His & Hers…

Diagnosis through the couple's perspective

As told by moms/ mums around the world

Compiled by Jennifer Jacob
2014

Our Stories

{Samuel}

California, United States

{Cathleen, Samuel's Mom}

Looking back, I think the universe was trying to prepare me for Sam all along. Down syndrome was never something horrifying to me—more a mystery than anything else. And although I didn't understand much about Down syndrome, I had compassion for people with it. There was a guy in my junior high and high school who, in retrospect, I think probably had DS—perhaps mosaic, since he didn't show *all* the signs, and perhaps with a dose of something else thrown in, as he had some anger issues in addition to his obvious cognitive delay. He was in the special-ed class, but we would cross paths on campus, and he was often lurking near where I was. He had a crush on me, I think, and I'm pretty sure it's because I was one of the few people who wasn't nasty to him. He scared me a little, because of his temper, so I can't say I befriended him, but I also wasn't mean to him. If he spoke to me, I replied to him. And I guess that was enough, because he spent the better part of six years hanging out in my general vicinity whenever our paths crossed.

Years later, I was talking to a friend about children with Down syndrome, as my friend was interested in Reece's Rainbow. And we talked about how, as odd as it sounds, we sort of felt like the universe (or God, if that's your belief system) should give *us* babies with Down syndrome instead of giving them to people who didn't want them, because we knew we would love any baby with all our hearts—disability didn't matter to us.

That's not to say I *wanted* to have a special-needs child—like anyone, I hoped for a "healthy" baby. It's just that I knew if I had a child with special needs, I was capable of loving that child as fiercely as I would love any child, and I was up to the specific challenges that go along with having a special-needs child.

My husband, Chris, and I had our first son, Theo, when I was 34. Theo turned out to be a bit of a handful—he was very colicky and eventually was diagnosed with high-functioning autism, among other things. But in many ways, he's a very typical little boy, and we love him dearly and decided to try for a second child. And when I was 37, we conceived our second baby.

It was a slightly harder pregnancy than my first—just a few more aches and pains, and it felt a little different. I loved feeling Theo move inside of me, but the first time I felt Sam move, I felt a little sick, and my first thought was, "Ugh, it's like I'm growing an alien in there!" I was aghast at the thought and immediately reminded myself that this was my *baby*, who I had wanted so desperately! I brushed aside the nagging feeling of something being slightly "off," and I soon grew to love the feeling of him moving inside of me, just as I had with Theo.

When I was seven months pregnant, I was at Target one day and picked up one of their "Target Picks" recommended books. It was *Expecting Adam*—a memoir about a woman who finds out her unborn son has Down syndrome, and she decides to continue her pregnancy despite a tremendous lack of support from her Harvard University colleagues and community. I mentioned to Chris, "I probably shouldn't read a book like this while pregnant with my own son, but what the heck—it looks good, and it's not as if our baby has Down syndrome."

And then, when I was 37 weeks pregnant, I took Theo to the library in our new town to try to get a library card. I left without a library card due to a paperwork mixup, but I wasn't sorry because I was absolutely furious. As we waited in line to get the card, the man in front of us was trying to settle an issue of overdue books. He was maybe in his thirties, and clearly he had Down

syndrome. He was polite, but it was taking him a while to try to figure out the overdue books and fine. The librarian, a woman in her sixties, was unforgivably rude to him. She acted impatient as he tried to settle his fine, and she rolled her eyes at me as if to say, "Can't he just *leave* already?" She was plainly disgusted with him, and I was appalled. This man, whoever he was, was obviously *trying* to be independent and handle his own library books, and she acted as if he wasn't worth the time of day. I was furious…and still with no idea that my baby, who would be born just days later, was affected by the same chromosomal anomaly as this man.

On our way out of the library, I picked up a community-activity guide for Theo, but when we got home, I realized I had picked up a special-needs booklet. I laughed at my mistake when I showed my husband later and said, "I didn't even know they *had* a booklet for special-needs activities. Guess we don't need this!"

Two days later, I almost missed my 37-week OB appointment. Theo had, unbeknownst to me, turned on the dome light in my car the day before, and effectively killed the battery. My pregnancy had been mostly trouble-free, so I thought about just skipping the appointment. But when AAA arrived and jumpstarted my car in time for me to get to the appointment after all, I went.

Our medical group provides only two ultrasounds per pregnancy—one at around 8 weeks and one at 20 weeks. But for some reason, at this appointment that I nearly skipped, the doctor cheerfully announced that she felt like doing a quick ultrasound, just to ensure that the baby was head down. "We know he is, but let's just check!" she said. And as it turns out, he wasn't—he was breech. And what's more, my amniotic fluid was low.

After a whirlwind couple of hours where the doctors debated performing a C-section right then, they decided I had enough amniotic fluid to continue the pregnancy, and they sent me on my way with strict instructions to return in two days for a fluid check and non-stress test (NST).

So Friday morning I was back, dutifully getting my fluid check and NST. The fluid looked okay, and the NST was fine. The ultrasound tech called in a doctor just to be certain, and he spent a very long time staring at my ultrasound and making notes and calculations. Endless minutes ticked by. He finally cleared his throat and said, "Interesting."

"What?" I asked.

"Was your first child small?" he asked.

I laughed. "Hardly! He was born at 38 weeks, and he was already 8 pounds 3 ounces!"

The doctor looked at me and said, "Interesting. This baby is measuring very small. I estimate five pounds."

