

# The Road to Diagnosis: Stories from Patients with Rare Diseases

*Personal stories from members of Inspire's online support communities*

# Introduction

By Brian Loew

About one in 10 people—30 million in the US alone—has a rare disease, and chances are someone you know is among them. The US NIH Office of Rare Diseases Research recognizes over 7,000 rare diseases, and about 30 new diseases are discovered each month. One of the key challenges for patients with rare diseases is getting an accurate diagnosis. This report is about that challenge.

In recognition of Rare Disease Day 2011, we asked Inspire members with rare diseases to tell us about the journey that led to their diagnosis. These are stories of an 81-year-old woman in New York, a 34-year-old man in Milwaukee, WI, a 68-year-old man in England, a four-year-old girl in the Netherlands, a 38-year-old woman in Victoria, Australia, and two dozen more. As you read these stories, I hope you will be struck, as we were, by what these patients share: an urgent desire to be heard, a willingness to share, and a desire to make the journey a little easier for others.

These are empowered patients who have learned a great deal and have much to teach us. As you read their words, please share this with others and help raise awareness about rare disease.

Together we're better,



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# Foreword

By Joyce Graff, VHL Family Alliance

When I was nineteen I married a wonderful, handsome guy with one blue eye and one grey eye. We knew that he had a disease called von Hippel-Lindau (VHL). We knew his father had died of it, and we knew it caused eye problems. But we knew little else. For the next 15 years we learned about VHL by “bumping into walls”—brain tumors beginning one year later, spinal tumors two years after that, kidney tumors more than ten years after that. We never met another person with VHL, though we once read a sensational article in the Enquirer about a woman with VHL in Switzerland who was mentally retarded. There were no scans, no way to see problems coming – we didn’t know we had a problem until we were in crisis. Our tongues were tied by the disease – we were new in Boston, and the only people we knew were his colleagues and their wives. He made me swear I would not mention it to any of the faculty wives because he was afraid of losing his job (and thus losing his health insurance). My close friends were in Memphis, Atlanta, San Antonio and Denver – I had a monster of a telephone bill. My husband died in 1977.



When my son was diagnosed with VHL in 1986, I set out to learn everything I could about VHL. It was not easy in those days before the Internet. As I sat one day in the Harvard Medical School library, surrounded by dusty old volumes of medical journals, I realized I was doing what a doctor would have to do to learn about VHL—and the reality is, doctors don’t have the time to spend their days in the library. A genetic counselor in Kansas linked me up with a family in Maryland also affected by VHL. After a one-hour conversation with Susan, both of us were struck by the power of what we had learned, just sharing our stories and our adventures with VHL. “What if we could get all the families together?” we asked. “Perhaps we could help give the doctors the clues that will lead to a cure for VHL?”

That quest—to get all the families together—has been the driving force in my life for the past 25 years. Today we have new tools, new technologies to help doctors find information about rare diseases, and to help families find medical care. Nonetheless, it is still hard, as you will see in the poignant stories that are shared below. And most of all, it is critically important to get all the families together. Just as there are support groups for the more common diseases in your local hospital, the people affected by rare diseases need each other at least as much, but it's not so easy to call a meeting. People with rare diseases are thinly scattered across the United States, and across the world. They are kept apart by well-meant privacy rules, by geography, by country boundaries, and by language. Instead of meeting in the hospital library, we meet in online support groups, like the ones hosted at Inspire. We listen to each other's stories, gain insights from others' experiences, share the names of helpful doctors, diagnostic tests, and treatments. We bring each other hope.

As we approach Rare Disease Day, every one of us needs to know the power of persistence, the power of collaboration, the power of asking questions. And not every person knows how to do it for themselves. If you don't need to do it for yourself or a loved one, perhaps you can help inquire for someone else. Just helping someone "Google it" can provide them with a tool that may well save his or her life. While each disease is rare, the experience of having a rare disease is not rare. One person in ten is dealing with some rare disease, or is still on a journey to arrive at a diagnosis. Please join us in the quest for hope.

*Joyce Graff is the founder and Executive Director of the VHL Family Alliance, a worldwide network of support groups for individuals and families affected by von Hippel-Lindau, a genetic tumor syndrome (<http://vhl.org>). She is a member of the U.S. National Cancer Institute Director's Consumer Liaison Group, and a past member of the Board of Directors of the National Organization for Rare Disorders. She has met with families and spoken about VHL in 28 countries on six continents.*



# “It is your life. Fight for it.”

Valerie Allen, 58, Fort Ashby, WV

I was going for a mammogram for a lump I found in my left breast, knowing it was left over from a car accident I wasn't too worried, When I got the results it said there were two lumps, one was in my right breast. Right then I knew it was cancer, but I thought it was a recurrence of my endometrial cancer, however it turned out not to be. After the Mammogram I was put thru a sonogram which confirmed the lump and then I was scheduled for a biopsy, all went extremely quick. Within a month I went from no problems to “you have leiomyosarcoma cancer.”

I had a stereotaxic biopsy and they sent it away so it took a week before I got the results and that was pure hell to go thru. But as I said, I knew the outcome. Just never thought it would be a “rare” type of cancer which is in a “rare” spot, my right breast. I felt rather abandoned by everyone when I had the lumpectomy completed and everything seemed just dropped. I am going for a second opinion due to this. I did not get any questions answered or any care as is suggested on the sites for leiomyosarcoma.

There are second opinions to be had and good doctors who listen and care are out there, go find them.

I am not giving up. My advice to others is do not take no for an answer and do not give up. There are second opinions to be had and good doctors who listen and care are out there, go find them. It is your life. Fight for it. NEVER GIVE UP, WE ARE ALL THERE WITH YOU, YOU ARE NOT ALONE.

# “I’m sick a lot, but on the good days, I see so much more of what’s important.”

Dawn, 35, Iowa

I was diagnosed with sarcoidosis in October of 2008. I began having problems with shortness of breath and chest pain two months prior to this. I would go to the doctor, get on antibiotics and inhalers and wasn't getting better. I was driving to work, I am a RN, and began having crushing chest pain, so went to the ER. My heart workup was fine, but they did a test called a D-dimer that may indicate a clot and that was elevated. So they did a cat scan of my lungs. There were several enlarged glands throughout the area between my lungs. They told me it could be lymphoma or sarcoidosis. So I waited for a bronchoscopy and that was unsuccessful due to too much blood to read any results. A few weeks later they went in through a cut in my neck and found out it was sarcoidosis. So it took about 3 -4 months from symptoms to diagnosis. I was started on prednisone and suffered from every negative side effects there were. My life was officially interrupted in a way that I had no idea of.

How many clinicians did I see? Wow. Went through four pulmonologists. My primary care provider told me it was in my head and I was stressed. One lung doctor told me to go home and exercise. One lung doctor said, “Take this prednisone and in four months you’ll never see it again. Your young and lucky.” I was so lucky that I was 40 pounds overweight, unable to hold a job because I was always short of breath and had so much pain. But “Lucky” (I guess that would be me) had this all in her head and since she was a nurse, knew too much for her own good. I

I can't give up...and there are times I want to throw the towel in. But there is some fighting desire so deep inside me that comes through when I think I can't be helped. NEVER LOSE HOPE.

I'm scared to see what the new CAT scan results will bring. I know it's spreading. I feel the pain. But I ask people to pray for me and I pray for others and then I tell myself, "Well, hold on for one more day and see what it brings."

was always on this Inspire website because I knew there was much more wrong and a lot of the members were going through the same thing. My family and friends did the best they could with this, but the medical community was not supportive really at all, until I went to an oncologist.

I had just been to the Mayo Clinic and they did no CAT scans and sent me home on iron and thyroid medication, which I don't even need now! The oncologist did a round of methotrexate, which failed miserably. One month after I got back from the Mayo Clinic and a few weeks before losing COBRA insurance, the oncologist found sarcoid in my right lung, my spleen and pelvis and I have a suspicious spot on my liver. So one bit of advice is just because the hospital has a "major" name, they can drop the ball too. Those CAT scan results were quite a big miss.

What advice do I have for people who are trying to find the right doctor and diagnosis? First, YOU ARE NOT CRAZY AND IT'S NOT IN YOUR HEAD! You will hear that. We all have. You know your body. Don't quit trying to find a doctor that you feel is competent. When you first meet one, don't bombard them with a thousand things, just be polite and firm and request all the testing that you have learned about. The most they can say is no. Know that they will, nine times out of 10, want to see what steroids are going to do. Everyone's experience is different. Ask about them and their side effects. Educate yourself with reputable sites. Start the SSI process as soon as you can if it starts too look like you're in it for the long haul. I suggest getting a lawyer. I'm on my third appeal with a hearing coming up and have a lawyer. It's up to you. And now since I don't have insurance and my sarcoid has come back with

a vengeance, it's now attacking my skin, find out what services your state has to offer. Right now, I've been approved to be seen at University of Iowa because I have zero income. I have no idea what they are going to do for treatment since prednisone and methotrexate didn't work (and I will let the Good Lord take me before I get on a course of steroids again.)

But I can't give up. . .and there are times I want to throw the towel in.

But there is some fighting desire so deep inside me that comes

through when I think I can't be helped. NEVER LOSE HOPE. There have been so many times with frustration and depression and feeling like a burden that hope is the only thing that you've got. Sometimes I wonder if they can't find a therapy or cure for me, then maybe my case will help eventually with the cure and I can say that I'm glad I never gave up. . .it's so much easier to die than live. This I have learned. And whatever God you have, grab on to it because it will be your lifeline more than you know. There will be times when no one in your life or on a support website understands and it's so scary, but if you have a God, He/It will carry you. I'm still here. And I'm scared to see what the new CAT scan results will bring. I know it's spreading. I feel the pain. But I ask people to pray for me and I pray for others and then I tell myself, "Well, hold on for one more day and see what it brings."

This is how my life is now. I grieve for the person I was before being diagnosed and I'm working on who I'm supposed to be now. I'm sick a lot, but on the good days, I see so much more of what's important and a little purpose of what I am. And that is to help find a cure to this ravishing disease. God bless you all as you walk the diagnosis journey.

I grieve for the person I was before being diagnosed and I'm working on who I'm supposed to be now. I'm sick a lot, but on the good days, I see so much more of what's important and a little purpose of what I am. And that is to help find a cure to this ravishing disease.

# **“If you do not understand, ask and then ask again if you need to.”**

**Twila Kelly, 57, Raleigh, NC**

By breaking silence about my experiences during the pre-diagnosis stages of my dance with scleroderma I stand in danger of violating the Thumper Doctrine. You remember Thumper. Right? He’s the adorable Bambi bunny who reminds us, “If you can’t say something nice, it’s better to say nothing at all.” Any musings about the internist who cared for me during those years does not invoke many words of praise. Quite the opposite. However, in this brief recount I will try to give her every benefit of the doubt—for my own emotional sanity. I do not have the time nor the energy for counterproductive anger.

I played flute. Not professionally but well enough to earn invitations for an occasional solo at a church or two. When I began to notice that my tone quality was deteriorating and that I could no longer play through a complete phrase I knew that something was seriously wrong with my breathing.

Over a period of months I twice reported this problem to my primary care physician (PCP). She recommended ibuprofen for, “a touch of pleurisy.” On the third complaint she completed a pulmonary function test. Results indicated a moderately severe impairment. When a number of in-house cardio tests came back normal, I assumed the problem would resolve itself and I did not pursue the issue. And neither did she.

More time passed and I returned with more breathing complaints—pain, breathlessness on climbing stairs and a persistent dry cough. An x-ray showed nothing amiss. She prescribed a medication for asthma. I embarked on a daily walking routine and began to lose a little weight. Neither the medication nor the new routine eased symptoms. In fact, they worsened.

A prescription for ADVAIR followed. While this medication eased the chest ache briefly, it provoked severe coughing attacks. These attacks produced yet another alarming symptom—distinctly blue fingers.

I returned yet again and asked for a referral to a pulmonologist. She seemed irritated and stated that I was out of breath because I was overweight. She went on to say that this was not a popular thing to say but it was the truth and that a pulmonologist would not find anything different. Even though somewhat shaken from her rant, I continued to insist on a referral because symptoms were progressing at such an alarming rate.

Literally within minutes, the pulmonologist identified interstitial lung disease. He suspected systemic scleroderma and immediately ordered tests, which confirmed his diagnosis. Since confidence in this PCP had by now come under serious doubt I reluctantly returned to her for further direction, but with my own list of rheumatologists. To her credit, she made the referral and forwarded my records—but not without a few parting shots about my lifestyle. I learned later she is no longer on staff at this group practice.

I am but one voice, among many, who has experienced some kind of frustration or some element of humiliation while dealing with a medical person.

While almost two years passed from the time of my first symptoms to the day I crossed the threshold of Duke University and found a team of caring, informed specialists, I count myself fortunate because I avoided the “dozen-doctor-bounce.” I had visited only two physicians before receiving an accurate diagnosis. However, the internist left scars. I avoid dwelling on her attitude, which often seemed scornful and belittling.

I am but one voice, among many, who has experienced some kind of frustration or some element of humiliation while dealing with a medical person. This is unfortunate because conversely I have met professionals who have dedicated their lives to the relief of suffering and offer tremendous compassion and understanding. I feel enormously fortunate to have a team of such individuals at my current place of care—Duke University in North Carolina.

I am a research patient, a role I view as critical in the advancement of our pool of knowledge. I give thanks daily for those who have gone before me. I always seek and am open to other ways that I can help raise awareness, act as an advocate, or help researchers learn more about systemic sclerosis.

I did take away a number of valuable lessons from my diagnosis experience: Not all doctors are created equal. Be your own best advocate. Research and learn as much as you can about your condition by using reliable sources. If you do not understand, ask and then ask again if you need to. Never give up.

May Thumper forgive me.

# **“As for advice about getting a diagnosis, I wish I had some magic formula for others.”**

**Jeanne, 68, Union City, CA**

My first symptoms of Sarcoidosis were in August 2003 and I was diagnosed in late 2008. So, it took about 5 years.

I saw my Kaiser PCP in 2003 for extreme fatigue, swollen ankles, a slight fever and raised blood pressure. She could not come up with a diagnosis and suggested I look at what I had changed in my life before the symptoms occurred. I remembered I had replaced the salt in our table shaker with Salt Substitute to help my husband after his heart surgery. I had no idea it would trigger my own autoimmune disease. After stopping use of the Salt Substitute I got somewhat better. But then in December I started experiencing eye problems. I saw an ophthalmologist who miss diagnosed my eye symptoms as iritis even though they symptoms exactly fit sarcoidosis and only sort of fit iritis. Treatments only sort of helped. I also saw several GI doctors and Ob-Gyn.

Then in 2006 I moved and saw a new PCP and ophth. At an annual check up my PCP noticed elevated liver readings and diagnosed Sarcoidosis via a lymph biopsy. At this time I was feeling quite well.

Then in 2010 when I was experiencing symptoms again my PCP said I was in remission. I have just changed insurance and will be seeing a new PCP tomorrow. Now that I have found this Inspire site I feel I have almost adequate information, but not from my doctors.

I have asked my Ob-Gyn if the abnormal Pap test and mammograms could be sarcoidosis and was told no. I have been told skin lesions could not possibly be sarc. I did have 1 skin lesion on foot biopsied and doc said it was not sarc, not cancer but some other kind of inflammation. So what kind? I still don't know. I have been using hydrocortisone 1% and it has reduced the size of one lesion but they are not going away. More are appearing elsewhere on my body.

Hopeful that new doctors will help explain all these weird symptoms that others have described as sarc.

