Complex Child E-Magazine

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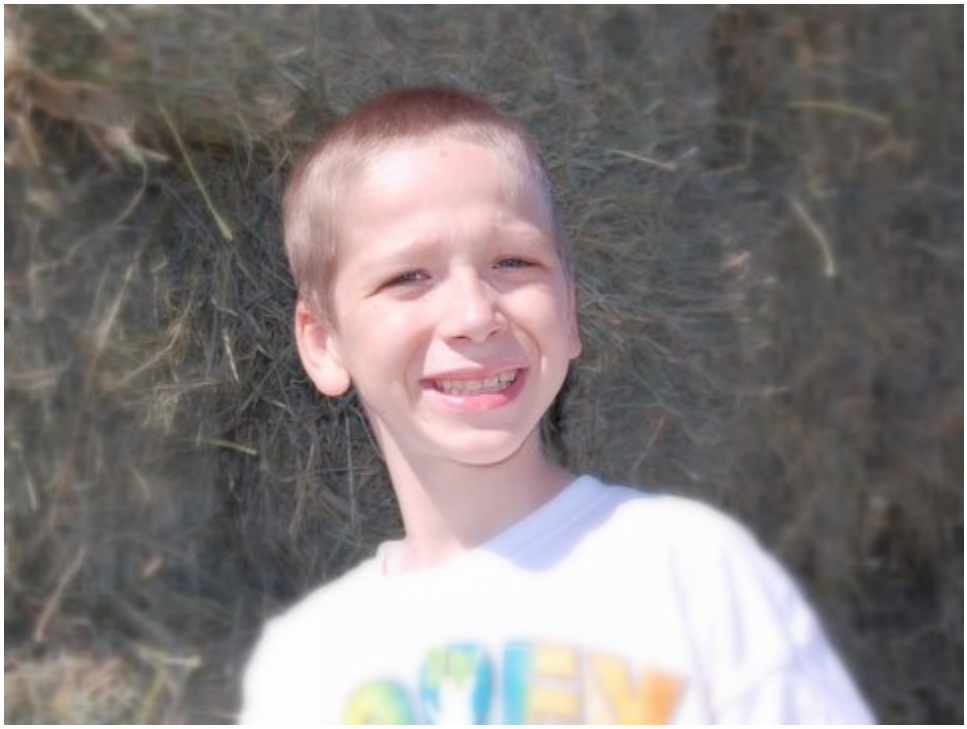
Dysautonomia:
A Minor Inconvenience Becomes a Devastating Disease

A mother's perspective on watching her son live with a life-taking disease

by Alison B.

Meet Andrew. Andrew is nine years old and will be ten very soon. He has blonde hair and blue eyes and lovely fair skin. He is a good looking young man with an amazing sense of humor, a laugh that is unbelievably contagious and a disease that is slowly, but steadily, taking him from us. This disease is a form of Degenerative Dysautonomia.



From Minor Inconveniences to Increasingly Frightening Symptoms

When Andrew was born, we thought he was a normal healthy baby, the youngest of four children. Within the first month of life, Andrew started to have difficulties with feeding and congestion. His breathing was three times faster than a normal newborn and his heart would go through periods of beating much faster than normal also. He was hospitalized several times in the first eight months of his life, mostly for dehydration. On more than one occasion we were told that he was simply the youngest of four, and was probably getting a lot of viruses from his siblings. It didn't sit right with us.

As the end of Andrew's first year neared, we were really beginning to think that something might be wrong. Between the ages of one and two his illness was much worse and he ended up spending more time in the hospital than he did at home. There were several diagnoses that we were given and literally hundreds of genetic tests that were run, but nothing gave an answer to why this little boy could be so perfectly well one day and turn around and be incredibly sick the next day. After living with an NG tube for most of the past 18 months, we, along with his medical professionals, decided it was time for a G tube. At the time, this seemed like one of the hardest decisions that a parent could ever possibly make. Little did we know what lay ahead.

Andrew today

Over the next many years Andrew's list of symptoms increased. He would have periods of wellness followed by periods when he would vomit to the point of fainting and lose his speech and all muscle tone. His intestinal system would periodically stop, losing all bowel sounds, and making it impossible to feed him through his G tube. He couldn't even keep down any fluids for hydration during these periods and required intravenous hydration. He would develop strange rashes and infections. He would lose the ability to pee and his bladder would fill to the point that it reached his belly button. He would cry and scream in what appeared to be pain. He started having seizures. His heartrate, breathing, and other vital signs become erratic. He was hospitalized over and over and over for these spells, usually once every three to six weeks.

As life threatening problems were mounting, so were the interventions that we were needing. By age five, Andrew was unable to be discharged from a hospital stay without oxygen in our home along with equipment for monitoring his condition and, shortly thereafter, in-home nursing care.

Moving Toward a Diagnosis

During one of these hospitalizations for a "spell," an observant doctor noted that Andrew's pupils were hugely dilated in comparison to his father's and mine and were clearly abnormal. It was then that the word "autonomic dysfunction" was first brought into our vocabulary.

I remember doing internet searches and almost all of what I found referred to what sounded like groupings of minor symptoms such as dizziness and fainting. It didn't seem to make sense to me since what I was reading sounded like it wasn't "that bad," yet what the doctors and social workers were telling us was that Andrew's disease was progressive. I would later learn that Andrew did not have simple autonomic dysfunction, a typically bothersome but manageable condition, but rather some form of Degenerative Dysautonomia. Andrew would likely not survive childhood. Whatever it was that he had was fatal based on what they were seeing, though we also ran into doctors that didn't think dysautonomia was his issue since he tested negative for the standard genetic markers.

