Periodic Paralysis and The MDA
“PERIODIC WHAT?…NO WE DON’T”

By Susan Q. Knittle-Hunter

This article is a difficult one for me to write. Thinking about the Muscular Dystrophy Association (MDA) in relationship to Periodic Paralysis (PP) and Andersen-Tawil Syndrome (ATS) makes me both very sad and very angry. I spent a great deal of time and effort and risked my life attempting to gain a diagnosis, with the hope that I would be able to receive medical help and support from the MDA. MDA spends money on research for Periodic Paralysis. They also pay doctors (neurologists) to diagnose and treat patients with Periodic Paralysis. It is a disease recognized by the MDA as “Other Myopathies” on their website page listed as “Diseases”:

http://www.mda.org/disease/

The following pages describe PP and ATS in great detail on the MDA Website:

http://www.mda.org/publications/Quest/q163infocus.html

http://quest.mda.org/series/focus-periodic-paralysis/focus-periodic-paralysis

http://quest.mda.org/series/focus-periodic-paralysis/periodic-paralyses

http://quest.mda.org/series/focus-periodic-paralysis/pp-it%E2%80%99s-all-muscles-not-head

I was devastated when I discovered that I was not going to be able to receive that help and support which I so badly need and deserve and they are being paid to provide to me. I am not the only one in my position. I know that many other people with PP are not receiving treatment either.

The MDA pays doctors (neurologists) who know nothing about PP and ATS. They do not recognize it. They do not understand the disease nor do they know how to diagnose or treat it. Many are using outdated information and therefore many people are not being diagnosed.

Some of them seem to be involved in a conflict of interest by receiving money from pharmaceutical companies, also, for trial medications and or research to treat PP. They are only accepting people who are genetically diagnosed and fit the criteria for
the studies. This excludes 30% to 40% of all people who have PP and/or ATS. The statistics clearly indicate that only 60% to 70% of the genetic mutations have been found for people with PP and/or ATS. So others without a genetic code are not being diagnosed or treated. One of these doctors was heard to say, as he excluded a woman who clearly has ATS and had traveled 100’s of miles, “It is better to under diagnose than over diagnose” to his assistant. So this woman goes undiagnosed and untreated.

These doctors only want pure PP/ATS. If anyone has other diseases coexisting with PP/ATS, they are also denied a diagnosis, as they will taint the pharmaceutical studies and data. So, not only do some of us have PP and/or ATS, but we have other diseases as well and are worse off than those with the “pure” genetically diagnosed cases.

Is this any way to treat deathly ill and desperate human beings who are in search of medical treatment?????? Why is the MDA allowing their doctors (neurologists) to mistreat, misdiagnose and turn away people who have diseases for which they clearly advertise to treat?

Some of the MDA Clinics with neurologists who do not understand how to recognize, diagnose and treat PP and/or ATS are: Portland, Oregon; Medford, Oregon; UCLA, Los Angeles, California; Spokane, Washington; and San Francisco, California. Most of these even denied on the phone that they treat Periodic Paralysis or Andersen-Tawil Syndrome. I had to send them emails with the information from their own website in order to educate them!!!

I called the MDA National Headquarters in Arizona and confronted someone from their clinical services office. She seemed upset to hear my story and was willing to help. She took notes and asked me to put my story and complaint in writing. I wrote a 27 page letter (which follows on page 4). I anticipated hearing back from her within a few days. It was more than a week, before I heard from someone else from her office. Another woman called and apologized for the bad experiences I had received from their doctors. Her solution was for me and my other family members to drive hundreds of miles out of our ways to go to other clinics.

What would be wrong with getting rid of those doctors and hiring doctors who know about Periodic Paralysis and Andersen-Tawil Syndrome? Doctors who have been trained appropriately? Doctors who know how to recognize the disease despite one’s age or other co-existing conditions? Doctors who are in a conflict of interest because of their dealings with pharmaceutical companies? Doctors who have a pleasant bedside manner? Doctors who do not have such large egos that they will not listen to their patients or be willing to learn from them? Doctors who genuinely care about the suffering of their patients? Doctors who take seriously the Hippocratic Oath they took”??
The Hippocratic Oath is an oath historically taken by doctors swearing to practice medicine ethically. Here are the six values which commonly apply to medical ethics:

- **Autonomy** - the patient has the right to refuse or choose their treatment.
- **Beneficence** - a practitioner should act in the best interest of the patient.
- **Non-maleficence** - "first, do no harm"
- **Justice** - concerns the distribution of scarce health resources, and the decision of who gets what treatment (fairness and equality).
- **Dignity** - the patient (and the person treating the patient) have the right to dignity.
- **Truthfulness and honesty**

http://en.wikipedia.org/wiki/Medical_ethics

I still hold out hope that the MDA will do the correct thing and either appropriately train (I will do it if they want to pay me) the existing doctors in charge of their clinics or hire new doctors. If not, I am requesting that they remove Periodic Paralysis as a disease that they diagnose and treat. There is no sense in disappointing and discouraging any more of us with this disease. Periodic Paralysis is already cruel enough to live with, without having to deal with the cruelty and indignity of the MDA doctors.

Maybe the MDA can put the money they pay these doctors and give for researching genetic codes, into another type of program; perhaps clinics designed specifically for Periodic Paralysis diagnosis and treatment. Hiring doctors who specialize in channelopathies rather than neurologists would be a great place to start. After all, it is not a neuromuscular disease. Neurologists are good only for ruling out the neuromuscular diseases.

The following is the letter I wrote to M@ Muscular Dystrophy Association – USA. I gave her as much information as I could about my diagnosis for obvious reasons and about what it is like to live with Periodic Paralysis: Andersen-Tawil Syndrome, because a few people at MDA believe it is a disease in which a person has "occasional muscle weakness". If that is all that happened, than we would not care for or need treatment!!!!!!! Periodic Paralysis is a devastating disease that can be fatal.
M,

Thank you for listening to me the other day on the phone. I hope you can understand my concern and sadness that my experience with the MDA in Oregon has been so unsettling; both in Northern and Southern Oregon.

