

Our Family's Experience Participating in the Natural History Study

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When my daughter, Macy, was diagnosed with Rett syndrome, we entered a new and often confusing world. There was so much to learn and adjust to. I dealt with "Why Me" and "Why Her" as I struggled to understand why my daughter had to endure what was happening within her. I decided to control the things I could and I immersed myself in this new world, deciding to learn EVERYTHING I could to help Macy. Little by little, day by day, I learned; about therapies, about wheelchairs, about g-tubes and about life with Rett. I also learned about the pharmaceuticals that were being tested in clinical trials. I really wanted Macy to be part of those clinical trials.

As I started to ask questions about clinical trials, I found that I didn't necessarily like all of the answers. I learned that Macy was too young to participate in the trials going on at that point. I was crushed by that and felt it was a huge road block to Macy's involvement in research. Then I came across the Natural History Study. Initially, I had no idea what that meant but, eventually, I learned that it was THE way we could make a difference and participate in research. We started where everyone should start. We participated in the Natural History Study.

What is this study and why is it so very important? Natural history studies are done for many disorders to gather fundamental information about the way the disorder presents and progresses over time. The Natural History study is a massive database of information about how Rett syndrome affects many aspects of our children's lives – including GI issues, breathing difficulties, seizures, and many more. That information educates the medical community in understanding Rett syndrome. It is also accessed by researchers and pharmaceutical companies and provides them with a wealth of data they use to develop treatments and ultimately a cure for Rett syndrome.

The Natural History Study takes place in 15 different locations in the US. We chose the UAB Rett clinic with Dr. Alan Percy. We contacted the clinic and the Program Coordinator, Jerry Childers, helped guide us through the steps needed to make an appointment. It was costly for us to travel, so I reached out to a local foundation for help with funding for travel. There was a LOT of paperwork but it was worth wading through it knowing we were part of something that would make a difference.

Our first appointment was April 4, 2017. Looking back, it was no different than walking in to any new doctor's office but to us it held the promise of answers to the many questions we had about Macy's condition and her future. We met with Susie, our dietician and then with Jane Lane and Dr. Alan Percy. We spent 4 hours at that appointment. That may sound long - and it was - but it flew by because I was so happy to finally have a Rett expert to listen to me and answer my questions. Not only did we get the guidance we needed in the moment but we knew we were part of something bigger.

We soon will be traveling again to the UAB Rett clinic for our annual visit. This time, my mind will be filled with not only new questions about Macy but with thoughts about all of my fellow Rett families that I hope will be inspired to join us and become part of the Natural History Study. Together, we will help unlock the mysteries of Rett syndrome and find treatments and even a cure for Rett syndrome.