As for advice about getting a diagnosis I wish I had some magic formula for others. I am still in the process of getting a full diagnosis.

# “We really researched online.”

Sally Timko, 57, Garden City, NY

My husband Denis, 60, was sick for two years. He was getting hard around his neck, shoulders and face. His doctor said he was anxious and should find a way to relax. He could not open his mouth wide, lift his arms up or make a fist. He began having swallowing problems. He was choking on a regular basis. He went for an endoscopy. He almost died because his throat was so restricted the doctor could not get the tube back up and he turned blue on the table. At that point we hit the ground running. That doctor thought he had a tumor in his throat and on and on. He was seen by rheumatologist panels, dermatologist panels and ENT panels. No diagnosis. After a deep tissue skin biopsy the pathologist told my husband to find a good dermatologist. He looked in New York Magazine at the top 100 doctors and found Dr. Andrew Franks, at NYU. He was triple board certified for rheumatology, dermatology and internal medicine, specializing in lupus.

We got an appointment and he diagnosed him on the spot with Scleromyxedema. An extremely rare disease of unknown etiology. 150 known cases worldwide. We were so lucky to have gone to this particular doctor. He had 4 other patients with this rare disease. What are the odds of that? We then went to the Mayo Clinic in Rochester, MN, to see Dr. Martha Lacey to confirm. A hematologist there who had experience with this disease. It is associated with monoclonal gammopathy, which can lead to multiple myeloma. The thought is that it fuels the hardening of the skin. Now, the treatment was the most difficult to get because the insurance company would not cover experimental treatments. Every treatment for this disease is considered experimental. He needed chemo and IVIG. His company had to pay for the first six months to show the insurance company that it was working. Again, lucky to have a job with a company willing to back you up. Not without some pleading on my part, I assure you. Thankfully it was working. He is now treated with IVIG every six weeks and is living a good life free of the problems associated with the disease.

We really researched online and I have gotten scleromyxedema mentioned on this web site. We also worked with others to get up a support web site, [www.scleromyxedema.net](http://www.scleromyxedema.net). This disease is often confused with Scleroderma and has nothing to do with that disease except that it may present similarly. However you can only get to the scleromyxedema page on NORD through scleroderma. I am trying to change that.

This is truly an orphan disease and needs all the recognition it can get.

# “How many people with rare diseases have doctors who cannot find out what they have?”

Ingrid R. Guerci, 60, Tarrytown, NY

I wonder sometimes how, if encephalitis has affected my memory so much, I can remember the day I first started feeling like something was really wrong with me and what it was like waiting in the waiting room at my local hospital. That was 12/18/95.

I live in Tarrytown, NY and had just returned from a week’s vacation in Florida. I called in sick the next day and never got out of bed. My eyes were crossing; I felt like I had a fever but didn’t check to see what it was; then, the scariest thing to happen to me was not being able to speak. I smoke cigarettes (no lectures, please) and told George that before I went to bed again I was going to have “a mustard”. He had no idea what I was talking about and that was when he realized something was really, really wrong. The next day I realized I had to go to the hospital. The nurses, then the doctors took my temperature, had me walk back and forth, do circles, etc. At first they thought I had a stroke because of the way I was speaking but I didn’t have any limp or weakness and my fever was up to 105. They didn’t know what to do so they contacted my doctor. She had no idea what was wrong with me, but whatever it was, it was serious enough so she then gave approval for a neurologist to check me out.

I may have some residual disabilities but since I came so close to dying, and spent so much time looking at only the negatives, I have now tried to focus on the positive aspects of life.

“Dr. R” has to be the best doctor I have ever known! One of his partners (another neurologist) had died from encephalitis a few years prior to his seeing me. I had a spinal tap, EKG, EEG and MRI. My fever was still over 105 degrees and I really couldn’t talk. I could answer yes and no questions by shaking my head. They admitted me, sent me to ICU, hooked me up to IV’s with penicillin, acyclovir (the doc figured if it wasn’t an infection, it was something that acyclovir could help) and packed me on ice (which I hated; I do not deal well with being cold; I have something called Raynaud’s Disease).

I know I came as close as you can to dying without really dying. The days just kind of went on. I was in the hospital on acyclovir for two full weeks; the first 5 days being in a private room in ICU. I went back to work part-time at the end of March 1996 (3 half days a week for about 2 months; 3 full days for all of the summer; and then full time after Labor Day). I have been fortunate not to have had any seizures. I am sure I am over the worst of it. I may not get any better than I am and hopefully, I won’t get any worse. We all have had different levels of this disease but we have all had the same disease and know what each other is going through. I feel so lucky to have a job where I am allowed access to the Internet. Our encephalitis online support group is at [www.inspire.com/groups/encephalitis-global/](http://www.inspire.com/groups/encephalitis-global/), and to find out more about it, email me at [encephgroup@aol.com](mailto:encephgroup@aol.com).

How many people with rare diseases have doctors who cannot find out what they have and treat it and how many of us are who do not have access to a computer? I may have some residual disabilities but since I came so close to dying, and spent so much time looking at only the negatives, I have now tried to focus on the positive aspects of life.

# “We have had noooooooooooooo luck with doctors.”

Bette, 62, Northampton, PA

My husband’s illness is so rare or bizarre he doesn’t even have a diagnosis. One doctor even said when he dies he would recommend an autopsy to find out what he had. Personally, I was quite offended by that comment. I have done tons of research and I think he has some kind, or numerous

kinds of autoimmune diseases. Off and on for 40 years he would have problems with his prostate and numerous UTIs. In early 2000 he began having memory loss and it was determined first that he had frontal temporal dementia, later changed to Dementia with Lewy Bodies. In 2005 he was referred to a urologist because of recurring blood and protein in his urine. Since there was no infection no antibiotic was given as had been done in the past by his GP. His dementia worsened immediately but he was still able to function normally until he had a severe prostate infection in 2007.

That infection left him unable to walk or care for himself and he had a bad rash on his back, purple marks on his hands, and open sores on his rear end. He was put on an antibiotic and he began walking and caring for himself until the antibiotic ran out and then it started all over again. I pleaded with every doctor to continue the antibiotic to no avail. After 3 months he was put in the hospital to search for an answer. None was found, but one doctor agreed to put him on a long-term antibiotic that he is still on today. He is still unable to walk

In my opinion, this information should be made known so others do not have to suffer alone ... It helped me a great deal finding out I was not the only one.

All I know is that he has something very rare and nothing any doctor has said has made any sense.

or care for himself but between the antibiotic and going on a gluten casein free diet, his rashes have finally disappeared. I have read if gluten has caused a neurological problem, they will not go away when gluten is eliminated. The doctors don't seem to know much about autoimmune diseases and gluten intolerance when it comes to neurological issues. The one he sees currently sees that he is somewhat better with the antibiotic and GFCF diet but I don't know if he agrees with "my" diagnosis. He just kind of just smiles when I tell him about my research and what I think has happened. I got a lot of my info from people on this site with autoimmune diseases.

When I began researching gluten I found it can cause the same abnormalities on a SPECT test that gave my husband the original diagnosis of FTD. I have also read that blood and protein in urine can be a sign of an autoimmune disease.

So, who knows? All I know is that he has something very rare and nothing any doctor has said has made any sense. We have had noooooooooooooo luck with doctors and I can identify with all those who not only have not received a diagnosis but have not been treated with any respect. In my opinion, this information should be made known so others do not have to suffer alone and maybe doctors will start taking people seriously and not just write them off. It helped me a great deal finding out I was not the only one.

# “My self-diagnosis with Wilson’s disease seemed to be a bit prophetic.”

Peter E. Richards, 27, Windham, NH

My first acquaintance with Wilson’s disease made in 2005, in the summer following my college graduation. I read about it, initially in “Anatomica”, then more so on the Internet. I asked whether the depression I had been experiencing, and, to be honest, lifetime procession of psychiatric symptoms, might have been caused by these errors in copper metabolism identified as WD. At that time, I had a large healthy handful of copper rich nuts and chocolate, and noticed I felt less well than I had previous to the consumptions of these “aliments.”

