Media Release

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AUSTRALIAN FIRST – Gene therapy prevents children with eye disease from going blind

Two Sydney siblings have become the first patients in the country to receive a novel gene therapy that has rescued their vision and holds hope for preventing them from going blind.

The ocular gene therapy, 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. Approved by the Therapeutic Goods Administration, LUXTURNA is used to treat children and adults with biallelic pathological mutations in RPE65, a rare mutation that leads to vision loss and blindness.

Seventeen-year-old Rylee and 15-year-old Saman were both diagnosed with Leber congenital amaurosis, a severe form of retinal dystrophy, in their first year of life. They received the life-changing therapy at The Children's Hospital at Westmead in late 2020 and early 2021. The therapy has stopped their progressive vision loss and led to some improvements in their vision.

The therapy was delivered as part of Ocular Gene and Cell Therapies Australia (OGCTA), a new collaboration involving the Genetic Eye Clinic at Sydney Children's Hospitals Network (SCHN), the Eye Genetics Research Unit and Stem Cell Medicine Group at the Children's Medical Research Institute (CMRI), and the Save Sight Institute at Sydney Eye Hospital and University of Sydney.

Professor Robyn Jamieson, Head of the Eye Genetics Research Unit at SCHN and CMRI, lead of OGCTA and Head, Specialty of Genomic Medicine, University of Sydney, said the therapy was revolutionary and would lead to transformation of care for patients with blinding eye diseases.

"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," Professor Jamieson said.

"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."

Children and adults born 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.

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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 real-world improvements in visual function has been quite remarkable bringing to life the rather dry clinical trials outcome measures. It is tremendously heartening to see the changes in vision capabilities for these first patients treated with LUXTURNA, Professor John Grigg, Head of Specialty of Ophthalmology, Save Sight Institute, University of Sydney and lead inherited retinal disease specialist in OGTCA said.

"As an ophthalmologist who has been caring for patients with Leber's amaurosis for many years and unable to offer any treatment, it is incredibly rewarding to now have the opportunity to not only give families hope but also be involved in improving their child's vision," Frank Martin, Clinical Professor in the Specialties of Paediatrics and Child Health and Ophthalmology at the University of Sydney said.

Associate Professor Matthew Simunovic, Vitreoretinal Surgeon, Sydney Eye Hospital and SCHN and Associate Professor at the Save Sight Institute, University of Sydney performed the first surgery and said the benefits of treatment should extend well into the future:

"This is incredibly delicate surgery in which LUXTURNA is injected under the retina, which in some patients can be as thin as a sheet of copy paper. Riley and Saman have had profound improvements in their vision, which mirror the results seen in the pivotal clinical trials. Importantly, such benefits appear to be sustained for many years – in fact, for as long as patients have been followed up. Successfully delivering the first approved gene therapy has been a fantastic team effort, and it underscores Australia's capability in this field" A/Prof. Simunovic said.

To date, this treatment has been used to treat four patients and while it can only be used to treat this specific form of retinal disease, it does provide significant hope that similar treatments will be able to be applied to other retinal disease genes in the future.

"This heralds a new era in transforming the lives of these people who otherwise have a life of blindness ahead of them and provides hope for more than 15,000 other affected Australians who live with some form of inherited retinal disease," Professor Grigg said.

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