I have worked diligently for many years to get a diagnosis and treatment for the ailment that has cruelly stolen the quality of my life. I had to give up a wonderful career as a Special Education Teacher. I had to sell, and move away from, a beautiful home in the mountains of Utah. 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. I am on oxygen 24 hours a day and the electrical workings of my heart are defective. I have tachycardia, bradycardia, arrhythmias to include long QT interval heartbeats (from which I can go into cardiac arrest at any given moment) and have had a heart loop monitor implanted in my chest.

The most difficult part of this, for me, is knowing that I may not have became this bad if just one of the doctors I have seen in the last 6 years in Oregon and the many years before, would have taken me seriously. One of the doctors I am most angry with is Dr Ali, the neurologist for the MDA in Southern Oregon. (It was suggested to me that I contact an MDA doctor by other Periodic Paralysis (PP) patients who have been successfully diagnosed and treated by them in other parts of the country.) His attitude of my being “too old” and his lack of understanding about diagnosing Periodic Paralysis correctly, set me back nearly six months, during which time I could have gotten the medical help I needed. When I saw him October 18, 2010, I was not as bad physically as I am now.

He started by telling me, “It is not usual for women of your age to be walking into a neurologists office and asking if you have Periodic Paralysis”. He told me he would not diagnose me with PP unless my potassium levels were below 3.5 during an episode. When I had the blood test done during an episode (on the wat home from his appointment…50 miles away), the soonest my husband could get me to the lab was about ½ hour after the episode started. The lab technician actually came to the car to take my blood since I was unable to walk, talk, etc. The results 2 hours later showed a problem with high creatinine levels and potassium was 3.8. He did not bother to call for 8 days, though he knew the results within 2 hours. Someone in his office called and said the
results were “unremarkable”, although there was a problem with the creatinine levels being too high. He never even bothered to let my PCP know of the problem. Luckily, I got a copy of it from the lab myself.

The doctor told me to get my blood drawn during a total paralytic episode. This is the result 25 minutes into it. He said that unless the potassium was below normal, he would not diagnose me.

<table>
<thead>
<tr>
<th>Test</th>
<th>Value</th>
<th>Reference Range</th>
<th>Units</th>
<th>LC</th>
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<td>132-143</td>
<td>mEq/L</td>
<td>UC</td>
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<tr>
<td>POTASSIUM</td>
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<td>3.5-5.1</td>
<td>mEq/L</td>
<td>UC</td>
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<td>6.0-20.6</td>
<td>mg/dL</td>
<td>OC</td>
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<td>SULFATE</td>
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<td>84-102</td>
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<td>OC</td>
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<td>gr/dl</td>
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<td>3.5-4.8</td>
<td>gr/dl</td>
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<td>A/G RATIO</td>
<td>1.6</td>
<td>1.1-2.4</td>
<td>OC</td>
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</tbody>
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ESTIMATED GFR Reference Range:
- GFR < 60: Chronic Kidney Disease, if found over a 3 month period.
- GFR < 15: Kidney Failure.
- For African Americans, multiply the calculated GFR by 1.21.
- GFR calculation is not valid for patients under age 18 years.
- For patients over age 70 please interpret results with caution as results have not been validated for this calculation method.

COMPLETED REPORT FAXED TO 10414880843 ON 10/19/2010 16:37
Pacific Time: LACDD
While in his office, my husband and I had tried to explain that potassium did not have to shift below normal to prove PP. His info was old. The following information will explain about potassium levels in Periodic Paralysis:

**Laboratory Studies**

During an attack, there is usually, **but not always**, a measurable fall in levels of serum potassium, but in some patients the K+ level may never fall below normal. Johnsen’s series of provocative studies recorded an episode of weakness of 11 hours duration provoked by a 0.3 mmol/l fall in the serum K+, and an episode of total paralysis of 19 hours duration provoked by a one point drop. During the attack there is urinary retention of sodium, potassium, chloride and water. Base your decisions on your patient’s strength and cardiac signs, not on serum potassium levels alone.

http://www.hkpp.org/physicians/hypokpp_er.html

**How low is low?**

The question of **how low** serum potassium must be to qualify as diagnostic of Hypokalemic PP is one that is constantly raised by physicians, and is the first question we will consider.

"Physiologic basis of flaccid weakness is inexcitability of muscle membrane (sarcolemma). Alteration of serum potassium is not the principal defect in primary PP; altered potassium metabolism is a result of PP. In genetic and thyrotoxic PP, flaccid paralyses occurs with relatively small changes in serum potassium, whereas in secondary PP serum potassium levels are markedly abnormal."

Naganand Sripathi, MD, Director of Neuromuscular Clinic, Assistant Professor, Department of Neurology, Henry Ford Hospital

“It becomes evident to those who read the entire body of literature that while some patients with HypoKPP exhibit serum potassium readings of 2.0 and below during episodes, others may experience paralysis and arrhythmias with K+ readings which are still within normal limits. The determining factor is not the level of the serum potassium written on the lab slip but the condition and response of the patient.”

http://hkpp.org/physicians/dxing_hypokpp.html

In primary HypoPP the K level is low during attacks. In HyperPP, K levels may be elevated; however, the K level remains within the normal range in up to 50% of cases (Plassart et al., 1994; Chinnery et al., 2002).

When I saw Dr. A, I hand carried all of my medical records with me and he had received 50 pages from my PCP. He did not seem at all interested in reading through any of it. It is interesting that he NOW wants to know my history and information.

What is disturbing to me is that he wants to know, “How you got diagnosed since the last time he saw you. He didn’t believe you had it.” (Words of C from the Eugene MDA office). He called my PCP and wanted to know. She did not have all of my information because she is new to me and did not request it, though I thought she had the report from the hospital. (My PCP called me and asked me to sign a release of information for Dr A so he could see the records from OHSU.)

When I spoke with C in a second conversation, asking her about the above statement, she said he just wanted to be “educated”, as did the other MDA doctors on the team; but I can’t help believe that he wants to try to disprove my diagnosis due to her first remark. I will not allow him to try to discredit and embarrass three professional, intelligent doctors, with integrity, who care about their patients and who went the extra mile to study everything they could about Periodic Paralysis: Andersen-Tawil Syndrome (ATS).

