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World first breakthrough for haemophilia at RPA

In a world first, Royal Prince Alfred Hospital researchers are among an international team to have developed a gene therapy for the life-threatening blood disorder, haemophilia.

The breakthrough, led by RPA's Professor John Rasko, (pictured with patient Mark Lee, his partner Shannon and daughter Violet), with researchers from Children's Hospital in Philadelphia, is a major milestone in the quest to cure the bleeding disorder.

The finding, published in today's *New England Journal of Medicine*, is the culmination of more than 20 years' work and opens the door to using gene therapy to treat more than 4000 other genetic disorders, including blindness.

"We are very excited about the results, as those people in our trial have previously had to live with the risks of spontaneous bleeding every day. To prevent potentially life-threatening bleeds they must typically inject themselves with clotting factors every few days," Professor Rasko said.

"This trial has targeted haemophilia B, which affects about 500 males in Australia (with about 100 having the severe form), but our next focus is targeting haemophilia A, which affects more than 2300."

The director of NSW Office of Health and Medical Research, Dr Tony Penna, said the discovery demonstrated the importance of supporting medical research in NSW.

"This extraordinary international collaboration could potentially save the lives of thousands of people around the world and was only possible because of the dedication and determination of our brilliant researchers," Dr Penna said.

"NSW has state-of-the-art gene and cell therapy facilities with outstanding clinician researchers like Professor Rasko who are leading this cutting-edge research and its translation into clinical care.

"This is a world first that all Australians can be proud of and highlights the need for both public and private investment in medical research.

The NSW Government is investing a record $1.25 billion over four years in medical research across the state.

Haemophilia B is an inherited disorder where blood does not clot properly due to missing or defective clotting factor nine (IX), affecting more than more than 500 males in Australia.

People with this disorder experience a wide range of bleeding episodes, usually into the joints or muscles. Episodes can often occur spontaneously, without an obvious cause, or as a result of trauma or injury. Over time, bleeding can cause severe arthritis, chronic pain and disability.

The research involved 10 adults who were injected with a gene therapy designed to produce the clotting protein Factor Nine (IX).

Trial participant Mark Lee, 38, has undergone infusions up to three times a week since birth and lost two brothers to complications from haemophilia B when they were children.

Since receiving the experimental gene therapy, he has not had any bleeds.

“This is life-changing for me. I spent my childhood wrapped up in cotton wool, unable to play football or do any of the things my mates could. I would always remind myself that there were people worse off than me, but it was still disappointing,” Mr Lee said.

“I have two daughters who are carriers for haemophilia, but now I know that if they have affected children, it will be one injection and they can live normal lives. This goes beyond our little family currently. It will have a positive impact on all generations to come.

“And my mum now knows she won’t see her only surviving son die from haemophilia.”

Professor Rasko has spent the past two decades fine-tuning the gene therapy and says it is the beginning of the end of this lifelong bleeding disorder.

“We now know how to beat the immune response to achieve what may be a permanent cure.”

The vice president of Haemophilia Foundation Australia, Daniel Credazzi, who has a son with haemophilia, said, even with current best practice treatments today, sufferers experienced painful bleeds causing long term damage to joints, muscles and organs throughout their lives.

“The real potential of a cure with safe and effective gene therapy is very exciting for people living with this chronic condition, and for their families.”

Spark Therapeutics, a biotech company attached to the Children’s Hospital in Philadelphia, is funding the commercialisation of the therapy.