Newborn Screening for Spinal Muscular Atrophy
Testimony of Cure SMA
Delivered by Mrs. Kristen Lasko

Meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
November 3, 2016

Good morning, Dr. Bocchini and members of the Advisory Committee. Thank you for the opportunity to testify. My name is Kristen Lasko, and my son Max has Spinal Muscular Atrophy. When Max was diagnosed, Cure SMA was there to help us. Cure SMA supports and directs comprehensive research that drives breakthroughs in treatment and care, and provides families the support they need.

On behalf of Cure SMA, my family, and thousands of other families affected by SMA, I am here to comment regarding the committee's nomination and evaluation process for candidate conditions on the Recommended Uniform Screening Panel (RUSP).

When my son Max was born, he received Apgar scores of eight and nine. Everyone commented on how he was such a "chill" baby, but we now know that he was exhibiting hypotonia. Sometimes it seemed like he had trouble managing secretions; I remember modelling a "cough" to try to get him to clear his throat. I worried, but family members assured me this was normal parental anxiety.

At one month of age, Max wasn't lifting his head on his own. Our pediatrician encouraged us to "wait and see." When Max's two month checkup came and he still wasn't lifting his head, a different pediatrician sent us to a pediatric neurologist. Three weeks later—after two consultations and a blood test—we received Max's SMA Type I diagnosis. We were devastated. The doctor told us that babies diagnosed with SMA Type I had a 50% chance of making it to age 2.

What sort of difference can early intervention make with SMA? All the difference in the world!

One week ago, a New Drug Application for a drug called nusinersen was accepted by the FDA – a first for the SMA community. Granted priority review, this takes us one step closer to an FDA-approved treatment for people with SMA like Max.

What would Max's life be like if we had learned his SMA diagnosis a little earlier? Max's friends who were fortunate enough to receive early treatment in a clinical trial are now lifting their heads, sitting up unassisted, moving their arms and legs against gravity, pushing their own manual wheelchairs, eating orally, standing with assistance, and breathing on their own.

If we had been able to learn of Max's diagnosis in the first few weeks of life, he may never have needed to go the hospital for seven weeks due to a cold that caused him to get a tracheostomy, G-tube, and six life-saving biomedical machines. With a pre-symptomatic diagnosis, Max could

have had pre-symptomatic treatments, which human data and animal model data suggest is most effective in SMA.

With a treatment on the cusp of approval, it is of utmost importance that SMA be added to the RUSP so that families can and get their child treatment as soon as possible.

The SMA community and I strongly urge the Advisory Committee to consider the forthcoming SMA screening nomination. SMA can be effectively diagnosed with a blood test. There is now a safe and efficacious treatment for SMA. And the earlier the intervention, the greater the benefit. I thank the Committee for the opportunity to speak to you, and I appreciate your consideration.