Media Release

The Sydney children's Hospitals Network
care, advocacy, research, education

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Gene therapy key in treating SMA

New research has found gene therapy may provide an effective treatment for Spinal Muscular Atrophy (SMA), a devastating and fatal genetic condition.

Results from the SPR1NT trial (part one), were presented overnight at the European Academy of Neurology (EAN) Conference. This trial investigated the use of Zolgensma, a novel viral vector-based gene replacement therapy. Fourteen infants, who were under six weeks of age and at risk of developing the most common and severe form of SMA, were treated before their symptoms started.

SMA affects the motor nerve cells in the spinal cord, causing progressive muscle weakness and preventing babies from being able to roll, sit up, crawl, walk and eventually breathe. Until recently, it was the leading genetic cause of infant death in Australia, occurring in 1 in every 10,000 births.

Sydney Children's Hospitals Network (SCHN) was the only Australian site selected to participate in the trial, and was one of the largest global recruitment sites, with four patients enrolled from SCHN. The recent introduction of SMA testing to the newborn screening program in NSW, funded by a $2 million investment from the NSW Government, made this possible.

The study, which followed each participant until aged 18 months, found that all children achieved the ability to sit independently (with 78 per cent achieving this milestone within the normal developmental window), all were alive and free of permanent ventilation and all had normal swallow function and were fed exclusively by mouth by 18 months of age.

The trial also showed that following treatment nine children were able to walk independently and all showed fine motor performance similar to babies without SMA by the completion of the study.

Site-based lead for the study and Paediatric Neurologist at Sydney Children's Hospital, Randwick, A/Prof Michelle Farrar, said the results of the trial was a potential game-changer for both clinicians and families affected by SMA.

"These results are extremely exciting and encouraging, not only are these children surviving but with this therapy, most are meeting the developmental milestones of any normal baby, which is unheard of," A/Prof Farrar said.

Zolgensma works by treating SMA at its root cause, inserting a functioning copy of the defective gene into the cells.

"By identifying these infants with SMA before the onset of symptoms, early results suggest we may have been able to take what was considered a lethal disease, and turn that around with a one-time, single dose infusion."

"It is giving life back to these babies and hope back to their families," A/Prof Farrar said.

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Since the introduction of SMA into the newborn screening program in 2018, more than 200,000 babies have been screening, which has helped significantly with early identification of the condition.

“We know that early identification is vital in the treatment of SMA and that is what the newborn screening program has allowed us to do. It has radically shifted our model of care and we are now in a position where we can rewrite the history of SMA,” Dr Farrar said.

In addition to the newborn screening program, the NSW Government has invested $25 million to boost the state’s capability to manufacture viral vectors – the key components of this type of therapy which holds great promise for novel treatments for other genetic diseases.

The second part of the SPR1NT trial, which explored the effect of Zolgensma in babies with milder SMA, is due to be presented later this year.

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Contact Details | Andrea Ford
Phone: 9382
Email: andrea.ford@health.nsw.gov.au