



An Added Twist...
Unique Stories of Diagnosis

As told by moms/ mums around the world

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In addition to not knowing about the 47th chromosome their child would have, these families encountered even further surprises as the adventure with Down syndrome began in their families.

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{A delayed diagnosis}

When I think back on those few weeks, they seem blurry already, but what I do remember are flashes. Flashes of feeling, thought, memory, perception. There was something like a persistent tapping that only I could hear. With each of those flashes, there was another tap. *Tap... Tap... Tap...* each time it was like a marble added to a scale, getting heavier and heavier, to show me something I didn't want to see.

It started with an unplanned third pregnancy. Maybe we had successfully spun the roulette wheel twice, but that was it. I worried about miscarriage. I uncharacteristically worried about car accidents, Listeria, everything. I never told anyone how unsettled I felt. How could I? It made no sense. The quiet *tap, tap, tap* had begun.



During labor, I couldn't quite place the difference, but it was there. I expected the pain, but it was as if being in labor was a one woman show, not an interplay between my body and the little life inside. When the time came to push, I felt like I was tearing myself into pieces. If I didn't keep pushing so hard, so intensely, I feared my labor would stop altogether and my baby would fade away. Later, I would learn that my baby's lower muscle tone most likely contributed to my feeling the need to push harder.

After he was born, the midwives and I peered into his little face pondering his resemblance to the family. I felt a strange pang of discomfort. Now I think that was the first time I saw a flicker of his features. There was something in his eyes and forehead that made me uncomfortable, and I spent hours staring at it. *Tap... Tap... Tap...*

He didn't gain as quickly as his sisters had. He was incredibly sleepy. It seemed that if I didn't wake him to feed him, he would just fade away. His jaundice lasted longer. Despite all this, he was all in the range of normal. But nothing felt right.

Then, I saw it.

I saw the faces of children I had seen at the grocery store, walking along the street, on pamphlets at the doctor's office. He had the face of a baby with Down syndrome.

That night I stared at pictures of hundreds of babies with Down syndrome. I learned that Down syndrome sometimes came with a set of physical markers. In the pitch black of night, I used the light from my phone to stare at his hands, his ears, trying to figure out if he had the markers for Down Syndrome. What the hell did "low set ears" mean? All babies have flattened nose bridges. If I smooshed his hand one way, it looked like he had the crease. Smoosh it another way, and the lines on his hand looked like mine.

But his face. I saw it.

The next morning, I said something to my husband. I watched closely. *Tap...* He made sure to immediately minimize it, but when pressed, he admitted, he saw it too.

I couldn't let it go. I started to read online forums, and blogs of parents who had children with Down syndrome. Each thing I read gave me a knot in my stomach. *Tap...* Even though he had no discernible major health issue, the picture they created in their descriptions somehow sounded exactly like my son. *Tap...*

I called our midwife. I felt the panic of someone standing in an open prairie, watching a storm on the horizon, with no place to hide.

Our midwife came. She said she was unsure. I held my baby and cried. And cried. That night, I went back and read my son's birth story. This was the first paragraph:

*I think the significance of the fact that you were born in caul
shouldn't be lost when I write about your birth. In fact, I
wonder if you're destined to float in life with a different
attitude and perspective than those around you. We will
have to see.*

I found the tapping noise. All those marbles gathered their weight and tipped the scale; I saw what had been there the entire time. It was my baby, telling me that he had Down

syndrome. Before we even went in to get a test done, I knew it was true.

With hindsight, I understand why no one saw it. He doesn't have many of the markers. No extra skin on the back of his neck. His ears aren't particularly low set. He doesn't have palmar creases on his hand. His muscle tone is actually quite good, but uneven.

I'm writing this now with those days behind me. I cried so much that my cheeks got chapped. The fear of potential pain gripped me. I could not bear a future of awkward moments, small and large slights, disappointments. Briefly, I regretted our choice to forgo prenatal testing. I thought about adoption. Then the guilt over being upset at all was overwhelming. Could he somehow sense my grief and feel unloved?



What I realized was that my grief came as a reaction to a world hostile to difference. As I scrutinized that grief, it gave way to an entirely new way of thinking. Why does our society value independence so much? Why is a person's worth measured by achievement? Why are some abilities valued over others? I had lived enough to know that independence, achievement, and ability did not equal happiness.

I can't choose all of the events in my life, but I can choose my experience of those events. I choose happiness. The choice is actually not very difficult. I have three beautiful children. An amazing husband. Wonderful family and friends. I have a kid with an extra chromosome. I know, as a mother, I must choose happiness for him now, so that he can one day choose it for himself.

~ Jisun, mom; 31; California, United States

Blogging @ <http://kimchilatkes.com/>

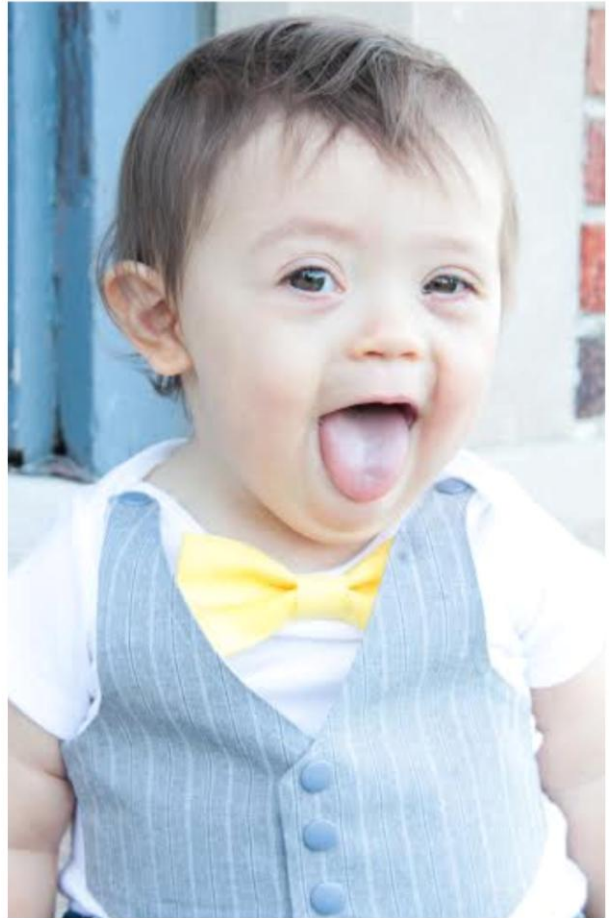
{A Mosaic Diagnosis}

I have always been a very optimistic, glass half full kinda gal. I had been very healthy thus far in life and have always taken it for granted. Our first child, Makenna Grace was born in January of 2009. Motherhood was all that I had ever dreamed it would be. That was until a summer day in June when we found out Makenna had a very serious cancer like disease called Histiocytosis.

After 2 years of treatment and a clean bill of health my husband, Kyle and I were finally ready to extend our family of three.

14 weeks into my easy and somewhat mundane pregnancy I got the terrible news that my baby had anacephaly and did not survive. Kyle and I were

absolutely devastated. How could this happen after all we had gone through with our girl, Makenna.



After months of mourning what was to be...we were again ready to try for our baby.

This time around I was considered high risk and monitored very closely with extra

testing and ultrasounds all the way up to 34 weeks. My baby boy was perfect. All of our tests and ultrasounds came back clear and we were confident we were awaiting the arrival of a healthy baby boy.

Then in the middle of the night unexpectedly at 38 weeks my water broke and before I knew it Kyle and I were dropping Makenna off at my in-laws and we were off to the hospital to have our baby boy.

After an easy labor and just a couple pushes my baby boy was here!! I was overcome with joy and relief. My boy was here. He was beautiful and healthy and MINE. After a couple hours of family visiting and celebrating Kyle, Colin(my beautiful baby boy) and I had a quiet night together.

The next morning I sent Kyle to work thinking "I've got this we are good, I've done this before and my boy is healthy".

After Kyle left for work Colin and I snuggled together and it was one of the happiest hours of my life. Just me and my baby boy together, all was right in the world.

About an hour or so after Kyle left for work the on call pediatrician came in and in one sentence changed our lives forever.

I was all alone and he came in making small talk and with no warning at all as if we

were chatting about the weather or last night's Cubs game he said, "Some of Colin's characteristics lead me to believe he probably has Down Syndrome".

Immediately my world was changed. Did he not know he scored a 9 on his Apgar...twice???

How could this be? Didn't he know all that my family had already been through? The rest of what he began to say was a fog. After a couple minutes I cut him off and said, "I'm sorry I don't mean to be rude but I can't do this right now. I need to call my mom". He said he understood and left the room.

The door closed and I was left alone with my boy. I had no idea what to do. I couldn't call Kyle. He had been through too much already and was out making business calls. It was as if my world had been turned completely upside down.

I ended up calling my mom who was on her way down with Makenna bringing me lunch. I broke the news to her through hysterical crying and she said she would be down just as soon as she could drop Makenna off with another family member. I sent Kyle a text asking him to come back to the hospital as soon as he could. I assured him we were ok but couldn't bring myself to call him in fear I would lose it on the phone. My father in law had just been at the hospital and was a medical sales representative and was going to make some calls around the hospital when he had left our room so my next call was to him. He was the first one to make it back to the hospital and I was so thankful he had

come back so quickly. Shortly after my mom and Kyle returned and all three of them were dumbfounded. They didn't understand and were as caught off guard as I had been.

A while later the on call pediatrician came back and explained his concern once again for Kyle, my mom and my father in law. This time there was a great deal of uncertainty in the doctor's voice. With every characteristic he explained to us that he had suspicions about he would then back pedal and comment on how unpronounced they were. One of Colin's hands had a Palmer crease but the other did not. His nose was kind of flat but not really....bla bla bla. I was so angry! Had this man just put us through hell and now was he changing his mind? Towards the end of the conversation he made the comment "Well if he doesn't have Down syndrome....he will always look kinda funny". I was beyond done with this man who had a thing or two to learn about compassion and bedside manner.

