

Periodic Paralysis and the Family “IT’S JUST THAT SIMPLE”

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On February 7, 2011 I had a heart loop monitor placed in my chest. This was done to keep track of my tachycardia and arrhythmias, including my long QT interval heart beat with the intention of possibly getting an implanted defibrillator in the future, and to assist in confirming a diagnosis of Periodic Paralysis (PP): Andersen-Tawil Syndrome (ATS). After the procedure, I was going to be sent to the intensive care unit and provocative tests were also going to be performed on me. The tests I was facing, were not going to be easy because I would end up paralyzed temporarily and they could possibly elicit deadly heart beats. I was not able to use any medications, due to the possibility of paralysis or the long QT heartbeat it can cause. I was administered only lidocaine topically and so was very much awake during the procedure. While I was lying on the operating table and Dr. P was inserting the monitor under the skin, he said to me, “I am so impressed with how brave you are to be doing this”. I started to cry and answered by saying to him, “I am doing this for my family.” “I have to get the diagnosis for them.”

I was willing to go through surgery with no painkillers or anesthesia and the other tests, no matter the risks, because I love my family, every one of them. So many of them have similar symptoms and need a diagnosis to receive the proper medications and treatment. I knew that once I had my diagnosis, the others could get theirs. I had to do it. I kept thinking of them as I became paralyzed on the operating table (from the lidocaine) and later in recovery after I was mistakenly given a saline IV drip. This sent me into the worse episode I had in my life. During the paralysis, my heart rate was over 140 bpm for an hour and then lowered a little for another hour. I feared I was dying, but if it meant a diagnosis was finally possible, I would do it again if necessary. I want all of them to be able to get treatment for their disease before they get as disabled and ill as I am. I did get my diagnosis that day at the age of 62.

As we know, a family is any group of persons closely related by blood, such as parents, children, brothers, sisters, uncles, aunts, and cousins, nieces, nephews, grandparents and grandchildren. A family is a group of persons sharing a common ancestry. A family is two or more people who share goals and values, with long-term commitments to one another. A family is all those claiming descent from a common ancestor; tribe or clan. A family is a lineage that can share a hereditary disease. My family is one of those. We share the hereditary channelopathy disease called Periodic Paralysis: Andersen-Tawil Syndrome. The family I am writing about is the descendants of Lahlee Duggins, my mother. She no doubt had PP: ATS, though she never knew it before she passed away a few years ago. She had been misdiagnosed with many things, however, with my diagnosis, it is clear what she suffered from all her life.

It probably came through her father, Louis Duggins, because my mother's sister, and 4 half sisters share some of the traits and symptoms as does their children, grandchildren and great-grandchildren. I can trace this disease through 6 generations. Louis Duggins and his 4 brothers all died of heart problems, some of them young and sudden indicating the possibility of the long QT interval heart arrhythmia. One was 41, he died during a paralysis attack when his heart suddenly stopped, and another was 51. Their only sister suffered most of her life with similar symptoms to my mother and me. My mother and her full sister had episodes of total paralysis and my mother was totally disabled from weak muscles before she died. I have two brothers who are as disabled as me and have episodes of paralysis but with no diagnosis, but we know now that they have ATS as well. Two of my children have symptoms as well as 3 of my grandchildren. One of my brothers' daughters has symptoms as does her children. The children and grandchildren, of one my brothers, have symptoms and characteristics. It is obvious that some who don't believe they have it are carriers, because their family members have some of the characteristics.

Family has always been important to me. I am the family historian and have been working on our genealogy for the past 40 years. I have three brothers; I was the only girl. My father had 12 siblings and my mother had 6 siblings. As a child, my aunts, uncles and cousins on my father's side were an integral part of my everyday life. Every holiday and vacation was spent with our extended family. Weekends always included at least one uncle or cousin. Some of my father's younger brothers lived with us after they left home, before they settled down with their own families. We had so much fun at each and every family gathering. We had at least 3 family picnics a year. We barbecued our hot dogs and hamburgers, ate great food, sucked on snow cones, laughed, teased each other, played games, sang to the music of our uncles' guitars, went for walks, watched the trains go by, ran up the hill to see when the next one was coming and listened to the adults tell stories of their childhood. On the outside we were a happy and loving family.

However, many of us had health problems. On my father's side there was back problems and diabetes affecting some of the uncles. At least four of them were in constant back pain, including my father. My mother was sick much of the time with many issues, but always put on a happy face. Though a few family members referred to her as being "hypochondriac". As she got older, more and more health issues appeared. We did not live near her family, but did have gatherings with her family once or twice a year. Her mother had a stroke at age 50. Most health issues were not discussed or known on that side because they were Christian Science followers and no doctors were ever seen.

As children, my brothers and I suffered from growing pains, leg problems, every childhood disease, tonsillitis, one brother had dyslexia and was behavior disordered, one brother had a speech impediment and problems with organization and one brother was the class clown and graduated at age 16. I wet the bed until I was 12, was overweight, had a foot that turned in, could not keep up with other kids on the monkey bars or any sports, could climb up a tree but could not get down, cried all the time, was very uncoordinated, began passing out at the age of 11 and as a teenager began to have pain in

my back and joints. My family looked on all of this as normal. We each had problems and our parents all had problems. It was just our life.