This prompted inquiries with my parents and G.P. My parents downplayed the likelihood that I might actually be onto something due to the perceived rareness of the disease, as did the G.P. Who, after administering a Liver panel, felt comfortable ruling out the disease.

Approximately three years later, in the summer of ‘08, I came back to the idea that I might have WD, as my psychiatric problems has worsened, as had my condition in general. Not only had the doctor’s tests been inadequate to ultimately rule out the disease (liver enzymes are not always elevated, nor are they when elevated drastically so), but my self-diagnosis with WD seemed to be a bit prophetic, as I was now, resulting of my forthright exchanges with psychiatrists, receiving a diagnosis of schizophrenia, which is identified by physicians in-the-know, as a symptom of WD. The only problem was that my belief that I had WD was now being identified as a symptom itself, a psychosis, since the psychiatrists believed I could not possibly have the disease, since my ceruloplasmin was not low, neurological examinations, urine copper, MRIs, liver panels, et. al. did not prove conclusively that I had the disease.

However, I had been during the time these tests were administered, treating my self with low copper dieting, and some zinc. Near the end of 2008, I was hospitalized. I managed to get a serum copper and a ceruloplasmin from a psychiatrist at the New Hampshire Hospital where I was admitted. The free copper was 17, which seemed to me well above the high normal mark of 10.

Four months later I was released from that hospital. The psychologist commented that besides me delusional belief that I had WD, I was doing quite well. I was referred to a doctor, who noted, basically, that, academically, I might have the disease, though he wasn't that concerned that I was suffering abuse from those psychiatrists who called me delusional and kept me hospitalized, and forced medication in attempts to cure said belief.

I stored up all the Abilify prescribed for the next 9 months or so. After seven months of this my parents had claimed they saw great improvements since I had left the hospital (which is precisely the time when the most psychotropics were in my blood). However, after I fled NH and my guardian, to return to Chicago where I had lived as I attended architecture school from '06-'08, my parents concluded that they hadn't believed I was better off without the psychotropics they had fought to administer to me.

As I was looking for a physician who could help me monitor my copper because I believed I had reached a Cu deficiency, I became very ill, I believe from varices, caused by my cold-turkey quitting of zinc, and went to the ER at NMH downtown. I was again hospitalized for the "delusion" Many doctors told me I couldn't have the disease. The head of hepatology told me I couldn't have WD because he said "a person with WD can't have a copper deficiency."

# **“Believe it or not, this different person is happy, happy with life.”**

**“clicquot,” 50, New York, NY**

I vividly remember the first time I noticed something was wrong. I was 49 years old, finishing my last semester of law school. I had stayed late after class one day. I was sitting still, confused. I asked the professor an off-base question.

The next symptoms came over the course of a few days. The world started to get a little fuzzy. While riding my motorcycle, I “dropped” it (tipped over) while nearly stopped. Twice. I became so disoriented that on my spouse’s advice I went to see my physician immediately. I was driven to his office (I normally take the subway). While in the office I was acting incoherently. My physician sent us to the ER.

We walked outside. This is when my disorientation became completely obvious: I clearly remember the scene. As I rounded the corner to go back to the car, I could no longer maintain my balance. I was disoriented. I stopped walking. A shopkeeper steadied me and held onto me. I was terrified and motionless. The next thing I remember is arriving at the ER and staggering to the admitting desk. There, I passed out. I vividly remember being wheeled into the ER on a gurney, nurses trying to get me to speak. My mind was racing a mile a minute and nothing understandable was coming out of my mouth. My mind was trapped.

Then I was in a coma, and I truly saw the proverbial light at the end of the tunnel. I am fortunate that I was almost immediately placed on IV acyclovir under the presumption that I had Herpes Simplex Encephalitis (HSE). Tests were performed. A few days later, I was out of the coma but not really here. I got slowly better. I was sent home. I stayed at home, not moving much, with IV acyclovir from a home health aide and care from my family. Many months went by and are gone forever.

I cannot give a precise answer as to how long it took to get my diagnosis. Perhaps an answer to this question should come from the physicians and other caregivers that made the diagnosis. I was, so to speak, not of this world when the diagnosis was made. The caregivers would know better than I exactly how long it took, but it was on the order of hours. Involved were my physician, the ER staff, and Neurology at NY-Presbyterian/Weill-Cornell.

From my post-coma, in-recovery standpoint, I know I had great care early on, and in that sense, diagnosis was either easy or providential. Otherwise, I would not be here in my current state. I have subsequently learned that the MRI done soon after admission showed damage consistent with HSE. My diagnosis of HSE is based on MRIs and my recovery after acyclovir. This diagnosis was made by neurologists at the hospital in consultation with my physician.

My subsequent recovery is also consistent with HSE: a lot of trouble early on with just about everything, but a slow recovery of most functions. Now, 20 months later, there is still disruption in short-term memory. This is both my self-diagnosis and diagnoses from physicians and family. But, here I am, functioning well enough to find my own way on the subway;

I am not the person I was 2 years ago, and I am certainly not the partial person I was 20 months ago. Believe it or not, this different person is happy, happy with life.

From my post-coma, in-recovery standpoint, I know I had great care early on, and in that sense, diagnosis was either easy or providential.

to make meals for myself and my family; to go to work full time in a demanding position; to enjoy life with my family and dogs. I am not the person I was 2 years ago, and I am certainly not the partial person I was 20 months ago. Believe it or not, this different person is happy, happy with life.

In summary, my diagnosis was reached by (1) my own recognition that I was not performing well; (2) my spouse's concurrence; (3) my physician's recognition that I was not myself; (4) a rapid response to my collapse and coma at a world-class medical center in New York City followed by a lot of testing. I don't think that makes it "easy" in any sense, or even lucky. Just aggressively followed by a great team including family and physicians.

# “I wish doctors were more willing to consult with others.”

Cathy Nevers, 55, Lawrenceburg, TN

I was diagnosed at 51 years old, in 2007, with PPMS. One year before I was diagnosed, I ran a half-marathon in Nashville, TN. I was in the best shape of my life and thought life was good! Until, the tingling on the bottom of my feet started and a feeling of numbness on occasion. This would go away after a while so I thought maybe I had a lower back issue and went to see my chiropractor. I tried this treatment for about a month and decided that was not working. I talked with a DR. friend who suggested I see a Neurologist.

I actually feel bad for the doctors since they are only providing an educated guess.

I scheduled an appointment however before the appointment my legs went numb while grocery shopping and I barely made it back to my car. I was hospitalized, all the necessary tests were run including a spinal tap. Results all came back MS. In the meantime, I haven't responded well to any of the typical medications and the MS is continuing to progress.

My Dr. is again testing me for other diseases including devic's. The testing for devic's can only be done at the Mayo Clinic in MN so it takes several weeks.

Sometimes I wonder if I even have MS. I feel like I'm heading in the wrong direction or maybe my doctor is? It is very frustrating when you call your doctor and he tells you, well, guess we know that steroids don't work for you anymore. When I ask next steps, he honestly doesn't know. I actually feel bad for the doctors since they are only providing an educated guess. I wish they were more willing to consult with others in their practices or wherever to get others opinions on specific cases.

# “I am hoping that other relatives will also be tested soon.”

Wendy Collins, 38, Victoria, Australia

My story actually started way back in about 1993 when I went for a medical for work. I was diagnosed with high blood pressure, and spent the next few years going through all sorts of tests and doctors trying to find out why I had this at my age (early 20's). Eventually 2 different doctors diagnosed cysts on my left kidney and a fatty liver. Not much I could do. I also saw 3 different dermatologists about some funny lumps on my skin that would hurt whenever I was cold. They all took biopsies and said there was nothing they could do. They named the lumps as piloleiomyomas.

