

- How old at onset were the youngest and oldest confirmed cases?
  - Youngest: 1 year old
  - Oldest: 80
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Presentation is typically unilateral; the fellow eye will almost **always** become affected

Onset is typically in the **2nd-4th** decades, with patients complaining of:

- Acute/subacute loss of acuity to < Snellen VA
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Onset is typically in the 2nd-4th decades, with patients complaining of:

- Acute/subacute loss of acuity to < 20/200
- Dyschromatopsia (usually red-green)
- Scotoma (usually cecocentral or central)

**Is the vision loss irreversible?**
LHON stands for *Leber’s hereditary optic neuropathy*

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Is the vision loss irreversible?
In most cases, yes. But a subset of pts demonstrate spontaneous improvement.
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What percent of cases comprise this fortunate subset?
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*Is the vision loss irreversible?* In most cases, yes. But a subset of pts demonstrate spontaneous improvement.

**What percent of cases comprise this fortunate subset?** 10-20
On LHON

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  - Scotoma (usually **location in VF**
  - Q
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Presentation is typically unilateral; the fellow eye will almost always become affected

Onset is typically in the 2nd-4th decades, with patients complaining of:

- Acute/subacute loss of acuity to < 20/200
- Scotoma (usually *ceccocentral or central*)
On LHON

LHON: Central/cecocentral scotomata
LHON stands for *Leber’s hereditary optic neuropathy*

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- Acute/subacute loss of acuity to < **20/200**
- Scotoma (usually **ceccentral or central**) (red - green vs blue - yellow)
- Dyschromatopsia (usually **red - green vs blue - yellow**)
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- Scotoma (usually cecocentral or central)
- Dyschromatopsia (usually red-green)

**Red-green!!?? I thought red-green was the inherited defect and blue-yellow the acquired defect. What gives?**
A

On LHON

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  - Dyschromatopsia (usually **red-green**)

Red-green!!?? *I thought red-green was the inherited defect and blue-yellow the acquired defect. What gives?*  
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.  
On the other hand, a **red-green** defect can be either acquired or congenital.
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*How can you tell if a red-green deficiency is acquired?*

1)  
2)  
3)  
4)
On LHON

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

On LHON
Classic DFE findings:
- ONH…telangiectasias
- ONH…pseudoedema
- Retinal arteriolar…
Classic DFE findings:
- ONH…telangiectasias
- ONH…pseudoedema
- Retinal arteriolar…tortuosity
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
Q

- **Classic DFE findings:**
  - ONH...telangiectasias
  - ONH...pseudoedema
  - Retinal arteriolar...tortuosity

- **Cardiac co-morbidity:**
  - On LHON
A

- Classic DFE findings:
  - ONH…telangiectasias
  - ONH…pseudoedema
  - Retinal arteriolar…tortuosity
- Cardiac co-morbidity: Wolf-Parkinson-White

On LHON
Classic DFE findings:
- ONH...telangiectasias
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Cardiac co-morbidity: Wolf-Parkinson-White

What is WPW?
• Classic DFE findings:
  - ONH…telangiectasias
  - ONH…pseudoedema
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• Cardiac co-morbidity: Wolf-Parkinson-White

What is WPW?
An abnormality of cardiac conduction
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

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

**What are the classic EKG findings in WPW?**
-- The PR interval is abnormally...[long vs short]
--
--
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

What is WPW?
An abnormality of cardiac conduction

What are the classic EKG findings in WPW?
--The PR interval is abnormally...short

On LHON
Classic DFE findings:
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Cardiac co-morbidity: **Wolf-Parkinson-White**

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

**What are the classic EKG findings in WPW?**
-- *The PR interval is abnormally...short*
-- *The QRS complex is abnormally...[wide vs narrow]*
Classic DFE findings:
- ONH…telangiectasias
- ONH…pseudoedema
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Cardiac co-morbidity: Wolf-Parkinson-White