But he said nothing more. And I thought nothing more about it, even when I Googled "low amniotic fluid" and saw that "chromosomal abnormality" was one cause. In fact, when Chris asked me what might cause low amniotic fluid, I laughed and said, "Chromosomal abnormality, apparently! But we don't have that, so who knows." I had declined all prenatal testing, since abortion would've never been a consideration for us anyway, but I just assumed that because my 20-week ultrasound had showed nothing amiss, our baby was developing typically.

Later that very night my water broke, so I went in for my C-section 16 days earlier than planned—at 37 weeks and 2 days gestation. The doctors on staff wanted to wait to perform my C-section until morning—both so they could get some rest and because I had eaten dinner and still had food in my stomach. But the baby had other plans. After two hours of labor (and still four hours before the planned C-section), I was hit with a *monster* contraction that wouldn't end. I was near tears with the pain, and then suddenly alarms started going off, and people swarmed into the room.

"What's happening?" I asked, terrified. Alarms everywhere. People everywhere, yelling at me to roll this way and that, turn over here and spread my legs there. An oxygen mask slapped on my face. I looked at the fetal monitor and saw that the baby's beautiful heart rate of 150-ish had plummeted to 34. Yes, 34. And it wasn't coming back up. Someone turned the monitor away from me as I began to sob, "Get him out! You need to get him out safely!"

The next thing I knew, we were racing down the hallway. "Code C in Labor and Delivery!" was being announced over the PA system. "Code C!"

"Is he okay? Will he be okay?" I kept asking. No one would answer. Finally, someone said, "That's what we need to find out. We're going to need to put you under full sedation for an emergency C-section." I wept.

But when they slid me from my gurney onto the operating table, a little miracle happened. The fetal monitor started picking up—that beautiful heartbeat was back! I wept in relief. They were able to do a standard C-section at that point, and within about 20 minutes, I heard the lusty cry of my newborn son and the doctor proclaim, "He's a bald one!" And I wept as I heard "APGAR of 9." He was safe. He was healthy.

And then the doctor attending the baby came over and bluntly announced, "I need to tell you this because you're scheduled to have your tubes tied. I'm concerned about some issues of tone with the baby."

"What? You mean jaundice?" I asked, thinking she meant *skin* tone.

"No," she replied bluntly. "Muscle tone. Your baby shows the signs of Down syndrome. So I need to know whether you still want your tubes tied."

Shock. Anger. This woman was acting as if my baby was somehow "broken," and I'd want to keep my tubes intact to try again for another "non-broken" baby. What did she mean: "Whoops,

this one's no good! Want to try again for another?" My protective instinct came out in full force, and I said icily, "Yes, please go ahead and tie my tubes. This is the baby I was meant to have."

Tears leaked out of my eyes as I lay there, my insides spread open for the rest of my procedure. They brought me the baby, and my heart soared and ached all at once. He was alive, and he was beautiful and precious and mine! But this thoughtless woman had just treated him as if he were less than perfect, and I knew in my heart that she would be only the first of many people to treat him that way. I knew the world could be a cruel and unaccepting place, and I had just brought a beautiful new life into a world that would likely be hurtful to him. I loved him with all my heart…but I couldn't help but wonder, was I selfish to have ignored prenatal testing and brought him into a cruel world? Would I have been a better mother *not* to bring him into the world? I felt confused and overwhelmed—brimming with love but battered by confusion.

And how *dare* this doctor mar the moments after my son's birth that way? I realize she was doing her job, but I still ache to this day when I think about it. She didn't even take a moment to congratulate us or to tell us he was beautiful. It was all about his disability, immediately. My beautiful, sweet baby was secondary to an ugly term called "Down syndrome." Was this how everyone would see him—as a disability or condition first, and a baby second? The thought overwhelmed me. I saw him as a beautiful shining light, and I wanted everyone else to see him that way, too.

Waiting for the baby to come back from being bathed and checked out was the longest 90 minutes of my life. I lay there in the recovery room, alone and with tears leaking down my face, Googling everything I could about Down syndrome on my phone. Not my best idea—the Internet can be a scary place if you're looking for information on Down syndrome! But when Chris (who I had sent with the baby) wheeled him into the recovery room 90 minutes later, I forgot all my fears. He placed the baby in my arms, and I held my sweet little sunshine for the

first time. He snuggled right down into me contentedly, and I felt peace along with my confusion. My son might have an extra chromosome, but oh, how I loved him already!

We named him then—we hadn't chosen a name before he was born. He is Samuel, which means "God listened." Because God *did* listen. I'm not a religious person, but I threw a prayer up to God when Sam's heartbeat was dangerously low, and He listened. And 18 months later, that name is even more appropriate, because God really, *really* listened. He gave us a baby who changed our lives for the better. He gave us the baby we never knew we wanted, but who we absolutely cherish and could not live without. Confusion and shock about his diagnosis are long gone, replaced with the brightest love I've ever felt.

People sometimes accuse us in the Down syndrome community of painting the condition with rainbows and unicorns—making it seem like something wonderful and special, and ignoring the concerns that go along with it. Truthfully, I don't think *any* parent of a child with Down syndrome ignores the concerns that go along with it. We're all well aware that yes, there are medical issues we may have to face—heart problems, digestive issues, higher incidence of leukemia, and so on. Believe me, we worry every time our child gets a funny rash or unusual blood-work results. And yes, we worry about people treating our child unfairly, due to his perceived differences. And yes, we wonder what effect our child's cognitive delays will have on his life long-term. But *every* parent worries—that is not unique to the special-needs community. Perhaps we just worry about different things.

