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Gene Therapy Delivered Soon After Birth Halts Progression Of Spinal Muscular Atrophy

Dell Children's Medical Center
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Dell Children's Newborn Patients Can Receive Gene Therapy for SMA, Radically Improving Their Chance of a Healthy Future

Dell Children's Pediatric Neuromuscular Disease Center is a

a specialty program within UT Health Austin Pediatric Neurosciences at Dell Children's. The center specializes in treating children with acquired and genetic neuromuscular diseases, such as spinal muscular atrophy (SMA), inherited neuropathies, muscular dystrophies, congenital myasthenic syndromes, and congenital myopathies.

Recent progress has been dramatic, especially for SMA, an autosomal recessive degenerative disorder of the spinal motor neurons. Untreated, SMA causes progressive muscle weakness, reducing the life expectancy for individuals with SMA type 1, the most common form, to around two years. The recent development of gene replacement therapy has dramatically improved this poor prognosis.

Veda Vedanarayanan, MD

"This is a major game-changer," says Dr. Veda Vedanarayanan, chief of the Pediatric Neuromuscular Disease Center at Dell Children's Medical Center.

To be fully effective, however, gene therapy for SMA must be administered early, before the child's spinal motor neurons deteriorate. Later treatment typically stabilizes the child's function but does not reverse existing dysfunction, so early diagnosis via newborn screening has become vitally important. Before newborns were routinely screened for SMA, most babies were diagnosed between three and six months of age, after they started to miss developmental milestones because their muscle condition was deteriorating. If newborn screening identifies the condition, prompt gene replacement therapy can be curative.

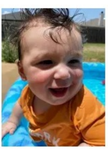
That's what Dr. Vedanarayanan provided for baby Noah, one of 35 patients with SMA who currently receive care at the Neuromuscular Disease Center, several of whom have undergone gene therapy. Noah was the first baby in Texas who was diagnosed with SMA type 1 by newborn screening, which added the SMA gene just 88 days before his birth.

"We could not have made this diagnosis by examining Noah as a newborn," said Dr. Vedanarayanan. "But with newborn screening, we can catch it really early and save a lot of nerve cells from deteriorating."

A month after birth, Noah underwent a single infusion of the drug Zolgensma—one of three available options and the one with the optimal chance for long-term benefit. The infusion provides a new copy of the gene that makes the deficient protein.

> "If he hadn't had this treatment, or if he were in a different situation, I would expect now, at his age, for him to be bed-bound, perhaps on ventilator support," said Dr. Vedanarayanan.

Today, Noah is an active kid who keeps his parents on their toes.



"More therapies for SMA are on the way," added Dr. Vedanarayanan, "including an agent to build muscles or muscle mass." Researchers are now investigating the feasibility of delivering gene therapy in a smaller dose directly into the spinal fluid."

"What I tell trainees and medical students is that gene therapy is going to be the next big step for genetic diseases," said Dr. Vedanarayanan. "In the future, we will be able to treat many conditions with gene therapy that cannot be remedied now."

You can bet Dell Children's will be at the forefront of that brighter future for kids all across the country. Stay tuned.

To Refer a Patient

For more about UT Health Austin Pediatric Neurosciences at Dell Children's, click here.

To contact the Pediatric Neurology Clinic directly, please call 512-628-1855 or fax referral forms to 512-380-7544.

About Dell Children's Medical Center

UT Health Austin Pediatric Neurosciences at Dell Children's, part of Ascension Seton, nationally ranked for its pediatric neurology and neurosurgery, serves families in Central Texas and its surrounding counties. In collaboration with UT Health Austin, the pediatric neuroscience program offers pediatric neurology, pediatric neurosurgery, pediatric rehabilitation, pediatric neuro-ophthalmology, and pediatric neuropsychology care. Whether the patient has epilepsy, ongoing headache pain, hydrocephalus, or another neurology need, our specialists deliver advanced, compassionate, and personalized care to get to the root of the problem. Dell Children's offers the most advanced neurosciences program for children in Central Texas, and we're the only Level 4 pediatric epilepsy monitoring unit. We also support research and provide training for medical students, as well as medical residents, in child and adult neurology, pediatrics, psychiatry, and other disciplines.

About our collaboration with UT Health Austin

UT Health Austin is the clinical practice of the Dell Medical School at The University of Texas at Austin. The collaboration between UT Health Austin and Dell Children's brings together medical professionals, medical school learners, and researchers who are all part of the integrated mission of transforming healthcare delivery and redesigning the academic health environment to better serve society. This collaboration allows us to provide you with a team of highly specialized providers who are at the forefront of the latest research, diagnostic, and technological developments.

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