I can understand the “team” needing to see my records and I could agree to that, if Dr Ali was not part of the “team”.

Another problem is that Chelsea had told me at least 3 times that I need to think about going to Portland for my MDA services rather than Medford. Her reasoning is that they know more in Portland. Well, I know different:

First, my daughter tried to get an MDA packet from the Portland office. The person she spoke with denied that they treated people with Periodic Paralysis. I, of course took care of that problem with an email.

Second, I have been seen by Dr. C and Dr. L at OHSU in Portland. They ignored the fact I was having a paralytic episode during the muscle biopsy being preformed by Dr. C (due to the lidocaine). He was too busy talking with Dr. L and one or two other people in the room. They were discussing Dr. C’s upcoming trip to Japan to do some lecturing. He was taking his son on the trip. I was choking and my heart was in tachycardia and my chest was tight and in pain….I was not hooked up to any devices for monitoring, but when they heard me, they asked what was wrong…..I could not answer…then Dr. L hollered…”Are you asleep?” I was finally able to barely answer..that I was having one of those episodes (paralysis) and that my chest hurt and my heart was pounding. No one did anything to help!!!! Just went back to their conversation. My husband had to help me afterwards due to my weakness.

When I returned a month later (had to drive 250 miles again) to get the results of the previous exam and muscle biopsy; Dr. L gave me the results. He said that nothing showed up on the biopsy and their conclusion was that I had nothing neuromuscular nor mitochondrial. (It is interesting that the test results indicated I had myopathy….changes
in size and shape of fibers…indicative of Periodic Paralysis.) My husband and I asked about the periods of paralysis that I was having. I told him I had one while I was having the muscle biopsy and asked him if he remembered hollering at me and me telling him about it? He denied even being in the room!! Then he dismissed me and said, “Go have a good time while you are here in Portland”. I told him that, “If I could have a good time, I wouldn’t be sitting in this consult room with you”. (I had episodes all the way there in the car and had them all the way home.) We asked him what to do and he said he was referring me back to my PCP and dismissed us.

The most disturbing thing about this is that when I was researching for the MDA Clinic in Portland, I discovered that he is the doctor for MDA who diagnoses Periodic Paralysis! How could I go back there after being treated like that???

Due to the doctors’ lack of concern, my diagnosis and treatment were set back almost a year. If I could have been diagnosed a year ago… I may not have become this serious.

_____________________________________________________________________

Here is the Muscle Biopsy Result:

OREGON HEALTH SCIENCE UNIVERSITY
University Hospitals and Clinics
3181 S.W. Sam Jackson Park Road Portland, Oregon 97201-3098
(503)494-5236 www.ohsu.edu/neuromuscular
neuromus@ohsu.edu
Muscle Biopsy Care Sheet
Because of your symptoms you and your doctor have decided that a muscle biopsy will help diagnose the problem that you are experiencing.

SURGICAL PATHOLOGY

SOURCE OF SPECIMEN:A Muscle Biopsy, Myopathy

Final Pathologic Diagnosis:
Skeletal muscle left quadriiceps, biopsy:
- Mild type 2 fiber atrophy
- Mild variation in type 1 fiber size
  - Mild increase in lipid
- Electron microscopy pending

Comment: An addendum will be issued when the electron microscopy results are available.

Case seen by:
G, M.D., Ph.D. / Neuropathologist
7/13/2010
Clinical History:
The patient is a 61 year old woman with a long history of weakness and difficulty walking. She describes episodes of weakness and separate episodes of ataxia. EMG and nerve conduction studies were normal.

Gross Description:
The specimen is received fresh from Dr. C, OHSU Department of Neurology, Portland, Oregon, delivered via courier on dry gauze over ice, labeled with the patient's name (initials SH) and medical record number 06218686. The accompanying paperwork identifies the tissue source as "L quad." It consists of a piece of red tissue with attached fat, and measures 1.2 x 0.8 x 0.5 cm. A portion of the specimen is fixed in formalin for paraffin-embedding as block A2, a small fragment is fixed in glutaraldehyde for possible electron microscopy, and the remainder is frozen for histochemical studies. Two separate pieces of snap-frozen tissue were received, measuring 1.0 x 0.5 x 03 and 0.8 x 0.4 x 0.3 cm. These will be kept frozen for additional studies if needed.

Microscopic Description:
Specimens Involved
Specimens: A: Muscle Biopsy, Myopathy

Histology
Tissue preservation good
Myofiber size: Variable:
Most fibers are between 50 and 80 microns in diameter. There are rare fibers up to 100 microns and scattered fibers less than 40 microns.
No increase in internalized nuclei
Angular atrophic myofibers
Comment(s): Mild
Round atrophic myofibers
Comment(s): Mild
Hypertrophic fibers
Comment(s): Rare

Inflammation: No inflammation present
There is no muscle fiber necrosis or phagocytosis. There is no endomysial fibrosis.

Histochemistry
Trichrome: Unremarkable with no ragged red fibers, nemaline rods or rimmed vacuoles
NADH: Unremarkable with no targets, targetoids, ring fibers or tubular aggregates
SDH: No mitochondrial changes
ATPase (pH 9.4 and pH 4.5): Normal checkerboard distribution of Type I and Type 2 fibers
Atrophy of both type I and type 2 fibers
Comment(s): Many of the type 2 fibers are mildly angular and the type 2's are generally smaller than the type 1's (25-70 versus 40-100 microns). The type 1 fibers are all polygonal, but vary in size.

Cytochrome Oxidase: Normal
PAS: Normal glycogen content and distribution
PAS-D: Digested by diastase
Oil-Red-O: Increased lipid content in both fiber types

Comment(s): Lipid in increased in both fiber types, more in 1's than 2's.

Acid Phosphatase: Unremarkable subsarcolemmal
Alkaline Phosphatase: Unremarkable vascular labeling
Non-specific Esterase: Unremarkable

Comment(s): There is a single angular, darkly staining fiber.

Myophosphorylase: Present
Myoadenylate Deaminase: Present

Electron Microscopy
Thick Sections: No abnormal subsarcolemmal deposits
No vacuolation
Myofibrillary apparatus appears unremarkable
Vasculature appears unremarkable

One micron thick plastic sections are stained with toluidine blue and PAS.

