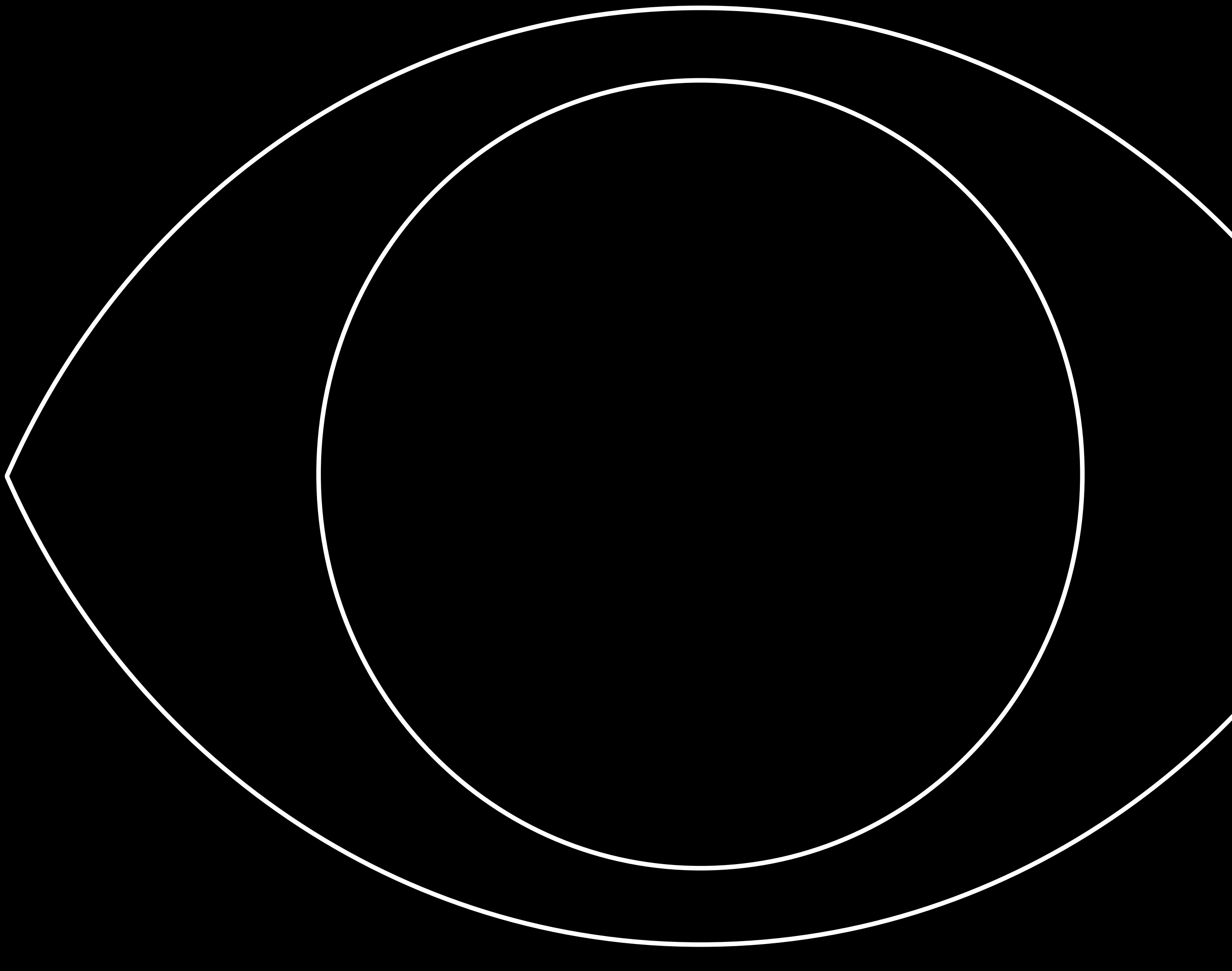
LHON

The guide for patients, by patients.
A guide to living with the rare eye disease.



Introduction

Dear patient,

You have just been diagnosed with LHON. Instead of the previous uncertainty, there is now a little more clarity, but certainly accompanied by justified worries, a flood of feelings and countless questions.

