

**“Seeing the Andersen-Tawil Specialist”
An Update on My Diagnosis
Periodic Paralysis: Knittle-Duggins-Alsop Syndrome
PP: KDA Syndrome
By
Susan Q. Knittle-Hunter**

It has been nearly a year since I got my diagnosis of Periodic Paralysis: Andersen-Tawil Syndrome (PP: ATS) and Calvin and I created The Periodic Paralysis Network. There have been many changes during this past year, and I felt the need to update our opening page, to reflect the changes. The following is a synopsis of our past year.

After receiving the diagnosis of Periodic Paralysis: Andersen-Tawil Syndrome (ATS) on February 7, 2011, I thought my life would take a turn for the better but I was wrong. Things got worse in many ways before improving somewhat. The first thing that happened was the doctor who diagnosed me decided he could do no more for me because I could not take diamox to treat the symptoms. I talked with my neurologist who told me that he too did not know what to do for me. And then in May my cardiologist told my husband and me that he could do no more for me either. This was due to the fact that my heart arrhythmias, tachycardia and bradycardia, did not fall into the “extreme” ranges on the heart loop monitor readings (above 180 bpm or below 30 bpm...mine fall into a range of 160 bpm to 41 bpm depending whether I am hypokalemic or hyperkalemic). Therefore I am not a candidate for a pacemaker or a defibrillator. Though my condition is serious, there is nothing any doctor can do because I cannot take any kind of medication either.

During that appointment in May, I told the doctor about my frustration and how my symptoms seemed to be getting worse and asked what I should do about my decline. He then decided to refer me to a clinic that specializes in ATS somewhere in the United States. He asked me to find it and then he would write a referral for me. To make a long and complicated story short and simple, I will explain that it was not until October 17, 2011 that I was actually able to see a specialist in San Francisco.

During those interim months, I received a letter from the genetic specialist in Germany. It stated that a genetic code had not been found for any of the known forms of Periodic Paralysis or Andersen-Tawil Syndrome in the studies of my blood. The doctor explained that the findings did not mean that I did not have a form of PP or ATS; it just meant I had a type that had not yet been found and named. (30% to 40% of all people with PP or ATS do not have a genetic code found.) The team in Germany continues to work on finding these unknown codes for the others without known codes and me. Those with ATS symptoms and without a known code are considered to have ATS Type 2. The symptoms are indistinguishable from those with the known codes.

The appointment in San Francisco lasted about 2 hours consisting of a medical assistant going over my medical records and medical history with me and then doing a physical exam. She left and conferred with the neurologist. He came in and reviewed some of the issues with me and also physically examined me.

His professional opinion was that I do have a form of Periodic Paralysis, but it is not a “**typical**” form of ATS. He said my symptoms were very subtle and I had some sensory issues that were not usual in ATS. He said that this form was obviously unique to my family and me. He joked that it would probably be named after us.

He was very kind and very willing to work with my doctors back in Grants Pass on a consulting basis. He also said he would write a letter to explain all of this to my doctors and to my family and that I would get a copy to share with them for their knowledge and for their doctors. (I have not received a copy of the letter yet) However, he did not have any real ideas that would help me, again because I am unable to take medications. He suggested a change in the amount of the potassium that I take and a change in the schedule of when I take it. He also told me that he was concerned with my lack of physical activity. He felt I should push myself a little more. Then we left feeling good about what we had discovered and the fact that we had a doctor who was willing to work with us.

As we traveled home we tried the change in amount and timing of my potassium. It did not help and I had more episodes than before. So I went back to taking it as I felt the need. I also tried pushing myself and ended up in a cycle of total weakness and in and out of paralysis for about two weeks. I have since stopped pushing myself.

I thought about his idea that because I had other things going on, that I did not have ATS Type 2. My thoughts were that maybe I have more than one disease and indeed have ATS Type 2 and something else or, most of my family members have more than PP going on, but that is probably part of this new or different type of ATS unique to my family. Maybe it is a combination of two things creating the variant.