The rest of our hospital stay was filled with tears, great sadness and guilt. I was sad for my child. Sad he might not have the full life I had wanted for him. Guilt that I wasn't happy during this time that was supposed to be full of joy. It was as if a black cloud had hovered over us and kept us from rejoicing and celebrating this wonderful gift we had just received.

The next few days, as we waited for Colin's blood tests to come back from Mayo Clinic to tell us for sure he had Down Syndrome, were so very difficult. I felt like I had been

robbed. I wasn't able to enjoy my new baby boy because I was mourning the baby I had planned on. I was grieving the loss of the baby I had dreamt and day dreamed about, the healthy "normal" baby boy.

When the test did come back assuring what I already knew in my heart of hearts...Colin indeed had Mosaic Down Syndrome. I spent the entirety of my maternity leave transitioning into our new reality. I spent late nights researching and learning all I could about this "thing" (DS) that was now part of our new life.

Colin has been very healthy thus far and we are so thankful for this. Colin has a bicuspid aortic valve which is a very common heart defect that we monitor but so far has not been an issue. We did recently find out that he will need hearing aids and since he is very verbal we were surprised by this.

Colin goes to occupational and physical therapy weekly and he is doing really well. At first I was overwhelmed by the appointments that seemed to consume the majority of our summer but then I remember that the reason for all of these appointments is to help Colin be the best that he can be! We have been so lucky meeting all the wonderful people we have in the last few months. We could not have asked for more wonderful doctors and therapists. It is comforting to feel you have a village of people working for you to help your child thrive.

I know that our journey has only begun but I am confident our story will only get better

from here. Although it has not been an easy first year I wouldn't change Colin for anything. He is the happiest, most loving baby and has added so much joy into our family. He is adored by his big sister, Makenna and virtually anyone else who meets him. I am thankful to have such a great support system and know that although Colin having Down Syndrome is not the plan I had dreamed and hoped for I thank God



everyday for blessing our family with my beautiful boy. I look into his beautiful Brushfield spotted brown eyes and am instantly reminded he indeed was "fearfully and wonderfully made".

~ Jessica , Colin's mom; 31; Iowa, United States

{A Missed Diagnosis}

A three hour car ride to Long Island, NY for my six months Well Woman Exam. Twice a year for me, sometimes three but today would be different. A new house, a new state, freshly engaged to be married but back to NY for the well planned, inevitable conversation with my long time Gynecologist. A baby, children or even the thought of having one, was not really in our future. My husband and I just did not have that drive or passion to be parents this day, or even our near future. Don't get me wrong, I like children, I really do. I have 3 great nephews, a few friends with cute kids here and there but was I meant to be a



Mom, were my fiancé and I really ever meant to be parents? Not so much. It was never really was much of a thought to us. We use to say, " We like kids but we love them more when they go home at the end of the night". My visit to the Gynecologist went almost just as planned. I sat in the waiting room for the usual hour, patiently and dreadfully waiting for my turn. My stomach was a bit queasy and the wait time seemed to be never ending. I knew today, that this visit was going to have a meaning and it was time for me to finally "get off the fence" per say. I guess I should explain. In medical terms, I had a 7 inch fibroid surrounded by a few more, living in the uterine-lining tissue outside and

growing inside the uterus. It sounds painful I suppose, but more of an unnecessary burden. These obnoxious fibroids, one mainly, has led to many unfortunate health issues since it began to take up residence in my body. Let's just begin with two pulmonary embolisms, seconds away from multiple strokes, a few hospital stays and not to mention ridiculous nonstop pain. Today, I will only have two sentences on my mind. "Just take it" and "Get it out, I'm done!" That was definitely the only words I was anticipating on saying but nature had a different plan on this day. You see, this day was supposed to be the day to end my 3 hour drives to the Gyno. This day was the day where we get the exact dimensions and location of that annoying monster of a fibroid. This day was supposed to be the day of the "conversation" where I explain to my doctor that I was ready to have them remove anything and everything, as long as it just took my pain away. Again, like I said, not having children, well we were OK with "it". My spouse and I would be "alright" because children were just not in our future. We were happy enough after 10 years, just finally being together, in the same state, a new home, with our dogs and engaged! But again, nature did not have that same plan for us on this day. "Jennifer", my name was finally called. "Right this way. Gown opening to the front and have a seat up there, the doctor will be in shortly". "Up there", the dreadful table but thank goodness! Finally, it's time. Wait, hold one second.. I'm bleeding! How humiliating! It definitely was way too early for that "time of the month", so why am I bleeding I thought. Just great! A 3 hour drive for nothing! The doctor finally came in and I immediately apologized. I explained to her that "my friend" came a bit early this month and we would have to reschedule the exam. Instead we went to her office and spoke about my next step and began to pick out dates and potential surgeons to do my long

waited surgery. We spoke about having a portion of my uterus removed and we spoke about not having children. My next step was to just have another dreadful internal ultrasound, which I would have done today but instead I will have to wait two weeks, post menstruation. We decided that we can basically do the next few steps in my own state, PA, which was great for having to save me time commuting. Over the next few weeks, I scheduled and had my internal ultrasound. The doctor confirmed the fibroid was at its largest and most painful peak. We chose a surgeon and scheduled my time off from work for the procedure and recovery.

