

Viral Vector Case Study: Mum's hope for future of gene therapy



Sydney mother Gemma Najem felt hope for the first time after her son Jonathan, diagnosed with Spinal Muscular Atrophy (SMA) as a newborn, was granted access to gene therapy and infused with a vital gene that was missing from his body.

This gene therapy utilises viral vectors, a key component of gene therapy which will be manufactured locally at a new commercial scale facility to be built at Westmead.

“When Jonathan received access to gene therapy at four and a half months, I yelled out in disbelief! It felt like a weight had been lifted off us,” Gemma said.

SMA is a genetic neuromuscular disorder, similar to motor neurone disease, causing irreversible muscle weakness. Roughly one in every 10,000 babies has SMA, and most parents don't know they're carrying the gene until their child is born.

Gemma says viral vector technology, which is a vital part of the gene therapy her son received has changed Jonathan's life.

“Access to this technology is giving him the best chance at mobility, strength and health. A couple of months before he turned two, he told me he can't walk but he wants to - with this technology enabling gene therapy treatment, he is now taking steps with his walker with the biggest smile on his face!

“It makes a huge difference in our day-to-day life as it has reduced the difficulties that are associated with his condition.

Gemma says plans to expand local production of viral vectors locally is crucial and would provide the best chance for Australians with rare genetic diseases to be treated with life-saving and life-changing technologies.

“For the families of children with rare genetic conditions, local production of viral vectors makes massive difference because it can fast track life-saving and life-changing treatments,” Gemma says.

“Timing of treatment is also critical with SMA - we watched American families have their children treated even earlier than our son, and with newborn screening and pre-symptomatic treatment, Jonathan wouldn't even need mobility equipment or intensive therapy.”

Throughout Jonathan's journey, the family has been supported by locally based Professor Ian Alexander, one of the world's leading gene therapy experts who is also part of the viral vector facility manufacturing project.

“Having access to gene therapy experts who are based in NSW has greatly assisted with Jonathan's quality of care,” Gemma said.

“It's made a world of difference and provided us with a sense of safety knowing he was being treated by world-leading scientists. It's made us feel confident we were getting the right advice and doing everything in our power to advocate for our baby boy.

“Jonathan is a cheeky, charming and determined toddler. While he has made progress since his treatment, in recent months he has taken strides forward in his progress.

“It's been a joy to us and loved ones all around the world to see his progress and we have more hope than ever before for his future.”