



Giving Thanks for Progress: The New SMA

By: Al Freedman

Twenty-one years ago - in 1995 - my 6-month-old son, Jack, was diagnosed with SMA. From that day forward, everything changed, especially my expectations for Jack's future. We were told there was no cure or treatment for SMA and that Jack would not live to see his 2nd birthday. Receiving this news was very painful for our family. It was very difficult for us to find any hope.

So we took our baby home and we did our best to learn how to care for him. I learned how to use a pulse oximeter, suction machine, and cough-assist. I've learned all about ventilator settings, wheelchair electronics, and feeding tubes. For years, I've driven around with a toothbrush and a packed overnight bag in the trunk of my car, always prepared for Jack's next hospitalization. Twenty-one years later, I feel very fortunate to have Jack, but the road we've travelled has been anything but smooth.

Twenty-one years ago - in 1995 - something else happened, too. Scientists discovered the gene responsible for SMA. Looking back, I didn't focus too much of my attention on this news. I was too busy taking care of Jack and trying to come to terms with the news that he would not live very long. But when I attended my first Families of SMA conference a few months later, a palpable sense of hope was in the air. Finding the gene responsible for SMA represented a significant step forward and provided new energy for our families and researchers.

Our SMA community back then was much smaller in number, but no less committed to finding a cure. Audrey Lewis, the founder of our organization, had been working tirelessly for years to raise awareness, support our families, and to fund research. I had heard that Audrey founded our organization in 1984, together with a small group of mothers of SMA-affected children, by holding meetings in her Chicago-area home. Audrey had truly made a difference, and progress had begun to be made on the research front.

Along with that renewed sense of hope, the researchers at our 1996 conference made it clear that the road ahead to finding a treatment or cure for SMA would be long and complicated. Years would pass before the discovery of the SMA gene would lead to any kind of treatment for our children. Because waiting for a treatment or cure would be far into the future, I tucked away the idea that Jack, or any of our children,

could be helped by the discovery of the SMA gene. The idea that a treatment for SMA could help our children was nothing short of a pipedream in my mind.

Twenty-one years have passed since the discovery of the SMA gene. Our Families of SMA organization is now called CureSMA. What started as a handful of parents meeting at Audrey's house in Chicago in 1984, has grown to thousands of families around the world. An entire generation of families affected by SMA has joined our community over these two decades. And with the announcement that the FDA is fast-tracking the approval of a new treatment for SMA, the road ahead now looks and feels very different for our families. With the help of nusinersen, many SMA-affected babies and young children are maintaining, and even gaining strength, rather than losing strength. This is the new SMA.

As I write this, Thanksgiving is just around the corner. As I reflect back on the past 21 years, I feel very grateful.

---I am grateful for Audrey Lewis and the many families who supported me when Jack was newly diagnosed with SMA. I've done my best to thank them by "paying it forward" to other families over the years.

---I am grateful for my son, Jack, who has defied the odds and has accomplished so much in his life.... regardless of whether he ultimately benefits directly from nusinersen or any other new treatment.

---I am grateful that our collective effort to raise awareness and funds to support SMA research has led to this significant research and treatment milestone. The pipedream of two decades ago is actually becoming a reality.

---But most of all, I'm grateful that the road ahead for newly diagnosed SMA-affected children will be more hopeful than the road my son has needed to travel.

As a community, we're all in this together. Let's take a moment to celebrate and be thankful for this extraordinary milestone, and continue to support each other as we move forward, with hope, into the future.