February 26th 2012 - A random thought.

That "time of the month" incident that occurred at the doctor's office only lasted one day.

I thought maybe stress, maybe it was the endometriosis, and maybe I'm pregnant.

Pregnant, ha! That would be impossible or would it? Ten years of intimacy and never once a scare. It had been Two years since my last pulmonary embolism, not being on Birth Control pills and not one scare. Pregnant?? Yea right! I remember this day like it was yesterday and the thoughts that went through my mind. How can I be pregnant right? I just had both a pelvic and internal sonogram of my uterus and my ovaries. I just gave another urine sample.. How can anyone not have noticed? Could it have just been missed because no one was looking? Well, 5 pregnancy tests later, the results were there..Black and White, Red line, Pink line, Plus signs, Digital, Yes! I was pregnant! Confirmation followed a few days later by my local Primary Care Physician, my surgery was canceled and my journey began.

March 23th, 2012 - My first visit to an OB.

The visit was pretty much routine. Height, beginning weight, prenatal vitamins, healthy eating tips and then the boot! I say the doctor "gave me the boot" and shipped me off to the Advanced Fetal Maternal Unit because of my age. 37 years old, with a medical rap sheet that is a mile long. Heart Surgery, PE's , Gastroperisis , Anemia, Vertigo, WPW etc.... oh and like they said "too old" .

March 28th 2012 - My first Ultrasound

Today I had my first sonogram followed up by my Sequential Screening, my non-invasive prenatal test blood draw and our first routine visit to the Genetic Counselor. I think back to that time now and I can't help but to wish that I would have maybe paid more attention, asked a few more questions or maybe just took home a few of those pamphlets hanging on the wall. You see, my whole life, childhood until present, I have always been misdiagnosed. It took years for the medical professionals to finally make the correct heart diagnosis, the correct gastrointestinal diagnosis and so on. On this day, 3/28/2012, our Diagnosis was "potential" Trisomy 21. "We just have to conduct some more routine testing to confirm" said the Genetic Counselor but let's go over your odds". Odds? 1 out of 5 chances that the baby that I am carrying , the baby that I did not plan for, the baby that slipped right in to my life, right before having a surgical procedure that would make it impossible to ever carry a child, may have Trisomy 21? Trisomy what? 21? "What is this Trisomy 21 nonsense all about?" That is all that went through my mind. Is that a way to make more money off of my insurance company? Another co-pay? More time off of work to test for this Trisomy21 thing? Was it because I was "too old"? Because I definitely didn't feel old. I play some sports here and there, I work out, I hang out until all hours of the night , I love happy hour after work, I still get "I.D'ed" when

buying a drink, I love going to music concerts. Old? I'm not "too old". This all must be routine right?

"Exactly what is too much fluid in the neck mean? **Nuchal fold scan?** Nuchal translucency screening test results? Chromosomal abnormalities?

So what was next", I asked. 16 Days of silence. 16 days of "What if's", 16 days of an unbearable weight on my shoulder, unspoken actions of confusion, two people that have been so much in love for 10 years, who plan to marry at the end of next month, who lay in bed as strangers, as cowards.

April 16, 2102 - My husband's Birthday and the day we finally received our results from the test.

A new, noninvasive test. A simple blood draw. Two vials of blood , 99.8 percent accuracy , results in 2 weeks you say? Why not?? Of course I will take those odds!

Give me that test!

The results were in. Our test shows no evidence of Trisomy 21, 13 and 18. **NEGATIVE.** If you have taken this test before and you have seen your results, you would know what I mean by the larger, bolded font in the upper right corner. **NEGATIVE!** That is right! Our child does not have Down syndrome! I knew they were wrong! Now let's get back in to wedding mode!! We're getting married in two short weeks!!

September 13, 2012 - The events of the next 48 hours, will forever change our lives .

Twelve Ultrasounds later, over 30 combined OB and Advance Fetal Maternal visits , tons of sono pics hanging on our refrigerator, cool tough name picked out, baby room

almost done and a "Surprise" baby shower scheduled for today and a 50th Birthday party out in Long Island that weekend. Well, once again, nature changed its course. Although my due date was October 6th, our little boy was ready to make his appearance sooner than expected.

39 hours later, After being induced, pushing through low amniotic fluid, fighting his way through fetal distress, 2 epidurals, an emergency C-section and on September 15, 2012 at 1:00 a.m., we welcomed our 5 pound bright blue eyed baby boy, Mason Lucas, in to our world and our journey began.

Mason was brought to nursery for body temperature regulation and I was brought to recovery with some minor medical issues. Mason stayed in the nursery until the next morning, where his temperature was slowly regulated. He was cleaned, changed , fed and swaddled by a few nurses. His Dad got to feed him and hold him while I stayed in bed recovering for the day. Pictures were taken, visitors came and went as did our doctors. Fast forward to what my husband calls the steady loud buzzing noise that occurs in your ears, after a bomb explodes. You know, that whistle noise that doesn't get any louder or less noisy. The one that you hear while you observe other people's mouths moving but you do not know exactly what they are saying. Yes, THAT noise. "We suspect that your child has Down Syndrome", queue in the 4 ft. 11inch doctor , with tons of make-up and cheap perfume. "Your child has some soft markers. He is kind of floppy, his eyes are almond shaped, and ears are a little low. He has what we call a sandal gap and a single crease in the palm of his hand".

