Good morning (afternoon), members of the Advisory Committee. Thank you for the opportunity to testify today.

My name is Amy Medina. My husband and I have two sons who are affected by spinal muscular atrophy. Mateo is five, and Javier is one.

Spinal muscular atrophy, also known as SMA, is the leading genetic cause of death for infants. On behalf of the SMA community and Cure SMA, I am here to comment regarding the urgent need for newborn screening for SMA.

Over the last decade, there have been significant advances in drug development for SMA, culminating in the approval on December 23, 2016, of Spinraza. Spinraza is an antisense drug that treats the underlying genetics of SMA.

With an FDA-approved therapy, newborn screening would allow infants born with SMA to immediately begin receiving treatment. Both human natural history data and animal model data suggest that early drug intervention is required for greatest efficacy in SMA.

Results from studies of infants treated with Spinraza show that presymptomatic infants treated with Spinraza develop more motor milestones than symptomatic treated infants.

My family's experience reflects this.

Mateo showed symptoms right at birth that included low tone, lack of crying and rapid breathing however we would not hear the words Spinal Muscular Atrophy until he was 1 month old. At that time we were told he would most likely never see his 2nd birthday and were give little to no hope. We transferred hospitals and found a doctor that was willing to educate us on SMA and fight with us to give Mateo the best life possible. A gtube surgery, many scary crashes which resulted in 911 calls and hospital stays and a trach surgery all happened in his first 7 months of life. Five years later Mateo has lost all movement except very tiny movement of his feet and a small curl of the side of his mouth to indicate a smile.

I had an amniocentesis with Javier and was prepared for him to have SMA. Javier was monitored closely throughout my pregnancy and at birth. I prepared for Javier to be similar to Mateo however he was given the opportunity to receive Spinraza at just 12 days old. Javier has gotten 6 doses. He is able to sit up on his own, he can roll all over the house, stands with assistance, eats everything by mouth, is very loud vocally, coughs on his own and does not need any type of breathing support. Javier has battled a cold at home and maintained his

oxygen saturations of 97 or higher, even while lying on his stomach to sleep. This is generally unheard of for SMA children as they are belly breathers and the common cold can most likely hospitalize them. Javier presents more like a typical child his age and people are often shocked when I tell them both my children have the same disease.

In conclusion, the SMA community strongly urges the Advisory Committee to take up consideration of the forthcoming SMA screening nomination with concerted focus on the availability of a treatment for SMA, the success of the technology in screening for SMA, and the demonstrated benefits of early intervention. I thank the Committee for the opportunity to address you today and appreciate your consideration.