“Sweet Claire was born in late 2013. She was the perfect addition to our little family, a sweet baby girl that was desperately wanted and loved long before we knew she was on the way. Claire was 6 pounds 13 ounces at birth, and had a textbook pregnancy and delivery.

Claire had some feeding issues from the beginning. She gagged easily, so she never accepted bottles or pacifiers. We tried every trick we could think of to get her to take an occasional bottle, but she flatly refused. When we introduced solids at 6 months, we really struggled. Claire gagged and choked almost every time she swallowed. We were convinced she was just a "mama's girl" that loved to nurse, so we didn't feel overly alarmed.

Claire met her milestones typically until about 6 months of age. She smiled constantly and always made great eye contact. She began crawling and standing with assistance at 7 months old, but then just seemed to stall out. Claire was always pretty quiet. She would coo and laugh, but never really babbled as other babies her age did. We thought she was simply quiet and introspective. She would "take it in" and seemed to enjoy the world around her instead of needing to be the center of our attention.

With the combination of the feeding issues with poor weight gain and delayed milestones we finally began to bring some concerns to our pediatrician at about 8 months. Thankfully we have a very smart and committed pediatrician who took our concerns seriously while reassuring us of Claire's overall good health and well-being. This marked the beginning of the long and difficult process of getting answers. We saw the GI doctor who ordered some labs and a swallow study and began occupational therapy with ECI to address Claire's gagging. We also started Claire on a drug called cyproheptadine to stimulate her appetite. We saw amazing results with this medication and Claire's eating began to improve. We were very excited that she began to gain weight again, and was finally up to 16 pounds at a year of age.

Claire was still not walking at 14 months, so we began physical therapy through Russell's hospital. Claire was able to pull up to standing but would fall very easily. She was completely unable to protect herself when she would fall and always seemed to land face first. Claire was able to bear her weight by locking out her joints, but had low tone and minimal functional strength. We also noticed that Claire was not meeting her verbal milestones. We began to wonder about her hearing, so she was evaluated by audiology and ENT. Claire failed her hearing tests and was found to need tubes and an adenoidectomy. At the time of Claire's procedure, her ENT noted that she had a very high arched palate but no cleft palate was noted. Claire said "mama" for the first time on Easter 2015 which we all saw as a precious sign that we were finally moving in the right direction.

Interestingly enough, with Claire's increased appetite came a new and troubling problem. Claire vomited all of the time and woke up almost every night around 3 am screaming. We tried tweaking the amount and timing of her medication, thinking she was waking up hungry. We also noticed that she was having numerous large, white-colored stools and her abdomen was huge. From 12-18 months of age, Claire hovered at 16-17 pounds. Our pediatrician sent us back to our GI doctor in June of 2015. He ordered a new set of labs and stool studies.

We saw a series of specialists as we searched for an explanation of Claire's global delays. All of our physicians took me seriously and worked toward finding answers for Claire. We saw a geneticist who was an amazing advocate for me. We did some baseline genetic tests which came back virtually normal, so she recommended a whole exome study "just to be sure." She felt confident we wouldn't find anything but we decided to pursue this test for the peace of mind a normal result would bring. We also saw a neurologist and had an MRI of Claire's brain performed. I agonized over the MRI because they had to put her to sleep and intubate her to do it. It was so much for such a tiny little person to go through, and I still felt like I could possibly be making too much of Claire's delays. The MRI resulted showing "delayed myelination" which the neurologist explained could possibly be our answer. We rejoiced over this result because we knew that myelination continues until around age 3, so there was still a great chance Claire would catch up. We were finding out answers that gave us hope!! Answered prayers!

The same week that Claire's MRI resulted, we received the results from Claire's GI lab work. I actually got a call on a Sunday morning from her GI doctor that "it looks like Claire might have CELIAC disease." This answer was as uplifting as it was overwhelming. Claire had a procedure to biopsy her small intestine less than a week later and her celiac diagnosis was confirmed. Removing wheat and gluten from our diet was going to be a huge adjustment, but it was something we could fix. I was thankful beyond belief because my hands finally had something to do. I poured myself into cooking and baking and reorganizing a gluten free kitchen.

Less than 2 weeks after we removed gluten from Claire's diet she began walking. Simply incredible. She hasn't thrown up once since we changed her diet and she began gloriously sleeping through the night again. We were so thankful and content. My prayers had all been answered and I was ready to ride off into the sunset of bliss with my sweet family. I had no idea that Claire's whole exome study was about to result with a diagnosis that would change everything.

A week before Claire's 2nd birthday, we had our follow-up appointment with her geneticist to discuss the results of her Whole Exome Study. We were sure that everything would result normally and as Claire's body healed from her months of poor nutrition she would catch up to her peers. Our genetic diagnosis was a shock to say the least. I wasn't at all prepared for it and it hit us very hard. Claire was diagnosed with a de novo (not inherited from Russell or me) single base pair substitution in exon 4 of her *SATB2* gene. This mutation occurred in a coding region that is highly conserved among many species, so it is in a region very important for brain and craniofacial development and is likely the cause of her global developmental delays and unique features.

What does *SATB2* look like? It is so rare that there are no cases exactly like Claire's! It makes it hard to compare her to others with mutations on the same gene because we don't know how great an effect a single base pair change will make. Other children with *SATB2* associated syndrome, or SAS, have been noted to have intellectual disability with absent or limited speech, behavioral problems, palate and dental abnormalities, and facial dysmorphism. We happen to see Claire as strikingly beautiful, but she does have a very unique smile and dental abnormalities.

Claire's verbal and intellectual abilities remain our greatest challenge. She has been in speech therapy for about a year now and has very limited speech. We love the sound of her voice and tell her so all the time. Claire will go extended periods saying nothing at all, but often is bubbly and babbles. Sometimes she even says amazing things, like "bubbles" or "I love you!" We haven't quite figured out the key to encouraging her to use her voice, but we are constantly trying and my greatest goal is to find a way for her to express her needs and desires. I pray constantly for her to find her voice! She has about 10-15 words that she can approximate, and is getting better every day with sign language. Claire's gross motor is improving steadily and she is truly a little lady on the go.

The most remarkable thing about Claire is her heart. She is love personified. She shines God's light so clearly you can't help but see it if you spend a moment in her presence. Claire's heart is pure gold and she lives in the moment, finding joy at every turn.

We have begun the process of deciding "what is next??" How do we help Claire to learn and grow with her unique strengths and abilities? What will life be like for Claire? We don't yet know the big picture, but we do know we are blessed by this precious child. We are proud that God chose us to be Claire's parents, even when the road ahead is uncertain and scary. Claire has taught us so much already about love and contentment, we can't wait to see where her journey takes us!”

Claire’s Mom