There is variation in fiber size and increased lipid similar to that seen in the frozen sections. There are no abnormal accumulations of PAS-positive material.

My electronic signature indicates that I have personally reviewed all diagnostic slides, the gross and/or microscopic portion of this report and formulated the final diagnosis.

Rendering Diagnostician: G M.D., Ph.D.  
Pathologist  
Electronically Signed 07/13/2010

**Muscle Biopsy**

“In all three varieties of periodic paralysis abnormalities are seen only during attacks; biopsies taken between attacks of weakness usually show little or no abnormality unless there is some degree of fixed weakness.

In biopsies of patients with longstanding periodic paralysis Weller and McArdle (1971)… The fixed myopathy found in some patients with periodic paralysis is associated with a typical non-specific features of the myopathy, including the variability in fiber size……” (Found at PPA Website)
Now, my present problem with MDA in Southern Oregon:

After the above issues I have discussed, I want to state that, I was finally clinically diagnosed on February 7, 2011, by a renal specialist in conjunction with an electro cardiologist, with “Periodic Paralysis, with a prolonged QT syndrome. It is most likely related to Andersen-Tawil Syndrome”. I must state here, that there is no other Periodic Paralysis with long QT interval heartbeat other than Andersen-Tawil Syndrome. The other name for Andersen-Tawil Syndrome is “Long QT Syndrome 7”.

This diagnosis was based on the following:

1. Everything else being ruled out (I have every record needed to be examined),

2. My episodes of partial and total paralysis which included tachycardia, bradycardia and arrhythmias to include long QT interval heartbeat as recorded on 2 24 hour Holter Monitor reading,

3. Oxygen levels dipping to the low 80’s during episodes as recorded on a 24 hour oximitry reading,

4. Characteristics which fit the syndrome to include:
   - Slightly webbed 2-3 toes,
   - Curved little fingers
   - Born with 6 teeth missing (2 eye teeth, 4 wisdom teeth)
   - Small mandible
   - Left foot has an extra bone

5. Family Characteristics (Displayed on a family genealogy flowchart with pictures and symptoms):
   - At least 7 other family members with similar symptoms
   - Across at least 5 generations

6. My complete medical history including, “Passing out episodes beginning at age 11”

7. Potassium shifting in low, high and normal levels causing partial and total paralysis.  
   (Based on Cardy Meter readings, charted and recorded.)

8. Ingestion of potassium citrate stopping paralytic episodes.

9. Several episodes of metabolic acidosis (in lab reports) combined with total paralysis,  
   high potassium readings and tachycardia, bradycardia, and arrythmias to include long QT 
   interval heartbeats and low oxygen levels.
The final diagnosis came at the hospital on February 7, 2011 and I am including my letter to my family about how it happened:

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 so afraid 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 probably 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.
Below please find the report from the hospital written by the Renal Specialist on February 7, 2011 in which I received my final diagnosis:

RVMMC
Consultation
Preliminary

PHYSICIAN REQUESTING CONSULTATION:

REASON FOR CONSULTATION: Periodic paralysis.

HISTORY OF PRESENT ILLNESS: This patient is an unfortunate 52-year-old female who reports a long history of episodic paralysis. She notes these symptoms began at the age of 11 when she began having periodic episodes of paralysis which were described back then as "passing out." She has had mild infrequent symptoms during most of her young adulthood. Episodes have typically been transient and are described as the patient suddenly beginning to feel tired and then "spacey," progressing to paralysis where the patient is unable to move, open her eyes, or speak. The patient's level of consciousness is unaltered, and she is able to recall events during these periods of paralysis. Her symptoms began to worsen somewhat approximately 2 years ago. She has had multiple episodes where she has had severe tachycardia with paralysis lasting for as long as 30 minutes. She notes the most recent severe episode occurred during Halloween, which she attributes to antibiotic therapy. She also notes that lidocaine has caused exacerbations, as have high-carbohydrate meals. She had a fairly severe episode of paralysis following Thanksgiving when she had significant carbohydrate intake. Her episodes are typically worse at night with less frequent episodes during the day. Most of her episodic paralysis lasts less than about 15 minutes, sometimes only 3 to 5 minutes. She recently underwent a neurologic evaluation which was negative for myasthenia gravis and porphyria. She recently underwent a Holter monitor which showed a prolonged QT interval. The patient notes that oftentimes she is able to abort attacks with potassium citrate. She typically takes 100-200 mEq at the onset of feeling weak. Usually she is able to abort episodes of paralysis. She has a remarkable family history for her mother and maternal aunt who both had episodes of periodic paralysis. She has two brothers who suffer with bouts of periodic paralysis as well. She has a niece and nephew, both of whom have had hypokalemic paralysis. There are multiple family members with webbed feet and micrognathia. Multiple family members have died from sudden cardiac death. On my arrival to the CVR today, the patient was conversing normally and then suddenly developed a profound paralysis which was flaccid. She was unable to open her eyes or verbalize. She was unable to withdraw to painful stimuli. The episode lasted approximately 40 minutes. She had a fairly marked tachycardia with a heart rate into the 140s during the episode. A stat potassium level was drawn and was 3.8. Her symptoms fully abated with the patient's mentation returning to baseline. She was able to recall the event. Over the last
several months, the patient has been on a low-carbohydrate diet and feels that this has helped minimize her symptoms significantly.

PAST MEDICAL HISTORY
1. Probable periodic paralysis with the patient undergoing neurologic workup which was negative for porphyria and myasthenia gravis.
2. Prolonged QT syndrome noted on Holter monitor.
3. Hyperlipidemia.

ALLERGIES: The patient is allergic to ALCOHOL, ALPRAZOLAM, ANESTHESIA, ANTIDEPRESSANTS, ATENOLOL, CAFFEINE, CIPROFLOXACIN, CITALOPRAM, CONTRAST DYE, DILTIAZEM, DUOXETINE, ZETIA, FLUOXETINE, GABAPENTIN, LIDOCAINE, METFORMIN, REGLAN, MUSCLE RELAXANTS, NARCOTICS, NITROFURANTOIN, PAINKILLERS, PSYCHIATRIC MEDICATIONS, SEROQUEL, RANITIDINE, SLEEPING PILLS, STATINS, TOLTERODINE, and TRAQUILIZERS.

