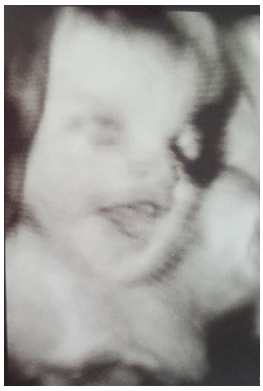
What would you do if you found out your child was born with a genetic disorder that no one else in the world has?



My pregnancy with Mia was as normal as it could possibly get. I had all the genetic tests, sonograms, glucose testing, and we even had a 4D scan done to get an extra sneak peek of our girl and everything was normal. We had no idea that anything was wrong. The day I went into labor was exactly a week before her due date. The contractions came on quickly and we were worried she would be born on I95 north. We got to the hospital and they rushed us to triage and were trying to get a heart monitor on me for her, but the contractions were so strong they very quickly transferred us to labor and delivery. Everything happened super quickly after that. The doctor came in and said that I had a bubble in the amniotic sac and without breaking my water the labor would continue. So obviously we told her to break my water. After that the nurse had me push 3 times and out Mia flew! The doctor barely had her birthing gear on before she and the nurse were catching her. They immediately took her over to the table to weigh her and clean her off. I heard them saying she "came out sunny side up". Basically, come out with her forehead looking up so they were saying that's why her head looked a little swollen. We didn't think anything about it and we're so happy she was finally here!

The rest of the day was filled with family visiting, tons of pictures, and lots of snuggles. By the time that night came, we were all exhausted. Because we have an older son, Stephan went home so they could sleep in their own beds. Mia went to the nursery to get some rest, and I tried to get a little sleep not expecting the next day to flip our worlds upside down.

The next morning about 6 they brought Mia into the room and we were snuggling, working on feeding her, and just enjoying our time. The nurse let me know that the pediatrician on duty would be around about 9 am for Mia's check-up. When the doctor came in to see her, she spent about 5 minutes in there looking at her, said she didn't like the size of her head, and she was concerned she wasn't eating enough so she was sending her to the NICU. I was in complete shock! She is 7.5 pounds!? How is this possible? I called Stephan hysterically not really clear WHY this was happening. They transferred her and told me that I'd be allowed in shortly once they finished checking her over. When we finally got to her room, she was in the NICU bed, hooked up to monitors, had IVs in her limbs, had been pricked and prodded, but was thankfully peacefully asleep.

Over the next couple of days, she had her entire body scanned pretty much from head to foot seemingly LOOKING for a problem. They did not find any, her lungs, kidneys, heart, brain, and liver all looked good. She had a large open soft spot, but really all babies do, so we didn't think much about it. We kept asking why they were keeping her, we figured out how to help her eat better, simply giving her support under her chin and on her cheeks and she seemed strong. She was slightly jaundice but really when you're in a windowless room who WOULDN'T be? We kept arguing with them about getting her out of there and no one said there was anything else to be concerned with. Finally, three days

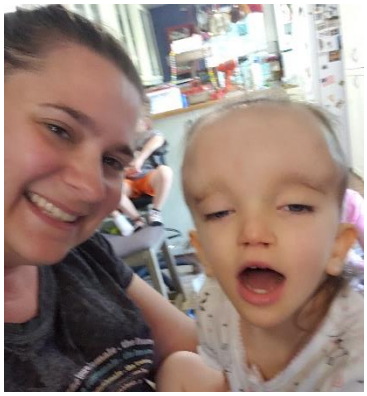
after she was born a genetic counselor came in to talk to us. Why does a genetic counselor want to talk to us?

When they came in, they sat down with me and said that they were concerned that Mia may have some sort of genetic disorder. They said that some of her symptoms made them think something was wrong. I couldn't understand what they meant, what symptoms? She's healthy, happy, she's eating more now, what could be wrong? They said things like her large soft spot, her large forehead, her trouble eating, her spitting up, her high palate, nystagmus, and various other smaller things that we didn't even notice. When we tried to ask what disorder, they thought she had they said they don't like to speculate on these things before knowing but they were thinking it could be something called Zellweger Syndrome. They took her blood and sent it off to the lab to be tested and told us they'd receive the results back in a few days. So, like any other parent who's been told this about their child we went and started researching everything we could about that. Everything we found was super scary and so much of it didn't seem like it was what our girl had.

