Chairman Huffman, Vice Chairman Gavarone, and Ranking Member Antonio thank you for the opportunity to testify today.

I am writing to let you know why SMA needs to be on the list for NBS. Twenty years ago my niece was DX with SMA type 1 by the doctors that will be present in the hearing today. Dr. Prior tested her blood to confirm she has SMA while Dr. Mendel saw her after her DX at the MDA Clinic. At that time my sister was told by the MDA doctors take her home and make her comfortable we do not treat type 1 SMA children. My niece is name is Madison Reed of Dublin Ohio. She was DX at 8 months - she was a typical 8 month old child except she did not meet any milestones! It took 8 months for doctors to figure out what was wrong with her. Sometime this still happens today. Knowing at birth what she may have had could have made a big difference in how she was treated during those 8 months of unknown mystery. However, back then they did not check for SMA on the NBS because there was no treatment for it. In 1995 they just discovered the gene and then in 1996 the Prior Lab at OSU discovered the testing for it. In 1997 Madison was born - to test for SMA NCH doctors were going to do a muscle biopsy but we lucked out when another doctor knew about the blood test. Yes right here at our local children's hospital they did not even know about the blood test. This is one big reason why SMA needs to be tested at birth. Another is we had no history of any family members having SMA on both side. Still more reason, Madison's brother is a carrier so when he meets a girl she would need to be carrier tested to see if she is a carrier. Even if she is not - there is still a chance of them still having a child with SMA.

Fast forward twenty years: In Dec 2016 for a nice Christmas present to the SMA community FDA approved the only drug treatment for SMA called Spinraza. Trials showed that giving the drug early to the worst type - hence type 1 - the babies were meeting milestones after milestones. For every 2 infants given the drug 1 infant was getting the placebo. Those not getting the drug was your typical SMA type 1 child - wasting away slowly losing motor functions - breathing, eating, reaching, moving period just laying there. Jackie Camboni was in the Gene Replacement trial at NCH - which is not fda approved yet - and also has Spinraza. Madison at Jackies age right now could never do what she is doing. Even though Jackie was given in the trial at 5 months she is achieving many milestones but not the true measure if given before 8 weeks. Those given either Spinraza and or Gene Therapy before 2 months show the best out come - almost completely normal. They can walk eat, speak, run, play - you would not know they have SMA. This is the biggest reason why SMA needs to be on the NBS list. While Spinraza is a treatment on the back up copy of SMA2 it still shows there is no major motor neuron loss. Gene Therapy actually goes after the missing main GENE - this will be the cure for SMA. Right now, Spinraza is the best thing we have to slow and reverse the progress of this disease.

Just to let you Madison is alive she is 20 will be 21 Feb 4th. nothing stops her not even this disease. She has a facebook page - Madison Reed - please check it out. Madison and my family raised over $1M dollars for SMA research at OSU and NCH. Money was used to test mice and other animals for the current gene therapy so data can be collected to present to FDA to start the first in human gene trial for SMA. Our family has and will do anything to make sure no family has to go through what we have been through for 20 years. If your child has to have SMA this is the time - we have a treatment Spinraza for sure and soon there will be a cure Gene Replacement for it.

When they did the trials for Spinraza they did not do it on any type 1 patient over the age of 6 months. The same is being done for gene trials. The reason was type 1 older children would produced many different varibles and fda would not like that. However, FDA approved Spinraza for all ages all types - Madison will be getting her 6th Spinraza dose in Dec. It has slowed progression of the disease and has helped in a few areas like breathing.

SMA needs to be on the list - knowing your infant has it can get them on Spinraza NOW.
Our journey has been long and frustrating but Madison motto is When you Believe Miracles Happen. Madison believed there would be treatment - she got the Spinraza. She also believes that Gene Therapy can not only help the new infants but can also help people like her. Madison believes NBS will be passed in Ohio so the next 45=50 families who would get the SMA DX will get to see thier baby grow up meeting milestones that make them a kid.
Here is a photo of Madison - during her 4 loading dosing of Spinraza.

Thank you
Michelle Aunt to the best kid who has SMA.

Michelle Worrellia