Prof Robyn Jamieson, Head, Eye Genetics Research Unit, Children's Medical Research Institute and The University of Sydney

Seeing the bigger picture: health economics of precision medicine for genetic eye disease

When Prof Robyn Jamieson began her career treating patients with blinding eye diseases, she faced the heart-breaking prospect explaining to parents that there was often no definitive diagnosis and no treatment options.

"We couldn't get a genetic diagnosis and there was no therapy," she recalls.

Now genetic testing is standard and Prof Jamieson, Head of the Eye Genetics Research Unit at CMRI and SCHN, is at the forefront of introducing revolutionary gene therapies with the potential to restore sight.

"This isn't like cancer, where there have been therapies in various areas for years," she says. "These patients had nothing. Their vision deteriorated and there was nothing we could do. So, the first gene therapy was a real icebreaker. And there are more gene therapies in the pipeline."

While the benefits to patients are easy to articulate, there had been no attempt to quantify how advances in gene therapy also translate into broader economic and societal advantages.

"These genetic conditions affect children their whole lives. If we can treat them and improve their vision, then they have a better opportunity to take advantage of education and they are better able to participate in the workforce of the future," Prof Jamieson says.

With a lack of specific evidence of the cost-effectiveness of the emerging ocular gene therapies, a Luminesce Alliance project was designed to gather and analyse the data.

The first study of its kind, it was a collaborative effort between CMRI, SCHN, The University of Sydney and Macquarie University.

The research team first established a new sophisticated model for quantifying the quality-of-life costs of genetic retinal eye diseases, before using it to work on assessing the benefits of genetic screening and gene therapy treatments.

Prof Jamieson says the research will ultimately result in data on the financial, societal and psychosocial costs of different retinal dystrophies, providing economic evidence to support future applications for genomic testing and new therapies.

Providing the first economic model of genetic blindness has potential benefits for other genetic

diseases, by enabling researchers to clearly show the cost savings that could be achieved through precision medicine. There are plans to expand the initial Luminesce Alliance-funded study across Australia.

"This work is critical," Prof Jamieson says. "If we can make a compelling economic argument showing the huge benefits of genomic investigations and precision therapies, then there is more chance that genomic medicine will be equitably funded and available across Australia.

"This initial project, and the ongoing work associated with it, will help us with all the therapies we are developing," she adds.

The study included detailed modelling on the health and economic impacts of impaired vision and blindness due to inherited retinal dystrophies. It will pave the way for further detailed studies investigating the benefits of the first gene therapy for genetic eye disease (Luxturna®) and other gene-specific ocular therapies.

Gene therapy breakthrough

Luxturna® is the world's first approved gene replacement therapy for an inherited blinding eye condition and one of the first gene replacements for any human disease.

It is used to treat children and adults with biallelic pathological mutations in RPE65, a rare mutation that leads to vision loss and blindness. People with a mutation in both copies of the RPE65 gene can suffer from a range of symptoms, including night blindness (nyctalopia), loss of light sensitivity, loss of peripheral vision, loss of sharpness or clarity of vision, and potentially total blindness.

Ocular gene therapy works by injecting Luxturna® under the retina and carrying a functioning RPE65 gene to replace the faulty one, thereby preventing some of these devastating symptoms.

The first patients to benefit were teenage siblings treated at the at The Children's Hospital at Westmead after the therapy was approved by the Therapeutic Goods Administration in 2020.

The therapy was delivered as part of Ocular Gene and Cell Therapies Australia (OGCTA), a collaboration involving SCHN, CMRI and Save Sight Institute, University of Sydney.

"Inherited retinal disease is a devastating diagnosis. Up until now, these patients suffered progressive vision loss that led to blindness and there was no therapy for them at all.

"But through new genomic diagnostics and the use of ocular gene therapy, we are finding that we have the ability to not only stop this ongoing progression but also help to improve vision for people who have RPE65-related retinal vision loss."

– Prof Robyn Jamieson