

“For the parents in this group...Regardless of our children having the same genetic make-up, they all will be their own little person. My hopes are that in taking the time to write this, even if one family picks up on something that may help them, it was worth my time!

I had a normal pregnancy for the most part. While pregnant, during my first ultrasound (4mo) the baby's head circumference was measured at 3 percentile. Because of this there was some concern by my physician but all else seemed norm. A second and third ultrasound was done at later stages and the head circumference still remained at an extremely low percentile. I was 35 years old when Sylvia was conceived and 36 years old when she was born. I used an IUD for birth control for 5 years before she was born as she was my third child, and we were not sure if our family was complete after my second.(I only add this because still to this date, it amazes me that it is never asked what kind of birth control or what medications a female may be on before or during pregnancy).

When Sylvia was born, although all of her features where norm, I knew that something was a bit different. I ran a small daycare in my home for 8 years, so I had experience with a lot of children including my own 2 born prior to Sylvia. As an infant she was very hard to settle. She cried a lot regardless of what position you had her in and Sleep was non-existent in her life.

Her birth weight was 7lbs 8oz. Her actual head circumference at birth was below the 2<sup>nd</sup> percentile and to date has never reached above the 10<sup>th</sup> percentile. She rolled at an early age, but seemed stiff as a board. She sat at an average age, but tipped over constantly and seemed to have no reflexes. It took her months to master sitting up without falling. I remember traveling to Florida during this time and our rental house had tile floors. We had to take the mattress of the bed and keep it on the floor for her to sit on! When she started to crawl, which was at a normal age, she dragged her right leg behind her instead of moving it as you should, and she moved as slow as a sloth. This was another big red flag to us that something did not seem right. She was very delayed in smiling, and laughing. At 10 months she was standing and cruising on any and everything, but it took her until 18 months to walk independently. During those 8 months, it was very challenging. She would let go of things, but had very little balance. She would tip over, strait as a board and did nothing to try to catch herself. For months we always had to be sure there was someone at her side to catch her as she tipped. When she began walking independently, if she turned her head side to side or looked up she fell. She seemed to vocalize a lot when she was younger, if you talked she would make noises back. At 14 months of age, that stopped and she became literally silent.

When she was 15 months old, I watched my daughter try to put a plastic ball into a play gumball machine and realized she could not co-ordinate that movement, and did not have control to let go of the ball. I found a pediatrician who would listen to me. 1 month later at the age of 16 months, I traveled 2 hours and spent 8 hours undergoing meetings with neurology, ENT and also an MRI scan. Her MRI showed very thin myelin structure (which was then thought to be the reason for her poor co-ordination). But everything else was fine. I was asked if it would be ok to run some blood work on her, just in case there was something else going on. I assumed they were looking at nutrition.

3 weeks later I received a phone call from her neurologist that to her surprise the genetic results were back (shocked, as I didn't even know a genetic test was done) and she had a microdeletion of 2q23.3-33.1. I was told from that neurologist that not really anything was known. She found 4 documented cases but didn't think this deletion was going to affect Sylvia because those cases all had a cleft palate and Sylvia did not. But I could google those cases if I wanted. So I did. I remember thinking this can't be my child. I remember crying. I didn't allow myself to "google" those numbers again for over a year. Time passed and I did one more time. I scrolled through pages and on the 5<sup>th</sup> page I came across a site that seemed interesting to me. It was a site for parents, called [www.rarechromo.org](http://www.rarechromo.org). I read page after page and realized this was my child. And I began to accept my child, instead of look at her every second of the day wondering what and who she would become.

Sylvia is now almost 7 years old. Here is a little bit about our journey with her so far.

The most significant thing we have learned:

At age 20 months we connected with a new neurologist at UW children's hospital in Madison, WI. She had her first EEG. We came in to have an outpatient 30min EEG performed. We were escorted directly to be admitted into the hospital instead of going home. Her EEG was very abnormal. We continued on for a 19.5 hour prolonged EEG. Although she never had a seizure during this time, it was still very abnormal. It was suggested to me that we consider a treatment plan for her, for seizures. I was too concerned about the possible side effects of the medication to begin the treatment. So we did not.

At age 2 years and 9 months I went back to that neurologist and we repeated the EEG with a 1 hour study. I was hoping that maybe she had "grown out" of the abnormal activity. She had not, and the EEG results were comparable to the first. Still I was nervous to start the medication.