As we grew up and left home and began our own families, we began to spread out across the country and were not close like we had been growing up. Priorities changed and many of us lost touch. Our parents' health declined through the years, especially my mother. My health was always an issue and so were two of my brothers during those years. One of my brothers did not seem to have any health issues, though his wife did. As we began to have children, many of them were born with health issues or developed them. As we got older and sicker, we began to communicate more than in previous years. Soon we discovered that many of our symptoms were similar. Two of my brothers and me, as well as our mother, were deemed disabled by the age of 51 after years of medical decline. Sadly, many of our children began showing these same symptoms.

It was clear that this illness was something hereditary. I had to find out what it was. It was now affecting a fourth generation. I was misdiagnosed over and over, but I continued to research our symptoms. It took many years, but I finally came to the realization that we had a disease called Periodic Paralysis. The type we had was a very rare type called Andersen-Tawil Syndrome.

Once I finally discovered the name of the disease that was disabling my family members and me, I set out to get diagnosed. As part of my plan to do this, I created a family flowchart listing all family members and their symptoms over 6 generations. I wrote the following as part of the chart for my doctors the doctors who are treating and diagnosing my family members.

This flowchart lists only the symptoms and characteristics of Andersen-Tawil Syndrome for each family member who has or had them. Unfortunately, many other diseases and conditions co-exist or co-existed with the listed symptoms and characteristics for many of these family members. They need to be sorted out and recognized for a correct diagnosis, so the family members who have Andersen-Tawil Syndrome can get the proper diagnosis. This is imperative so each family member with Andersen-Tawil Syndrome can receive the proper medications, treatments and assistance from the proper organizations and so they can take part in the studies for Andersen-Tawil Syndrome.

Some of the other diseases have masked the Andersen-Tawil Syndrome and have been the cause of misdiagnoses such as fibromyalgia and multiple sclerosis. Odd effects of certain medications have also caused symptoms resembling other conditions such as ataxia and seizure-like episodes. These misdiagnoses have led some family members to be in their 50's and 60's now (and some already passed away) still attempting to find out what has disabled them by gradual muscle weakness, partial and total paralysis and heart problems since their childhood or teen years.

Some of the co-existing diseases include: degenerative disk disease; osteoporosis, especially of the spine; arthritis, especially of the spine; peripheral poly neuropathy; type 2 diabetes; restless leg syndrome; asthma; strokes; depression; allergies; migraines; learning disabilities; and cyclic vomiting syndrome.

This family has been plagued with serious health problems and disabling conditions through 3 generations and is now beginning to manifest itself in a 4th generation. Please help this family by considering the strong possibility of Andersen-Tawil Syndrome.

It has recently been discovered that another branch of this family also has these conditions and characteristics. Due to a time restraint, this family was not added to this report. If needed, this information can be added at a later time.

The flowchart and attached letter were instrumental in my final diagnosis and I hope it will help the others in my family in need of a diagnosis.

Part Two: Period Paralysis: My Family Dynamics and a Lack of Understanding

Unfortunately, there are members of my family, who do not understand or who think some of us who are ill are hypochondriacs or malingerers. There are also some members who do not believe they have it, but clearly they are symptomatic. There are others who do not believe in a hereditary disease in our family, but they are carriers because their children and grandchildren are symptomatic and have some of the characteristics. There are others who stay away because they do not want to watch my decline and that of other family members. There are those who believe we need only to have “good thoughts” and we will get well. Others believe prayer is the answer. Others believe it is best to keep my grandchildren away for fear that the children will see us and be afraid that they will end up like us, by the power of suggestion. I was even given a book by a family member explaining that all of my illness was due to choices I had made in a prior lifetime and that I chose and welcomed it on a subconscious level.

This is so disheartening to those of us who are sick and disabled. We are suffering and watching our children suffering with fear for our grandchildren now. We are not liars. We did not choose this. We did not ask for it. Prayer will not cure it. Good thoughts will not make it go away. Keeping my grandchildren away will not keep it from happening to them if they were born with it. The power of suggestion will not disable them. I am sick and continue to decline and may die soon. Do not stay away too much longer. Please do

not keep my grandchildren away; they will miss knowing me due to a baseless fear. They need to know me and understand the disease...they may have it. Some of you have all the symptoms, please get help now so you do not get as sick as me. Some of you do not understand heredity...it is scientifically based and proven. This disease, Periodic Paralysis: Andersen-Tawil Syndrome, is an inherited disease. One has it only if it was passed on to them through one or both of their parents.

To my family:

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 vision, digestion, breathing and my heart. I must be on oxygen constantly and cannot exert myself in any way. The electrical workings of my heart are defective. I have had a heart loop monitor inserted in my chest to monitor the tachycardia and arrhythmias, that includes long QT interval beats. I now spend my days in a recliner, unable to walk farther than across a room. I must use a motorized wheelchair for anything farther. If I did not have the help of my husband, I would have to live in an assisted living program.

