



VIRTUALMEDICALPRACTICE, LLC **the future of medicine is now.....**

Before I became pregnant, I had this vision of what it would be like to be a parent...what it would be like to hold my child for the first time...what it would be like to watch him/her grow, laugh smile, and thrive.

Little did I know that my vision was quite different from what the reality would be...and that, while I would see my children do all these things – heartache would also come as I watched my children suffer, live with chronic illness, and wade in a sea of uncertainty as we searched for the light of Hope in the darkness of the unknown.

While both my children had many medical issues, my youngest child, Sasha seemed to have the worst of it. She was born fighting for her life, and while some of her issues resolved, new issues began to sprout up and take residence within her.

As time went on and nearly 2 years passed, it became crystal clear that ‘something’ wasn’t right with her. Appointment after appointment, all the ‘best of the best’ had never seen anything like her and didn’t know what to do with her. It was quite scary to be in the “Mecca” of the US (Boston) and to be told that they had no clue what was wrong with your child – and they didn’t know the best way to treat her.

My son, Will also struggled with a litany of medical issues, but he seemed to physically handle these issues better than his sister...but still the search for answers continued.

My medical issues (which I have had all my life) continued and worsened as well – my primary care was convinced that there was some type of energy storage issue, but I was too focused on what was going on with my children to stop and take care of myself and I never stopped to think about the connection between my issues and theirs, until last year. One of Sasha’s physicians suggested that I might have mitochondrial disease and he wanted to run some tests. The first of the tests came back without answer....but with each passing month, he was more and more convinced that this was, in fact, mitochondrial disease.

Living with uncertainty is something that you never ‘get used to’ but you do learn to accept it and all of the confusion that comes along with it. We had a fantastic physician up here for Sasha who was trying to piece it all together, but he too also suggested an outside view might be helpful when looking at my daughter and myself together.

Frustrated and determined to get an answer, not only for myself, but my daughter, I began looking online to find a specialist that sees adult patients with mitochondrial disease as well as children. I came across Dr. Kendall’s site and sent a fairly lengthy email off asking about her practice. Within a few hours, I received a response asking for my number in order to discuss things more in detail. I responded to the email and almost immediately, I got a phone call from Dr. Kendall! I was more than impressed with the speed in which there was a response and the fact that she took time out of her day to call and discuss our complicated family over the

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phone. She was kind, patient, and thorough – completely focused on the best path for us to travel in getting our answers. She didn't rush me off the phone and took time to listen to all the details before she offered her opinion. She agreed that she felt we were on the right path (about this being related to mitochondrial disease) but she wanted me to wait on muscle biopsy as she felt we might yield answers doing less invasive testing. I told her I had the POL-G1 testing done and was waiting on the results, and she agreed that there were a few other tests to run before we jumped to muscle biopsy.

Sasha and I were scheduled to see her down in Atlanta at the end of March – which was just about a month away from our call.

We headed down to Atlanta with hope that we would at least find answers for one of us. On March 31st, my life changed forever.

As I was led into the exam room by her husband (he runs the practice with her), there she was, in the room already waiting for me with our medical records on the table next to her. She was kind, sweet and prepared for what would turn out to be a marathon appointment session that covered my life and Sasha's life in less than 4 hours.

We started off with me. The first words out of her mouth were, "Hi Stefani, so great to meet you. I have spent some time with both your records and your daughter's records and also the POL-G1 mutation that was discovered in you last week. I have printed up some information for you about the POL-G1 mutation and I wanted to let you know that this mutation that you have is a recognized and disease causing mitochondrial mutation."

I paused for a moment and I clarified with her, "So, that means that I have mitochondrial disease?" and she said, "Yes, it does." There was a pause....and, for a moment – it felt like the world around me stopped. While it has been speculated that I too could have mitochondrial disease like they suspected in Sasha – it never seemed real. There's been so much 'talk' about it, but I didn't expect that I'd get official confirmation before Sasha did. I said to her, "So, it's not all in my head?" and she said, "No...and it never was."

I guess I had hoped that I was just a hypochondriac or that I was making more out of my symptoms than need be – I mean, wouldn't that be the easy answer?

My heart sank...because, in that moment....everything changed. Despite the fact that this was a genetic mutation that I have carried with me from birth, discovering the reality that this disease was active in my body made everything so real, so scary, so unpredictable. I was reeling....and trying SO hard to focus on the appointment that was continuing on in the midst of the chaos going on in my head.