At age 3 years 8 months we repeated the EEG with a 1 hour study. We saw a different neurologist that was recommended to me by a close friend, and was closer to family so that when we traveled for appointments my other 2 children had a place to stay. Sylvia never had a seizure during that study. However that neurologist was extremely concerned about the activity he saw in this short time. We came back in 2 months later for an overnight 24hr EEG. This time he hand calculated the times that she showed abnormal activity. In a 24 hour period 70% of the time she was having abnormal activity. It was then explained to us, that for a person or a child to retain what is learned, with abnormal activity that high it was nearly impossible. The "magic" number would be below 40%. If we could find a medication that she responded to get her to that level, it may be possible that she would learn and retain what she was learning. 1 week later we started her on Keppra. Our goal was to get her to 7.5 ml but we took it slow over the course of 6 months and only increased her dosage by .5ml at a time. With each increase we would see behavioral challenges escalate for a few days then taper off. No other side effects however.

5 months later we had her first follow up EEG after starting Keppra. Her abnormal activity was below 30% of a 24 hour period. Shortly after that we noticed that her memory was better. She would put a toy away at night, and in the morning remember exactly where she put it. She could sit and concentrate longer. When she developed a new sound, it turned into a word that we did not only hear once, she would bring it back over and over until eventually it was a full word that

she used and remembered. At age 5 years 10mo she had her last EEG. This was a 2 night/3day study. We used a portable machine, so she could move around after hook up. Again fantastic results. Now moving forward she will only have a neurology apt. annually w/o an EEG, and we will be adjusting her Keppra as her weight increases.

About 9 months ago, we had an experience that allowed us to truly test how the keppra is working. I noticed that Sylvia's eyes were drifting frequently, like they did when she was young (3see below). She became completely silent, and unaware like she just was not with us. It went on for 4 days and I was devastated. I was going to call her neurologist to discuss and my husband mentioned to me that when he picked up her keppra prescription the week before they told him they had changed the manufacturer of the medication. He did not think anything of it. I called our neurologist and he informed me that generic companies are allowed to dilute their medications up to 30%. He has Sylvia on such a tight dosage that the new manufacturer had become ineffective for her. We immediately called the pharmacy; they ordered in the old manufactured and within 24hours of switching back, we got her back. I always wondered if maybe she was just hitting new milestones, and the med had nothing to do with us. This proved to us how much the keppra was and is helping her.

#### **Other findings etc.:**

At age 18mo she had her first set of tubes put in her ears after failing a hearing test. She has had a total of 4 different sets in, to help prevent sinus infections. It has been over a year since her last so we are hopeful that after this set we will not need to replace. She also had her adenoids out at age 4 ½ and this has seemed to help with cutting down on colds etc.

At age 3 ½ I did an intensive 5 week hyperbaric oxygen therapy treatment with her. Twice a day, for 5 weeks. We did see that she seemed to become more alert, and had an easier time coordinating movements. By the end of the 5 weeks she said bye with a hand wave at the same time. However, after we completed treatments, that faded.

At age 4 years 10 mo. she attended a craniofacial clinic at UW children's hospital. It was learned that she has a high arched palate; with so little muscle development that it would be thought that she was just born with tissue, no muscle. However the muscle is physically there, giving us hope that she may develop speech. She also was diagnosed with severe verbal apraxia during her speech assessment there.

At age 4 she was seen by a developing pediatrician at the Waisman Center in Madison, WI. She was concerned about Sylvia's restless legs during her sleep; it was something that I brought up. She ordered bloodwork including her Ferritin level. She shared with me that there is an association with low ferritin levels and restless leg syndrome in children. We received the results and although her iron levels were ok, her ferritin level was extremely low. We increased her iron, and it made a difference in how much she moved around in her sleep.

#### **Eyes**

Sylvia has intermediate exotropia in both eyes. When she was young, her eyes wandered everywhere throughout the day. It was worse when she was tired. We patched but it did not change things. At time throughout the day, they would drift like she was daydreaming and we

would snap our fingers etc. to try to snap her out of it. When she started on Keppra for her abnormal EEG we saw a SIGNIFICANT change in her eyes. They no longer wandered and within a couple of weeks of being on the med, she seemed to gain control over the muscles in her eyes. Her ophthalmologist and neurologist felt that the abnormal activity was constantly fatiguing her during the day, causing her to lose muscle control in her eyes. Once the abnormal activity decreased, she was not as fatigued and could hold more control.

### **Dental**

Her teeth were very late at coming in. They are all in the correct place but the 4 front teeth are abnormally large. Last year we had x-rays done for the first time and it shows that she only has a total of 8 adult teeth that have formed, and they are placed to come in in very unusual areas. It is expected that she will only get these teeth, and we are anticipated a lot of dental work to reconstruct her teeth as she is older. She has always been very defensive in brushing her teeth, yet is able to get through a dental cleaning every 6 months. Just in the last 2 weeks she has shown interest and is animated that she brushes her own teeth, and she brushes them forever!