If anyone knows what all this means, it is people who have gone through such a journey themselves. That‘s why patients like Niklas, Jan and Martina have given us invaluable insights into their lives with LHON – and how they cope with everyday life. They explained to us that LHON brings with it many new challenges. And at the same time, they proved that you can also learn new things, find new hope and take different paths. Together with the patients, we have decided to change their names and use photos that only show them as representatives. Because sharing experiences is something very personal.

This brochure was created in collaboration with Chiesi GmbH. At two workshops, LHON patients shared their ideas and views with us. After all, the most important thing for LHON patients after their diagnosis is an honest opinion from people who know what is important.

People who live with the condition from day to day are at the centre of everything we do. They need assistance and support on their journey towards living a good life with LHON.

We hope that this guide can help you with your own journey.

Chiesi
global rare diseases

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“I’m Jan. I was 17 at the time, at first it was just a dot in the centre of my visual field that shouldn’t really have been there. I didn’t think much about at the time but it was still there two weeks later.”

Hi, my name’s Niklas. A few weeks after my 22nd birthday I asked “So what is LHON then?” It all started a few months earlier, with colours seen from my right eye looking paler than from my left. So I went to my ophthalmologist for a checkup. After a number of other tests the genetic test came up with a diagnosis of Leber’s hereditary optic neuropathy. I’d never heard of it.”

“I’m Martina. With me it started in my early 50s. My vision suddenly became more blurred and the first thing I thought was “now you’re getting old.”

What exactly is LHON?

The first sign of Leber’s hereditary optic neuropathy (LHON) is mild visual impairment, which then very quickly becomes incredibly severe. It can sometime lead to a complete loss of vision. The condition affects men in particular – they are four times as likely as women to have it. It usually starts between the ages of 15 and 35, though it can occur at any age.

Background

LHON is a rare hereditary disease. The name of the disease comes from Professor Theodor Karl Gustav Leber who described it at the end of the 19th century. We’ve known since the late 1980s that LHON is caused by mutations of the mitochondrial genome. Mitochondria are small components of the body’s cells that are responsible for providing energy. This process is impaired in the retinal cells of people with LHON. This reduces the supply of energy and increases oxidative stress, which stops the optic nerve from doing its job, which is to send visual information from the eye to the brain. In the end this causes severe visual impairment.

Initial symptoms

Your sight gets worse quickly and relatively suddenly. It is mostly painless and affects just one eye at first. Our brain is trained to replace missing details in our visual field. So although you may have difficulties seeing out of one eye your brain will replace what's missing – this is referred to as the "compensation effect". In many cases you won't see any signs of the condition to start with.

Some people describe what they do see as like a picture with a black spot in the middle. You might find it harder to distinguish contrasts and colours and find bright lights increasingly uncomfortable to look at. But people's experiences of LHON vary greatly. Then, after a few weeks, these problems with your sight often spread to your second eye. In just a few weeks your vision can get far worse, and you might lose your sight almost entirely after just a few months.

Professor Klopstock and his team in Munich see around 150 LHON patients a year at their day clinic and in their research projects (register studies, natural history studies, therapy studies). Most patients come for a checkup once a year. They tend to see around 12 new LHON patients a year.

Fact check with Professor Klopstock

The ten most important questions

Professor Thomas Klopstock is a specialist neurologist working mainly on mitochondrial diseases, which include LHON. He has been interested in hereditary diseases caused by mitochondrial mutations since his student days. His research focuses mainly on these diseases and the development of new treatments. He has been assisting LHON patients and their family members at the Munich University Clinic for years along with his team as well as colleagues at the Ophthalmic Clinic. In this “fact check” he answers the ten most important questions from LHON patients.

1

What causes the hereditary disease LHON?

LHON is a hereditary disease caused by a mutation of mitochondrial DNA. There are three classic mutations that are responsible for 95% of cases. The effects of the mutations vary, particularly in terms of how the disease progresses. LHON is inherited via maternal DNA. Cases may also occur in family members but don’t always do so.

2

What does “penetrance” of a mutation mean?

