Perfectly Me
Advocate. Educate. Legislate. For Down Syndrome!

LOGAN'S STORY

When the doctor offered us the Integrated Prenatal Screening during one of my first prenatal appointments, my husband and I agreed to it without much thought. I think we even replied with the cliché response of wanting to know "just incase" to help us prepare if we needed to. Being young and healthy we assumed we would not need to worry ourselves about it, based on my age the chances of having a child with Down syndrome were 1 in 769. I did the blood work and ultrasound, and like many other pregnant ladies, promptly forgot all about it.

Fast forward a few months, and the doctor informed us that my blood work has been flagged as high risk. She sounded a bit concerned but still the chances of the baby having Down Syndrome was only 1 in 55, "that's less than a 2% chance," she said while handing us the information for a rush appointment with a genetics counsellor at a hospital two hours from home. As scary as it sounded we were still optimistic everything would come back clear.

At about 20 weeks pregnant we travelled to London to meet with a genetic counsellor who explained how the screening process works, and what triggered the positive screen result. She spoke a bit about what Down Syndrome is, offering a medical definition of the condition and correlated facts. She offered some information on potential next steps including a more detailed anatomy scan, another screening test (the Non-Invasive Prenatal Test), or amniocentesis if we wanted a definitive diagnosis. She also explained what further options existed beyond testing - including the option to terminate our pregnancy. We clearly stated termination was not an option we were interested in hearing about and she immediately noted that in our file. After our meeting we opted to complete the NIPT to have a more accurate screen result.

The following weeks were a blur of continuing on with our regular routines while also trying desperately to find out as much information as we could. We looked for information from Down Syndrome organizations, from other parents who had been in our place, from families with a connection to the Down Syndrome community - if the information was out there, we were trying to find it because we had not been given the information from our medical team.

After what felt like an eternity (but was more like 14 business days), our counsellor called to confirm that our baby has tested positive for Trisomy 21, more commonly known as Down syndrome. These were the results of the Non-invasive prenatal testing (NIPT) which were to have an accuracy of~99%.

We were shocked. We were sad. We were mad. It did not seem fair. There were tears and fears and lots of Google searches (which contributed to more tears and fears) but our love for this little baby never faltered. We worried about his health – heart conditions, certain types of cancer, hearing impairment, the list of potential health issues just kept growing. We worried about the future – would he make friends? Would he be accepted? Would people tease him? Would he live with us forever? Fear of the unknown was so difficult.

My pregnancy was instantly deemed high risk so my prenatal care was transferred to a larger hospital two hours away. The genetic counsellor met us at our first appointment with our new doctor. She brought along a welcome package from a local Down Syndrome Association, as well as two different resources - one based in science and research and one of stories from those within the Down Syndrome community. It was the perfect mix to help us prepare and reassure us that a Down Syndrome diagnosis was nothing to be afraid of.

Our prenatal diagnosis story is a positive one. After stating, at our first meeting, that termination was not an option it was never mentioned again. We never felt pressured, nor shamed or guilted by those charged with caring for me and our baby. Our medical team encouraged us to enjoy this pregnancy just as we had always planned. They spoke to us as typical soon-to-be-new parents - they asked about the nursery, about maternity photos, about babymoon plans.

Even after baby was born the medical team continued to be supportive. We heard "congratulations," not "I'm sorry." We heard "he is perfect," and "he looks just like Dad." We heard all of the things you want to hear after delivering a baby. We are lucky that we had such a positive experience. We know from others that this is not always the case. Even within the same hospital we delivered at, parents have been made to feel badly over their choice to choose life with an extra chromosome. That some medical professionals will unfairly push their out-dated, inaccurate or negatively biased views onto parents.

That is just one of the reasons why we advocate for our son and the entire Down Syndrome community. Parents receiving a potential diagnosis deserve to know the facts. Relevant, current, and accurate facts so parents can make a truly informed decision.

We wouldn't change our son for the world, but with help we can change the world for him.

Subjective:

Objective: