

Personal Story

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Periodic Paralysis: Andersen-Tawil Syndrome: An Overview With History/Attacks/Symptoms of Susan Hunter

I have had episodes of partial and total paralysis for many years. During the episodes, my potassium shifts are low (hypokalemia), high (hyperkalemia) and within the normal ranges (normokalemia). Due to several misdiagnoses and a lack of proper diagnosis and treatment for over 50 years, I have become totally and permanently disabled with weak muscles throughout my body including those involved with my breathing and my heart. I must be on oxygen constantly and cannot exert myself in any way. I have had a heart loop monitor inserted in my chest to monitor the tachychardia and arrhythmias which include long QT interval beats. Although, I have become totally disabled, most people with the condition will not.

My Story

As a child, I was considered “sickly” by my father and mother. The “sickliness” continued into my adult life and caused me to retire in my early 50’s from a teaching career when I was deemed “disabled” by my doctors and Social Security. Through the early, difficult years, I bore and raised 4 children, each with their own weaknesses and illnesses. (One passed away at the age of 5). Each pregnancy was difficult and a 5th was forced to be terminated due to severe illness and weakness. The doctor told me I would die if I didn’t have an abortion.

Since retiring, I have continued to gradually decline. I now need to use an electric wheelchair part of the time and suffer from periods of partial and total paralysis.

I have been diagnosed and misdiagnosed with diseases from MS to Inherited Peripheral Polyneuropathy. I have been tested and studied for diseases from Friedreich's Ataxia to ALS and Parkinson's. A muscle biopsy ruled out mitochondrial diseases and myopathies. Neuromuscular diseases have also been ruled out.

Many other members of my family suffer from similar symptoms and are also undiagnosed. These symptoms can be traced back to my mother and her father. One of my daughters has symptoms also, making 5 generations that I know of for sure.

By a process of elimination and based on my symptoms and those of other family members, I now finally know what has made me ill my entire life and has affected other members of my family. We have a familial type of Periodic Paralysis (PP); Andersen-Tawil Syndrome (ATS).

Although it is not a true muscular disease, Periodic Paralysis is listed under the umbrella of diseases of the muscle because the muscles are affected and weakened during episodes of partial and total paralysis. It is recognized by the Muscular Dystrophy Association and research is partially funded by them. It is a very rare disease with only about 2,700 cases in the United States.

One nurse practitioner once told me, when discussing a diagnosis, that we were looking for a "zebra" because it wasn't a typical "horse". Andersen-Tawil Syndrome is not only a "zebra" but a very rare "zebra". I have the difficulty breathing, speaking and swallowing and heart arrhythmia (occasionally) during some attacks. This is considered rare, so not only do I have a rare "zebra" but an extremely rare "zebra": Andersen-Tawil Syndrome. Only 100 people world-wide are diagnosed with ATS.

The symptoms have changed over the years and looked like many diseases, because of the odd things that most medications prescribed to me, to treat my symptoms and diagnosed conditions over the years, did to me. I have had symptoms from passing out to ataxia; over active bladder to inability to handle anesthesia; weak legs to total body paralysis; low blood pressure to high blood pressure, polyneuropathy to no neuropathy; diarrhea to severe constipation; hypoglycemia to type II diabetes; mitral valve prolapse, to no mitral valve prolapse (when diagnosing numbers were changed); constant nausea to overeating; sleeping too much to not being able to sleep; numbness and tingling of hands and feet to extreme pain in hands and feet; constant tightness and pain in the calves that never goes away to very few headaches; sleeping pills keeping me awake and beta blockers causing ataxia; statins making the tightness and pain in calves excruciating to muscle relaxers causing seizures; etc.....

These symptoms and changing of symptoms have been very confusing to myself, my family and the medical professionals in my life and made a diagnosis difficult. With all of the testing done and everything ruled out and now being off of all medications, the real symptoms have finally emerged and it is very obvious what has plagued me for my entire life.....Periodic Paralysis, possibly Andersen-Tawil Syndrome.

I have studied the things that cause the paralysis and the things to do to stop them. I have changed my diet and began to ingest potassium citrate when I feel an episode beginning. I dilute the 99mg capsules in a cup of water and drink about ½ of the contents. Within minutes the episode will stop and I begin to feel well again. This lasts for about 2 to 4 hours before I need to do it again.

I continue to have episodes during the night and wake up at different times, paralyzed. When I am able, I take the potassium citrate and I will usually fall asleep feeling well rested when I wake up with little or no paralysis.

(On October 31, 2010, after taking an antibiotic, I went into metabolic acidosis and began to have periods of high potassium; hyperkalemia and shifting of potassium in the normal ranges causing total paralysis. I was unable to take the potassium to stop the episodes. I changed my diet to a ph balanced diet and went on oxygen and as of Jan 2011, I am back to the low ranges of potassium shifting. I now take potassium citrate to control my paralytic episodes. I have them only during sleep now)

I was finally diagnosed on February 7, 2011 at the age of 62. I felt the need to do this, for my siblings, children, grandchildren and great-grandchildren. I would love for them to avoid what I have been through and get help early to learn how to live with and treat this disease so that they will be able to live a much fuller and more enjoyable life than I, my mother, my grandmother, brothers and daughter live/lived.