Fast forward to 2004. I was on maternity leave with my then 8-month-old daughter. I had developed a cough that would sometimes be so bad I would vomit from coughing so much, so, as I had time off from work made an appointment to see my GP about

I had developed a cough that would sometimes be so bad I would vomit from coughing so much.

this cough. I was thinking along the lines of a chest infection. My GP was wonderful. He said my chest was clear and sent me off to have all sorts of blood tests to cover everything possible, from perhaps a weird disease from animals to gall stone problems. After these results came back with not much to show for them, he sent me off for an ultrasound. This showed a mass in my left kidney (a-ha - covered by the cysts?) and within a month I had weaned my daughter of breast milk, written my will, and had surgery to remove my left

kidney with a 10 cm tumor in it. Apparently if it hadn't been found within a year or so it would have been too late to remove surgically. Oh. . .the cough disappeared as soon as the kidney went!

As soon as the kidney was removed, my blood pressure went back to normal, as it is now, 5 years on. I believe that there was definitely some link between the cysts, the tumor, the blood pressure and the kidney. My high blood pressure was never given a reason (before the tumor was found).

The saga continues—I went back to my dermatologist to have one of my piloleiomyomas removed because of the pain, and updated my surgical history with him. He found the link between piloleiomyomas and kidney cancer and referred me to a genetic counselor who organized to have my DNA tested in the US for HLRCC. When the result of that came back, we found out that I was the only one at that time in the Southern Hemisphere diagnosed with this.

Since then, family testing has begun with my siblings all clear, and one aunt positive. I am hoping that other relatives will also be tested soon to find out if they are at risk of this rare disease.

# “Now that we have a diagnosis, we can continue to explore treatments.”

“mom\_traps,” Sunderland, MA

My daughter is 17, and was finally diagnosed with a genetically caused periodic fever syndrome (TRAPS) at age 15 after over 10 years of tests, multiple hospitalizations, and missing lots of school and social stuff along the way.

Genetically caused periodic fever syndromes are caused by an inherited genetic abnormality and can only be diagnosed by genetic testing. The most well known of these is FMF (Familial Mediterranean Fever, which is actually a misnomer, as it is not restricted to individuals of a specific ethnicity), but there are several others. Some people with these genetic abnormalities exhibit no symptoms, some have symptoms only intermittently, while others are severely affected. Episodes can be triggered by physical or emotional stress or exposure to infection (cold, flu, etc.), but for some people, they just appear like clockwork without an apparent trigger.

My daughter began exhibiting symptoms when she was four, and these recur roughly every 6 weeks. She has high fevers, severe abdominal pain, joint pain, and can experience pain in other areas such as her heart, lungs, eyes, etc.—it all depends where the inflammation occurs during the episode. In between episodes she is easily fatigued and often nauseated. Diagnoses have run the gamut: “mystery virus,” strep, appendicitis, “something she’ll grow out of,” juvenile arthritis, lupus, leukemia, Lyme disease, Crohn’s disease. Our pediatrician treated her for Lyme, but when the symptoms returned, she was totally baffled and basically threw up her hands and said, “I don’t know what this is.” She did not offer any recommendations or refer us to another doctor, she just went back to doing test after test after test.

I just want to stress that sometimes you have to push to get to see the right doctors, and get the right tests.

Since my daughter's symptoms were mostly abdominal pain and fever, I looked for and found a pediatric GI, but we could not get an appointment with him for six months. So, when she had her next severe episode, I took her to the emergency room of the hospital that he was affiliated with, and he saw her the next morning. She stayed in the hospital for three days, had multiple tests including endoscopy and colonoscopy but found nothing conclusive. A couple more years passed with many more episodes and many more tests, and our GI

also finally threw up his hands, but guessed that it had to be something like an autoimmune condition. He sent us to a pediatric rheumatologist who was familiar with periodic fevers. She immediately did genetic testing, and contacted a doctor at NIH when the results showed a genetic abnormality known to be associated with the periodic fever syndrome called TRAPS. We travelled to NIH in 2008 and they confirmed the diagnosis.

Since diagnosis, my daughter has been taking prednisone for flares, but it has become less effective over time. She has fewer fevers, but more severe and prolonged episodes of inflammation and just feeling "crappy" (nausea, fatigue). We may be looking at trying Enbrel (injections) but this could make her more susceptible to other infections, some possibly serious. Since she hopes to start college in the fall, and live in a dorm, this could be a recipe for disaster.

In the meantime, we try to make sure that she stays as healthy as possible by getting regular exercise, plenty of sleep, and eating well. But, since she is trying to be a "normal" teenager this is not easy, and stressing-out is just part of being an adolescent girl. All this plus lack of sleep are par for the course in college, so it is going to be a real challenge for her to be successful there.

My daughter's disease has shaped who she is today: She is a very determined young woman who, in spite of her illness, has managed to always be an A student (even now, attending a community college for her senior year of HS). She works part-time, tutors classmates in math and physics, is on the ski and tennis teams at school, and just applied to 10 (yes, 10!) colleges. She has never told anyone about her illness until this year, and she has never used it as an excuse for doing anything less than her best. I watch her endure pain, exhaustion, and nausea, almost every day and still keep trying. Some days she cannot get out of bed. Other days, she does, but I often don't know how she gets through the day. I know I couldn't do what she does.

Anyway, I just want to stress that sometimes you have to push to get to see the right doctors, and get the right tests. Genetic testing is expensive, and we were lucky to find a doctor who went to bat for us with our insurance company so we could get it done. Now that we have a diagnosis, we can continue to explore treatments. There is no cure for TRAPS yet, but when one does come, we'll be ready.

# “You have to do your own research.”

“GrahamL,” 68, United Kingdom

Oct. 2009: Blood in urine - CT scan showed tumor in left kidney. Jan. 2010: removed radical nephrectomy—unusual type collecting duct. I showed surgeon benign skin lumps which I knew were unusual and called “piloleosomething” as I had one diagnosed some 25 years ago. July 2010: Wanted to find exact spelling of “piloleiomyoma.” I Googled and found HLRCC connection. Now having genetic tests on the FH Gene. You cannot expect a rare disease to always be diagnosed if it is not being looked for, and you may have to do your own research. I have had good support since I found the connection.

July 2010: Wanted to find exact spelling of “piloleiomyoma.” I Googled and found HLRCC connection.

# “Don’t accept a pleasant diagnosis too easily.”

“Roadwalker,” Berkeley, CA

My daughter was born with ash leaf marks on her legs. Her pediatrician said they were areas of hypopigmentation (which explains nothing), so we didn’t investigate further. Much later, we found out that these marks are associated with tuberous sclerosis, a genetic disorder that can cause tumors in many of the major organs. When my daughter was 9, she exhibited signs of hydrocephalus and was diagnosed with a large subependymal giant cell astrocytoma. This is the type of brain tumor that is associated with tuberous sclerosis.

Most of the tumor was surgically removed, but the neurosurgeon never directly said she had tuberous sclerosis. It wasn’t until two years later, when she had a shunt failure, that a different neurosurgeon told us that “of course” she had tuberous sclerosis. And it wasn’t until then that we began the series of periodic tests that are essential for every patient with tuberous sclerosis: CTs, MRIs, and ultrasounds of the brain, the kidneys, the eyes, and the lungs. We often wonder whether the neurosurgeon would have been able to remove the entire tumor if her tuberous sclerosis had been diagnosed early on.

Do your research, and, if necessary, get another opinion.

So, my advice is to be suspicious: don’t accept a pleasant diagnosis too easily, do your research, and, if necessary, get another opinion.

# “NF was practically unheard of back then.”

Artie DeMonte, 53, Pleasantville, NY

I was diagnosed 40 years ago when I was 13. If NF is considered “rare” today, it was practically unheard of back then.

