

## What is it?

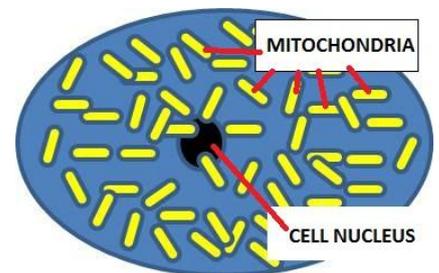
Leber's Hereditary Optic Neuropathy (LHON) is a rare medical condition which can cause the loss of central vision in one or both eyes. People affected by LHON cannot see things in the center of their field of view, but will probably be able to see some things "out of the corner of the eye" by using peripheral vision. This means that they may be able to walk around without bumping into furniture, but will not have enough detailed vision to read, recognize others at a distance or drive a car.

The German eye specialist Theodore Leber first wrote about LHON in 1871, but we knew almost nothing about it until the late 1980's when researchers discovered the way in which LHON is inherited in a family. Theodore Leber also named other, completely unrelated, eye diseases such as Leber's Congenital Amaurosis (LCA), so it is very important not to get them mixed up.

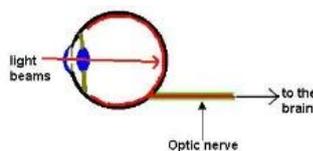
LHON is a genetic disorder, which means the underlying cause is in the affected person's inherited genes – LHON is not caused by a germ or bug and cannot be passed on from one person to another like the 'flu.

## What causes LHON?

LHON has been linked to a number of changes in DNA – the chemical that passes information from one generation to the next about how our bodies work. These changes are all in the DNA of special parts of every cell, called Mitochondria (pronounced mite-oh-kon-dree-ah). Mitochondria are the 'power plants' of every cell, where energy is released from food so the cell can do its job. Although most of a person's DNA is held in the nucleus of their cells, the mitochondria contain some DNA as well. The LHON changes are in this small amount of DNA carried inside the mitochondria.



Although we know that someone affected by LHON will carry one or more of the LHON gene changes, the vast majority of people carrying a LHON gene will not be affected by it. Only a small number of families carrying a LHON gene have someone who has lost eyesight because of it. Even if a family member has lost eyesight, most people in that family will probably not lose eyesight due to LHON.



We do not yet know exactly why some people carrying a LHON gene lose eyesight.

We know that central vision is lost because of damage to the nerve cells at the back of the eye – the Retinal Ganglia Cells. The eye may be focusing a picture correctly onto the retina at the back of the eye, but the Optic Nerve can no longer send correct signals to the brain. We also think that the LHON genes cause a huge drop in the amount of fuel available to the nerve cells. However we don't know why most people, especially women, carry a LHON gene without losing eyesight.

## Treatments for LHON

Unfortunately, there is no proven cure or treatment for LHON yet.

Researchers are investigating some possible treatments, and some are showing very hopeful early results, but no treatment offers a definite cure at the moment.

### Drug Therapies – Idebenone or EPI-743

These are man-made chemicals, taken daily in tablet form. Researchers think that these drugs help the “damaged” fuel-producing mechanism in the mitochondria work more efficiently.

### Gene Therapy

This technique tries to supply working copies of the “faulty” mitochondrial genes directly to the eyes of an affected person.

### “Mitochondrial Helpers”

Some people take food supplements which are meant to help the function of mitochondria, such as Co-enzyme Q10 or combinations of vitamins and minerals.

Some food supplements are dangerous in large doses and should only be taken with supervision from your medical practitioner.

## Possible Triggers of eyesight loss

Because LHON is so rare, it is difficult for doctors to find any definite patterns to show what actually triggers the loss of eyesight. There are medical case studies describing individual patients, and a few large-scale studies trying to identify triggers.



Smoking seems to dramatically increase the risk of losing eyesight for someone carrying a LHON gene. Heavy drinking may also be a trigger, but the evidence is less clear.



Some other possible triggers have been reported in case studies:

- Anaemia/Vitamin B12 deficiency
- Treatment with certain drugs (Cyanocobalamin, Ethambutol, Sodium Nitroprusside, Tadalafil / Viagra, Antibiotics like Erythromycin / Telithromycin)
- Severe illness or psychological stress
- Illegal drugs such as Cocaine/Ecstasy.
- Physical injury/Trauma



## Living with LHON

Being affected by LHON is a huge emotional blow, both for the affected person and his/her family. As well as getting physical help such as special computer software, magnifiers and so on, people need support to deal with the situation.

LHON is a rare disease, but is found all over the world, and there is now an active self-help and support community on the Internet. This warm and friendly group of people have shared experiences, and are working with specialist medical researchers to help us move towards the cure. Take a look at the last section of this booklet for some contact information.

### [Aids which can help](#)

Even if someone has some sight, Mobility and Daily Living training helps rebuild confidence and independence. A white cane and/or guide dog can be a great help at times.

### [Computer adjustments and accessibility](#)

Some computers, tablets and phones have built-in accessibility programs like Apple Voiceover and Zoom. There are settings in Microsoft Windows that can make it easier to use a PC, such as changing the mouse pointer visibility, Using a desktop or laptop computer is made a lot easier by learning to touch type.

There are several screen reading and braille output programs for computers, so it is a good idea to get specialist advice and try them out to see if one of them suits you.

