Q

- LHON stands for Leber's hereditary optic neuropathy
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 from mothers to both males and females.
LHON stands for Leber’s hereditary optic neuropathy.

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

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?*
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.
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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?**
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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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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?
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This is not yet known, but estrogen seems to play a protective role
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Presentation is typically unilateral; the fellow eye will **always** become affected.
On LHON

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

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

Weeks to months to 

Yes, intervals as long as 8 years have been reported.
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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?
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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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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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**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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Is the vision loss irreversible?
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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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- Acute/subacute loss of acuity to < 20/200
- Scotoma (usually red-green location in VF)
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Onset is typically in the 2nd-4th decades, with patients complaining of:

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- Scotoma (usually cecocentral or central)
On LHON

LHON: Central/cecocentral scotomata
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Onset is typically in the 2nd-4th decades, with patients complaining of:

- Acute/subacute loss of acuity to < 20/200
- Scotoma (usually cecocentral or central)
- Dyschromatopsia (usually red-green vs blue-yellow)
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Red-green!!!?? I thought red-green was the inherited defect and blue-yellow the acquired defect. What gives?
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*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)
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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...

On LHON
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
Classic DFE findings:
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- ONH...pseudoedema
- Retinal arteriolar...
Classic DFE findings:
- ONH...telangiectasias
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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.
LHON: Progression of ONH atrophy
Classic DFE findings:
- ONH...telangiectasias
- ONH...pseudoedema
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Cardiac co-morbidity: 

On LHON
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Cardiac co-morbidity: Wolf-Parkinson-White
Q

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

*What is WPW?*
An abnormality of cardiac **conduction**
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...[long vs short]*
--
--
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 vs narrow]
A

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

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Diagnosis: Blood assay for **mDNA mutation**
On LHON

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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?
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Which carries the lowest likelihood of spontaneous visual recovery?
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Treatment:
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

Treatment: None, unfortunately