What is WPW?
An abnormality of cardiac conduction

What are the classic EKG findings in WPW?
-- The PR interval is abnormally… short
-- The QRS complex is abnormally… wide
What is WPW?
An abnormality of cardiac **conduction**

*What are the classic EKG findings in WPW?*
-- *The PR interval is abnormally...short*
-- *The QRS complex is abnormally...wide*
-- *The QRS complex onset is...[classic descriptor]*
A

Classic DFE findings:
- ONH...telangiectasias
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Cardiac co-morbidity: Wolf-Parkinson-White

What is WPW?
An abnormality of cardiac conduction

What are the classic EKG findings in WPW?
-- The PR interval is abnormally...short
-- The QRS complex is abnormally...wide
-- The QRS complex onset is...‘slurred’
WPW: Slurred onset of the QRS complex
Classic DFE findings:
- ONH...telangiectasias
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Cardiac co-morbidity: Wolf-Parkinson-White

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An abnormality of cardiac conduction

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-- The PR interval is abnormally... **short**
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*WPW renders pts prone to what abnormal rhythm?*
Classic DFE findings:
- ONH...telangiectasias
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Cardiac co-morbidity: Wolf-Parkinson-White

What is WPW?
An abnormality of cardiac conduction

What are the classic EKG findings in WPW?
--The PR interval is abnormally...short
--The QRS complex is abnormally...wide
--The QRS complex onset is...‘slurred’

WPW renders pts prone to what abnormal rhythm?
Supraventricular tachycardia (SVT)
- Classic DFE findings:
  - ONH...telangiectasias
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  - Retinal arteriolar...tortuosity

- Cardiac co-morbidity: Wolf-Parkinson-White

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
On LHON

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

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

Myotonic dystrophy
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

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

What are its other ocular manifestations?
- --
- --

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?
Q/A

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Cardiac co-morbidity: Wolf-Parkinson-White

On LHON

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

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

Myotonic dystrophy

What are its classic nonocular findings?
- Cardiac conduction issues--
- Myotonia--
- Characteristic 'hatchet' facies--
- Frontal balding--
- Low intelligence--

two-word description
On LHON

- Classic DFE findings:
  - ONH...telangiectasias
  - ONH...pseudoedema
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Myotonic dystrophy: Christmas tree cataract
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**On LHON**

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- Bilateral symmetric ptosis--
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- ‘Christmas tree’ cataracts--

What are its classic nonocular findings?
- Cardiac conduction issues--
- Myotonia --
- Characteristic ‘hatchet’ facies--
- Frontal balding--
- Low intelligence--

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?
Q/A

- Classic DFE findings:
  - ONH...telangiectasias
  - ONH...pseudoedema
  - Retinal arteriolar...tortuosity

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

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**On LHON**

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

**What are its other ocular manifestations?**
- Bilateral symmetric ptosis--
- Pigmentary retinopathy--
- ‘Christmas tree’ cataracts--

**What are its classic nonocular findings?**
- Cardiac conduction issues--
- Mytonia--
- Characteristic **facies**--
- Frontal balding--
- Low intelligence--

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?
Classic DFE findings:
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**Cardiac co-morbidity:** Wolf-Parkinson-White

In a nutshell, what sort of condition is myotonic dystrophy?
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What are its classic nonocular findings?
- Cardiac conduction issues--
- Myotonia--
- Characteristic ‘hatchet’ facies--
- Frontal balding--
- Low intelligence--
On LHON

Myotonic dystrophy: Hatchet face and frontal balding
For more on myotonic dystrophy see slide-set O21

Myotonic dystrophy: Hatchet face and frontal balding
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

On LHON

What do CPEO and KSS stand for in this context?
--CPEO:
--KSS:
Classic DFE findings:
- ONH…telangiectasias
- ONH…pseudoedema
- Retinal arteriolar…tortuosity

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

On LHON

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?