The following letter was written to my family after the first visit to my electro cardiologist in January of 2011.

Dearest Family,

I saw the electrocardiologist today. I will try to make this as short and as easy to understand as I can, although it is complicated.

It looks like I have a serious heart condition secondary to (caused by) a channelopathy:

(Channelopathy: A disease involving dysfunction of an ion channel.

Channelopathies are known to involve the ion channels for potassium, sodium, chloride and calcium.)

Periodic Paralysis is a channelopathy.

(A group of diseases marked by episodes of muscular weakness or flaccid paralysis without loss of consciousness or sensation.)

Andersen-Tawil Syndrome (ATS) is a channelopathy disease and a form of Periodic Paralysis. It is hereditary.

Periodic Paralysis is also considered a rare form of Muscular Dystrophy and recognized by the Muscular Dystrophy Association. When one is diagnosed with Periodic Paralysis...any form.... you can receive testing, medical treatment from the MDA doctors, equipment, etc, from them for free or in conjunction with your insurance in most cases. This may be an option for those of you with no insurance for diagnosis and treatment.

The dysfunction of the ion channel in my body has caused potassium to shift into my muscles for most of my life, causing partial and total paralysis. Now that I am 62 (at least 50 years of this in my body), this has caused severe damage to my muscles (can no longer walk more than a few steps) and many of my organs. The electrical part of my heart has been affected and causes me to have a too fast heart beat, several types of irregular heart beats, and a long QT interval heart beat. This all gets worse when I am in paralysis (several times a day and during the night). This combination keeps me out of breath and in a very weakened state. It is not treatable for me. I am not a candidate for a pacemaker (the doctor told me). I have been put on oxygen, 24 hours a day, to ease some of the symptoms. Sudden cardiac death is the most devastating complication of the long QT syndrome due to Ventricular Fibrillation (V Fib).

Because of this, the cardiologist says my heart needs to be monitored 24 hours a day, 7 days a week. So on Feb 7, he is going to put a heart monitor of some kind in my chest under the skin that will record my heart beats and send them to him. In three years it will be replaced for another three years, etc. He discussed the possibility of using a defibrillator if need be (later)....I believe it is implanted.

He is somewhat knowledgeable about Periodic Paralysis, but not about Andersen-Tawil Syndrome. He believes that it is a good possibility from my symptoms and the family members' symptoms that it is ATS. He is going to study everything he can about ATS between now and the procedure and said that he hopes to give me a "definitive diagnosis" then.

He talked about how most doctors will never see a case of Periodic Paralysis in their life time, and to see someone with ATS is even more unlikely, (why it has been misdiagnosed all of these years) but he is really excited about it and is very anxious to find out if there is any way he can help me/us.

I told him I most wanted a diagnosis for YOU, MY FAMILY. I want all of you to know about this disease and get the appropriate treatment so you don't end up like me. I have very little quality of life left. I told him I wanted all of you and the children to come, to have better than this.

So, for now, it looks like we have a diagnosis of probable Periodic Paralysis and probable Andersen-Tawil Syndrome, from an electrocardiologist. This is my gift to you.

For me, I am extremely ill and will not get well (his words). He said that there is a chance that he may be able to improve the quality of my life somewhat....but no guarantee.

For all of you...if you have symptoms, please let your doctors know that this runs in the family (Especially the long QT Interval). Please learn all you can about it. You, your children and grandchildren may very well have it. Please consider getting genetic testing. A doctor in Germany is doing genetic testing for all of the Periodic Paralysis Diseases. My blood has already been sent to him. He does the testing for free, if we pay the postage. The more of us that send blood to him, the better chance they have of finding the marker for our family.

For those of you who don't have it or don't believe you have it, please know that you could be carriers and may have passed it on to your children.

Grandma Lahlee was the probable carrier and it probably came from her father's side (Duggins), as some of that family also have symptoms. So, any descendant of Lahlee Duggins Knittle may have this or be a carrier and /or carry some of the characteristics. Butch, Brud, Kristen and Shari most likely will also be diagnosed eventually. They have most of the symptoms. Several others are also displaying problems with potassium, palpitations, tachychardia, fainting, muscle weakness and have some of the characteristics, like webbed toes.

Several studies are being done and our family may be able to be studied and possibly get in on trials for medications, treatments, etc. I will keep you informed about this.

The cardiologist today, says I now need to see a nephrologist (kidney specialist). We will be setting that up right away. On the 27th of this month I will return to the neurologist who also believes I have Periodic Paralysis. He may also diagnose me as definite on that day. We are looking for a new Primary Care Physician who knows about this disease and can coordinate all of my treatment and needs. The cardiologist told us we need a doctor who deals with channelopathies, but they would only be available in large cities and medical centers.

Please let me know if any of you want to know more about the disease, etc. I have much to share.

For now, I will just say I love each one of you and hope for only good health for you.

Hugs and much love, Mom, Grandma, Aunt Susie, Susie, Susan, Suse

The following is the letter written after my procedure and my final diagnosis.

