Nov 5th 2010

Six weeks ago a bomb was dropped. It tore through our home, devastated our lives and left us in a state of complete shock. Our 8 month old daughter, our perfect and beautiful angel, has Spinal Muscular Atrophy. To make things even worse, she has type 1, which is the most aggressive of the four types. Before receiving this diagnosis we couldn’t tell you what SMA is, we had never heard of it, never met anyone affected, even though one in forty people is a carrier and one in six thousand has the disease. My husband and I are both carriers, which means that we are missing one of two available Survival Motor Neuron 1 genes. Our children have a one in four chance of being born with both genes missing and, therefore, having the disease. Jessika was born with this disease. She is now 9 months old; she will never walk, run or dance. She cannot sit or crawl; she cannot lift her arms to feed herself unless she’s lying on her back. She will need a feeding tube and machines to help her breathe as her respiratory muscles begin to weaken. She will need extra special care everyday for the entire span of what could be a very short life. But she is happy and she is surrounded by love. She has the most beautiful smile, a tricky smile, like she’s just told a joke and is having a little laugh. She sings and chatters all day long and loves when her older brothers sing and play with her. Every day that we spend with her will be a miracle, every hour a blessing, every minute filled with joy.

Our SMA journey...the diagnosis Nov 8th, 2010

On September 30th, 2010, we walked out of the Montreal Children's Hospital, our lives forever changed.  We had just met with our Neurologist and received the results from a blood test taken two weeks earlier.  The test was for a rare genetic disease called SMA, the results were positive.

  Our daughter, Jessika, had started regressing just before she turned 5 months.  She moved her legs less than she had before, she showed no interest in rolling over but we were not concerned.  We told ourselves that Jessika was simply taking her time, developing at her own pace.  We saw nothing that gave us cause to worry, especially since we have other children, all of them having different developmental patterns, all of them healthy in mind and body.

Jessika was, and still is, happy. She gives smiles and love freely and generously; she coos, babbles and giggles; she is very sociable, responsive and alert. Our baby girl was nursing better than my son had, was healthy in every way that we could see. We started her on solids; pureed bananas, rice, oat and barley cereals, carrots, squash, apples, all the usual first foods with great success. Jessika loves eating and we are still introducing her to new and exciting flavours. But we began to grow suspicious of her lack of movement and took her to a chiropractor at the insistence of my parents. The chiropractor told us that Jessika was hypotonic, that she had low muscle tone. After that initial session it seemed that Jessika became a little more mobile so we continued seeing that chiropractor for a few more weeks. There was no major improvement, Jessika still seemed unwilling to move her legs but, again, we didn’t think it could be anything overly severe because she could move her feet and toes.

David, a friend of ours and my son’s Taekwondo Master, asked how we were all doing one day after class. I told him that we had been taking Jessika to see a chiropractor, that we were a little concerned by Jessika’s lack of mobility but that there seemed to be some improvement. He gave me a number to an osteopath that he knew personally who could give me a second opinion. I made an appointment, which we ended up cancelling; David had sent an email urging me to take Jessika to the Children’s Hospital to see a neurologist. He had noticed that Jessika had tongue tremors, something we, ourselves, had missed and that this could signify a neurological issue. I decided to take Jessika to the hospital, spent seven long hours describing her symptoms to paediatricians, nurses and specialists, and watched the nurses poke needles into Jessika’s arms, feet and hands to extract blood from veins that were so deep and impossible to reach until finally they needed to go into a vein high on her forehead. I met with our neurologist, she explained that they were testing for SMA and did I know what this is? She gave me a basic description, told me it wasn’t a good prognosis at all, and told me they hoped to have the results the following week.

That brings us back to the 30th of September. That cold and rainy day, that terrible day, I cried all the way home from the hospital. Jeff made phone call after phone call, telling our friends, employers and family the news. We cried a lot those first few days. How could we not, we had just been told that Jessika would never sit on her own, she wouldn’t crawl or walk, she would lose the strength to use her major muscles, her respiratory muscles would weaken and, eventually give up, the muscles used for swallowing and speaking would deteriorate as well. Our baby girl, her life once full of so many possibilities and so much promise, has a terminally degenerative genetic disease. Jessika has Spinal Muscular Atrophy type 1.