Through the past years of my physical decline, I have had to give up my career as a special education teacher, my hobbies to include hiking, walking, swimming, exercising, fishing, camping, traveling, shopping, cooking and baking. I had to sell, and move away from, a beautiful home my husband and I built in the mountains of Utah. I had to move far away from my family in order to live in a better climate. I can no longer drive our car. I have lost many friends, because I could not keep up with them or entertain any longer.

However, the hardest losses to deal with are those of my family. I have lost contact with or have a strained relationship with family members who did not understand or did not want to watch my decline or who thought I was a hypochondriac. And, I have lost the connection I once had with my grandchildren because I can no longer keep up with them or continue a meaningful relationship with them or they are being kept away.

Please understand what it is like when I am paralyzed:

“Usually, I wake up in the morning and I am paralyzed. I find I can’t move. I can’t open my eyes. My mouth is open. I can’t breathe through my nose. I have urges to swallow but can’t so there is a choking sound in my throat every few minutes. Sometimes my heart will race or beat irregularly. My breathing stops at times. I don’t have any or much feeling in my body. My mouth is very dry. I cannot speak.

As I begin to come out of it, my mouth will start to get saliva, my eyes will open but I can only see what is in front of me, since I can’t move my head. Sometimes my eyes will jerk around when I first open them, usually

jerking up. My body will sometimes jerk a little. Sometimes there is a big breath my body will take

Sometimes, I will go back into it. My eyes close, I feel very hot and all the symptoms return. Sometimes there will be a few jerks as I go back into it.

During all of this I am awake and am aware of everything going on around me. Sometime I begin to cry, due to the frustration, and fear. I can feel the tears running down my cheeks.

If I have these at times other than upon waking, the symptoms are the same. I get a strange sensation of heat body wide, usually beginning in my back. My eyes will close and then my body goes limp. I may have a few jerks as I am going limp. My mouth will open and I am in it...unable to move, speak or open my eyes.

Sometimes, I don't go too deep. It is all the same but I am able to open my eyes and can speak a little with a tight tongue and tight lips. My mouth is still open, however. I can't move my body.

Once one of these begins, it may last up to 45 minutes to several hours, or can be as short as about 15 minutes, if it is a second or third one in a row.

It takes about 15 to 30 minutes to come out of it all the way. I am always left with lingering weakness for many hours that can linger into days. Speaking is difficult. Walking is difficult. My arms and hands come back sooner than my legs. I begin to get feeling back in my body. I can move my lips. I begin to breath thru my nose again. It is difficult to speak or move but it gradually comes back. Speech is very difficult, my lips don't want to move. My tongue is difficult to move. I will suddenly have an urgency to urinate. If, at this point, I get help to the bathroom, I am like a rag doll, especially my legs. My arms flail, like a child just learning to stand and walk; balancing herself.

For many hours, I remain too weak to do much of anything but sit up in bed or sit in a recliner. I must use my walker or a wheelchair.

I have a long QT interval heartbeat that can happen while I am in this state. If it does happen, I can go into cardiac arrest and die.

Why would I make it up, pretend, or lie about it? Why would I choose this or make it happen? All the prayers in the world will not make it stop. "Good thoughts" will not keep it from happening. Why would I have given up so much?

Please try to understand the truth of this horrible and debilitating disease. I did not choose it. It is real. You may have it. You may be a carrier. Your children may have it. The

sooner you get it diagnosed, the better chance you have of not becoming disabled. I love you and got the diagnosis to help you, your children and grandchildren.

I would like to address the remainder of this article to others who have periodic paralysis. I am in contact with many people whose family is much like mine. Periodic Paralysis is a cruel disease. It is one that is difficult to diagnose, though it doesn't need to be. Doctors, for the most part, who do not understand it, look at patients with it as hypochondriacs, malingerers or faking it. If the doctors treat us that way, is it any wonder our families may feel the same?

Some family members stay away or are very limited in their interaction with you, because they cannot face watching your decline. They may feel helpless. Some just don't know what to do or say. Some don't care or are in denial.

Others may not understand, for whatever reason. They may not have read about it. They may misunderstand. They may be stubborn about issues such as hereditary. They may not want to know. They may not care.

Many family members are living in fear that it may begin to manifest in themselves or their children. Others actually have the symptoms but are afraid to face them. They will have to sooner or later. I will be there for them or my website with all the information they may need.

I thought perhaps when I finally got my diagnosis that I had become vindicated and validated to my family. I thought they would believe me and thank me. Except for a few members who already believed me and supported me, I was wrong. The silence remains disheartening.

The way I handle it is to provide as much information as I can to them. I have the papers with my diagnosis and am happy to share them with those that are trying to get diagnosed. I am developing a website for them and others to refer to as they have questions. I try to maintain communication with them, however strained it may be. I am here for them and let them know. Beyond that, I can do no more.

The loss of our families seems to be another cruel reality of Periodic Paralysis, and as one of my brothers says, "It's just that simple!!".