Interview with Avery's mother

Would you please briefly introduce yourself?
My name is Shannon, I'm from Georgia in the United States. My husband, Sterling, and I live in Alabama on the border with Georgia with our daughter Avery, our dogs Bear and Roscoe, and cats Dixie, Oscar and Catniss. Avery is our one and only miracle child we waited years to have.

How did you find out that Avery had SMA and how old was Avery at that time?
We got her results through newborn screening and she was the first baby positive for SMA on this screening in Georgia. She was five days old when we got these results. She was almost a month old when we got secondary test results to confirm the diagnosis.

What were the available treatment options when Avery was diagnosed?
Avery was diagnosed in April 2019 so Spinraza was available but Zolgensma became FDA approved by the end of that May, very soon after the secondary test results came back.

How did you hear about Zolgensma and what made you decide to prefer this treatment?
Zolgensma came on our radar after lots of research and then our hopes about its effectiveness were solidified after joining some Facebook support groups and seeing the testimonies from those in the trials. We felt it was worth the difficulty obtaining for the potential life long efficacy with just one dose.

When and where did Avery receive treatment?
We tried to get into presymptomatic trials with no luck but were contacted by a doctor in Massachusetts about the managed access program. We made the 1,000 mile trip soon after in June 2019 for preliminary tests and then again in July. She received the treatment in Boston on July 17th, 2019.

Could you share your experiences with the treatment and the after care?
The treatment itself was uneventful after the IV was placed. The IV placement was very difficult with her being a tiny 3 month old. After it was placed everything went smoothly and Avery napped, ate and played for the rest of our stay. After treatment things were difficult with her tolerating the prednisolone and she did experience the common low fever the first few days. We found a better formulation of the drug about five days afterward that she could keep down better and used omeprazole to help with the vomiting and reflux. Overall, I'd definitely say the whole experience was hard but well worth the lifelong benefit.

When did you notice developments after treatment and what were they?
Avery never showed outward symptoms and was very strong leading up to treatment. In a matter of days after she sat independently very briefly in a tripod position at 3 months old! She soon began rolling and was sitting and crawling independently by 5 months old. Today just shy of 16 months Avery has hit all her physical milestones typical of a child her age.

What is your opinion on the different treatment options Spinraza, Zolgensma and Risdiplam?
I think Zolgensma is the best option where possible with Risdiplam being a good supporting option or standalone treatment where Zolgensma isn't possible. We are considering Risdiplam as her doctor believes it may help improve swallowing function which is Avery's main struggle. Spinraza was a breakthrough for sure and has saved many lives but I think it will phase out in years to come if/when Zolgensma and Risdiplam become more globally available.

How did the Zolgensma treatment changed the life of your family?
Zolgensma changed everything, literally everything. Avery was projected to be type two and I'd like to think she would have been on the stronger end of the spectrum based on how strong she was pretreatment. Regardless, she would have never walked and probably wouldn't be able to eat orally based on her difficulty swallowing that she's still facing. This is a girl who loves to run all over the place, climb things and get into all kinds of trouble she wouldn't be able to do without Zolgensma.

What is your opinion on the necessity of the SMA newbornscreening?
I cannot stress how important it is. It is life altering! We would have never known Avery had SMA or anything wrong at birth without that test. She appeared perfectly healthy and so, so strong! It would have taken a sudden decline in movement or constant respiratory issues to have a clue something was wrong and at that point the damage is done. It's such a sneaky, quick disease. With amazing treatments available there's no reason not to test!

How will SMA treatment look like five years from now, in your opinion?
I think Zolgensma will be the ideal treatment for babies and toddlers with Risdiplam being preferred for older patients. It may not be in five years,but I definitely think CRISPR type treatment will be in use in our children's lifetime.