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A Mother's Intuition and a Message for Physicians

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A mother's intuition can be a powerful and insightful guiding force, and yet far too often it is ignored or severely underappreciated by medical professionals. I never felt this feeling as strong as after the birth of my first child, a son we named Jonah.

My husband and I were married for several years before we decided to have children. We were not anxious and thought we would just let nature take its course. Getting pregnant turned out to be far more difficult than anticipated, and we were in the process of exploring in vitro fertilization when our miracle happened.

I can still recall that overwhelming sense of joy when I discovered that I was pregnant. Every mom remembers that feeling – it's miraculous! Unfortunately, our first pregnancy was not meant to be, and I miscarried at seven weeks. It was a tragic loss, and to this day, I wonder how life would have been different if this event had not occurred. Despite this, we conceived again one year later.

The pregnancy was easy. Mine was one of those rare stories of no nausea, no cravings, no swelling, no sleep disruption and nominal weight gain although the baby was growing fine. Everything was progressing nicely up until week 31 when I was rushed to St. Joseph's Hospital in London due to bleeding. I was admitted into hospital and placed on bed rest.

In the early hours of the morning on December 24th, I woke with contractions and realized that my water had broken. I was told that I was at an increased risk of delivering the baby early. The doctor called my husband telling him to come to the hospital right away. Before he could arrive, I was being rushed into a delivery room. My husband arrived with moments to spare and was holding my hand as the doctor delivered our precious baby boy. Labour and delivery lasted just over an hour, and before we knew it, my husband and I were parents of a seven week premature baby boy.

Our son weighed in at exactly four pounds. He was immediately taken from us for examination. He was very fortunate not to have any immediate health issues and was breathing on his own. The baby stayed in the Neonatal Intensive Care Unit at St. Joseph's Hospital for only a few days and then was moved to a unit called Progressive Care. Our son remained there for just under a month and was then moved to an area called Care By Parent. I stayed in the hospital with the baby the entire time to promote breast feeding and parental bonding. At six weeks of age, our son was strong enough to leave the hospital.

As first time parents, we were given lots of advice from family and friends. As with most new moms, I was faithfully reading books on child development and caring for my newborn. I was also comparing stories with my sister-in-law who had just been blessed with her first daughter one week before Jonah was born. Very early on, I noticed that Jonah was not "textbook perfect." I attributed the delay in achieving developmental milestones to Jonah's prematurity, but there was a part of me that kept wondering if there was something more.

At visits to the doctor during that first year, my husband and I would question Jonah's progress and query things that seemed unusual. Our doctor repeatedly told us that it was normal for premature babies to take longer meeting milestones, and we were encouraged to relax. But it wasn't just hitting those milestones that bothered us. There was something about Jonah's inability to connect with us and his world around him that was puzzling. He was also a very restless little guy. Words like content and peaceful were never used to describe Jonah. My intuition was trying to tell me something, but what was it?

As a newborn, Jonah slept a lot. People would say, "Enjoy it – take advantage of the time." I took no comfort in those statements. To me, this was troublesome. Was this intended as a sign that something may not be quite right? When Jonah was awake, he cried a lot and was incredibly hard to console. This was especially true when he woke from his afternoon naps. He

would cry so uncontrollably, and there appeared nothing I could do to appease him. One day, I decided to take him into the bathroom and let him cry while I took a shower. Soon after turning the shower on, the crying subsided. The sound of fast running water soothed him – but why? I resorted to taking our son into the bathroom on a daily basis and turning the tap on full blast. The number of required trips to the tap to hear racing water was soon being used to define our day. I also noted that Jonah required frequent changes in his environment. My husband and I started planning our day around Jonah’s need to regularly transition from one place to another. We were constantly on the move – going for walks, to the park, visits to grandma’s house, excursions – to fill the day and keep the peace. While others would giggle at these stories and find them amusing, I was deeply troubled with the reality that this just didn’t seem normal.