My pediatrician always found my CALs (which I had from 4 years old) interesting, and he’d rigidly document them. He knew they were more than birthmarks, but didn’t fully understand their significance. He knew the CALs indicated something, but what?

Then, one day, he found my blood pressure dangerously high and, as he was giving my body a detailed “going over” to track the CALs, noted a tumor growing under the skin on my back.

He sent me across the street to the ER where they did a biopsy. Shortly thereafter the report indicated the tumor was benign but of unknown origin. Dr. Kunz was still very concerned about the blood pressure and wondered if there was a tumor pressing against an artery somewhere. He began working to have me go in to Montefiore Hospital in the Bronx for testing.

Now, Scotty—the character from Star Trek—liked reading technical manuals in his leisure time. Dr. Kunz was like this with medical journals. One evening as he was sipping a Scotch and browsing through a journal, the title of an article caught his eye: HYPERTENSION AND CAFE AU LAIT MARKINGS IN TEENAGERS. He could

A surgery was scheduled, and a 16-hour operation was performed the following January.

hardly believe his good fortune and devoured the article. Halfway through, he was confident I had NF, “discovered” by Fredich Daniel van Recklinghausen in 1882. He called my parents at eight o’clock that evening to tell them of the suspect diagnosis.

A week later, I was in the hospital. After 12 days of intensive and often very uncomfortable testing, not only was NF confirmed, but as Dr. Kunz suspected, there was a fibroma pressing against my right renal artery, constricting blood flow. A surgery was scheduled, and a 16-hour operation was performed the following January.

So, from the time my first tumor was found (with the elevated BP) to my diagnosis was astonishingly fast—a mere three weeks. And this was decades before genetic testing and doctors being significantly more aware of NF today than they were when I was diagnosed.

I was diagnosed 40 years ago when I was 13. If NF is considered “rare” today, it was practically unheard of back then.

# “The doctors admitted that for a long time they didn’t know what to do.”

Els Harbers, 29, the Netherlands

This is the story of Maaïke Harbers. She is now a 4-year-old girl that goes to a regular school. But there have been times that we weren’t so sure that she would make it to this point.

Maaïke was crying a lot when she was a baby. When she was 3 months old her weight started to drop back. She didn’t gain enough for her age.

When she was 6 months old we went to a pediatrician for the first time. They found some abnormalities in the blood, but didn’t know what they meant. A lot of different tests were ordered, but it took a while before all these tests were performed and we had all the results. And the end result was that she didn’t have any of the things they tested for. We (accidentally) changed pediatricians. The first time we saw her she proposed to do some other tests. The next day she called us. She admitted that she was shocked by looking at our daughter. The tests she suggested the other day she had already planned by reading her chart, but now she thought she had to rush things. Maaïke was now 13 months old and hasn’t gained any weight since she was 9 months. She was 7 kilos [15.4 pounds] and her development has been put on hold. She could hardly roll

I just hope that sharing stories leads to contact with people who can help us forward. Sometimes it’s really frustrating that even doctors don’t really know anything about the disease.

from her back to her stomach and you could see the trouble she had just to lift her head. You could make her sit, but in less than 10 minutes she would fall to a side and couldn't get up anymore.

When this pediatrician put a rush on the examinations we also had periods that Maaïke was admitted to the hospital for a week to do a lot of tests in a short period of time.

The pediatrician consulted a specialist (hematology) at another (university) hospital. The specialist took over the care of our daughter. We saw several specialists and they admitted that this was a strange, difficult case. When Maaïke was 16 months old we started tube feeding. This was really good for her and she soon her development fitted her age. This didn't mean we made it. We still didn't know what caused her problems.

The doctors noticed that Maaïke's red blood cell count reached a really dangerous level. Even though they didn't know the cause, they knew they had to start treatment. So when Maaïke was 21 months old we started bleeding. At first we had to drain blood every week to lower her red blood cell count. When the blood became better we went to once every 2 weeks. When Maaïke was 26 months old, the doctors finally had an idea about what could be wrong. They needed just some genetic blood tests to verify the suspicion.

They were right this time. When Maaïke was almost 2.5 years old she was officially diagnosed. She has inherited a wrong gene from both the parents on exactly the same spot of the VHL-gene. Therefore she doesn't make a protein that involves the production of EPO. She makes EPO 24/7 and this never stops. Because of this she makes way too many red blood cells.

The disease she has is known by the name Chuvash Polycythemia. She has almost exactly this disease but the abnormality in the gene on mother's side is slightly different. Now we treat her with bleeding and she appears to be healthy. We can only hope this treatment will stay sufficient.

P.S.: The doctors admitted that for a long time they didn't know what to do and that Maaïke's life has been in danger for quite a while.

I just hope that sharing stories leads to contact with people who can help us forward. Sometimes it's really frustrating that even doctors don't really know anything about the disease.

# **“If your doctor still doesn’t listen or refer you to someone else, FIRE him/her.”**

**Ella Martin, 75, Southside, AL**

It’s difficult to know exactly how long it took to get a diagnosis of neurosarcoidosis.

Several years ago I was diagnosed with Spasmodic Dysphonia. About 2003 they started Botox injections into my vocal cords. Over the past 25-30 years I had V-meningitis 3X. Verified with spinal tap. I was a week in the hospital each time, that I don’t remember. Periods of time, for days or weeks, after released from hospital that I cannot recall, but, in appearance was functionally normal. I have had a history of migraines from about 1970-1990. A number of my siblings and children have them. I’ve had a long history of sinus infections and UTIs. In 2002, after semi-retirement at age 67 I worked in my daughters greenhouses each year from Jan.- May. My PCP was giving me antibiotics for UTI. One after another for a year. I went to a Urologist to see why I was having so many infections instead of just treating the symptoms. He prescribed me Macrochantin 100 mg. to take 1x daily and said it was not an antibiotic and come back in a year. I went back in 13 months and had not had a UTI the whole year. He said I could start on 3 weekly. It was during that time I started having shortness of breath, fatigue and muscle pain and weakness in various parts of my body for short periods of time.

I went to my PCP with a list of my symptoms and when she came into the office and saw the list, she said, “you come in here hurting here and hurting there and I have 15 minutes to spend with you”. I walked out and haven’t been back. I got another PCP. He was very concerned with my symptoms and ordered a number of test. Pulmonary Function done at local Hospital came back showing Asthma. He started me on ADVAIR Disc for 2 months. No good. Meanwhile having cardiac test, etc. All negative. Lab work, all negative.

To anyone trying to get a diagnosis I would say: 1. Never let them tell you it's in your head. 2. Keep a log of your symptoms whether you think they are related or not. 3. If your doctor still doesn't listen or refer you to someone else, FIRE him/her.

days. The MRI showed "possible infarct on pons and/or brain stem but, could be an artifact". Back to the Neurologist for the results on the syndrome and MRI. The syndrome was negative and the neuro said the infarct was an artifact and nothing significant on MRI. At least they were honest and said, "something was wrong but they didn't know what."

I was still working in the green houses in 2009. I would work a few hours and stop and rest for an hour or 2 and try it again. A pulmonary Dr. did another pulmonary function and said I had no sign of asthma. He said CT of chest showed nothing significant to go back to my PCP. He sent me to a Neurologist (two working together.) They did an EMG and muscle/nerve conductive test. They thought I had Myasthenia gravis and started me on Mestinon, but, said they would have to verify with lab work that takes 6 weeks. That test was negative. They sent for another test that may be a syndrome that mimics MS. During that time I had a dizzy spell and temporary blindness in my right eye for a few seconds. I went to my ophthalmologist and he diagnosed me with Glaucoma, Optic Atrophy and Uveitis. This was Jan./2010. He ordered me drops for the Uveitis and Glaucoma. My PCP ordered a brain MRI for the Optic Atrophy. I started double vertical vision for several

They sent me to a Neuro Prof at UAB,AL and he redid the EMG and Muscle/nerve conductive test and did a muscle biopsy in my bicep that same day. The next day, Feb.20, 2010, he called me and said I had Sarcoidosis in my nerves and muscles, (Neuro-Sarcoidosis) and was calling me in a prescription for 40 mg of Prednisone. I had him repeat it twice and spell it for me. I had never heard of Sarcoidosis. I have found that most medical workers including doctors know nothing or little also. When I first told my ophthalmologist that I had Sarcoid he said, "mostly younger people than you have it and mostly black". I had to show him the Neuro's evaluation (that had already been sent to him) before he believed me.