And the truth is that along with the worries come joy and beauty. Sam has taught us so much already. He has taught us how to celebrate the tiny joys and triumphs—the ones that we never even stopped to consider before having a child with Down syndrome. *He took a bite of solid food—rejoice! He finally pulled to standing at 17 months—celebrate! He picked up a cup of water and purposely dumped it on the floor, then set the cup back down—what awesome fine-*

motor skills! He has taught us how to slow down and simply go with the flow, rather than focusing on the next big milestone or goal. *He'll walk someday—when it happens, it happens*. He has taught us that there is no one-size-fits-all approach to life and that the best-laid plans are meant to be torn apart and scattered to the wind sometimes. He has taught us to love unabashedly and freely, without reserve—who can resist a toddler that gives any random stranger a full-body smile, complete with million-dollar grin, waving arms, and kicking feet? Sam greets everyone as if he's just met his biggest hero—it's completely awesome. And having Sam has introduced to us a whole world of fascinating, diverse, and wonderful people—both those with Down syndrome and those who love them.

If a future pregnancy were still an option for us, would I do prenatal testing this time around? Yes, I would—only to be prepared in case the baby had a serious medical condition at birth. We dodged a bullet with Sam—he was physically healthy from birth, but that isn't always the case in babies with Down syndrome. But do I wish I'd had prenatal testing when I was carrying Sam? Not for a second. I am *so* glad we chose to skip it, because if I had known I was carrying a baby with Down syndrome, I would've spent my pregnancy terrified of the unknown. In the abstract, I knew I could love and care for a child with Down syndrome, but when it comes down to real life, I would've been scared out of my mind as I waited for him to be born. Instead of enjoying my final pregnancy, I would've been a nervous wreck, because I didn't *really* know what Down syndrome was like. Now I do—and I know there's nothing to fear about Down syndrome itself. There's concern about the related medical issues, but most of those can be dealt with—even heart surgery is somewhat routine now. But when you talk about Down syndrome itself, I now know that it's not scary; rather, it can be beautiful in ways you never imagined, and your life can be so very enriched because of it.

Sam is now 18 months old and the light of all of our lives. Chris has commented on numerous occasions that Sam has made him a better father to *both* of our sons. I am happier and more

content than I have ever been in my life, and Sam is a big part of that. And Theo—well, Theo has just blossomed as a big brother. My impatient, volatile, brilliant little five-year-old slows down and becomes a tender, compassionate, patient teacher for his little brother. He delights in Sam, and Sam is the first person he wants to see every morning and the first person he wants to share things with.

And Sam himself? He's a happy, silly, sweet, and, yes, temperamental little boy. He is a determined little monkey who aspires to steal anything he can find and throw it into the toilet. He loves music and will dance like crazy if Theo puts on the Spice Girls. He's fascinated by our dogs and a big devotee of *Baby Signing Time*. Oh, and he hates therapy. He will attempt to pull every clever ploy in the book to get out of physical and occupational therapy, whether that means flirting with his therapist to try to distract her with hugs, crawling away as fast as he can, or sticking out his lower lip and throwing a full-blown tantrum. He's a master at avoiding what he doesn't want to do. In other words, he's a very typical toddler!

Thank you, Samuel Ames, for opening our eyes…and continuing to open them every day.

{Chris, Samuel's Dad}

It's difficult to revisit the moment my son was diagnosed with Down syndrome. Or, at least, the moment the doctors told us they were concerned about some "tonal issues." You see, the moments before that one were joyous and full of relief, as we'd witnessed the birth of our son after a harrowing delivery. The moments when most parents start to wonder about their child's future, choose his name, or figure out whose eyes he has. Typical moments.

Birth…

My wife had always told me that if she was going to have kids, she wanted to do it before she turned 40 because the risk of DS was so much higher after that age. When we conceived our second baby, she was 37; it looked like we were in the clear. We even opted out of genetic testing because the odds of DS were low and the miscarriage risk was comparatively high. (She was showing signs of early menopause before the pregnancy, so we didn't want to risk losing what might be our last chance to have a baby.) It's not to say we went into it blindly—we discussed the possibility of DS, and we were clear that we'd love our child no matter what. Not that we *expected* anything to happen, mind you.

That night, we drove to the hospital around midnight, after my wife's water broke and contractions started. The baby was breech, so we were looking at a C-section. After about an hour of waiting in the prep room, though, Cathleen had a really long, odd contraction, and the

baby's heart rate plummeted. The doctors and nurses intervened quickly, whisking her away to the operating room. No one knew what was going on, only that they were both in danger. All I could do was stand out in the hallway and pray for their safety. Soon enough, everything stabilized. A nurse took me to my wife's bedside, and a few minutes later we listened as the doctor announced we had a healthy son.

The nurses carried our son into view, and then over to various machines to weigh and measure him. He looked just like the 3D ultrasound image we'd had taken a couple months earlier—much like his brother, in fact—and had this quivering little cry. As my wife was getting stitched up, I went over whispered to him that it was OK and held his hand. He settled down when he heard me, and I breathed a huge sigh of relief, knowing my son was safe and everything was indeed OK.

Diagnosis…

I went back to check on my wife. The doctors were preparing to do her tubal ligation, which we'd arranged well in advance because we considered our family complete now. But one of the doctors suddenly interrupted and said she had concerns about some "tonal issues," so we might want to reconsider. We weren't quite sure what she meant until she clarified that he had "markers for Down syndrome." Our life changed in that instant, and we had to start making decisions immediately. We simultaneously responded that we wanted to go ahead with the ligation, rejecting the doctor's implication that we might want to keep our options open because this baby was damaged, and telling those in the room that DS didn't matter—that our son was a welcome member of our family, perfect as he was.