"OK, So he has Asian eyes? Well so does my sister, my nephew and most of my mother's side of the family". Oh so he also has a single palm crease". "Look doctor so do I! Have you seen my Dad's ears?" Oh I can show you a picture of my other sister's huge sandal gap toe too, she can send me one, if you just give me a second. "YOUR SON HAS DOWN SYNDROME, we just ordered some tests to confirm but we already know just by his markers"

Exits the pediatric doctor on duty.

Enter the Cardiologist. "Great your here!!" as I greeted him. "Is my son's heart ok?" Throughout the pregnancy I was concerned about Mason's heart. I thought he may have inherited my heart defect but no issues were ever found "Did you do an EKG yet? Does he have Wolfe Parkinson's White like me?"

"Your son's PDA valve has not closed and he has a slight murmur but nothing to be alarmed about. It is very common with BABIES THAT HAVE DOWN SYNDROME." said so nonchalantly by our Cardiologist.

"Our baby does NOT have Down Syndrome . I have all of my medical records with me. Just give me a second I will show you my non-invasive prenatal test results. You know the new test. The one that our genetic counselor said had 99.8 percent accuracy, which is only a small difference from the Amniocentesis. Hold on, I will show you!" "I will show you my last 12 sono pictures too! "

Doctor -"I have been a cardiologist for many years. I have seen many babies like your little boy. YOUR SON HAS DOWN SYNDROME".

Exits the Cardiologist.

24 hours later Mason was moved to the NICU. While they continued to say he had Down Syndrome, he began presenting signs of Breathing Apnea, slow heartbeat, low platelet count, temperature issues and jaundice.

Mason remained in the NICU for a week as I stayed in ICU for a collapsing lung and the potential to clot. Every day became more of a challenge to prove the doctor's wrong. I sat in my hospital bed day and night just researching every site I can find on the internet. My sad, confused husband was torn between waiting by my bed side or Mason's incubator in the NICU. More pictures were taken but these pictures were almost of a different baby. Pictures were taken of Mason's sandal gap , of his almond eyes , his palm of his hand and whatever else I can capture just to so I can hear someone say that the Doctors were wrong. The waiting period for the positive FISH test was followed by a longer, less hopeful wait time for the Chromosome Karyotype test results.

The comments and questions were the biggest heart breaker:

"Are they sure?"

"I don't see it."

"Can you get a second opinion?"

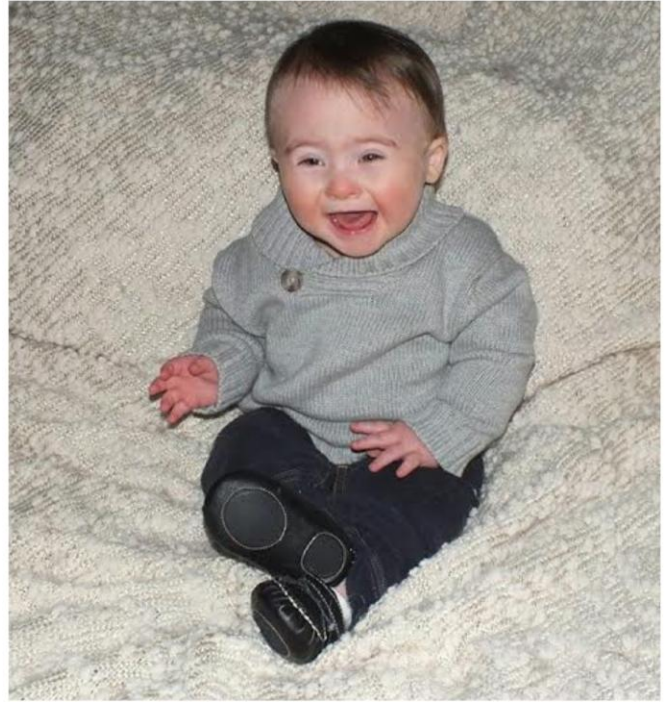
"Will you keep him?"

"I'm sorry."

Not only did they add more doubt to my thoughts but they gave me a feeling of failure. I was so worried about Mason's health during my pregnancy from the medication that I was on. I worried about his heart. I worried that my blood thinning medication may have caused delays. I worried because he never showed his face during our Ultrasound visits. I worried that the coffee clerk may have slipped me caffeine rather than decaf once or twice. I worried that I may have had too much dairy. I worried about Autism but I hadn't thought about Down Syndrome after my test results came back as a "Negative." A week later, both Mason and I had finally received clearance to be discharged from the hospital. I was not really sure what that meant for us at that time. We did not have the best support from the hospital. Our doctor's now seemed to be our enemies and our baby was not who we expected. Sure, he was absolutely beautiful and he was getting stronger and he did not have all of the potential health issues that the internet said he would. He did not really have the face of the "googled image" children with Down syndrome. He smiled, ate well, he had a strong hand grip and he rolled over. Maybe our child can beat this thing, I thought. The doubt remained for a very long time. I cried for weeks for so many different reasons. I thought about how disappointed my husband may have been with me. I thought about how I "caused" Mason's Down syndrome. I thought about his future, our future. The feeling of never wanting children resurfaced. Should I leave them? Will my husband leave us? The emotions were a never ending roller coaster.