### **Orthopedic**

She has muscle weakness in her ankles. When she was younger, right when she started walking we used SMO's on her. However she seemed to struggle more with walking having them on. So instead we used orthotic toddler tennis shoes, and had molded orthotic inserts made for her. We still have custom inserts molded, so that it helps give extra support in the areas needed, and helps hold the ankles straighter. Last summer she did break her ankle on a simple bouncing on a trampoline.

### **Sleep**

As mentioned above Sylvia wasn't very good at sleeping! As an infant she would only settle at night for about an hour, and then she would wake. This went on all night long for about the first 1 ½ years of life. This time was very hard on our family, as I was exhausted all the time. After starting her on Keppra she slept through the night within 24 hrs. of starting the med. Now she either sleeps through the night or wakes once. If she wakes once, regardless of what time it is she thinks it is time to be up. She usually comes into my room with her iPad in her hand, because that is the first thing she gets to do in the morning. Regardless of it is 1am or 5am, I remind her it is night time, set down the iPad and go back to bed with her in her room. Then she falls asleep again until 6:30 or 7:00am. Also after increasing her iron, her sleep became less restless. Routine is the key for her. We keep her bedtime early even now, 7:30pm and hardly ever waiver from that. If we do, we see her behavior become more challenging the next day. She also still takes a nap 3-4 days a week. We found a sound machine a must for her, it calms her and blocks out the household noise.

### **Eating**

I nursed Sylvia until 1 year of age. Her intake was very little, however she did not seem to have a difficult time nursing. She started solids at the age of 5 months. As a toddler her appetite was very minimal. We used a special tomato highchair because it had foot rests that were adjustable and helped her keep grounded so she didn't move around as much. When she was younger she

was very distractible, so at times we would have to feed her just to be sure she actually ate. She was capable of eating independently, but because it took her so long I think she lost interest. She stayed in the high chair until age 3, and then we transitioned her into a special tomato booster seat. It helped her stay positioned correctly, and had a restraint on it to keep her seated. At age 5 her appetite increased, and she truly began eating age appropriately amounts. We now have her sit legs crossed in the chair, as she is less wiggly. At times we still put a homemade seat belt around her to the chair, so she will sit through a meal. Her weight is currently at the 35<sup>th</sup> percentile. And height is at 50 percentile.

### **Play**

Sylvia has always been extremely social. She loves to play with kids and adults of all ages. She loves princesses and anything pink. She tends to play with toys that a child a year or two younger than her would play with. In the last 6 months she has begun to use her little dolls and figures to play out scenes. For example she will hold them by each other and say “hi” then “play” then pretend they are playing together. Her biggest challenge with play is that she does not like to do it independently. If we are at home she struggles keeping herself occupied. Even if I set her up with play-do etc. she will whine and pull on you relentlessly until someone is at her side. We utilize respite care in our home when I am at work, and sometimes even when I am at home, just so I can get something done. She is very active and plays on the playground like her classmates. She can throw a ball now, but still cannot catch. She learned to ride a trike at age 5, and we just modified her big girl bike with fat tire training wheels, pedals with straps and a supported back on her seat and are anxious for her to be able to move into that.

### **Academically/cognitively**

Sylvia is delayed globally. Looking back at some of her assessments an example would be that at age 22 months, on the Rossetti scale she ranked 0-3 months for language expression, the same for comprehension, and 6-9 months for play. At age 2 years she ranked 15-18 months for personal/social skills, 9-12 months for cognitive, 6-9 months for fine motor. Her last evaluations were at almost age 4 for speech. She ranked equivalent to a 2 year old for auditory comprehension and a 14month old for expressive communication. We have seen Sylvia have the most gains in the past year and are scheduled for another evaluation in a few months. We are using the eval for insurance purposes (to help gain more therapy).

### **School**

Sylvia entered into Birth-3 at 16 months of age. This is a program in Wisconsin that brings therapists etc. into your home. At age 3 she transitioned into early childhood at our elementary/middle school. She was in that classroom for 2 years with 3-4 other special needs children. Children from the pre-school room were brought in daily for center time to work with her. At age 5 she attended the regular pre-school class with an individual aide, and did the same for kindergarten. She will do the same, and be mainstreamed for 1<sup>st</sup> grade with an individual aide. This year she has shown an interest in numbers and letters. She can recognize 70% of the alphabet, her colors and her numbers 1-10. She can write her name, and trace other letters. However, she is still considered significantly delayed. Since she entered school she has 3 (60min) speech sessions a week, 2 (60min) OT, and 2 (60min) PT. We have not been successful on getting private therapy covered through our insurance beyond what she gets in school.