But that was on the outside. On the inside? I hurt—terribly. I tried to put on a good face, but it was an empty gesture. I just hurt. And I was afraid, of what I knew about DS and what I didn't know. The bottom line was that for five minutes, our son *was* typical to us. I don't mourn what he

would have been—I mourn what he actually *was*, in my mind. That child was real—I talked to him and held his hand—and he was pulled away as soon as I met him. And so much of me still hurts for that loss. But when I remember we almost lost him, it hurts a little less.

That night, we named our son Samuel because it means "God listened." He did that night. He has all along.

Now…

Sam is 17 months old now and recently appeared in his first commercial for Gymboree. Yes, a commercial. We certainly thought he was cute, but how many babies have a modeling contract and work permit before they can crawl? The other day, he pulled to standing on my wife's knees, cackling as he fell back on his bottom and babbling "ma-ma-ma-ma-ma" in my wife's direction. Sam is a true joy, and he's made me a better husband, father, and person in so many ways. And for him, that's typical.

{Ella}

South Australia, Australia

{Mandy, Ella's mum}

I thought I was ready for it. That's the main thing I remember about the day I received the news that would change my life. I had said to my husband Leighton, about an hour before that I was fairly sure that our newborn did have Down syndrome, and felt like I was ready to hear that. But as it turned out, I wasn't. When the midwife said, "We have the preliminary results back, and I'm sorry, it's positive for Down syndrome", I felt like the whole world stopped. I looked down at the floor, and said something like "OK, thank you", and as soon as she left, I cried like I have never cried before, standing over the plastic hospital bassinet, clinging to Leighton, and feeling more broken than I've ever felt in my life.

I recalled sitting at my pre-natal appointment a few days after my 20-week when the midwife read from the ultrasound report "A tiny echogenic focus is noted in the left ventricle, which in the absence of any other abnormality is of doubtful clinical significance". The Doctor explained that it could be a soft marker for Down syndrome, but they saw it quite often in ultrasounds and almost always meant nothing. My 12-week NT scan had been fine, as had been my bloodwork at that time. It did concern me a bit though, and at my next appointment, four weeks later, I asked the Doctor whether I should be having further tests. I felt like if my baby had Down syndrome, I could cope with that, but I would prefer to know now. He said that it was extremely unlikely that it meant anything, and wasn't worth having further tests. I felt comfortable with this, but never quite forgot about it through the remainder of my pregnancy, and even on the day I was in labour, decided to take a really good like at this baby once she arrived, to make sure there was nothing I could see which might indicate Down syndrome. Even though it was in the back of my mind for those months, I never really believed that it might happen. After all, I, and

my family, lived inside that golden circle, protected by the fact that those sorts of things happened to "other people".

The day of Ella's birth, the day before her due date, arrived. After a relatively easy labour and delivery, this beautiful baby girl was placed on my chest at 8:58pm. Yes, she was beautiful, but she didn't quite look the way I had expected. I remember staring at her with amazement, joy and love, as I had stared at my other children upon their arrivals, but there was another feeling too, an uneasy, something's-not-quite-right-here type feeling.

Once the Doctor arrived (about half an hour after Ella), he checked her over and I mentioned to him about the 20-week ultrasound report, and asked whether he saw anything which might indicate Down syndrome. He told me that there were a few things he was concerned about, but nothing worth worrying about tonight, and that we'd talk more tomorrow. I told Leighton about this conversation when he came back, but he felt that this baby was perfect, and that I was worrying about nothing.

The next morning, the Doctor came back to check Ella out, and when the midwife brought her back, she said that he did think she might have Down syndrome, and that they needed to take some blood samples and send away to find out for sure. They said we would have the results in the morning. We were very calm then, just nodding and saying "Yes, OK, no worries". Not long after, Leighton's parents came in and as soon as we told them, I started to cry, as though saying it out loud made it real.

The following morning was probably the longest morning of my life. We waited and waited and waited some more. Eventually I went to the nurses station to ask whether the Doctor was there, and we were told that he'd been and gone. One of the midwives came to our room shortly after, and delivered the news that we'd been dreading.

So here we were, in a hospital room, and trying to comprehend that news that life as we knew it would never be the same. I felt like all the plans I'd made, all the dreams I'd had for this baby were gone, and had been replaced by some scary and awful future. I was very scared, sad, angry and overall, felt ripped off. I'd had a plan for how this was all supposed to go, and this was not part of my plan. I heard a newborn in the next room crying and wondered why hadn't that baby been born with Down syndrome. Why my baby, why me?

Gradually, in the days, weeks and months that followed, we began to heal. The most important factor in my healing was the beautiful girl in my arms. I could certainly see that in some ways, a pre-natal diagnosis would have meant that I could have processed a lot of the grief and anger and pain before Ella's arrival, but I feel fortunate that in my darkest moments, I could look at her, hold her and know that everything was going to be all right.

{Leighton, Ella's Dad}

Late in the winter evening of June 18th, 2009, in a small country hospital in regional South Australia, my family was to change in a way that we never anticipated nor planned for. On the arrival of a large, gurgling and healthy baby girl, a mother's intuition requested verification that her baby was OK. The response from the doctor was measured, yet we could sense the trepidation in his voice, "Just enjoy your beautiful daughter tonight and I will see you all tomorrow morning". Both my wife Mandy and I thought that she did look a little different. Not much was said on that night, but Mandy and I reminded each other, that her scan at week twenty through the pregnancy, revealed a minor shadow on her heart. At the time we questioned the doctor, who indicated that they see this many times, and we should not be concerned; this opinion was supported by a 1 in 700 chance of our daughter having Down syndrome.