As each day passed, we grew stronger. I grew stronger. I began to see Mason as a beautiful angel that fought his way in to this world through so many obstacles. I thought about how close I was to not knowing he even existed, as mentioned in the beginning of

the story. I thought about how he managed to skate under 12 sonogram radars and how he managed to cheat his way through a fetal DNA test. Weeks passed and more and more I began to finally realize that Mason was meant to be here. I was meant to be his Mother and we were all meant to be a family. He may not be the baby that we expected or the child we ever wanted but he is more than what we have ever imagined and we are filled with so much love for him. I am going to end this Birth story here, on one note... Some people say that special children like Mason are gifts from the Angels above. I do believe that Mason is a gift. I believe he did chose us and I believe he has every right to walk on earth just like any other typical child. I know that there will be some obstacles ahead as we pass the ones that have occurred but we are prepared. We are ready and prepared to get through them as strong and as tough as Mason was when he was fighting to get in to our world. We can only hope to be as clever and strong as he was .After all the happiness he has brought us, we owe him that much. I love my little boy to the moon and back and although some days are rougher than others, I am so glad he found us.



~ Jenny, Mason's mom; 37; Pennsylvania, United States

{The Mixed-Up Test}

My parents always told me that our dreams signify either a fear or a desire. When I was pregnant with my second son, I had a vivid dream about a little girl. I knew she was mine, and she was beautiful. She had her hair in two pigtails and had glasses. She also had Down syndrome. My mother reassured me, “It’s just fear honey, the baby is fine”. At my 20 week ultrasound we found out we were having a boy, but the doctors

found two markers
for Down syndrome.

Our little boy had a
bright spot in his
heart and one in his
bowel. My heart
sank. I knew he had
it. Why would I have
dreamt about this



little girl with Down syndrome? An amnio confirmed Luke had typical chromosomes. Hesitantly, I confessed my dream to my husband. He couldn’t believe it. He had the same dream as me. He dreamt of a little girl, his daughter. They were standing on the sidelines of a football game. She had Down syndrome.

When I became pregnant with our third and last child, my husband and I were nervous but thrilled. This was an unplanned surprise. We had already decided that two boys were the right amount of kids for us. They were 3 and 5, we were out of diapers! Our

lives were getting more manageable as we both work full time. But this was happening and we embraced it. After the initial shock wore off we were beyond excited and grateful to welcome a new addition to our family.

My pregnancy was relatively uneventful except for my own personal issues, like gestational diabetes and thyroid problems. All ultrasounds of our surprise bundle were beautiful, then on my 34th birthday we found out what we were having, a baby girl. I was going to have a daughter. This was the best birthday present I could have asked for. My husband had tears in his eyes, he was convinced it was a third boy. I was so grateful and I couldn't believe it was really true. I spent the remainder of my pregnancy planning and daydreaming about this little girl.

At 12 weeks I had a quad screen done as well as a new test called maternit21. It is a new non-invasive blood test that can detect trisomies through the mother's blood. I was excited about this new test since it posed no risk to the baby. At my 16 week appointment my doctor gave me the news: baby is healthy, negative maternit21 test and quad screen is 1:450 for Down syndrome. I had better odds than someone else my age. I was so elated to hear our baby was healthy.

As she grew in my belly, her head was extended backward and touching her back. It was disturbing and no one could tell us what it meant. My doctor explained it could be nothing or could be a neurological or osteo issue. She was also in an oblique position. My friend at work immediately googled "hyper extended neck on baby" and the first

thing she saw was a baby with Down syndrome. She asked me if the baby could have trisomy 21 and I immediately brushed it off, there's no way.

Then at 37 weeks I became sick with bronchitis. Our little girl was moving less and so I was sent in for non-stress tests and biophysical profiles. I had three in one week and I knew in my heart that she needed to come out. Something wasn't right. Then on January 3rd the ultrasound tech found that the chambers of her heart were different sizes. There was a pediatric cardiologist in the office that day and he took a look, confirming that her ductus has closed prematurely. They needed to get her out, and soon. We were scheduled for a c-section the following day.

January 4th, 2013 is the date of my daughter's birthday. This day will forever be burned in my memory as the most stressful, exciting, miraculous and life changing days of my life. My husband was by my side comforting me the whole way. As she was lifted from my body, I did not hear her cry. My husband barely got a glimpse of her when they whisked her away behind the heavy double doors. A few minutes later they called my husband back to see her. I was so scared. When he opened the door I heard music to my ears, a beautiful girlish cry was coming from the other room. My daughter. She was crying and is OK. I couldn't wait to hear all about her. I reminded my husband to take a picture. I couldn't wait to lay my eyes on her and to meet her. It felt like a lifetime waiting for this moment.