MEDICATIONS: The patient takes Mirapex 0.5 mg 2 tablets q.2-3 hours before bedtime.

SOCIAL HISTORY: She is married. No alcohol or tobacco use. She is retired and disabled.

FAMILY HISTORY: Her family history is as noted above with multiple family members with prolonged QT syndrome and periodic paralysis as well as webbed feet, micrognathia, short stature, and deformities involving digits.

PHYSICAL EXAMINATION
VITAL SIGNS: Blood pressure 140/66, pulse 102.
HEENT: Extracranial movements are intact. Pupils are equal, round, and reactive to light.
NECK: Supple.
CHEST: Lungs are clear to auscultation bilaterally.
CARDIAC: Exam reveals mild tachycardia. No rub.
ABDOMEN: Benign.
EXTREMITIES: No lower extremity edema.

DIAGNOSTIC STUDIES / LABORATORY DATA: Earlier today, her potassium was 3.8. Note the potassium of 3.8 was obtained during an episode of paralysis. Laboratories from earlier this morning: Sodium 140, potassium 4.0, creatinine 0.58, CO2 25.

ASSESSMENT AND PLAN: Periodic paralysis with a prolonged QT syndrome. It is most likely related to Andersen-Tawil syndrome. Her family history is extremely compelling with multiple family members with prolonged QT.
The following is my medical history (Written before my diagnosis):

 syndrome and sudden death as well as many first-degree relatives with periodic paralysis. There are multiple family members with webbed footing and micrognathia and other features consistent with the Andersen-Tawil habitus. The patient's exam during her episode of periodic paralysis earlier today was inconsistent with a conversion reaction. Periodic paralysis is known to occur with normokalemia as well as hypokalemia and hyperkalemia in the Andersen-Tawil syndrome. Provocation is contraindicated due to the risk of inducing malignant arrhythmias. Thus at this time the patient will not undergo provocation testing due to the inherent cardiac risk. She will be started on acetazolamide 250 mg b.i.d., as there has been clinical reports of improving patient's weakness and preventing attacks on this therapy. Her electrolytes will need to be monitored fairly closely. She will follow up with me in clinic in 1 week. She will be discharged to the intensive care unit, as provocation was not felt to be warranted.
Periodic Paralysis: Possible Andersen-Tawil Syndrome: An Overview
With History/Attacks/Symptoms of Susan Hunter

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 4 generations that I know of for sure.

By a process of elimination and based on my symptoms and those of other family members, I believe that I finally know what has made me ill my entire life and has affected other members of my family. I believe that we have a familial type of Periodic Paralysis, possibly Andersen-Tawil Syndrome.

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 doctor 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 arrythmias (occasionally) during some attacks. This is considered rare, so not only do I have a rare “zebra” but an extremely rare “zebra”.)
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, polynuropathy 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 excrutiating 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.

(As of October 31, 2010, after taking an antibiotic, I went into metabolic acidosis and since that time have had periods of high potassium; hyperkalemia and shifting of potassium in the normal ranges causing total paralysis. I can no longer take the potassium to stop the episodes.)

I would like to be finally diagnosed and be tested for the familial types of Periodic Paralysis. I would like a diagnosis so I can receive the proper treatments from my primary care physician (and for her knowledge) and so I can receive the proper emergency care treatment for my sometimes life-threatening symptoms during serious episodes of paralysis (breathing difficulties, tachycardia and arrythmias and chocking due to swallowing difficulties) in the ER. I feel the need to do this, also, 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.

In this paper I discuss my symptoms from childhood until the present.
Other Family Members with Similar Symptoms:
(All undiagnosed and a question mark to their doctors)

Maternal Grandfather (Deceased)
Maternal Great Uncle (Deceased)
Mother: Lahlee (Deceased)
Aunt: Barbara (Deceased)
Brother: Raymond
Brother: William
Daughter: Sharon
Niece: Kristen

Effects from Use of Potassium Citrate

Over the past 2 months, I have been taking potassium citrate orally when a paralytic episode begins. Much to my surprise and relief, it has helped to stop, ease or lessen the episodes, especially the total in 1/2 cup of water orally. It only takes a few minutes to feel the much appreciated results!!! I take it every 2 to 4 hours or when needed.

Three weeks ago, I began a high protein/no carb, sugar or salt diet as recommended for HKPP patients. I have felt even more improvement and less partial episodes. I have lost 9 lbs.

The times I have had total episodes in the past 8 weeks, I can trace them to OTC medication, accidently eating a carb and exercise intolerance. (Excluding the ones I have every night and the ones I wake up in every morning.)

I do live with many aborted attacks, and continued weakness, however. paralytic episodes!!!!!!! I take 50 to 99mg (Written October 2010)

Total Paralytic Attacks since I Began to Take Potassium Citrate:

08/19/2010 begin potassium citrate…almost immediate results….stops attack….becomes aborted attack

08/24/2010 ate ice cream….had total episodes during the night

09/08/2010 terrible episode started about 12:45…lasted hours….had used a glycerin suppository about 8:00am…my therapist, Rosie Watne, saw me in it….stopped all meds except mirapex…tried to stop it but the restless leg was too bad…. 

09/20/2010 begin Atkins diet…almost immediately the partial episodes ease up…. 

09/24/2010 increased mirapex from 1 ½ to 1 ¾ …a total episode that night…
09/26/2010  lost 3 lbs

09/29/2010… after walking too much on a trip… the next morning…. total episode in car…. on the way home

09/30/2010…. no carbs or sugar at all… having aborted episodes… trying to stop them

10/01/2010… flu shot in afternoon

10/02/2010…. spent time preparing simple dish for gathering…. accidentally ate pasta at gathering….. in evening and during the night and in morning total episodes.. and horrible pain, especially in hip, back and thighs…. I believe the pain was from the flu shot…

10/06/2010.. have lost 8 ½ lbs since began Atkins diet

10/08/2010 evening after dinner… no sugar, carbs, etc … took nexium for acid reflux and within minutes it was one of the worst attacks I ever had… lasted about 2 hours…. this time I could open my eyes, but nothing else… Total weakness thru the night and into the morning….. Terrible pain in hips, buttocks and lower back in the morning…

10/09/2010  weak all day from the episode the day before.