In order to clarify this issue, I have named this form of Periodic Paralysis that is unique to my family. I am now calling it Knittle-Duggins-Alsup Syndrome or KDA Syndrome. So from now on, it will be referred to as Periodic Paralysis: Type KDA Syndrome.

I have traced it back to the Duggins line and to the Alsup line. Both families have similar symptoms going back several generations. My mother’s parents, L. Duggins and Q. Alsup married and had two daughters, both with symptoms. L. Duggins married two other women and had more daughters. Most of those women seem to display some of the symptoms and characteristics, as do some of their children and grandchildren. Q. Alsup remarried and had another child. That child does not seem to have similar symptoms, but his daughters and granddaughters do. Therefore, at this time I need to include both lines. My mother married a Knittle. Three of their four children are affected, and the grandchildren and great-grandchildren of the one who does not seem to have it, do seem to have symptoms and characteristics of it. It is the Knittle children, grandchildren and

great-grandchildren who are most affected, so I felt the need to use the Knittle name, thus, Knittle-Duggins-Alsup Syndrome or KDA Syndrome.

Since returning from San Francisco, I have finally located a primary care physician who actually wants to work with me. This is exciting since I have been through three of them since last December. We had to leave the first one due to her lack of appropriate care and the other two doctors left town due to being overloaded with patients. The doctor we found is in another town about 40 miles away near my specialists and two better hospitals. She has a background in genetics and is young and energetic. She practices in a clinic, with 81 other doctors, which is open 7 days a week. They have their own lab and radiology department and are open in the evenings as well. This will afford me the option of getting urgent care when I need it without going to the ER and easier access to a doctor when I need one.

I also have a special power wheelchair being built for me. It will include a seat that can recline at different angles to help when I go into paralysis and to alleviate the problem I have with sitting up straight in a chair. It is very taxing on me and can cause paralysis. It will have special cushions to help alleviate the pain I feel constantly. Being in the chair will aid Calvin when he has to help me during an episode of total or partial paralysis. It will give me some independence that I do not have right now.

Home health care is now an option also as part of my management plan. I have had to use them 2 times this year and will be able to call on them as needed for blood draws and other necessary medical needs that arise.

I am feeling very positive about my medical care now, but it has taken a very long time and much work to get to this point. My situation, shortly after receiving my diagnosis, looked grim due to the fact no doctors could do much for me. But, I feel better knowing that I have some good doctors who want to help me and some other great resources for my medical care. I feel the need, too, to tell others that there are good doctors and services out there so please do not give up looking.

My condition remains untreatable with medication, surgery, heart defibrillators or pacemakers, therefore my heart condition and muscle weakness including my breathing muscles will continue to progress. However, it is manageable at this time, with a proper PH balance diet, supplements, potassium, oxygen therapy 24/7; and by avoiding the triggers that start the paralysis and all exertion and stress; and by monitoring my vitals to include potassium levels, blood sugar levels, oxygen levels, body temperature and blood pressure.

I am continuing to work on my book and should finish it in the next few months. I hope to get it published and will use some of the proceeds from the sales for The Periodic Paralysis Network. My dream is to be able to incorporate into a 501 3c non-profit organization so that we can begin to accept donations. With the money from the book sales and donations, I would like to be able to provide a free kit with a cardy meter and the other necessary medical tools to any family and/or individual who needs it to monitor

their vitals for better management of this disease. I would like some of the money to go towards educating doctors on how to properly identify, diagnose and treat PP. Finally, I would like to see some of the money help those that need to travel long distances to see specialists.

I now am writing a daily blog and anyone is welcome to follow my progress. Contact me if you would like to join. It is private.

Calvin continues to research and add his findings to our website and our Facebook discussion group. We also provide as much assistance and support as possible to others with PP and to their families.

Thanks for joining us here at The Periodic Paralysis Network.