I vividly recall another day when there was no appeasing Jonah’s inconsolable crying. I was pacing the house, cuddling my son and rocking him in my arms. My frustration level was at a peak, and I felt desperate. My husband had already stepped outside, not able to tolerate the screaming anymore. I laid Jonah on the bed momentarily as I could feel tears welling up in my eyes. Almost immediately, the screaming stopped. My husband ran into the bedroom to see what was “wrong.” “What happened to Jonah? Why did he stop screaming?” I looked at him puzzled and said, “I just laid him on the bed and he stopped.” That was the first time I realized that Jonah had some aversion to touch. Being embarrassed to tell this story, I mean what child does not take comfort in his mother’s arms?, when I finally did share this with my doctor, he laughed and said, “He probably just needed a break from you.”

I don’t know how many times my husband and I rushed our son to the emergency department (ER) during the night due to uncontrolled crying. As a mom, you try everything and then start to worry that maybe the problem is medical. The staff in the ER would give me that “her again” look as they checked Jonah over but never found anything conclusive.

At four months of age, we discovered Jonah had a hernia. My husband and I were hoping that this was the reason for Jonah’s unsettled nature. Following surgery Jonah suffered an unusual reaction to the anaesthetic. He laid on a gurney, eyes wide open and screaming out as though completely horrified. I ran to him and grabbed him tight, hoping that my presence and the sound of my voice would calm him. Jonah looked right through me as if I was a stranger. There was no connection at all. The doctors had to administer codeine to calm him down. Again, in consulting with the doctor I felt dismissed when he said, “some people just don’t handle anaesthetic well.”

At 19 months, our doctor connected us with physio and occupational therapy services through Thames Valley Children’s Centre. The first visit was memorable but not a pleasant one. While waiting for our appointment, a receptionist approached me with a clip board of papers to

complete. Attached to the clip board was the poem, “Welcome to Holland” by Emily Perl Kingsley (1987). I immediately felt a chill run through me. You see, I know this poem. I have used this poem in my work at a developmental service organization. I have given this poem to family members who had children with intellectual disabilities. Why was I now on the receiving end? What did this mean? “This is just our standard application” I was told, but immediately I felt a lump in the pit of my stomach that just ached.

Soon after, we started receiving home visits from an occupational and physiotherapist. They too told me that premature babies often take a little longer to reach developmental milestones. One day, one of the therapists was commenting on Jonah and saying how he reminded her of a little guy with whom she used to work. That’s all it took for me to probe deeper. “What similarities do you see? Did this child have a diagnosis?” This was the first time I learned of Williams syndrome. The therapist had not even reached her car, and I was on the internet googling Williams syndrome and scouring websites. I was frantically reading characteristics of this diagnosis while running a “yes” and “no” checklist in my head. What I discovered is that Williams syndrome is a rare genetic disorder that can only be confirmed through a blood test called a Fish Test. There are specific unique requirements for the test, and I ran off the information as quickly as I could. After telling my husband what I had discovered, I made an appointment with the doctor. I reviewed the information I had gathered and asked that the test be done. My doctor smiled, shook his head and reassuringly stated, “This is not necessary. Jonah is fine. You need to just relax.” “That may be,” I said, “but just do this for me. I need peace of mind.” Reluctantly, he made arrangements for the test.

It took only days for the results to come back. The results were conclusive ... Jonah had the diagnosis of Williams syndrome. My husband, my son, our newborn daughter (Olivia) and I were at my parents’ home when we got the news. I remember asking, “Why?? Why Jonah??” I remember my husband holding me and telling me it would be okay. I remember looking at my mother in desperation, “What are we going to do?” My mother grabbed my shoulders, looked me straight in the eye, and with confidence and strength in her voice she said, “We’re going to deal with this. That’s what we are going to do.” Later that night, my husband was putting the children down to sleep, and I had a moment to myself. I felt calmness come over me, and peacefulness entered my heart. I realized that I now had the answer for which I had been searching – my intuition had been right all along. I knew I could deal with the known; it was the unknown that had terrified me. I also stood firm in the belief that not a diagnosis, nor a label, nor a document on the internet was going to define my son.

When I met with the doctor following the diagnosis, he apologized for ignoring my intuition and not taking my concerns seriously. He also openly admitted that he knew nothing about Williams syndrome and asked for the website from which I was gathering information. This

website proved to be very helpful as there are risks associated with the heart that are often common in people with Williams syndrome. The doctor referred us to a developmental paediatrician for an assessment, a paediatric neurologist and a paediatric cardiologist.