Throughout the pregnancy we had decided that our daughter would be named Lily, but this baby did not look like a Lily and somehow, Lily just did not feel right, so we settled on Ella. As Mandy and Ella settled into the night, I returned home, very concerned for my new daughter and the unforseen changes that a potential diagnosis would have on our family.

Arriving home I jumped straight onto the computer and googled, images of new born down syndrome, and an image of the baby I had just left appeared on my computer screen. With little sleep I returned to the hospital the following morning after a harrowing night to find my daughter in Mandy's arms.

Mandy and I discussed our thoughts about our baby having Down syndrome, prior to family and friend visiting to hospital. In our minds we believed that our daughter may have Down syndrome.

She was not the daughter we expected, yet we loved her from the minute she was born. The daughter that we expected was no longer, the baby we had was not Lily; tears were shed as we mourned the loss of our daughter. Yet we had this beautiful baby, Ella, requiring our immediate attention and love.

As our other children and family came to visit the hospital on the Friday morning, Mandy and I showed little emotion and kept quiet on Ella's possible diagnosis. We could feel the whispers in the corridors of the hospital grow as we waited for the return of our doctor. Midwives and nurses would visit throughout that morning, as we would question them whether Ella had Down syndrome and when we would expect to see the doctor. We learnt that to confirm Ella had Down syndrome, that blood test would have to be transported to Adelaide's Women's & Children's Children Hospital 3 hours away and tested. Our family had started to suspect that something may be wrong with Ella. My mother and father, who had driven from Adelaide to see Ella that morning took the blood samples to Adelaide immediately for testing, as these results would provide Mandy and I with the necessary confirmation over the weekend. I don't think we could have waited over the weekend and into the next week without knowing.

It was not until the afternoon on the Saturday that we were informed that preliminary results had indicated that Ella had Down syndrome. And it felt that our world had crashed around us.

As Ella's father, I felt that now was the time to step up and inform all family members and friends, as Mandy was struggling with coming to terms with not only losing the daughter that she expected, but now having a baby with Down syndrome. People reacted in different ways, with statements like "special kids go to special homes" and "kids with Down syndrome are great, they are always happy". Although well intentioned, these perceptions and statements failed to alleviate our loss of the daughter we were expecting. Mandy and I did not feel special in any

way; we were normal people living our normal life, and now I question the word normal and it's meaning.

Once we had our diagnosis, immediate my thoughts raced to, will my daughter have friends, will she go to school, will she play sport, will she walk, will she talk, will she be teased, will she get married and on and on and on. Only fathers in my situation can understand what I thought and how I felt.

Prior to leaving the hospital, we were provided with the contact details of a family who lived 45 minutes from us, who had a 3 year old daughter with Down syndrome. I spoke with this family and asked if she would visit us with her daughter when we got out of hospital. The day after Mandy & Ella came home from hospital, Sam and her daughter Dakota came and visited us. Dakota was bright, responsive, she walked, talked, had selected her clothes for the day and Mandy and I could see a positive future immediately - it was what we exactly needed at the time.

By the time Ella was six weeks old, we had joined a support group two hours away in Victoria and were attending a fortnightly early intervention program at the South Australian Down syndrome association in Adelaide. The people we have met through these groups have now become our closest of friends as we all now belong to a new family.

Ella is truly a inspiration to me, our family and friends, I witness the positive effect that she has on others and the better person she has made me become. I am under no illusions that her having Down syndrome will result in issues presenting themselves at times, but we are becoming increasing more capable to handling these issues.

Down syndrome does not define Ella. More importantly she is a four year old girl that is funny, moody, affectionate, canny and manipulative, much the same as her three sisters and brother.

Ella had just turned 4 and has loads of personality, a fiery temper and a sweet, loving side. She goes to child care three days a week, and will start Kindy very soon. We have been lucky in regards to Ella's health; while she's had a few extended hospital stays with respiratory infections, it's nothing like what some of her little mates have dealt with.

Ella continues to surprise us with what she's capable of, and she continues to challenge to myths & stereotypes about kids with Ds. She has taught me many lessons in the past four years, and I think we have many more to come!

{Gianluca}

Illinois, United States

{Jenny, Gianluca 's Mom}

When I found out I was pregnant with my second baby, I was ecstatic and surprised! We had been trying for a year, and recently decided not to focus on getting pregnant since we were just so busy. But there I was, pregnant!

I was 36 years old, which they called advanced maternal age, and I joked that I was an elderly pregnant lady. We knew we wanted prenatal testing; I was a planner and wanted to know everything! I had to go to great lengths to get the quad screening approved through my insurance company, and eventually we had the results that we had a reduced risk for Down syndrome (1:538).

Three days after the testing, a series of complications began. At 13 weeks I almost lost my baby due to a 6 cm tear in my placenta and spent 6 weeks on bedrest. At 22 weeks my baby was diagnosed with an atrial septal aneurism. At 36 weeks, I failed a biophysical and was hospitalized for observation.

At 39 weeks, my water broke and I was so relieved. It was time to have this baby and put the stress and worry behind us. Well, as much as you can do that as a parent, but it seemed the big stuff would be behind us.