The penetrance of a mutation determines how often it actually results in symptoms. LHON mutations have relatively low penetrance, so people carrying this mutation will not necessarily contract the disease. Around 50% of men with one of these mutations get the disease, compared with just 10% of women.

3

Why is it more common in men than in women?

Our research into LHON is also looking at this imbalance between the sexes. It seems that female sex hormones provide a certain level of protection against the onset of the hereditary disease.

4

Is there a drug to treat LHON?

We now have idebenone, an active substance that intervenes in the disease mechanism and protects mitochondrial function. Although it cannot cure the condition it does improve the eyesight of around 50% of patients treated with it.[1] Even this means they can read an average of seven more lines on the eye chart than before.

5

Can you describe the normal progression of the disease?

LHON progresses in a way that is unusual for a hereditary disease and tends to be the same for most patients. In most cases people have difficulties seeing out of one eye after a few days, followed a few weeks later by the second eye. They often go from 100% vision to less than 5% in a very short period of time.[2] They also find it increasingly difficult to perceive colours and contrasts. We register patients with LHON on the mitoNET register (www.mitoNET.org) and enrol them on a very detailed “proLHON” study in Munich. This will help to improve prognoses as it allows us to develop options for catching and treating the disease early on. Another LHON register is maintained by PRO RETINA Deutschland e.V.

6

How often do you need to go for a checkup once you’ve been diagnosed?

I recommend going for a checkup every three months in the first year. In the second year you just need to go every six months.

7

Can I do anything to protect myself if I am a carrier of this type of mutation?

Yes, there are triggers (sometimes referred to as “trigger factors”) that can make it more likely that you will get the disease. A study of the potential trigger factors for LHON identified smoking and too much alcohol or drugs as the most important triggers. Smoking makes it more likely that you will get the disease in the first place but also that it will progress quickly and be more severe once you do get it. So you should definitely not smoke if you are a carrier of the mutation. Consuming too much alcohol will have the same effect. Having an occasional glass of wine or a beer isn’t a problem. But drinking lots of alcohol every weekend or every evening puts you more at risk of developing the disease.

8 Do I need a special diet or a complete transformation in my life?

People with LHON need to have as healthy a diet as possible with lots of fresh fruit and vegetables. These contain antioxidative substances that have a positive effect on health. One example is the so-called “Mediterranean diet”.

You may need to make other changes to your life but this depends on how good your eyesight is. For example, you can keep up sports activities – or anything as long as you can manage, really. And there are also aids that can help you cope. Your life can certainly be worth living even if you have LHON!

9 What about my siblings and other family members?

Carriers won’t always get LHON but I recommend talking to your immediate and wider family if you’ve been diagnosed with it. You can explain to them that not smoking or consuming too much alcohol early on can have an effect on whether they’ll get the disease. We have been researching into LHON at our clinic for a long time and patients without symptoms but who are carriers of the mutation can also enrol on the study. This helps us obtain even more information about the disease.

10

Do you have any tips on how to learn to deal with the diagnosis?

Of course the diagnosis will come as a shock at first, and the first few months are difficult. But after that many patients cope quite well with the disease. They find ways of living with the diagnosis by using smartphones and other aids. And they’re not completely blind, of course! The sight loss is severe but they can still find their way around, you can see a lot using magnifying aids, for example. In most people it is only the central vision that is impaired and they generally still have peripheral vision. Out of a total of 200 patients with LHON I know just two who need a guide dog and a stick.

Test results and what they mean

Here’s Martina’s experience:

“I was rushing from one doctor to another for ages, so I almost felt a sense of relief when the diagnosis finally came. But that only lasted a short time and then I really despaired. I wondered what LHON would mean for me – would I have to take early retirement? And what about my daughter – was she at risk as well, or might she need to care for me?”

Jan reported:

“The doctors thought it was an injury to the optic nerve and gave me cortisone treatment, but that didn’t help. When I was diagnosed with LHON it was like the whole world came crashing down.”