Hello Family and Friends.....I made it through one of the toughest days of my life.....I had a heart loop recorder inserted in chest with only lidocaine to numb it, because I cannot tolerate any meds. I told them the lidocaine would send me into paralysis, so they used a type without epinephrine and felt assured it would not cause paralysis. As luck would have it (and I knew it) 1/2 way through the procedure, I went into paralysis. By that time the device was already implanted. The doctor and rest of the team were watching my heart doing it's thing (tachycardia, arrhythmias) saying things like, "Look at the huge T-waves" and reading off numbers that I didn't understand and ohing and awing. He proceeded to explain my disease to them. Then when I was able to answer questions, they all began to ask me questions about it...remember.... what I have, Andersen-Tawil Syndrome.... only 100 other people, world-wide, have been diagnosed with.

As they were ready to take me to recovery, I noticed an IV drip in my arm. I got horribly upset and asked what it was. They told me "saline". I swore and told them I was not supposed to have that. They then removed it. What I did not know was that it had been on me the entire procedure. I thought they had just hooked it up and then took it off after I told them that.

I went back to recovery and was doing fairly well, except for slipping in and out of small paralysis episodes.

My heart doctor had put together a "team" for the rest of the procedures they were going to do for a confirmation of how my potassium shifts and how to treat it. The plan was to put me in ICU and have a kidney specialist direct the testing and an Intensivist to monitor all signs and symptoms and be there to treat my symptoms which could include my heart stopping.

I called Shari to tell her how well I was doing and tell her the plan to pass on to everyone.

Just as the kidney specialist showed up to tell me the plan and ask me a few questions, I began to have trouble answering his first question. I slipped into the worst episode of paralysis to date. My heart began to beat at a sustained heart rate of 130 to 140 bpm for over the next hour. My blood pressure was at 168/80. I felt like an elephant was on my chest. I could feel and hear my heart racing and the horrible pain from it. I could not have any pain meds or any meds to slow my heart because it would have made it worse. Everyone was in astonishment watching the heart monitor and not knowing what to do. Calvin was very upset and I thought I was dying. I could hear everything but was unable to open my eyes, or speak. I could not move, just hear everything and feel all the pain and pressure. The doctors kept saying they had never seen anything like it and the nurses kept touching my hands, arms and face, telling me they were sorry they couldn't help me.

(The saline drip caused this...if you have PP/ATS never let them put a saline or glucose IV in you)

My heart finally began to slow down over the next hour. I was finally able to open my eyes and could speak a little. Finally, I was doing very well. The doctors decided to proceed as planned and told us the plan to put me in ICU and load me with carbs to start the paralysis again and then test my potassium levels through the paralysis process, etc.

They got me upstairs and had me ready to begin the next phase. Calvin left to go home and get some much needed rest. The kidney specialist came in at that point and said, "We have decided not to proceed with the testing and are sending you home now." I asked why? His reply floored me....."We don't need to do anymore testing...there is no doubt you have Andersen-Tawil Syndrome (ATS). Everything we have read tells us that you could die if we do the testing we were

going to do, if you have ATS. We are going to give you Diamox to treat your paralysis symptoms, but because you have ATS it may not work for you....but we want to try". I thanked him and told him I already knew that but was taking the chance for the little hope I had that the meds might help me...otherwise there is not much else I can do for a better quality of life.

I already had the diagnosis of Periodic Paralysis (PP) from my neurologist, but now I had the diagnosis of the type of PP.

He told me how humbled he felt to be diagnosing me. He said he was just an ordinary MD..nephrologist...not like the big and powerful doctors who wrote all the info on PP and ATS back East. But after seeing what happened and going over all the facts and by the process of elimination and studying all of the latest research I had presented to them and the family flowchart I had put together with all of the family medical history and all of your input....including Kristen's toes!!!.....he felt he had no other choice. He told me I had done all the work and had done an excellent job of presenting it all to them. Without that, I might never have got the ATS diagnosis also. He had never heard of ATS before, none of them had, but they had been researching and studying it.

Thank you Family, for answering the little questionnaires' I sent out and all of your input and patience with me over the years.

He has dictated a report which each of you will get. You can then give it to your doctors. My children have a 50/50 chance of having it and of passing it on. All of you must get checked. The Long QT interval heartbeat is nothing to ignore.

Thank you all for your best wishes and kind words and thoughts.

I've heard it referenced by some people with the disease, that Periodic Paralysis is our "friend". I must disagree with this ideation. Periodic Paralysis is not our

“friend”. After all, a friend does not leave you totally paralyzed. A friend does not leave you unable to walk. A friend does not leave you unable to talk. A friend does not leave you in fear. A friend does not take away your quality of life. We cannot treat it as our friend. We must treat Periodic Paralysis as an enemy that we must battle daily, minute by minute; second by second. By doing so, it is possible to keep our “foe” regulated in order to decrease the number and severity of paralytic episodes and the amount of possible permanent damage. We must walk a constant “tightrope” of sorts.

["Walking the tightrope" \(managing and treating the disease\).](#)