[Sherri Kroll and son Jonah](#)

Those appointments introduced me to the stereotypical ignorance and disrespect of people with disabilities. I was appalled when a highly respected “professional” said to me in reference to my son, “He’s quite retarded you know.” Another so called “professional” stated, “I’ve seen this kind before,” meaning kids with the diagnosis of Williams syndrome and then wrote in his report, “While in my office he (Jonah) cried continuously, was very uncooperative, even hysterical”. My twenty years of experience in the developmental services sector gave me the confidence and strength to confront these kinds of statements

head on and correct such derogatory language. I often found myself thinking about those families that had no experiences upon which to draw, and how frightening, belittling and hurtful those comments are to parents who are already under tremendous stress and shock.

What has surprised me most of all on this journey has been how little information is offered to families when they discover they need help. When your first contact as a new parent is the family doctor or paediatrician who knows nothing about the diagnosis, you are really at a loss. No one takes your hand and guides you through this process. Families, who are likely stunned, confused, scared and feeling helpless, are left with the challenge of seeking out answers to questions they never dreamed they would have to ask. The good news is that support is out there, and services for children (unlike services for adults) are relatively rich. However, and unfortunately, you don’t know what questions to ask and what to look for. I was surprised to learn that Easter Seals offers an Incontinence Grant for kids who still require diapers beyond typical years owing to a disability. Never in my wildest dreams would I have gone looking for a grant for diapers. It would not have dawned on me that something like this might be available.

Other examples of available services include: special therapies, services and programs, financial assistance, tax credits, Registered Disability Savings Programs, Henson Trusts, and so on which are unknown to most people. I have met so many families that are exhausted just getting through the day that they cannot even fathom searching out activities, services and supports. It can be an overwhelming chore. Thank goodness for the support of our family, as well as my

connections and insights as a result of my professional career which has enabled me to manoeuvre through the various systems, but even so, I still find myself running in circles at times.

Today life is good. Jonah is now approaching his tenth birthday. His sister Olivia recently celebrated her eight birthday and my husband and I are both, well let's just say older and wiser. Jonah attends a fantastic school and continues to learn and progress. A couple of years ago we made the difficult decision to move Jonah into a segregated classroom. I fought tooth and nail to keep him in an integrated environment, but at the end of the day, I realized that I was fighting the battle for me and my personal views rather than what was in my son's best interest. One highlight from school was a singing performance in last year's Talent Contest where Jonah sang Jason Mraz's song – "I'm Yours." He stole the show, and I wept with pride as the students clapped to the beat, cheered him on and greeted him off stage with high fives. Jonah may spend part of his day in a segregated classroom, but he lives a very integrated life.



Jonah and sister Olivia

Outside of school, Jonah takes music lessons and has registered for horseback riding. He stills loves being on the go, and our family rarely misses a community fair, parade or event. We regularly go the park, library and on shopping excursions. Due to our faithful attendance, we are recognized and greeted by name at the Children's Museum, Story Book Gardens and most local stores. Jonah loves celebrations of any kind and being in the company of others. He truly is a great kid (as is his sister whom I would be remiss not to mention). Jonah has taught our family patience, added much humour to our lives and is the greatest role model of unconditional love. I am probably the only mother of a ten year old boy who gets greeted every morning with an endearing smile, the best squishiest hug you could imagine

and a pat on the back while Jonah says "Hello my Princess."

What I have learned along this journey is that you have to find strength and keep strong. There will be battles to face and sometimes the challenges can seem insurmountable, but put your head and heart in the right direction, and your feet will follow. I encourage families to connect with others, ask lots of questions, be continual learners, don't be afraid to ask for help and take it when you can get it. I also encourage doctors to take time to listen, trust their patients and to be ever sensitive to a mothers' intuition.

For a similar story to Sherri Kroll's, look at this free website access from AAIDD article on Fragile X

http://journals.lww.com/neurologynow/Fulltext/2011/07050/Fragile_X_Syndrome_Research_into_Fragile_X.17.aspx **Neurology Now**: October/November 2011 - Volume 7 - Issue 5 - pp 37,45–49. doi: 10.1097/01.NNN.0000406989.86920.67

Fragile X Syndrome: Research into Fragile X Syndrome, a common cause of inherited intellectual disability, is starting to generate treatments Gordon, Debra M.S.