In the spring of 2004 Andrew came down with a very bad cellulitis infection at his G tube site. He got extremely sick very fast and his body's autonomic system, which should automatically respond to stress within the body, couldn't do what it needed to do. We don't know exactly what combination of his issues is to blame, but the thought is that due to a massive drop in blood pressure and perfusion to the kidneys over only a couple of hours, his kidneys suffered a devastating blow and he went into acute renal failure. We were asked, based on the fact that he had a mysterious degenerative disease, did we want to do dialysis or did we want to just keep him comfortable and let him go? We chose dialysis and to fight for his life. Andrew spent four weeks on dialysis and it was nothing short of a miracle that his kidneys began to function again and began to recover from the serious blow they had taken.

During this hospital stay we were introduced to a doctor who went beyond guessing that Andrew had dysautonomia to actually proving he had a form of dysautonomia. When Andrew had recovered and was back on his feet, we arranged to meet up with her and her team to undergo a whole realm of tests designed to put stress on the body and especially the autonomic nervous system.

Testing and Diagnosis

In a healthy person, the body reacts with chemical and nerve responses whenever you undergo stress. For example, if you slam on the brakes when a child runs in front of the car you are driving, within seconds your heart is beating faster, your brow and armpits are sweaty, your blood pressure would be higher, and your legs are likely shaking. These are standard responses of your autonomic nervous system to a fearful situation. Apart from giving us the shakes during times of stress, the autonomic nervous system also does many things to keep you alive. For example, if you fall into a cold river, you shake and shiver because your autonomic system knows you must generate heat to survive.

Andrew had multiple tests to challenge his autonomic nervous system to respond. Each test would stress his body in a different way. Where you would expect to see a response, however, Andrew's body did not respond. This "evidence" from his tests was then sent to top dysautonomia specialists in New York and Rochester who were asked if Andrew had dysautonomia. The resounding answer was most definitely yes.

Andrew has an unidentified form of Degenerative Dysautonomia clinically identical to Familial Dysautonomia, a genetic disease found primarily in Ashkenazi Jews. The type of dysautonomia that Andrew has is very rare, with less than 400 cases at this time, and you will not find much information on the internet, which primarily references milder forms of autonomic dysfunction such as POTS, or autonomic issues that are part of different diseases such as mitochondrial diseases. Andrew's form of dysautonomia is part of a class called Hereditary Sensory and Autonomic Neuropathies (HSAN), a collection of genetic diseases that vary in severity but are all characterized by loss of pain perception and temperature regulation.

Andrew's HSAN most closely mirrors the severest form of Familial Dysautonomia, also called Riley Day Syndrome, but he is negative for the mutation associated with the disease. This makes his presentation even more rare and the road ahead even more unknown. Andrew's dysautonomia is an unknown degenerative disease that, each year, takes more pieces of the precious boy that we love.

Treatments and Decisions

We began to use symptom management medications that are used in children who have Familial Dysautonomia. These medications and protocols have greatly enhanced Andrew's quality of life in that we can now help his body through his "autonomic crises" at home, instead of in the hospital for weeks at a time as we did when he was young.

Andrew currently relies on IV fluids daily, G tube feeds, and IV medications. He has a surgically placed IV catheter in his chest and his bladder drains into a diaper through an opening in his abdomen called a vesicostomy. He is on oxygen frequently and relies on BiPAP every night to keep him breathing. His heart rate slows and his blood pressure drops very low, requiring intervention to stop a devastating event. He has a greatly inhibited pain response now and has suffered burns to his skin and cuts to his eye without any awareness of injury since he does not respond to surface pain. His development has been greatly affected and, at the age of almost ten, in most areas Andrew functions more like a preschooler. In reasoning and self care, he is more like a toddler.

Due to the way his dysautonomia affects his intestinal system and the fact that he has an indwelling IV line, he has suffered from several life threatening sepsis infections. His endocrine system is becoming affected more and more, causing him to be unable to release the stress hormones that his body needs, and requiring us to give the hormones to him intravenously. His blood sugars are erratic as the messages to release insulin at the appropriate times are not occurring correctly, causing highs and lows in blood sugars. His ability to eat and to swallow is becoming more and more impaired, and his ability to walk and move is deteriorating as the sensory nerves in his body are being destroyed.

We have learned to live with nurses in our home on an almost daily basis. Andrew's bedroom is a mini ICU ward and our medication cabinet would match any pharmacy that I know. My husband and I have both earned, or should have earned, honorary RN degrees. While the first big decision we made was to get a G tube, we are now confronted regularly with, "how many interventions do we add?" "How far do we go?" "What is best for Andrew?" And, "what life saving measures do we wish to have taken?"

Through the world of the internet I have met a handful of other children who have dysautonomic diseases at the extreme end of the spectrum similar to Andrew's diseases. These children probably have diseases that are nameless, what doctors call orphan diseases. Every affected child has the amazing ability to persevere through life-threatening events, sometimes on a daily basis, and to steal the hearts of almost all who meet him or her. Their diseases may be unknown, but the children who have severe Degenerative Dysautonomias will not be forgotten as their will to survive is unforgettable.

The author with her son Andrew

Alison is mom to four children (and a dog) ages 15, 14, 12 and 9. She lives with her husband and children in Canada. Alison is a stay at home mom. She can be contacted via email at boet64@telus.net