After a quick and easy labor, my son was born and I was ready to feel relief. But he wasn't crying so of course I was worried. They assured me that he was fine, his APGAR scores were

great, and there was nothing to worry about. When they handed me my son I was instantly in love and so relieved that he was here. They took him away for the usual pediatrician evaluation, and we sent out messages to friends and family that our perfect son had arrived. I could finally exhale. My parents arrived with our daughter and we were so happy to see them all as we'd been looking forward to Luca meeting his sister. Our family would be complete.

Shortly after the nurse returned Luca to us, the doctor came in and abruptly asked everyone to leave the room. He sat down on the bed and told us that Luca exhibited soft markers for Trisomy 21, Down syndrome. I was in shock. He showed us the palmar crease on his hand and said it could be a sign, but could also just run in families. We immediately studied our own palms and willed our creases to merge. He showed us the sandal gap and we ripped off our socks and swore we had a big gap between our toes as well. He explained that the shape of his eyes and his lower tone were also soft markers, but he really could not make the diagnosis. He said that in some cases, he could tell the parents with confidence in this conversation that their baby had Down syndrome, but he didn't have that confidence in this case and would need the results of a blood test (FISH test) to be sure. The results in 3 days, our first day at home, so they would call us sometime that day. I immediately panicked about how we would keep family away until we heard the news because I was so emotional and unsure and scared, and just didn't want anyone to know until we were certain. I snapped out of it when he told me not to worry, that we would love our baby no matter what. Anger surged up in me at him for saying this to me. I never doubted that I would love my baby. It just seemed like such a cavalier statement to make at the time. I think he was trying to reassure us, but it stood out as rude.

Our heads were spinning. At some point in the conversation my mom forced her way into the room. Later she told me she suspected Luca had Ds from the way he felt when she held him. She's a retired nurse, so I knew she was right, even though I didn't want to believe it. She knew

that when the doctor kicked them out and looked so serious that something was up. I was glad she came in because I was in such shock that I didn't know what to say or do.

My mind flashed back two years prior, when we were at a birthday party and there was a little boy there with Down syndrome. As my friend shared that his diagnosis came at birth, I remember thinking- that would be the worst. To be told at birth that your child has Down syndrome. And here I was, sitting in this hospital room, facing a Down syndrome diagnosis. I wanted to know why this was happening to us. How could this be our story? At some point, in the quiet of the night, as I held my son in my arms, I knew why this was happening to us- we were the right parents for him. We could do so much for him and others with Down syndrome. Suddenly this fight roared up within me and I knew I just wanted to be his proud mother.

When my daughter came into the room, she climbed right up into bed with Luca and me, demanded to hold him, and gave him the biggest hug. My heart melted. Holding back the tears, I knew in that moment I had so much to learn from my kids. She showed me what true love looked like and I held them both so tight at that moment.

We weren't able to go home as planned because Luca wasn't maintaining his body temperature. As we waited to find out the results, my husband and I spent all our time with Luca. We would stare at him, pray over him, snuggle him extra long and just let our hearts connect with his. I wavered between believing he had Ds, and believing he didn't; exploring both sets of emotions. We bonded with him in such a special way those early days which was very important.

While we were sitting by him in the SCN one night, an overnight nurse started up a conversation with us. When we shared that the FISH results were not yet in, she blurted that her friend was a genetic counselor and it was unbelievable the things they find in those tests; all sorts of chromosomal anomalies that you wouldn't believe; stuff that kills babies. Our hearts sank and

we were scared. She was behind a computer and couldn't see the terrified looks on our faces. It was in that moment I realized I didn't care if he had Down syndrome. I just didn't want to lose him!

It was a Friday morning, I had just gotten out of the shower and was blow drying my hair when our pediatrician came in. She had a serious look on her face so I stopped with half a head of wet hair, and sat down on the couch next to my husband. She then told us she had the results of the blood test, and Luca tested positive for Trisomy 21, Down syndrome. That was it. She asked if we had any questions. Just as she finished talking, my mom called and I started crying as I spoke. She told me everything would be ok and that we can handle this. I'll never forget that confidence and immediate support.

As the news spread about Luca's diagnosis, we were overwhelmed by the love and support that came our way. Our families, close friends, coworkers, friends we hadn't spoken to in years, and friends who were already part of the special needs community all reached out to share their love. I was welcomed into a Facebook group of moms who have kids with Down syndrome born in 2012 or 2013. It was an incredible comfort to be able to connect with moms with children around Luca's age, who were in the same stages of processing as I was. I learned quickly that this was going to be a place I could share some of my deepest, darkest, and brightest thoughts without judgment. That group was, and is, a lifeline to me.

In the beginning, I was fixated on the potential medical issues children with Ds may develop. Since the birth diagnosis was such a surprise after a rocky pregnancy, I had this creepy feeling of constantly waiting for the other shoe to drop. I read about everything from reflux to leukemia. There was so much to learn and be aware of and I did not want to miss a single cue.

It took me a solid three months to process Luca's Ds diagnosis. In those months, life felt like a rollercoaster. Not only were we grappling with what Ds would mean for Luca and our family, but

we were trying to adjust to life with two children, in the midst of sleep deprivation. That was a tough few months!

For the next 2 months, we sorted out some early medical issues. Everything was very benign- Luca had reflux, laryngomalacia (floppy airway) and plagiocephaly which earned him a cool "space" helmet. I learned quickly what being my son's advocate meant as I had diagnosed each of these ailments and pushed my concerns forward to the doctors. I felt good that I was doing right by my son.