10/10/2010 spent an hour cooking breakfast in the am…. After lunch… had Atkins drink… first time since starting diet… took just a few minutes…. went right into it. Lasted about an hour… could open my eyes sooner….. partial paralysis of legs and mouth (trouble speaking) for several hours…. weakness continues for rest of day…. 

10/13/2010 lost 9 lbs.

( 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 not able to take the potassium to stop the episodes. I changed my diet to a pH balanced (Alkaline-Acid) 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.)

1/18/2011… have now lost 22 pounds. I have added more complex carbs back into my diet. After the antibiotic reaction in October, my episodes of total paralysis have increased back to several a day and at night during sleep. They are now due to potassium shifting into the high ranges or shifting within normal ranges. I have had increased problems with breathing during them and upon exertion. Tachycardia has increased and I have long QT interval heartbeats which showed up on EKG’s. I have been put on oxygen.

3/11/2011… have now lost 25 pounds. I am on oxygen 24 hours a day. I have had a heart loop monitor implanted in my chest. I have been diagnosed with Period-Paralysis:
Andersen-Tawil Syndrome. I was put on Diamox but it caused more episodes so had to stop taking it. I am eating an alkaline diet. Since I have been on the diet I have not had a paralytic attack since three days after the heart procedure; during the day. I continue to have them at night. I believe my diet and using the oxygen are reducing the episodes during the day by preventing my body from going into metabolic acidosis. Sleep is a natural trigger and I may never be able to stop the episodes at night.

My Diagnosed Conditions
Fibromyalgia
Restless Leg Syndrome
Osteoporosis (Bone Crush in Spine)
Degenerative Disk Disease
Arthritis of the Spine (Cervical)
Type II Diabetes
Extra Bone in Left Foot
Neuroma in Left Foot
Basal Cell Carcinoma (Multifocal) Nose 2 X’s
Cataracts
Tachycardia
Angina.
Arrhythmias: PVC’s and PAC’s
Small left venticle
“Nonspecific mild ST Segment abnormalities of the inferior lead”
Abnormal T waves
Long QT interval beat
Small Vessel Ischemia of the brain Stroke (recent brain infarct on MRI)
High Cholesterol/Triglicerides
Diverticulitis
Acid Reflux
Hiatal Hernia
Chronic, severe constipation

My History

Physical Problems (As a child)
Could not do the monkey bars (no arms above head)
Climb tree could not get down
Fell over small cliff, could not climb up small hill
Legs and feet hurt when skating
Difficulty climbing uphill
Poor at all sports, weak clumsy
Could not jump gymnastic horse
Clumsy
Overweight
Tripped often
Left foot turned in
Growing pains
Wet the bed until 12
Bladder infections
Always crying
Age 11 collapsed/fainted in church 2 times
Could never keep up with peers walking or running
Illness and Medication Problems (As a child)
Deathly ill every time I got antibiotic shots
Every childhood disease extremely bad symptoms
Parents called me “sickly” as a child
Poor immune system

Physical Problems (As an adult)
Episodes of angina, tachycardia and arrhythmia
Episodes of passing out
Legs hurt when skiing
Accidently hit my knee on two occasions and passed out immediately each time
Difficulty walking uphill
Difficulty walking up stairs
Could do nothing with hands and arms above my head
Hand cramps when doing things like writing
Hand staying cramped, unable to open
Hand and wrist collapse when holding pan, etc
Foot cramps
Hands and arms numb during night
Legs and feet numb at night
Wake up unable to walk in am or during the night…need to hold onto wall
After sitting, riding in car, etc, trouble standing and walking
After walking a little, legs get weak, difficult to walk
Balance problem…need cane
Numbness in legs and feet
Episode of paralysis in 1985 at cabin couldn’t walk, talk, lasted about 48 hours
Left arm and hand numb for a month
Could not walk up stairs….had to pull self up by arms…horrible pain and weakness afterwards…Can’t walk straight…veer to the side
Weakness in hands….can’t open jars, trouble grasping doorknobs
Episode of legs feeling like I was wearing very tight tights and total body weakness
(never the same after that) Started using a cane trouble walking off and on after that (Labor Day 1999)
(1999) Episode where legs lost feeling and I fell to floor…EMG next showed slight poly peripheral neuropathy
(1999) Had to give up teaching career
Considered Disabled, Awarded Disability for Fibromyalgia, Degenerative Disk Disease and possible MS.
(1999-2010) Continual, gradual decline with periods of total body-wide weakness to episodes of total paralysis. Progressed from need for walker to wheel chair to electric wheelchair due to exercise intolerance.
(2008) Discovered misdiagnosis of MS
(2010) Tested for ALS, Atypical Parkinsons, Friedrech’s Ataxia (due to symptoms that looked like ataxia from different medications) Tested for Myopathies and Mitochondrial Diseases (Muscle Biopsy) All Neuromuscular and Mitochondrial Diseases ruled out. Muscle biopsy shows only mild variations in size and shape of fibers and increased lipids.

Illness and Medication Problems (As an Adult)
1968: 2 car accidents. Both times put on muscle relaxers, etc. Could not get out of bed or walk. In and out of consciousness. Could not care for children…the same as above. Must have been the medications causing the episodes of paralysis. Once off of meds, recovered

1974: Pregnancy that had to be terminated due to severe illness. Doctor didn’t know what was wrong, but said I would die if I did not have an abortion. I could not get out of bed or walk. Had to crawl to the bathroom and back, could not eat, could not take care of children. In and out of consciousness. After abortion, I got well again.

Every pregnancy, morning sickness from beginning to end. Always passed out, then discovered pregnancy.
Any medication given to me, caused some type of reaction. Usually, passing out, blood pressure dropping.