## **Potty training**

She is just starting to show a little awareness when she goes, but no interest in sitting on the potty, and when she does we have not been successful yet on having her go.

## **Speech/communication**

We started to use sign language with her around 15 months. A lot of hand over hand and made signing times videos part of our daily routine. When she started early childhood education at school her preferred choice of communication was signing. She knew over 100 signs but then slowly she stopped using them, as she started to make sounds. Now she will still sign and say a word or sound together. Her signs are modified so she can do them and not the exact ASL signs. Before we started Keppra she would make some sounds. Occasionally we thought we would hear a word, but then never hear it again. That changed and she began slowly forming words. The first time she would say a word it would be extremely drawn out. Bow for example was like saying boooooooooowwwwww, for several seconds. After she said it for over a week, it would shorten until eventually just bow. Now words come easier and sometimes she says something out of nowhere and it is completely clear. Last night she said tilla for tortilla and I had never heard her say it. She thought it was funny and said it over and over. Other words come out and you know what she is trying to say but it is all mixed up. There is no combination of words yet, just single words. She has been using Prologue2go for 2 years now on an iPad. She needs prompting to remind her to find the picture of what she is telling us. She tends to use it more in school than at home, but our house is a crazy busy home and we don't always take the time to direct her to her "talker" and she is good at getting her point across of what is needed. I will be honest we have been tough on her with speech. I have worked with speech therapists that would take her favorite things and put them in clear plastic containers, out of reach and make her make a sound before they would give them to her. For example her stuffed animal mini mouse. We would allow her to see it (it makes her furious when anyone other than her has it) and we would hold it above her head so she could not get it, in a clear plastic container...and she would scream for it, we would help her make the sound m and didn't give it to her until she did, which could take a whole session. But once she made that sound, it stayed. I recall a session where her speech therapist brought in a real bunny and wouldn't let Sylvia touch it, but had it within reach, until she made the sound B. Sylvia is very stubborn, so it has taken us doing things like this to teach her she can make those sounds.

At her current age of 6.5 years, I can safely say that currently Sylvia has about 100 plus words that she says complete. She can say the full word, and the general public would understand her. Most other words she says the beginning sound. When you prompt her she can say the end, but she can not co-ordinate the beginning, middle and ending together. That is the severe apraxia diagnosis. Even with that said...many days I look at her and it is still very hard to imagine that I will someday have a full verbal conversation with her, or that she will co-ordinate 5 words together to make a complete sentence. But only time will tell.

I think it is true to say that teaching your child to communicate (verbal, signing or using a device) will have to become part of your day, actually part of almost every moment you are with your child. But it will just become norm. Your child will walk into the room, and when you look at them you will know exactly what they want, what they need. But you can't just give it to them-you have to make them work for it somehow if you want to see your child make progress.

## **Behavior**

For the most part Sylvia is enjoyable to be around. At times she shows aggression towards mom & brothers but seldom around anyone else. We did come across something that seems to affect her however. Red dye. She would randomly have bouts of hitting, biting, yelling. I have always been a label reader, and cautious of what she eats. At home we took out all artificial colors in her diet, especially red dye. I began asking for ingredient lists when we are out an about. But she would still sometimes have these bouts. Even though we requested no artificial colors at school, the children bring in the snacks/treats and it is hard to monitor. Last year Sylvia had a sinus infection and was treated with an antibiotic. When I picked it up it was pink. I was frustrated (really why does red dye have to be in that?!) and must have not wanted to deal with it, so I used it. The next four days were horrendous. We were called from school to get her because her behavior was so aggressive. We took turns with her at home, keeping her in a room away from her siblings because she was out of control with them. We stopped the antibiotic, switched to a dye free option. It took over 48hours for her to return to norm. Her teachers at school, and our family felt that it was not a coincidence, and she is that sensitive to the colors.

At her current age of 6 years 10 months...

We have learned a lot. For parents that are just learning of their child's diagnosis, and for others that have known. My biggest words of advice...get an EEG, just to be sure there is no abnormal activity going on. And secondly, I remember when Sylvia was first diagnosed, I sat on the ground playing with her and I would just watch her, my mind going everywhere wondering what she would do in life, what she would be capable of doing. I think I lost months of my life worrying, and wondering about those things. Time slipped away on me, and I wasn't there for the rest of my family. I recognized that I needed to start living in the moment and not worrying about tomorrow. I began to enjoy Sylvia for who she was. She has changed our life, and changed the lives of many people who know her and work with her."

*Sylvia's mom*