LHON stands for Leber's hereditary optic neuropathy. 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, with patients complaining of:

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A

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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

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

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

80-90 are male; 10-20 are female
● LHON stands for *Leber’s hereditary optic neuropathy*

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How are mitochondrial disorders inherited; ie, what is the pattern?
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How are mitochondrial disorders inherited; ie, what is the pattern?
Maternal; ie, women pass it along to all their biological offspring.
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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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Given that LHON is a mitochondrial disorder, why is its strong male preponderance unusual?
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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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Q/A

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This is not yet known, but estrogen seems to play a protective role
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Maternal; i.e., women pass it along to all their biological offspring

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

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What is the typical time interval between initial and fellow-eye presentation? Week 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
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Onset is typically in the **2nd - 4th** decades
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What is the typically-cited age range of onset?
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What is the typically-cited age range of onset?  
10-30
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**Onset is typically in the 2\textsuperscript{nd}-4\textsuperscript{th} 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**

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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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- Acute/subacute loss of acuity to < Snellen VA
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  - *Is the vision loss irreversible?*
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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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- Scotoma (usually **location in VF**)

Q
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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. 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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\textit{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

- Classic DFE findings:
  - ONH...
  - ONH...
  -
A

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

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

What is WPW?
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
- Retinal arteriolar...tortuosity

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

**What is WPW?**
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--The PR interval is abnormally...short
On LHON

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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**]
On LHON

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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...short
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--The QRS complex onset is...[classic descriptor]
Classic DFE findings:
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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...short
-- The QRS complex is abnormally...wide
-- The QRS complex onset is...‘slurred’
Classic DFE findings:
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**WPW renders pts prone to what abnormal rhythm?**
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-- *The PR interval is abnormally…short*
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**WPW renders pts prone to what abnormal rhythm?**
Supraventricular tachycardia (SVT)
Q

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

- Diagnosis: Blood assay for...
Classic DFE findings:
- ONH...telangiectasias
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Cardiac co-morbidity: Wolf-Parkinson-White

Diagnosis: Blood assay for mDNA mutation
Classic DFE findings:
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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
- 14484

11778 is most common.

11778 is associated with the poorest ultimate vision.

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

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?
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?
14484

highest

Which carries the lowest likelihood of spontaneous visual recovery?
14484
- Classic DFE findings:
  - ONH...telangiectasias
  - ONH...pseudoedema
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- Cardiac co-morbidity: Wolf-Parkinson-White
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- Treatment:
On LHON

- Classic DFE findings:
  - ONH...telangiectasias
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  - Retinal arteriolar...tortuosity
- Cardiac co-morbidity: Wolf-Parkinson-White
- Diagnosis: Blood assay for mDNA mutation
- Treatment: None, unfortunately