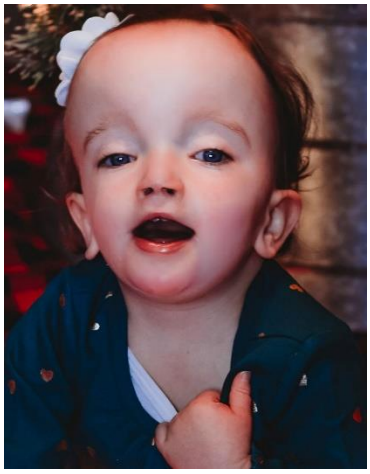
She was finally released from the hospital 5 days after being born and even though we were super nervous about the unknown we were so happy to finally all be home together. Two days after that we received the results we had been waiting for. She did NOT have Zellweger syndrome. Ok, awesome, so what DOES she have? "A Chromosome 14q22.3q23.1 deletion.". Ok, so that doesn't sound too bad, what does that mean? "Well, we aren't entirely sure because she is the only person who we can find with this specific deletion.". After not comprehending what they were telling us it boils down to this; she is missing 29 genes on this specific "arm" of her genetic makeup. Unfortunately, they only know what 18 of those genes do, leaving 11 that we have no idea what will be affected. Neither Stephan nor I have this so they said this is "one of those things that just happen". They have continued to follow Mia, checking in with us every so often but unfortunately do not have any new information for us.

For the first six months of her life, we slept with her in between us in our bed in a wedge that kept her upright because she started throwing up hours after eating and we were concerned she would choke in her sleep. We saw several different gastroenterologists to try and get this under control and with medicine, lots of patience, and lots of changes of clothes (for all of us) she was finally able to not choke. This did not mean she didn't have these issues anymore (she still does to this day but not as severe thankfully) but it did mean the threat of aspirating on it was low. We almost made the decision for her to have a Nissen fundoplication (a major surgery that would have changed her life even more), and thankfully we decided against that.

She's been closely monitored by a neurologist and neurosurgeon and recently had a major skull reconstruction to help begin the process to close her large soft spot and reshape her forehead to give her more protection for her eyes. She was under anesthesia for 6 hours and we had a 5-day hospital stay following. She will have to have at least one more surgery to finish the process in about 6-12 months.



We have been working with Early Intervention (EI) in our county and were thrilled when she finally started crawling at 21 months old. Physically and mentally, she is significantly delayed. She is three years old and does not walk independently, does not talk much more than single small words (mama, dada, ball, Elmo, etc.), still needs to be fed chopped up baby food, and drinks milk only from bottles. We do not know how many other challenges she will have and hope to be able to provide her with all the support and assistance she needs as she grows. Because she is the only person in the world to ever have her specific deletion, we can not do much more than continue to address the problems as they arise and be as prepared as possible for the "unknown". Our occupational therapist with EI sent me the information about a family support group which I absolutely wanted to join ASAP! We had some family struggles going on, so it took me a week or two after receiving the information before I was able to join one of the Zoom calls. I am SO GRATEFUL that she sent me this information and that I was finally able to join in on a Tuesday Momma's and Poppa's Connection call! I look forward to connecting with these other families each week to compare notes, laugh, cry, and just have a few moments to "be with", even virtually, these people who can understand some of what we're going through! It has been an amazing outlet both to receive tips and ideas of things to do for Mia but also an opportunity to help others who may be earlier in their stages of EI. We are in the process of switching out of EI to Child Find due to Mia turning 3, but I am so grateful to still have this connection and realize how invaluable this has become to us!



Recently we found out that she has slight albinism in her eyes which is one reason why she seems sensitive to bright lights, even when she wants to stare at the sun through the windows.

We are blessed that Mia is the sweetest, most laid-back little girl. She has taken on all these challenges and still just loves people, specifically to bounce on anyone who will let her, and loves to laugh and smile. Since her surgery, she has regressed slightly with things that she was starting to do prior, but we are hopeful that she will bounce back to her old self soon!

We know she will do great things and look forward to continuing to watch her blow through milestones!