# Patient story: Baby M

A few days after Baby M's newborn heal prick test, his parents received some devastating news: their perfect little boy had inherited the most severe form of the genetic condition Spinal Muscular Atrophy (SMA).

While he had not yet developed symptoms, they were told he would progressively lose the ability to roll, sit up, crawl, walk and, eventually, breathe. In 9 to 10 months, his condition would be fatal.

One of the options available to them in NSW was to take part in an international clinical trial of a gene therapy for SMA, with the aim of assessing its safety and efficacy. This investigational medicine was given as a single injection with the gene delivered inside a viral vector.

Baby M's parents and research team hoped that the science would offer him the chance of a healthy life. The family made the difficult choice to enrol their baby in the gene therapy trial, trusting that the science and medicine would be positive.

One year later, Baby M is reaching his milestones and defying his prognosis; he is sitting independently, breathing, feeding and swallowing normally, and is learning to walk.

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

In future, it is likely that gene therapy will rewrite the outlook for other babies in Australia who are born with SMA. Baby M's parents have generously supported the research program to produce evidence looking at the acceptability, health economics and implementation of this new model of care for SMA, so that this can change health practices and inform sustainable health policy.

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