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We went over family history, my history, past issues and current symptoms. We talked about protocols – ER protocols, fasting protocols (which is now a direct admit the night prior), surgery protocols, illness protocols as well as getting it set up to have access to IV fluids in the home setting as needed. These are things I just never thought I would need to have to think about...and here I am, at 32 facing the harsh reality that these will be a part of my life from here on out. That being said, I am hopeful that having these protocols in place will make things easier for me and help me to feel better. She said a priority is definitely to get the autonomic issues and the GI issues addressed immediately...and I agree.

We discussed her protocol letter – stating that mine will be about 4 pages long...it will explain what they need to do and what they need to watch for as well as a brief description about the disease. She will be in contact with my PCP and we will begin paperwork for the long road ahead. She wrote a prescription for the ‘Mito Cocktail’ (which is just a bunch of vitamins in mega doses) and I will be starting it as soon as I return home – knowing this will not be a ‘fix’ but it could possibly help me to feel a bit better. We will be starting the long process of disability and she will be writing for a wheelchair for me to be able to use when it is needed...all of these steps take a LONG time to complete and it is wise to begin now, when I am not in dire need than to wait until the last minute and be struggling and desperate for things I cannot attain.

We talked about Sasha’s history, she watched her videos and we discussed her muscle biopsy. She told me that there is a shift in the position many of the mito specialists have on this topic as they are finding that this is not a ‘consistent’ testing method – that many families are having inconclusive, false negatives and even some are having false positives through this method – which is invasive, costly, and – for some patients, risky.

The shift is the focus on the genetic discoveries – which, once you pinpoint the genetic mutation/deletion/change, you have a better ability to focus on clinical trials for specific mutations within that and we’ll be able to track effectiveness of any future treatment options more efficiently this way.

Like her doctors up in Boston, she feels that Sasha fits the bill for Mitochondrial Disease – but we need to find the actual link in order to say it for sure - we are currently waiting for genetic testing to come back that will hopefully give us more answers. She said that there isn’t much else that it could be given the fact that so many organ systems are involved. Again, while I knew this to be the likely possibility, it just made my heart sink even deeper because I know the road ahead will not be easy.

We spent some time talking with her husband about the letters and the next steps and then headed back to my cousin’s house.



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Our care with Dr. Kendall didn't end when we left her office – before we were on the plane back to Boston, I had a standard protocol letter and some reference material sent to me via email, and within 2 weeks I had everything I needed (clinic note, PT referral for wheelchair, disability letter, specialized and individualized protocol letters). She arranged for me to see a local GI for my issues and she also had touched base with my PCP and Sasha's metabolic doctor. Even though she had many other patients to coordinate care for, it felt like we were her only patients...and, while we didn't leave with great news, we left feeling like we had the support we needed to get us moving in the right direction.

I can't say enough about Dr. Kendall or her husband - they were so wonderful, informative, and kind to us - giving us all the time we needed for our questions and understanding that the news we were receiving was not exactly easy to digest. I highly recommend her to anyone out there who is looking for a mitochondrial disease specialist and I have recommended her already several times to many of the people within the mito community who have reached out and asked if I knew of a good mito specialist. She not only offered care that was top notch, but she treated us like we were HUMAN and not just another file folder on her desk. This is a rare quality to find within the medical community – especially when the cases are so complex, time consuming, and unique.

In the months since our meeting in Atlanta, I've had several incidents where I have emailed her and received immediate response, and when necessary – a phone call to discuss matters. It again solidified the feeling that we weren't just another 'file' we were worth the extra time and effort...and that we mattered...

All I could think of on the way home was how our visit here while informative and overwhelming – changed everything and changed nothing at the same time...

We are still the same mom and daughter that we were before we walked into her office...we still have the same love for each other that we had before our appointment...and we still have the same goals and dreams that we had before the doctor shared the diagnosis.

It does change the future in terms of how things are handled and what lies ahead...but it changes nothing about who *I* am or who *Sasha* is or the resolve we have always had to live each day to the fullest while making the most positive impact on each other and the lives of those around us.

My resolve to live as normal a life as possible is strengthened by the knowledge that it is almost certain that Sasha also has this disease.

While the truth of this is that it will not be easy at times and that it will be scary at times as well...but she needs to see me facing each day with hope, promise, and joy...so she knows that 'once you choose HOPE...anything is possible.'

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