From the time of my fatigue and extreme muscle weakness and SOB until my diagnosis was about 2 years. I think during that time I saw 10 doctors. Eight of them specialist in their field.

With it being in my muscles and nerves I already had Glaucoma, Hearing impairment, Digestive Disease caused by the muscle not working in my bile duct, which, by the way. . .was several more test and doctors that I saw during that time. It's in my sinuses and thyroid glands. I have hand and head tremors. I still walk without walking aids, but very carefully. My right hip and thigh hurt profusely when I put my weight on them. Always the extreme fatigue. Going to the mailbox and back makes me feel like I have concrete blocks on my feet. Nearly ever symptom fluctuates. . .hearing, vision, body temp. (not fever) I get hot than cold, swelling, BP, fatigue and mood swings.

I look like a blimp from the prednisone which they are now trying to wean me off, and I'm on Imuran. Both have terrible side effects. I can't tell which are causing certain symptoms the meds or disease.

To anyone trying to get a diagnosis I would say: 1. Never let them tell you it's in your head. 2. Keep a log of your symptoms whether you think they are related or not. 3. If your doctor still doesn't listen or refer you to someone else, FIRE him/her.

When you live in your body you know when it's doing something out of the ordinary. When I told my second PCP that I had muscle pain sporadically in different

places. I told him I didn't want to hear the word Fibromyalgia. He told me it was probably pulled muscles to go home and take Tylenol. If I had not been on a church mission trip with him I probably would have slapped him (just kidding). But I did tell him after the diagnosis.

For now, I feel very confused and depressed over my care and treatment. I am using a Pulmo-Sarcoid Spec that I have to drive 60 miles one way to see. I don't think he knows for sure what to do with a Neuro-sarc patient. I mean I have different symptoms than a Pulmo-sarc patient. The Neuro-Sarcoid doctors are very few and far between. My local Drs., all but one, appear to know little if anything about Sarcoid. With every symptom, I have to go to a different Dr. I try to keep copies of reports from each one so they will be in synch. If I ask them to fax the reports some time they do and sometimes not.

When you live in your body you know when it's doing something out of the ordinary.

# “Stay strong and don’t believe everything you hear or read.”

**Beth Nolan, 37, Hudson Falls, NY**

I am the mother to a nine-year-old girl named Hannah. Hannah is our only child, and the light of our lives. Everyone who meets her comes to love her. Hannah’s friendly demeanor and easy way about her makes her a joy to be around.

This past May, my husband and I noticed Hannah “didn’t look right.” Her legs looked heavy and she constantly had a tired, puffy look to her eyes. A few days after Memorial Day, I noticed I could not find Hannah’s anklebones or kneecaps; the swelling was so bad in her legs. I took her to her pediatrician that afternoon and was told Hannah was suffering from nephrotic syndrome. Nephrotic syndrome is a disorder in which the kidneys are damaged, causing them to leak large amounts of protein from the blood into the urine.

We were referred to a specialist in pediatric nephrology in Albany. Hannah began a high dose of steroids in the beginning of June. We were told that this medicine would take care of the nephrotic syndrome, and Hannah would be fine. Unfortunately, that did not happen.

Hannah’s protein never came down after 6 weeks on the steroids. Her specialist ordered a biopsy of Hannah’s kidney. The results showed our biggest fear. Hannah was not suffering from simply nephrotic syndrome, but instead Hannah has a disease called Focal segmental glomerulosclerosis (FSGS).

Focal Segmental Glomerulosclerosis (FSGS) is a disease that attacks the kidney’s filtering system (glomeruli) causing serious scarring. Very few treatments are available for patients with FSGS. Most commonly patients

are treated with steroid regimens, most of which have very harsh side effects. Some patients have been shown to respond positively to immunosuppressive drugs as well as blood pressure drugs which have been shown to lower the level of protein in the urine. To date, there is no common effective treatment or cure and there are no FDA approved drugs to treat FSGS.

Hannah is on many of the drugs listed above, but has shown no signs of improvement to date. As the kidneys begin to scar, over time they lose their function and begin to shutdown. Hannah is facing dialysis and eventual transplant if this disease continues this course. We took her to Boston this past August to a specialist at the Children's hospital. There, our worst feared were confirmed. Hannah mostly likely will reach end stage renal failure in 5 years.

I am working hard at getting Hannah a third and fourth opinion. I am trying to educate myself on feeding her better. This has been so crazy for my husband and me, but we are coming to terms and trying our best to keep Hannah healthy.

My advice would be to stay strong and don't believe everything you hear or read. Everyone's story is different. Don't give up, and keep searching for the answers.

I am working hard at getting Hannah a third and fourth opinion. I am trying to educate myself on feeding her better.

# **“If you have a feeling in your gut that something is wrong with you, there probably is.”**

**Heather R. Morgan, MSW, 40, Dunnellon, FL**

I have neurosarcoidosis and I was diagnosed in August of 2009. When I wrote the first draft of this journal entry, I wanted to concentrate on how lucky I felt receiving my diagnosis in such a short time. After letting my thoughts percolate for a while, I realize that would have been disingenuous. When I was lying in my bed, acutely ill, the only thing that made me feel better was crying. Not only because my eyes were so dry, but because I felt incredibly hopeless that I would ever find out what was wrong with me!

Getting neurosarcoidosis was the worst sickness I have ever had. I know now that I had meningitis, but at the time that I was walking around with a fever, muscle pain, and headache, no one knew what was wrong. Going to the lab to have my blood drawn, then going to the doctor, this is the sickest that I have ever been. I was missing months of work. My husband worked out of town. I was responsible for running the household and taking care of our son. It was incomprehensible that I wouldn't be able to manage that and my part-time job. One night when my leg just “gave out” on me while I was cleaning in the kitchen, I realized that I could not manage my responsibilities safely any longer. The fear of not knowing what was wrong, combined with the fear of never knowing, and the fear of never getting any better drove me to get help from my husband. He had to quit going to work also.

Finally, I had an MRI of my brain. Then a bronchoscopy, and a lumbar puncture. This confirmed my diagnosis. I saw only my primary care provider and a neurologist to get my diagnosis. Many of the people I have met on Inspire.com spend much more time and see many more doctors to get their diagnosis. Speaking only for myself, after I found out that I had sarcoidosis the lucky feeling stopped. I was relieved that I knew what was

going on with my body and its name. The most common alternative diagnosis is lymphoma. But, for me it was like going from the fire to the frying pan. My booby prize was a year on high doses of prednisone, the front-line treatment for sarcoidosis. I am ashamed to say I wished I had lymphoma because then I would have had the big C. People can relate to cancer. Heck, most people cannot even pronounce sarcoidosis! I am sure that many people with a rare disease can relate to this feeling.

Don't accept that your symptoms are all in your head, or that there is nothing to be done to make you feel better.

I didn't feel that I had adequate information and support after my diagnosis, but I was able to search the Internet. I was able to find the sources of support that I needed from friends, family, and others on Inspire.com. After a year of treatment, and lots of social support, I am doing well physically and emotionally!

For others who are still trying to get diagnosed, my advice is never give up! If you have a feeling in your gut that something is wrong with you, there probably is. Work with your primary care doctor, then get a second opinion if your primary doctor doesn't agree with you. Be an active participant in your own healthcare, not a passive recipient. Don't accept that your symptoms are all in your head, or that there is nothing to be done to make you feel better. You can get better, there is help out there, and sometimes it just takes a little longer to find it.