The journey to a diagnosis

It isn’t easy for the ophthalmologist to diagnose LHON. The symptoms are consistent with lots of other diseases such as malignant eye lesions or even multiple sclerosis. So many patients find themselves rushing from one doctor to another. Sometimes it’s just a matter of chance that one of them realises the symptoms might be caused by a hereditary disease and arranges for a genetic test to be done. And people with LHON often have to wait a long time before they get the certainty the genetic test brings.

And now?

Many LHON patients have a feeling of emptiness immediately after the diagnosis, or are completely overwhelmed. Some have described it as feeling as if they are falling into a deep hole. That’s entirely normal. You just need to learn how to live with these fears and all the thoughts the diagnosis brings. Some people start to research the disease, some talk about it and some people become withdrawn. You’re perfectly entitled to ask for time to process the diagnosis, as it involves strong emotions. Take time to process any fears, concerns and perhaps anger that come up – at your own pace. You might wonder what it will mean for you and your life. It takes time to process the diagnosis.

Your nearest and dearest ...

The diagnosis will have an effect on you and your family whatever age you are when you get the disease. It means big changes for your roles within the family in particular. If you’re an adult when you are diagnosed, perhaps with children of your own, its significance will be different than it is for younger people. You might wonder whether your children will have to care for you now, or you might suddenly feel much older. If you’re young when you’re diagnosed and perhaps planning to leave home shortly you might worry you will be treated like a child again. Its significance for you and your life, as well as your family, will be different for everyone involved. If everyone can talk openly about what matters to them and what they will need it will make the next few weeks and months a little easier.

First steps after diagnosis

Jan’s story:

“I really wanted to stay optimistic. I had lots of energy, which wasn’t always positive. I was very angry and unhappy about the diagnosis. But then I thought “how can I channel this energy properly to make it work for me?”

And Niklas says:

“After the initial shock I wanted to find out how other people were managing with the condition. I got to know Mehmet, who was diagnosed years ago, on an online forum. Talking to him gave me a new perspective and I gradually got back on top of things. I started making music again. And I found some information about opportunities for studying on the internet despite my visual impairment.

LHON isn’t just about visual impairment. It involves so much more than that, which you only gradually realise. The main thing, alongside the painful adjustments you have to make, is definitely the loss of independence. Lots of everyday things are harder when you can’t see well. That can put significant psychological strain on patients. So it’s better if you have people around you who are actively supporting you. When you can’t manage routines or journeys without help any more you can rely on your family, friends and other helpers. So being open to offers of help from other people is an important step in your new routine.

How can I put it?

Think about the kinds of support you might need in advance, and then talk about your thoughts and the limitations you might face. And tell people what you don’t want as well. Whoever you’re talking to, you decide how much you want to tell them yourself. The same applies to new people you meet. Actually, being as open and honest as possible about your condition can make things easier for you and the person you’re talking to. Even if odd questions come up or someone says “I’d never have known”, talking about it can clear the air and make your eye problems less important, even if you’re within a relationship.

Even when your illness is really getting on top of you, you don’t need to hide away. Having one or two people around you who you can call anytime really helps.

What kind of help am I entitled to?

With LHON your vision may be so impaired that you not only have to organise your daily routine differently – you may be entitled to financial support. This covers aids, for example, or you may be certified as severely disabled. There are also lots of different kinds of training, and learning support assistants for schoolchildren, for example, who can make it easier to live with the disease. As well as your ophthalmologist there are specialist centres for eye diseases and patient organisations which have people you can contact for advice. They can also help you with other questions, on family planning or perhaps changing jobs or careers, for example. If you feel you're not coping well with the situation you can also go to a therapist for psychological support. It's sometimes helpful to talk to someone not directly involved. Health insurance providers will usually cover the costs. It's best to talk to your health insurance provider beforehand.

What can I do myself?

Various factors can have a good – and bad – effect on LHON. These range from adjusting your lifestyle to making organisational changes and working on your personal attitude to the disease.