After he met her, he came back to my side. The look on his face said it all. He looked devastated. Through his tears he told me they suspected she had Down syndrome. I told him “there’s no way, the test, I took the test, and it was negative! She doesn’t have it!” I sobbed uncontrollably. I worried about my husband. I thought at that moment he was going to leave me with my three children. He was done having kids. This was my fault. He will never walk her down the aisle. What about my sons? This would change their lives too. It was all too much. My dream of having a daughter was shattered. It’s amazing the amount of irrational thoughts you can have in such a short period of time. Then I said to him *“she’s our girl, and we’re going to love her”*. And he agreed.

That day my husband reminded me of our dream. It’s like we were being prepared for this moment all along. This baby girl is meant to be ours. When I looked into her beautiful blue eyes with sparkles of white my fears melted away. When she looks at me it’s like she’s peering into my soul. She is more than I ever could have imagined. She is the light of our lives. Her brothers love her more than words can describe, and everyone who meets her is mesmerized by her. We named her Ellie, which means warrior, sun ray, and light. Little did we know four years ago that our dreams were not brought on by fear. They were in fact a desire. They predicted a happy ending we never could have dreamed of.

Ellie is a smiley 8 month old. She amazes us with how much she is growing and learning. She is now able to sit up on her own and is working on crawling. She loves all kinds of foods, but her favorite is applesauce and yogurt. She can’t stand pureed peas! She knows how to give hugs and kisses now too, which melts our hearts every time.

She is babbling up a storm and when she's upset she says "dadada" to her daddy's delight. Health-wise she has no issues and was cleared by the cardiologist at 2 months old. I thought our life was over when we received the diagnosis, I was 100% wrong. I am back at work full-time and am able to work from home on Fridays. Ellie loves to go boating and feel the wind on her face. She loves to giggle when her brothers tickle her. If you would have told me 8 months ago how "normal" our life would be I wouldn't have believed it. Except now our hearts are more open. We have joined a club that we didn't want to be a part of. I now see that we are the lucky ones. I'm proud to be a part of this club, I am proud she is my daughter. Our lives are better because of Ellie and I'm so grateful. I can't wait to see what Ellie can accomplish in her life. I know her future is bright. As my mother-in-law says, "having Ellie in my life is like winning the lottery" and I couldn't agree more.



Author's note: After some digging, I found that the Maternit21 test was never completed. My blood sample was never sent to the lab for testing, hence the negative result.

~ Tiffany, Ellie's mom; 34; Oregon, United States

Blogging @ www.our3lilbirds.blogspot.com

{Double Diagnosis}

I'm a Control Freak, a Perfectionist! I admit it! I've always needed things to be done my way. I had what I considered to be the perfect life. A nice house, a great career, an amazing husband and a beautiful, perfect daughter. And if things weren't perfect enough, the day before my 10 year wedding anniversary, I found out I was expecting Identical Twins. How PERFECT! How could I be so incredibly lucky?? How could I have been so blessed as to have been given TWO babies? Identical twins are so rare! I hit the jackpot! What an incredible gift!

The weeks following the initial ultrasound were pure bliss, that is, until an ultrasound at 12 weeks revealed that both babies could possibly have Down Syndrome. My heart sank. Surely this was a mistake. I wasn't even 35 yet! Down Syndrome didn't run in the family, I ate a very healthy organic diet, and I was a good person, dammit!!! A very nice person!! What horrible thing did I do to deserve to be punished in this way? Why was this happening to ME?



I knew what Down Syndrome was, and it wasn't good! Some of the grocery baggers at the supermarket had it. I would greet them with a strained "Hello" feeling sorry for them. I've even seen some babies at the park with it. Those poor babies, I couldn't even look at them, smile at them like I did with other babies in strollers because I knew their mothers would think I was doing it out of pity. Poor mothers. Down Syndrome was not good, at all! It was the worst thing that could possibly happen.

My husband and I decided to confirm the diagnosis via a new, highly accurate, non-invasive blood test. We waited two grueling weeks for the results. They came back positive for Ds. We

were devastated. I cried, a lot. I cursed the heavens above for this cruel joke. How could I have been given such a beautiful gift, then have it turned into something so abnormal? What would our family say? What would our friends think? What would our babies look like? What would their futures be like? I thought of all the times I looked away from the “poor babies” with Ds and felt tremendous pity for their mothers. Now I was one of them. My perfect life was now soiled and imperfect.

I was in pain. I was mourning the loss of the children I thought I would have. I had no control over this! Seeking some sort of solace, I reached out to several Ds Pregnancy support groups on the internet. I posted my introduction, reluctant and shameful, “Hi, I’m Venessa and my babies have Down Syndrome”. I expected condolences, virtual hugs, some advice for not bursting into tears in front of my daughter. Instead, hundreds of people responded with “CONGRATULATIONS!”. They all said nearly the same thing, that children with Ds are a blessing, they would change my life and the lives of the people around them in the most amazing and beautiful way. That they were perfection. “Perfection”, really?