DDD site: <http://www.ddd.uwo.ca/resources/syndromeswilliams.html>

U.S.A. National Institute of Neurological Disorders and Stroke (NINDS), William Syndrome Information Page

<http://www.ninds.nih.gov/disorders/williams/williams.htm>

Definition of Williams syndrome (WS). US National Institutes of Health. Last updated: 9 Sept 2008.

What is Williams Syndrome?

Williams Syndrome (WS) is a rare genetic disorder characterized by mild to moderate mental retardation or learning difficulties, a distinctive facial appearance, and a unique personality that combines over-friendliness and high levels of empathy with anxiety. The most significant medical problem associated with WS is cardiovascular disease caused by narrowed arteries. WS is also associated with elevated blood calcium levels in infancy. A random genetic mutation (deletion of a small piece of chromosome 7), rather than inheritance, most often causes the disorder. However, individuals who have WS have a 50 percent chance of passing it on if they decide to have children. The characteristic facial features of WS include puffiness around the eyes, a short nose with a broad nasal tip, wide mouth, full cheeks, full lips, and a small chin. People with WS are also likely to have a long neck, sloping shoulders, short stature, limited mobility in their joints, and curvature of the spine. Some individuals with WS have a star-like pattern in the iris of their eyes. Infants with WS are often irritable and colicky, with feeding problems that keep them from gaining weight. Chronic abdominal pain is common in adolescents and adults. By age 30, the majority of individuals with WS have diabetes or pre-diabetes and mild to moderate sensorineural hearing loss (a form of deafness due to disturbed function of the auditory nerve). For some people, hearing loss may begin as early as late childhood. WS also is associated with a characteristic "cognitive profile" of mental strengths and weaknesses composed of strengths in verbal short-term memory and language, combined with severe weakness in visuospatial construction (the skills used to copy patterns, draw, or write). Within language, the strongest skills are typically in concrete, practical vocabulary, which in many cases is in the low average to average range for the general population. Abstract or conceptual-relational vocabulary is much more limited. Most older children and adults with WS speak fluently and use good grammar. More than 50% of children with WS have attention deficit disorders (ADD or ADHD), and about 50% have specific phobias, such as a fear of loud noises. The majority of individuals with WS worry excessively.

Is there any treatment?

There is no cure for Williams syndrome, nor is there a standard course of treatment. Because WS is an uncommon and complex disorder, multidisciplinary clinics have been established at several centers in the United States. Treatments are based on an individual's particular

symptoms. People with WS require regular cardiovascular monitoring for potential medical problems, such as symptomatic narrowing of the blood vessels, high blood pressure, and heart failure

What is the prognosis?

The prognosis for individuals with WS varies. Some degree of mental retardation is found in most people with the disorder. Some adults are able to function independently, complete academic or vocational school, and live in supervised homes or on their own; most live with a caregiver. Parents can increase the likelihood that their child will be able to live semi-independently by teaching self-help skills early. Early intervention and individualized educational programs designed with the distinct cognitive and personality profiles of WS in mind also help individuals maximize their potential. Medical complications associated with the disorder may shorten the lifespan of some individuals with WS.

Canadian Association for Williams Syndrome (CAWS)

<http://www.caws-can.org/>

CAWS' goals are: To support research into the educational, behavioural, social and medical aspects of Williams syndrome.

To increase society's awareness of CAWS so individuals with Williams syndrome and their families have a resource available to them.

To become visible to the medical scientific, educational and professional communities by providing information on Williams syndrome.

There is a section for new parents.

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The Williams Syndrome Association

<http://www.williams-syndrome.org>

Information for people and families living with Williams syndrome, as well as doctors, researchers and educators. Includes a road map for parents, growth charts and health care guidelines.

The Williams Syndrome Comprehensive Web Site

<http://www.wsf.org/toc.htm>

Information for new parents and physicians includes medical, genetic (FISH test), dental, and behavioural guidelines for parents with a young child and families with an adult with WS.