Further information, tips and a checklist für helpful documents are also available on our website: https://lhonaware.com/patient-home/

Do:

- eat more vitamins, particularly A, C and E as well as selenium
- improve your diet with more fruit, vegetables, good oils, occasionally fish
- get involved in endurance sport: anything that gets you moving
- schedule in breaks
- get advice on the financial help you're entitled to
- complete and send off applications for financial support
- find out about aids and apply for them
- inform your friends, family and boss or colleagues

Don't:

- smoke
- drink too much alcohol
- get into situations with too much stress
- look at dazzling lights
- do anything that puts you under pressure

Treatment and expectations

Martina thought:

"I had high hopes when I found out there was a treatment. But at the same time I was afraid of what I would do if it didn't improve things at all."

Jan is convinced:

"I think it's important not to underestimate your abilities. And to stay optimistic. Hey, 2% eyesight is still 2%!"

The LHON fact check includes some information about treatment options (p.11). Many patients go through phases of hope and fear before treatment and even during it. Unfortunately there's no way back to the "old normal", even with treatment. As with any treatment, it's important to keep taking your medication regularly.

Your ophthalmologist can give you information about treatment options and explain what you can expect. You can have an open discussion with the specialists.

What can I expect?

Whether the treatment works depends on lots of things, such as which specific mutation is responsible for your disease. So it’s important to manage your expectations – you can involve your family members in this, too. There is no guarantee that the treatment will be a success: it won’t necessarily achieve the desired outcome, or it might only improve your eyesight a little. Your ophthalmologist can give you information on how effective the treatment is likely to be and what you can expect from it.

☺ Living with LHON

Martina reports:

“At first, I was worried it would be worse after the treatment. Then I really started to think about my lifestyle and pay more attention to my diet as well as myself and my own needs generally.”

Jan says:

“I flew off to China for four months! I had to plan everything carefully to make it work. Things went wrong but it was the best experience of my life. I managed to do it, I got a girlfriend, I coped with everyday life in a foreign country, I even went cycling at night! The best thing about this foreign trip for me was that I had a social life there and an everyday routine that suited me. What did I learn from my China trip? I can deal with problems, sort them out and set myself new challenges.”

After a few months you might find you just see the outlines of people, objects or screens, and taking the train to see your granny might seem impossible. However, we human beings are capable of amazing things, despite having poor vision – or even because of it. We can overcome changes and limitations by thinking up new ways and new approaches. Obviously, everything will be just a little bit slower, partly because you need time to adjust to the changes in your everyday life. And being patient with yourself is one of the hardest lessons to learn, particularly to start with. So the main thing LHON patients need is time to accept their disease and adjust to their new life.

Will everything be different?

Once you've begun to process the diagnosis you can go on to bigger issues such as what living with LHON will be like, where you have to make changes and what will stay the same. With aids and support you'll probably be able to continue your job or your studies, but you might also want to try out something new. You never know, you might find something where your eye disease is less of a problem.

You'll come up against your limits time and again, sometimes you might even be angry with yourself if everything takes a bit longer. Try to go easy on yourself, as you would a good friend. After all, you wouldn't blame a good friend if they stopped being able to do things. Everything takes time, not least accepting and making changes.

Will the disease take over?

Once you are diagnosed with the disease it will also start to take up lots of space in your life. Your thoughts and conversations constantly revolve around it, you have to organise lots of things, make changes, get help, and you quickly start to feel that the disease has taken over. But you're so much more than the disease. It affects your eyes, but remember your ears, your voice and your mobility are still the same as ever. And your mind can help you too – our thoughts can affect our mood. It's up to you how you deal with your disease. Treating it as a challenge makes it easier to cope with your new everyday life.

Living a whole life – even with LHON

Whatever your plans, whether it's going abroad, starting an apprenticeship or changing jobs, there is almost always a way to do it despite the disease. You won't be able do it without help from other people, but there will definitely always be someone to support you. Whether it's help organising somewhere to stay when you're abroad, applying for aids to make sure the new job works out or even applying for a place at university. If you're still at school it will be a disappointment not to be able to get your driver's licence like many others. But why shouldn't you be able to go cycling, skiing with a guide or try out other things?

☞ Tips and tricks for everyday living

Niklas’ experience:

“Honestly? I had pictured my life differently. I now think people are right when they say “life is what happens while you’re busy making plans.” I’ve developed new routines for my everyday life and I rely on lots of aids. If I’m poring over books for my studies I use my smart reading lens.”