**Anesthesiology Problems** (As an Adult)
After surgery for nose reduction after a car accident 1972
After abortion 1975
After D&C 1975
After tubal ligation 1976
During colonoscopy 1983 (had to stop, never was able to finish it)
Before colonoscopy and after 1990
After hemorrhoid surgery 1990
(Blood pressure drops, becomes paralyzed, unable to speak)

**Testing Materials** (As an Adult)
Dyes for MRI’s …Kidney
Injection during Spinal Taps

**Lidocaine Problems** (As an Adult)
Muscle Biopsy 2010
Mole Biopsy 2010

**My Medication Reactions**
(Ataxia is most likely partial paralysis)
Metformin: Tremors and ataxia
Albuterol: .Tremor, ataxia and paralysis episode
Diltiazem: Ataxia
Seroquel: Seizures, paralysis episodes
Flouxitine: Seizures, paralysis episodes
Cipro: Partial paralysis of legs and painful tight calves
Macrodantin: Partial paralysis of legs and painful tight calves
Septra: Ataxia, 10/31/10 paralysis
Ranitidine: Ataxia
Detrol LA: Ataxia
Reglan: Tardive dyskenesia
Atenolol: Low blood pressure, passing out
Psychotropic medications: seizures
Narcotics: Unconsciousness, nausea
Pain Killers: Low blood pressure, passing out
Tranquilizers: Low blood pressure, passing out
Sleeping Pills: Become hyper, stay awake
Alcohol: Fall Asleep/paralysis within minutes
Muscle Relaxers: Muscle cramps, partial paralysis
Antidepressants: Weakness, tiredness, low blood pressure
Statins: Lower leg cramps, leg weakness, partial paralysis

**Things that Bring on My Episodes**
Stress (good or bad)
Cold/Heat
Most Medications/Anesthesia
Fatigue
Waking up
During Sleep
Falling Asleep
Exercise (After Exercise)
Walking (After Walking)
Exertion (After Exertion)
Standing too long
High Blood Sugar
Sugar
Simple Carbs
Unknown

My Symptoms During Episodes

“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, though usually, there is no problem with my heart. 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 an hour, or can be as short as about 10 minutes, if it is a second or third one in a row. Sometimes they last for 3 or more hours.

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.

It is difficult to know what brings these episodes on. I know that sleep has something to do with some of them, but not all of them. I know that sometimes, when I wake up during the night with an urgency to urinate, I am coming out of one, because I have all of the symptoms previously discussed. My arms and hands and legs are numb and feeling is just coming back. My mouth is tight and dry. Walking is difficult.”

(May 28, 2010)

The following is how I manage my symptoms daily:

How I manage my Periodic Paralysis Symptoms: Partial and Total Paralysis
Walking the Tightrope:

The following list/plan was put together after trial and error in my own quest for treatment and management. I had no doctor assisting me and gleaned as much as possible on the internet and in discussion with other people who live with Periodic Paralysis and Andersen-Tawil Syndrome. It is essential that I do everything possible to avoid the paralysis because when I am in it the tachycardia begins. When it begins, the long QT interval heartbeats start and I can go into cardiac arrest. I must constantly walk a tightrope.

There are many triggers that set the partial and total paralysis into motion:
- simple carbohydrates,
- carbohydrates,
- most meat,
- salt,
- sugar,
- caffeine,
- medications including over-the-counter medications,
- exercise,
- rest after exercise,
- sleep,
- stress (good or bad),
- dehydration
- and?

Things that I do to relieve my symptoms:
- avoid triggers
- following a proper alkaline ph balanced diet, eating from the farm; not the factory,
- take no medications including over-the-counter medications,
- avoid stress,
- no exercise,
- no exertion
- get plenty of rest
- stay well hydrated
- monitor vitals
- take potassium when needed (if low potassium)
- take sugar as needed (if high potassium)
- 24/7 oxygen

Diligently monitoring my vitals assists me in keeping PP in check. I use several pieces of medical equipment for measuring your vitals (all purchased out of pocket…no insurance would pay for it):
cardy meter,
finger pulse oximeter,
blood sugar monitor,
stethoscope,
wrist blood pressure monitor,
a thermometer and a digital
ph balance reader
litmus paper.

These items are necessary for Calvin, my husband and caregiver, to monitor me while in paralysis so he knows if it is time to call an ambulance for my heart, breathing, blood pressure or choking issues. We monitor round the clock also to know which direction my potassium shifts for proper treatment.

I hope the above information can help you to understand just what this disease has done to my life and that of my family members and what it does to others who have it. I hope you can also understand how horrifying it would be for me to have some doctor enter the scene at this point in my life and attempt to discredit me, my doctors and my family. He had his chance to diagnose me and help me, but he dismissed me with little regard for my feelings and that of my husband.

There is another point I need to make about the MDA doctors in Oregon, both Northern and Southern. C suggested that because of my knowledge and understanding and experience with this disease and Andersen-Tawil Syndrome, that perhaps I could “educate” the MDA doctors and others about it. I can do that. My husband and I are writing a book about it and we are developing a website:

http://www.periodicparalysisnetwork.com/

The following passage is our mission statements:

The Periodic Paralysis Network was created to provide a “hands on” approach to understanding the disease, getting a proper diagnosis, managing the symptoms, and assisting caregivers and family members. We will attempt to discuss issues relating to Periodic Paralysis in practical language. Our hope also is that the medical professionals dealing with individuals with Periodic Paralysis may come to our site and learn more about how to recognize, diagnose and properly treat their patients in a timely manner.

We are only at the beginning stages, but hope to be up and running as soon as possible. As it is, at this point, we do have quite a bit of information already posted that can be useful for understanding, diagnosing and treating Periodic Paralysis. It is straightforward and anyone can contact us for more in-depth information. We are not giving any medical advice, just simple ideas and plans that can help the processes along. These are things we have learned by trial and error along the way.
That being said, I was expecting to find doctors at the MDA who could help me and others with PP and ATS; doctors who knew about and could diagnose PP and ATS; doctors who might be well read on the latest medications and treatments for PP and ATS; doctors with knowledge of the latest medication trials and studies and doctors with open minds and a willingness to learn. I did not find that. Maybe my personal experience can change that. I would hate to have another person with PP be treated as I was treated and end up as I have ended up.