### [Magnifiers](#)

We now have a huge range of magnifiers for close up work and distance reading. The right type of magnifier can make a big difference, for reading, working or getting around.

### [Talking Books/Devices/Audio Description](#)

Many books and newspapers can be found on disk or downloadable format. As well as this there are lots of talking devices such as clocks, watches, timers, scales and so on. These make a big difference to independence.

More and more movies and TV shows provide Audio Description, a commentary describing key things on the screen. There are talking program guides on some TV services.

### [Sports and Hobbies](#)

People affected by LHON can still do a range of hobbies and sports including Golf, Cycling Sailing and Skiing.

### [Education and work](#)

Having LHON can make school, college, university or work more difficult, but people with LHON do go on to complete their education, and hold down a job.

There is much more about living with LHON on the Internet, and you can talk with many other people about their personal experiences.

## Who can be affected?

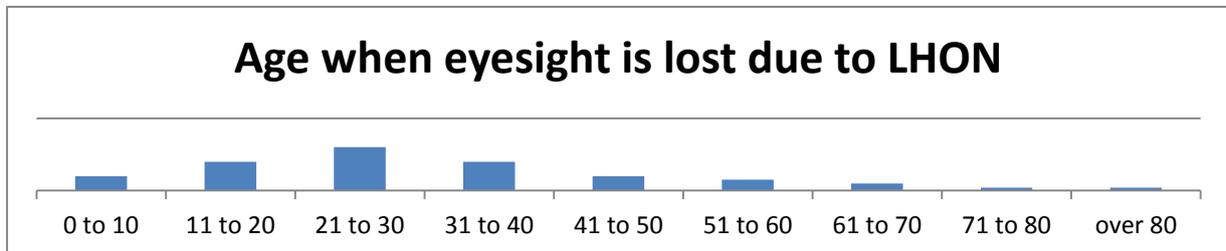
Anyone carrying one or more of the LHON genes is at risk of losing their eyesight, but only around 1 in 30,000 people carry a LHON gene.

The "Typical" course of LHON is for a man to be affected in the late teens, early twenties or thirties. Central vision is blurred and lost in one eye over a period of a few weeks, then the vision in the second eye is lost. The time when vision is getting worse is called the "Acute" phase, and it usually lasts for one or two months. Once the Acute phase is over, there is usually some useable peripheral sight.

Women can be affected as well. There are about 9 men affected for every woman affected.



Eyesight can be lost at any age, from infancy to people aged over 80.

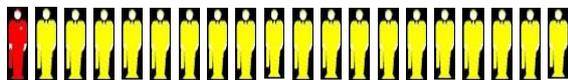


## Can the eyesight recover without treatment?

A small number of people recover eyesight after being affected by LHON. This can happen even years after the Acute phase has ended. Just as we do not yet know why some people lose their eyesight, we do not know why some people recover. Affected children are more likely to recover than affected adults.

Almost all cases of LHON involve one of three 'Primary' gene mutations: 11778, 3460 and 14484. The chance of recovery is different for each of these genes:

Untreated Recovery Rate for 11778 mutation: 4% (<1 in 20)



Untreated Recovery rate for 3460 mutation: 22% (< 1 in 4)

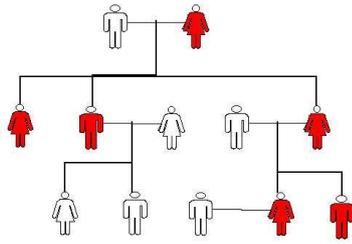


Untreated Recovery rate for 14484 mutation: 37% (< 2 in 5)





## How a LHON gene is inherited



All of the LHON causing gene mutations are in DNA held in Mitochondria. As everyone receives all of their mitochondria from the mother, the LHON genes cannot be passed on by a man. A woman carrying a LHON gene will pass it on to her children, and they will be at risk of developing LHON.

At present we cannot identify which people carrying a LHON gene will lose eyesight. Once someone in the family loses eyesight due to LHON, then we know that maternal blood relatives will be carrying a LHON gene.

The risk of losing eyesight for male blood relatives carrying a LHON gene is estimated at 50% (1 in 2) and for females is around 10% (1 in 10).

The best advice for reducing the risk is to avoid smoking and other possible triggers.

## Where can I find out more?

This is a very brief leaflet summarising patient information on LHON. You should consult your medical practitioner or qualified specialists for medical advice and information.

As LHON is so rare, the biggest patient community is on the Internet:

FACEBOOK GROUP: [Leber's Hereditary Optic Neuropathy](#)

YAHOO GROUP: [LHON Members Yahoo group](#)

INTERNET SITES:

[LHON Home Page https://sites.google.com/site/planetleeder/lhon](https://sites.google.com/site/planetleeder/lhon)

[LHON.ORG - a website by an affected family in the USA.](#)

[LHON.INFO](#)

An excellent book containing the experiences of several families affected by LHON:

“Born with a Bomb Suddenly Blind from Leber's Hereditary Optic Neuropathy” by Valerie Byrne Rudisill

### Acknowledgments

This booklet was produced by James Leeder with the help of:

Members of the Leber's Hereditary Optic Neuropathy Facebook group

Researchers at the [Wellcome Trust Centre for Mitochondrial Research, Newcastle University](#)