Jan explains:

“People keeping asking me “do you need glasses?” It does get on your nerves sometimes. What I really need is my mobile phone. That’s essential. I have a girlfriend who has an eye disease as well and can’t see much, she always has three mobile phones with her just in case.”

If you have a visual impairment there are lots of normal things at home that you can’t do as well as you used to. Whether it’s reading envelopes, the display on your mobile phone or the setting on the oven, you quickly enter a state of helplessness or even dependency on other people. LHON patients also realise their limitations when they are out and about if they want to find their way around without help. There are lots of different kinds of aids for this, technical, practical and sometimes really simple ones. Digital solutions are really helpful for people living with LHON. As well as helpful apps, there are some simple aids that are still used. We’ve listed the most important ones here:

Technical aids

Screen readers: these help you to continue reading whatever you want, and can even read aloud to you.

Accessible smartphones: many are already accessible, or have settings to enable this. Mobile phones have voice-over functions and all sorts of add-ons for people with a visual impairment that really help.

Apps etc: there are specialist apps such as “Be my eyes” or “KNFB Reader” that provide help on the spot when you are out and about. The DB Navigator for travelling by train, the barcode app for identifying products or the GRETA app that gives an audio description of films in cinemas also remove some of the restrictions you face.

Siri, Alexa or Google Assistant: Devices with voice assistants are a real help at home! You don’t need to search for music on difficult to read displays and you just need to say a few words to order a pizza or even control your heating.

Simple practical aids

Magnifying glasses: come in large, small or takeaway sizes and are a classic reading aid that will help you read the smallest of small print.

Low vision filter glasses: protect your eyes against bright light better than ordinary sunglasses. The lenses are specially tinted to filter out harmful light. Health insurance providers will usually cover the cost of the aid.

At home

Tidiness is half the battle: keep your health insurance documents in the top drawer, hats on the tray, keys on the hook – having a system for storing all your stuff at home and sorting it into categories will save time trying to find it as well as making you feel safer.

Please don’t change anything! Ask the people around you to stick to your system. And if you live with other people, ask them not to tidy things away or put them anywhere else without your permission.

Label household equipment etc.: labels and signs make routine things like the settings on the dishwasher or which bin to use for the rubbish much easier. For example, you can draw a red arrow against the washing machine programme you always use, or stick a big coloured sheet of paper on every rubbish bin.

Good lighting! Make sure all parts of your home are lit in a way that suits you. That helps you avoid hazards and improves your wellbeing.

Where to get help

In Germany there are a number of eye centres, institutions and clinics that specialise in the diagnosis and treatment of LHON. You can find a complete list of the 14 current addresses online at www.pro-retina.de/netzhauterkrankungen/lhon/zentren. Or simply scan the QR code below.

There are also two associations which provide help and support for people with LHON and their family members: LHON e. V. and PRO RETINA Deutschland e. V.

They also have self-help groups, which can support you in your everyday life as well as helping you to feel you are not alone with your disease. Talking to other patients often opens up new prospects for yourself and new friendships. But they will always respect your personal situation, too.

Addresses

You can find all 14 addresses here. Simply scan the QR code on your smartphone or go to www.pro-retina.de/netzhauterkrankungen/lhon/zentren.

Understanding LHON even better

We’ve written this brochure to give you some initial insights into the disease. You’ve heard from LHON patients themselves about what they went through around the time they were diagnosed.

For further information, topics and tips please visit our website https://lhonaware.com/patient-home/

Sources:
1 Catarino CB et al. J Neuro-Ophthalmol 2020; 00: 1–8.
2 Newman NJ, Biousse V, Newman SA et al. Progression of visual field defects in Leber hereditary optic neuropathy: experience of the LHON treatment trial. Am J Ophthalmol 2006; 141: 1061–1067. doi: 10.1016/j.ajo.2005.12.045.

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We’d like to finish by asking Jan to speak:

> “I’m now convinced that setting yourself new challenges is the way to grow and overcome your limitations.
>
> Don’t get discouraged – you can do anything in spite of LHON, and you should do whatever you want.”