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# MEDIA RELEASE

**New study provides Australia's first snapshot of families at risk of blindness from rare genetic eye disease**

- Researchers, a genetic counsellor and person living with Leber Hereditary Optic Neuropathy are available for interview.

New research has revealed for the first time the number of Australian families affected by genetic mutations that cause the rare genetic eye disease Leber Hereditary Optic Neuropathy (LHON) – and the risk of going blind from the disease.

The research, led by Professor David Mackey AO from the Lions Eye Institute and University of Western Australia and Dr Isabel Lopez-Sanchez from the Centre for Eye Research Australia (CERA) and University of Melbourne, is published in the *American Journal of Human Genetics* today.

The study, which reveals that 96 Australian families are affected by the genes, presents the most up-to-date statistics of the disease in the world.

It also reveals the risk of losing vision from the disease may be less than previously reported.

The new information is expected to help families affected by LHON make better informed family planning decisions.

It will also aid studies researching why some people are at higher risk of vision loss from the disease and help identify patients suitable for future clinical trials into potential cures.

**What is LHON?**
LHON is a rare genetic eye disease that affects the optic nerve. It is caused by changes in the DNA of the mitochondria – the powerpacks that provide energy to our cells.

However, it carries an uncertain diagnosis as even though family members may have the same genetic mutation their vision will not be impacted in the same way.

Very few people with a LHON mutation will experience vision loss. However, some may experience sudden and permanent vision loss – with blindness sometimes occurring in a matter of weeks.

“People affected by LHON never go completely blind, and a small percentage may recover some vision, but the vast majority will not be able to drive, read or recognise faces,’’ says Professor Mackey.

“For 30 years I have worked with eye specialists around the country and the research team at CERA to be able to assemble some of the most accurate data about LHON risk anywhere in the world.

“Although we are still working to find treatments for LHON, knowing exactly the risk for vision loss allows us to design better clinical trials.

“If the federal government passes legislation to allow mitochondrial donation, we will be able to give family members accurate risk data so they can make an informed decision about opting for this new treatment.”

**Study results**
The new study found 96 Australian families currently have the gene and 355 people are currently living with vision loss because of it.

It also shows the overall risk of losing vision if you have the LHON gene is 17.5% for males (one in six males) and 5.4% for females (one in 20 females). This is significantly less than the popularly quoted risk of 50% for males (one in two males) and 10% for females (one in 10 females).

Lisa Kearns, a research genetic counsellor and orthoptist at CERA has worked with LHON families for 20 years, says LHON has traditionally been known as a young man’s disease.

“However, the study’s findings confirm LHON can also affect a smaller number of women, older adults and younger children,’’ she says.

**Research impact**
CERA Principal Investigator Dr Isabel Lopez Sanchez says because LHON is very rare, some ophthalmologists may have never encountered a patient with the disease.

“As a result of our study we want them to consider LHON could be a possibility if a woman, younger child or older adult has lost their vision, to avoid a delayed diagnosis or even misdiagnosis,” she says.

Dr Lopez Sanchez says the study will provide researchers with a comprehensive database of people they can ask to take part in future clinical trials and studies aiming to prevent or treat vision loss.

“It will also help us design accurate trials and studies, which is very important, as that will help us develop efficient treatments,” she says.

**Read the study**
Isabel G. Lopez Sanchez, Lisa S. Kearns, Sandra E. Staffieri, Linda Clark, Myra B. McGuinness, Wafaa Meteoukki, Sona Samuel, Jonathan B. Ruddle, Celia Chen, Clare L. Fraser, John Harrison, Alex W. Hewitt, Neil Howell, David A. Mackey Establishing risk of vision loss in Leber Hereditary Optic Neuropathy. *American Journal of Human Genetics* https://doi.org/10.1016/j.ajhg.2021.09.015

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**Attachments**
LHON fast facts
Case study of Duncan Meerding, who lives with LHON

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**About the Lions Eye Institute**
At the Lions Eye Institute, we make a difference to people's lives through excellent patient care and by pushing the frontiers of science to discover new treatments and cures for eye disease. The Lions Eye Institute spans the dual complementary pathways of research and clinical care, bringing together a globally recognised team of researchers and clinicians who continually build on each other's discoveries, knowledge and expertise, to deliver sight-saving treatment and care around the world. The quest for knowledge and its life-changing applications for patients is what drives our work.

**About the Centre for Eye Research Australia**
The Centre for Eye Research Australia (CERA) is an independent medical research institute based at the Royal Victorian Eye and Ear Hospital in Melbourne and affiliated with the University of Melbourne. We're deeply committed to conducting eye research with real-life impact and finding ways to prevent each other going blind. As an international leader in eye research, we use our world-class knowledge and expertise to achieve better treatments and faster diagnosis of eye disease. Our goal is to prevent vision loss and, ultimately, find cures to restore sight

## Attachment 1

## Fast facts – Leber's Hereditary Optic Neuropathy

- The genetic eye disease risk is automatically passed down from a mother to all her children.
- It may or may not cause vision loss in a person's lifetime.
- Latest statistics show the overall risk of losing vision if you have the LHON gene is 17.5% for males (one in six males) and 5.4% for females (one in 20 females).
- If vision loss does occur, it usually strikes in young adulthood.
- Scientists don't yet know why some people with the LHON gene lose their vision and others keep their vision.
- The vision loss usually only affects the central part of the eye, with peripheral vision (side vision) maintained.
- The central vision loss makes reading, driving and recognising faces difficult.
- Vision loss is sudden, permanent and painless. It usually strikes first in one eye and then, days, weeks or months later, the other eye.
- People with LHON have normal vision until the disease strikes.
- There is currently no treatment or cure for LHON, but emerging technologies, such as gene therapy, are making treatments and cures possible in the future.
- Professor David Mackey is an ophthalmologist who has worked with LHON patients for more than 30 years. He published pioneering work on the prevalence of LHON in the 1990s and helped discover some of the causative mutations for LHON.
- CERA Principal Investigator Dr Isabel Lopez Sanchez is a basic scientist mitochondrial biology and disease who is currently researching why some people with the LHON gene lose vison while others have good vision for life.

**Attachment 2**

**Case study – Duncan Meerding**

It was just an average Friday morning when Duncan Meerding, then 18, realised he had a major problem with his vision.

“I couldn’t see the TV screen,” recalls Duncan, who is now an award-winning furniture and lighting designer in his 30s.

“I was moving my hands about for perspective and I noticed there was a giant blind spot in my left eye. I was like, ‘Ah, I could just be overtired’.”

But it wasn’t a simple matter of fatigue. After visits to doctors and eye specialists, the diagnosis came in: Duncan had LHON. Within eight months, he also lost central vision in his other eye.

He recalls losing his vision was, initially, “psychologically and physically difficult to deal with”.

“Luckily, I had great support from doctors and my Uncle Doug, who also has vision loss from LHON. Because of that support, I came to realise having LHON wasn’t the end of the world – it was the beginning of something new,” he says.

While welcoming the new study’s findings, Duncan encourages anyone with LHON or a similar condition to carve out a life in the here and now.

“Live your life as someone with vision loss; don’t put off your life,” he says. “If and when treatment comes along, that will be great but, in the meantime, learn how to adapt.”

Duncan’s story also brings back memories for Professor Mackey, who has been researching LHON for 30 years.

“I remember seeing Duncan as a little boy when I first started research in the 1990s and met his Uncle Doug.’’