For the next 3 months, we enjoyed our son!!! Yes, it was finally at 5 months of age that we were starting to hit our stride! We had been working with a wonderful Early Intervention team for 2 months, and we were starting to think about his 6 month goals. That was when some worry started to creep back in.

Luca had been developing very close to the typical milestone timeline. Our goal had been to try to keep him as close to the typical timeframe as possible. At 6 months he was sitting up, rolling over from front-to-back and back-to-front, working on supporting himself on all fours, babbling, smiling and laughing all the time. But it seemed like he was starting to plateau. He started putting his hands in his mouth quite a bit, which prompted us all to suspect he was teething. I thought this seemed a little odd as I had read that kids with Ds may teethe late. And my daughter did not break her first tooth until she was 9 months old. I figured he was starting to feel those teeth early, and this would be a long teething process We started to have to move therapy session times around to try to catch him when he was more awake and not as bothered by his teeth.

Teething aside, we were about to go on vacation with some friends, and looking forward to introducing them to Luca. We were excited to get away as a family to do something "normal" and enjoy the beach together! Luca was so unusually cranky, I wanted to be certain he wasn't

developing an ear infection, and took him in for one last check-up. The pediatrician cleared him and said there was no reason not to go on vacation!

Three days into the vacation, our world changed once again. Only we didn't know then quite how significantly. Luca started demonstrating an odd movement. He would throw his arms up and roll his eyes back. It was all very quick, like a startle. My stomach was in knots. I had watched videos about Infantile Spasms in those first few months because I wanted to know what to look for if Luca developed them. By the end of the trip, Luca was experiencing clusters of spasms. I was on the phone with his pediatrician while we were at the airport returning home begging for an appointment for the next day. Our flight home was diverted to the airport in Milwaukee, which was where Luca's specialists were, so I got off the plane with a medical emergency and brought him to the Emergency Room.

My mom and aunt met me in the ER and we worried deeply about what we were going to learn. The team there was very nice and explained that we had nothing to worry about. I showed them a video of the movements and Luca also did a few himself in the moment, and they assured us that these movements were not indicative of anything harmful for Luca. They sent us home with direction to have our pediatrician order an MRI and EEG for Luca. They couldn't do it themselves due to some insurance restrictions. I felt much better that we were given such a positive discharge.

The next day, the pit in my stomach returned as the frequency of the clusters increased. I fought hard to get in to see a pediatrician the next day, and finally, at 5pm when she saw the video and Luca demonstrate a spasm in the office, she was on the phone with the Children's hospital getting us admitted for a potential Infantile Spasms diagnosis.

On June 27, 2013, at nearly 9 months of age, Luca was diagnosed with Infantile Spasms (IS), a catastrophic seizure disorder. Hallmark symptoms of IS are developmental delays, regression,

and mental retardation. Within a week of the diagnosis, we saw Luca lose nearly all of his milestones. We saw our son go from a smiling, laughing baby to a silent toddler who couldn't hold his head up. Loosing Luca's laughs and smiles hurt us the most.

Typically, children with Ds respond quickly to IS treatment, and are less likely to relapse. We faced his diagnosis with tenacity and convinced ourselves this would be a blip on our medical radar. Luca, however, took his own path once again, as he broke through 4 medications and developed 2 other seizure types. He spent time in the hospital every month for the next 6 months, was ambulanced twice, went through testing for metabolic disorders and mitochondrial disorders, and developed pneumonia which earned him a feeding tube. We longed for the days when he just had DS.

A few weeks after his first birthday, Luca smiled again. We had initiated the ketogenic diet and adjusted his medications and were cautiously optimistic that he was gaining seizure control. October 24, 2013 was the last time we saw Luca experience a seizure.

I have learned through his first year of life that the Ds diagnosis is a slippery slope. I spent 3 months processing, which I certainly needed, but now wish I had spent just truly enjoying my baby. I lived my life waiting for the other shoe to drop, rather than enjoying the moment. I realized that the developmental delays and cognitive impairment associated with Ds are just not a big deal to me.

When I worry about what the future will hold for Luca and our family, I realize it isn't a Ds diagnosis or an IS diagnosis that send me spinning. It's being a parent who is head over heels in love with her son and family and only wanting the best for them. I'm thankful to have my son, no matter what path his chromosomes send us down.

With all of the worry, fear, concern, sadness, and stress that has come with Luca's medical issues, I have to share that there has been incredible joy, love, support, selflessness, generosity

and kindness brought into our lives. Our families will drop anything to help us out, our friends started a campaign to raise funds for Luca's care, my Facebook moms have sent cards and gifts with touching sentiments only they can share, a former colleague and her friends ran a marathon in his honor, and I've heard from old friends who found themselves down this special needs path offering me their support and shoulder to lean on if I need it.

At the end of the day, I focus less on the superficial and more on the real now. I've learned to truly take life one moment at a time. I'm curious to see what the future holds, but I stay in the now. And I never underestimate the healing powers of a pumpkin beer and hot shower.