**CPEO/KSS**

**CPEO:** Chronic progressive external ophthalmoplegia
**KSS:** Kearns-Sayre syndrome, a variant of CPEO

What do CPEO and KSS stand for in this context?

- CPEO: Chronic progressive external ophthalmoplegia
- KSS: Kearns-Sayre syndrome, a variant of CPEO

Myotonic dystrophy

Leigh syndrome
Q

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

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

On LHON

Myotonic dystrophy  CPEO/KSS  LHON  Leigh syndrome

What do CPEO and KSS stand for in this context?
--CPEO: Chronic progressive external ophthalmoplegia
--KSS: Kearns-Sayre syndrome, a variant of CPEO

Briefly, what is CPEO?
On LHON

- Classic DFE findings:
  - ONH...telangiectasias
  - ONH...pseudoedema
  - Retinal arteriolar...tortuosity

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

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**Myotonic dystrophy**  
**CPEO/KSS**  
**LHON**  
**Leigh syndrome**

**What do CPEO and KSS stand for in this context?**
--CPEO: Chronic progressive external ophthalmoplegia  
--KSS: Kearns-Sayre syndrome, a variant of CPEO

**Briefly, what is CPEO?**
A mitochondrial disorder characterized by progressive, symmetric paralysis of the extraocular muscles commencing in childhood.

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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?
CPEO: Symmetric ophthalmoplegia

On LHON
Q

- Classic DFE findings:
  - ONH...telangiectasias
  - ONH...pseudoedema
  - Retinal arteriolar...tortuosity
- **Cardiac co-morbidity:** Wolf-Parkinson-White

On LHON

- Myotonic dystrophy
- CPEO/KSS
- LHON
- Leigh syndrome

What do CPEO and KSS stand for in this context?
--CPEO: Chronic progressive external ophthalmoplegia
--KSS: Kearns-Sayre syndrome, a variant of CPEO

Briefly, what is CPEO?
A mitochondrial disorder characterized by progressive, symmetric paralysis of the extraocular muscles commencing in childhood

Which muscles are affected first?
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

On LHON

What do CPEO and KSS stand for in this context?
--CPEO: Chronic progressive external ophthalmoplegia
--KSS: Kearns-Sayre syndrome, a variant of CPEO

Briefly, what is CPEO?
A mitochondrial disorder characterized by progressive, symmetric paralysis of the extraocular muscles commencing in childhood

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

CPEO: Progressive ptosis
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

On LHON

What do CPEO and KSS stand for in this context?
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Cardiac co-morbidity:

Myotonic dystrophy  CPEO/KSS  LHON  Leigh syndrome

What do CPEO and KSS stand for in this context?
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Briefly, what is CPEO?
A mitochondrial disorder characterized by progressive, symmetric paralysis of the extraocular muscles commencing in childhood

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

KSS has a classic triad—what is it?
CPEO + pigmentary retinopathy + cardiac conduction abnormalities
CPEO: Pigmentary retinopathy
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

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

On LHON

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? 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.)
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

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

On LHON

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?

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.
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for
A

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

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

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

**On LHON**
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

What are the genetic positions for the three most common mutations?
11778, 3460 and 14484
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

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

Which is most common?
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

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

Which is most common?
11778

Which is associated with the poorest ultimate vision?
11778

Which carries the lowest likelihood of spontaneous visual recovery?
11778
Classic DFE findings:
- ONH...telangiectasias
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- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

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Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

**On LHON**

*What are the genetic positions for the three most common mutations?*
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Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

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**Q**

**On LHON**

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Diagnosis: Blood assay for mDNA mutation

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Diagnosis: Blood assay for mDNA mutation

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

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Which carries the lowest likelihood of spontaneous visual recovery?
11778, 14484
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoeedema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

Treatment:
A

Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudopoeodema
- Retinal arteriolar...tortuosity

Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation

Treatment: None, unfortunately