I looked over the flood of photos they shared with me of their adorable little babies, their toddlers, teenagers, adult children, their beloved aunts, uncles and cousins, all with Down Syndrome. They were photographed on vacation, at school, at parties. Just like us, they were living normal lives. It all seemed so positive, no doom and gloom. In my research I found out that Ds is a chromosomal abnormality. It can happen to anyone, at any age, of any race, regardless of their diet or whether or not they were “a good person”.

Then I found a film on YouTube called Dakota’s Pride. The film depicted so many wonderful adults with Ds that were excelling in all areas of life! They were athletes, business owners, public speakers, newlyweds! They were just like anyone else! There was no reason to pity them, no reason to look away!

At the next ultrasound I watched my babies, now confirmed to be boys, bounce around, playfully pushing each other. I could see their little arms, legs, hands and feet. Their beautiful profiles.

Their hearts, beating. My boys. I loved them from the minute I found out I was expecting. They were my children and I was their mother.

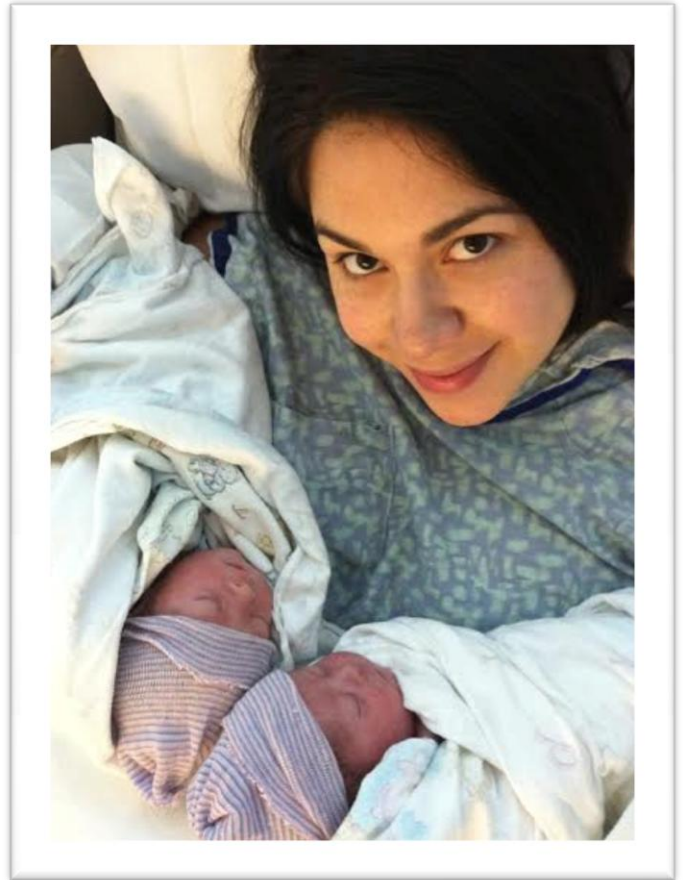
In that moment I decided to let go. Let go of the grief. Let go of the guilt I felt for the weeks I spent crying. For letting the antiquated information I had about people with disabilities cloud my vision of “perfection”. For letting the doctors fill my head with doubt and question whether or not my children were worthy of coming into this world because of their extra chromosome. Down Syndrome was not the worst thing in the world. They would have a future. My children would go to school, and parties and vacations! They would wake up on Christmas morning and open the gifts that Santa left. They would climb into our laps at bedtime to hear a story. They would chase each other in the back yard in the summer and fight over toys. They would ask for extra ice cream on the rare days that we ate it. They would live a life of limitless possibilities, a life full of LOVE!

I let go of the sadness and chose to be happy. I was about to embark on a great journey. I was going to have two children! And although the diagnosis of Down Syndrome wasn't anything I could control, my attitude and actions were. I had to be an advocate and the best mother I could be for them. So I began to celebrate them, Julian and Noel, two human beings that were coming into the world. I bought matching outfits, decorated their nursery and had a spectacular baby shower (which I planned, of course)! I did all the things any mother would do to welcome her new baby into the world.

I admit, I was scared, even terrified at times. Like a giant wave, the due date was fast approaching and my life was about to change forever, ready or not! Sometimes I would lie awake at night wondering how I would feel when they were finally here. Would I still be happy? When I looked at their faces and saw the almond shaped eyes, characteristic of Ds, would my heart break all over again? Would I cry out of grief and never stop?

On January 9th the wave finally came crashing in. My little boys made their debut, kicking and screaming! I frantically searched their face, looking for confirmation of Down Syndrome.

They had my chin, my husbands lips, my daughters ears and beautiful almond shaped eyes--such BEAUTIFUL eyes. I fell in love, a kind of love I had never known before. I cried...tears of joy! My boys were gorgeous. My boys were healthy and strong, not everyone is that fortunate. I WAS lucky, I WAS blessed! I held those two precious beings to my chest and thanked them for coming into our family, for choosing me to be their mother.



Soon after the delivery, a pediatrician cautiously approached my husband. He said he suspected the boys may have Down Syndrome. My husband, proudly holding his sons in his arms said, "Yes, we know. We've known for several months." The doctor smiled and said, "Well, it looks like they came to the right family then." Yes, yes they did. The perfect family.

~Venessa, Julian and Noel's mom; 00; New York, United States