In one last statement, I would like to address the treatment my brother received when he saw the MDA doctor, Dr. P S at UCLA in Los Angeles, California. He was also told he was “too old” and diagnosed as having diabetic neuropathy. My brother’s endocrinologist disagrees since it is not even clear that my brother has diabetes. Dr. S also has a lack of understanding about how to diagnose ATS. He said he would only see my brother again if I had a genetic diagnosis. I have been diagnosed clinically.

As far as diagnosing ATS simply based on genetic testing, it would exclude 30% to 40% of people with the Syndrome.

I have sited two articles below, which explain this.

### How Common Is It?

Andersen-Tawil syndrome is a rare genetic disorder; its incidence is unknown. About 100 people with this condition have been reported worldwide. Type 1 accounts for about 70% of all cases of Andersen-Tawil syndrome. Type 2 accounts for the remaining 30% of cases of Andersen-Tawil syndrome.

**Genetics & Inheritance**

Andersen-Tawil syndrome type 1 is caused by mutations in the KCNJ2 gene. The cause of cases of Anderson-Tawil syndrome type 2 is unknown.

http://www.inheritedhealth.com/condition/Andersen-Tawil_Syndrome/37

ATS is caused by missense mutations or small deletions (Plaster et al., 2001; Tristani-Firouzi et al., 2002; Ai et al., 2002; Andelfinger et al., 2002; Donaldson et al., 2003; Hosaka et al., 2003) in KCNJ2, encoding the inwardly rectifying K channel, Kir 2.1 (Plaster et al., 2001), in approximately two-thirds of the affected individuals (ATS1) (Plaster et al., 2001; Tristani-Firouzi et al., 2002; Donaldson et al., 2003). The molecular lesion(s) have not been identified in ~ 30% of subjects including kindreds not linked to KCNJ2.

http://brain.oxfordjournals.org/content/129/1/8.full

The following is the criteria for making a clinical diagnosis:

[27]
Table 3 Diagnostic criteria for ATS

(1) A clinically definite diagnosis requires two of the following three features:

   a. PP
   b. Prolonged QTc interval or ventricular ectopy (identified on ECG or Holter)
   c. The typical ATS facies including:
      Low set ears, ocular hypertelorism, small mandible, fifth digit clinodactyly, syndactyly

(2) Alternatively, a diagnosis may be made with one of the three features above and an affected family member meeting two of three.


Based on the above criteria, I was diagnosed by meeting a, b and c. Therefore, my brother could be diagnosed based on a, b or c and my diagnosis.

In a paper written by and just updated by Dr Rabi Tawil himself, it is stated that Type 1 and Type 2 are indistinguishable in how they are manifested. They can't find any difference in people with Type 1 or Type 2. The cause is the difference; in Type 1 the cause is known, in Type 2 the cause has not yet been found.

**Here is the research:**

**Periodic Paralysis: Andersen-Tawil Syndrome Type 2**

**Type 1 and type 2**

Two types of Andersen–Tawil syndrome are distinguished by their genetic causes.

- Type 1, which accounts for about 60 percent of all cases of the disorder, is caused by mutations in the *KCNJ2* gene.[1][2]
- The remaining 40 percent of cases are designated as type 2; the cause of the condition in these cases is unknown.

The protein made by the *KCNJ2* gene forms a channel that transports potassium ions into muscle cells. The movement of potassium ions through these channels is critical for maintaining the normal functions of skeletal muscles which are used for movement and cardiac muscle. Mutations in the *KCNJ2* gene alter the usual structure and function of potassium channels or prevent the channels from being inserted correctly into the cell membrane. Many mutations prevent a molecule called PIP2 from binding to the channels and effectively regulating their activity. These changes disrupt the flow of potassium ions in skeletal and cardiac muscle, leading to the periodic paralysis and irregular heart rhythm characteristic of Andersen–Tawil syndrome.

Researchers have not yet determined the role of the *KCNJ2* gene in bone development, and it is not known how mutations in the gene lead to the developmental abnormalities often found in Andersen–Tawil syndrome.

[http://en.wikipedia.org/wiki/Andersen-Tawil_syndrome#Type_1_and_type_2](http://en.wikipedia.org/wiki/Andersen-Tawil_syndrome#Type_1_and_type_2)
Molecular Genetic Testing (Written by Dr Tawil himself)

Andersen-Tawil Syndrome
LQT7, Long QT Syndrome 7, Andersen Syndrome. Includes: Andersen Syndrome Type 1, Andersen Syndrome Type 2
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“Gene. KCNJ2, encoding the inward rectifier potassium channel 2 protein (Kir2.1), is the only gene known to be associated with Andersen-Tawil syndrome type 1 (ATS1).

Other loci. To date, no other loci have been identified to account for ATS (termed Andersen-Tawil syndrome type 2, or ATS2) in the 40% of kindreds not linked to KCNJ2.”

Genotype-Phenotype Correlations
Individuals with clinically defined ATS are phenotypically indistinguishable, regardless of the presence of a KCNJ2 mutation (ATS1) or absence of a KCNJ2 mutation (ATS2) [Tristani-Firouzi et al 2002, Donaldson et al 2003].

In a single large kindred with the KCNJ2 Arg67Trp mutation, periodic paralysis was observed only in men, cardiac symptoms only in women, and congenital anomalies in both [Andelfinger et al 2002]. However, this apparent sex-limited bias in clinical presentation has not been confirmed [Donaldson et al 2003, Davies et al 2005].


I am requesting that my brother can be seen in another clinic in the Los Angeles area, not associated with Dr. S, namely at Loma Linda. His endocrinologist has written a prescription to be seen at an MDA clinic for diagnosis of “probable Periodic Paralysis”.

At this point I would like to say that I will not be actively seeking assistance from MDA any further. However, my brother, William J (Los Angeles, California) and my daughter Sharon (Portland, Oregon) and several other family members who are very ill, will (Ohio, Texas and Salt Lake City). My hope is that they will receive the treatment and respect they deserve.

Thank you, Sincerely,

Susan Q. Knittle-Hunter