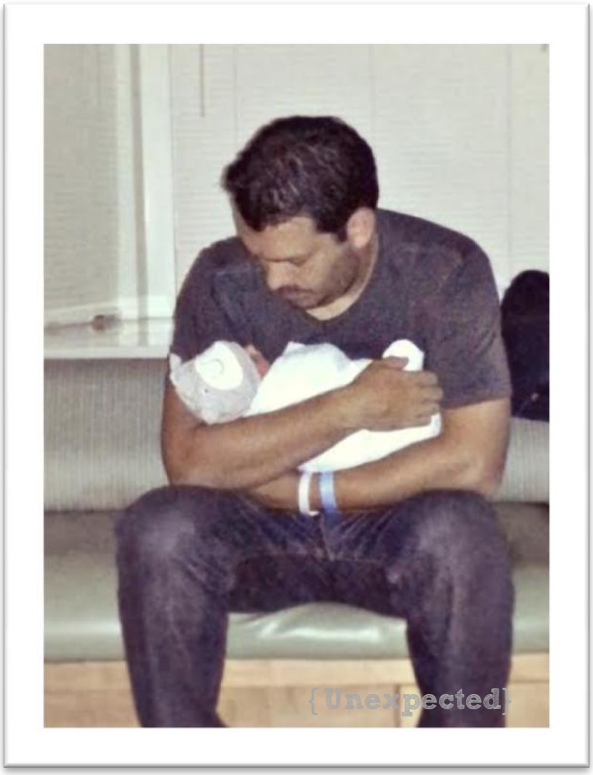
{Nick, Gianluca's Dad}

"How are you doing with all of this?"

That's the signal that a conversation about Trisomy 21 is starting, and it means I better change my mood and get ready for a down and dirty honest conversation about our son, Gianluca.

My wife, Jenny, usually utters the above sentence while waving her hand around like Obi-Wan Kenobi trying to Jedi-mind-trick me into switching topics in our conversation.

It usually works, but not without some resistance on my part. I am certainly not some mindless storm trooper, but eventually the façade has to crack because that's how a marriage works, and it's especially true when dealing with a child like Gianluca.



Before that, though, Jenny has to force her way through my canned responses of "I'm OK. You?" I choose to deflect the question and give a standard response that harkens back to my days as a PR guy. Jenny usually continues to push it, and I give her a real answer because she's my wife and deserves the truth, and one of the reasons I married her is because she WILL push me and not settle on something that sounds like it came from a press release.

Trust me, I have a mental database of safe answers that would sound much more fitting coming out of a coach at a press conference or a politician trying to tip-toe around a potential controversy.

Then there's the reality, and that's exactly what Jenny wants. As my wife, she deserves it … and so does Gianluca.

So, how am I doing? It depends on the day; some are good and some aren't. However, Gianluca's smile and gut-busting laugh makes it all fantastic, helmet and all.

When I think back to what it was like learning that our second – and most likely last – child was born with Trisomy 21, I just remember how misinformed we were. We had no idea what we were getting into, and no life experience with it. We found out after birth, and it hit us hard.

We knew there would be challenges, and we knew we would be up to face them together. One thing about us; we're strong and even stronger together.

Still, it was a rollercoaster of emotions, but only if the coaster went crashing to the ground as expectations and dreams were met with uncertainty and confusion.

We knew nothing. We just knew he would be different, and the emotions I felt were some of the rawest since experiencing the death of my Dad due to cancer in June 2000. You don't forget that feeling, which was brought back several years ago after Uncle Tommy's passing.

This time, we were starting not ending, and there were a lot of questions.

We have loving family and friends who showed tremendous support from around the world. It was truly overwhelming to get calls, emails, texts and whatever other correspondences sent to us. We knew we weren't alone in this. That's something we cherish, and will never forget, and it helped drive us.

Throughout our hospital stay, I was frequently in touch with my cousin Mary Jo, who is a doctor at Mount Sinai in New York. She would go on explaining to me what Down Syndrome was, and

how Gianluca was going to be great and we were going to do everything we could to make sure of that.

Mary Jo's defiance to my ignorance included a telephone call with my sister Marcella and brother Giuseppe. Marcella is a middle school Italian teacher, and she was telling me about her experience with students who had Trisomy 21.

"They can learn a foreign language?!?!?" Yeah, I was that clueless.

After Mary Jo and Marcella set me straight, the switch was flipped.

Jenny and I would have long, difficult conversations throughout our hospital stay. There were tears, hugs, kisses, anger, sadness and finally smiles.

For all the raw emotion, I was able to draw the line; "We're still his parents. We have to do everything we can to help him."

You want to know how I'm doing? That's it right there. Every day that goes by, I just look at him as my amazing son. One day, I started to wonder what life would be like if he didn't have Down Syndrome. I stared at him, he looked at me, smiled and burst out with his laugh. I just said to him, "You wouldn't be Gianluca, and I WANT Gianluca."

He's our son and I can't imagine life without him, extra chromosome and all. Just like I can't imagine being without Jenny and our daughter Gabriella, whose unique personality keeps us on our toes and brightens up the room.

I would do anything for all of them, and I do.

Trisomy 21 may be something we have to overcome in some respects, but we'll do it together as a family, and that includes our extended family and friends. It may have surprised us, as life threw us a curveball, but we'll persevere and he'll be even better for it.

We can sit around blaming things; whatever deity you prefer, the doctors, each other, the randomness of the extra chromosome. You can waste your time and energy focusing on things that don't matter because you want answers or to place blame. That's wasted energy that should be focused on him, and the family. I don't have time for self-pity, and I don't have time to worry about what brought us here. We're here, so look forward to the challenges that we need to overcome.

That determination drives me. However, I don't want to think about the future when nothing is written. When I do, I make sure to pull myself back to reality and stay focused on the little guy.

Fantasy is for baseball and football, not raising kids.

We can influence this and help him achieve his potential, and we do that on a daily basis because he's our son, and we love him very much. I wouldn't want it any other way,

I'm doing great, and there's no need for tricks.