# “I find sharing experiences and information extremely important to my well being.”

“libbo,” 81, New York

I was on vacation in France when my feet began to swell. It was worse in the daytime since the swelling went down overnight. When I returned home I went to see my internist and she assured me it was from all the walking I did in Europe. The swelling was constant but since it didn't really interfere with my daily activities I chose to ignore it. Then suddenly I began having excruciating cramps in my calves. My fingers also began cramping. Back to my internist and now she prescribed a muscle relaxant. During a routine blood test my internist noted a rise in my cholesterol and now she wanted to increase my cholesterol lowering drugs.

One day I noticed a large amount of “bubbles” in my urine. I went on line and got my first inkling that I might have a problem with my kidneys. I reported back to my internist, and for the first time she ordered a urine test. Needless to say, she is no longer my internist.

The swelling occurred on my 79th birthday. I was diagnosed with MCD 9 months later. I am now 81 1/2. I have a new internist and a nephrologist that I see regularly.

Although my nephrologist seems to be doing right by me, I still have a multitude of questions that he is either too busy to answer, or perhaps he feels that I ask too many questions.

My initial treatment was with prednisone, which I tolerated very well. I went into remission after 8 months and relapsed 9 months later. I went back on prednisone (a smaller dosage for a shorter time). I'm in remission once again. I am on a 5mg maintenance program with prednisone and have been in remission for almost a year.

I have been trying to connect with other older MCD patients to no avail. Most of the people on the nephcure site have FSGS and they are all a good deal younger than I am. Although my nephrologist seems to be doing right by me, I still have a multitude of questions that he is either too busy to answer, or perhaps he feels that I ask too many questions.

At any rate I would love to connect with people who are MCD only patients. I find sharing experiences and information extremely important to my well being.

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# **“If you fight the fight, and become your own advocate, you have a fighting chance.”**

**“Adoniasservant,” 46, Sparta, MI**

Let’s face it. We all have a story. All of our stories are different. The reason I tell my story, is so others may be able to have a different ending to their story.

My mother started having medical problems around five years ago. She complained to her primary care physician who told her nothing was wrong.

Two years after she began complaining to her PCP she went to a routine OBG/YN appointment where they discovered she had too much protein in her blood. Something that was flagged at here PCP office for over two years without further tests being done.

**We all have a story. All of our stories are different.**

After the protein was discovered her life was a whirlwind. She was scheduled for every test imaginable, saw many doctors in oncology, kidney doctors (by this time her one and only kidney was

failing), and by the time she was done with her doctors appointment she was told she 1. Was in kidney failure 2. She had Multiple Myeloma 3. She could get a bone marrow transplant (but probably would not survive) and my favorite. . .4. “You have about 1 1/2 years to live. There is nothing you can do, go live your life.”

If my mother had gone home and “lived her life” she would be gone now. I selfishly am glad that she is fighting and hope that for all of you.

My mother has since been diagnosed with Primary AL Amyloidosis. She was told there was no treatment. She refused to listen to that and “googled” treatments. She began her first treatment at Mayo Clinic and that did not work for her. She continued, on her own to find another treatment. She contacted the hospital in Boston who referred her to Dr. Jeffery Zonder in Detroit, MI. Treatments began and her condition has improved.

Although she has since lost both kidneys and is on dialysis, she has not given up. Treatments have been stopped for a while but she is considering starting up again in March.

If you fight the fight, and become your own advocate, you have a fighting chance. If my mother had gone home and “lived her life” she would be gone now. I selfishly am glad that she is fighting and hope that for all of you.

# “Am I just one of the lucky ones who has a little bit of everything?”

Tracy Jones, 44, Mustang, OK

I went to the ER in June '01 for kidney stones the IV was hurting but was told it was just the pain meds. Well, I am one of the lucky ones that it only happens to in 1 out of a million people the medicine reacted with my sympathetic nerves after 8 months of horrible pain and swelling where I could not move my hand, color changes in my hand, temperature changes, and doctor after doctor visits, I was finally told I had complex regional pain syndrome also known as RSD.

Then, in '06, a lot of other things started happening to me. First, severe weight gain without changing anything I was doing, and then the pain all over my body. I hurt so bad i could some days I could not walk from one room to another in my house. My muscles hurt so bad, then the left side of my face started caving in. I look worse then a stroke victim now after 4 years only to be told you have nothing else wrong with you—just quit eating can't tell you why your face looks like that. My pain doctor who helps me with my RSD knew something else was wrong but he is not supposed to be treating me for all my other problems. Then in '07 they said we think you have ovarian cancer so they removed my ovaries only to find it was not cancer but they had so much inflammation they looked as though they had a tumor. Then they found a nodule on my thyroid but said it is small and your levels are fine, don't worry. By Oct. '08 the nodule is now the size of an egg so they removed half my thyroid but said levels are fine, don't worry. Well I am still having every symptom of hypothyroidism—hair falling out, eyebrows leaving, fatigue, hurting all over but my PCP would not listen.

My muscles hurt so bad, then the left side of my face started caving in.

Finally, in 2010, my pain doctor said he would order labs came back and sent me to a endo who finally started me on thyroid meds. They worked for about 3 months then back to where I was. This endo said just give it more time well I decided I am tired of being put off and finally found a great endo who found I am not only hypothyroid but have very low cortisol. In fact I was one point away of being in a coma since then we have also found I have double the growth hormone in my body. So, you put all this together it says should be a pituitary tumor but mine came back negative, so I go back the end of January 2011 to see where we go from here. Also I found out what my face was by using the computer and talking with my pain doctor to figure out it is Parry-Romberg syndrome. I did find a plastic surgeon to confirm the diagnosis only to have my insurance deny helping me fix it. I have seen three rheumatologists who have no idea what Parry-Romberg syndrome is and will not even help me with it. Also I have been diagnosed with ankylosing spondylitis which is kind of rare in women. So if it is not right I will have it trying to see how it all goes together, or am I just one of the lucky ones who has a little bit of everything?

I have also started a support and awareness group here in Oklahoma for RSD. My site is [www.oklahomarsdcrpssupport.com](http://www.oklahomarsdcrpssupport.com). I am always trying to help others.

# “I have never met another person with Myofibrillar Myopathy.”

“KeepsGoing,” 63, Cleveland, OH

I have a very rare type of Muscular Dystrophy called Myofibrillar Myopathy and probably a rare type of Parkinson’s plus disorder called (MSA-P) Multiple System Atrophy - Parkinson’s type.

Here are some of my diagnoses:

1995 – Borderline Diabetes; 2006 – Fibromyalgia; 2008 - Parkinson’s disease; 2009 – Neck Extensor Myopathy (Dropped Head Syndrome); 2009- Myasthenia Gravis; 2009 - Myofibrillar Myopathy; 2006 & 2009 –Cataracts; 2010 – Peripheral Neuropathy; 2010 - Diabetes; 2010 – probable MSA-P

I had an EEG, where they said I had Myasthenia Gravis and probably another Muscle Disease. It ended up that through a Muscle Biopsy and Genetic testing, they said I had a very rare Autosomal Dominant form of Muscular Dystrophy which is under the umbrella of Limb Girdle Muscular Dystrophy. Besides, hip and shoulder weakness it can cause Lung, Cardiac and Neuropathy problems. I had also been diagnosed with Peripheral Neuropathy. Because I wasn’t responding to the medications for Myasthenia, they removed that diagnosis. And after seeing a Rheumatologist after getting the PD dx, he said the Pain I was in was from PD and not from Fibromyalgia.

Through it all, I seemed to be progressing with my Parkinson’s disease more rapidly than other PD patients. The Meds didn’t seem to be working and my balance was very bad. I seemed to be falling a lot plus some bladder problems. I ended up being sent to a Movement Disorder Specialist, because my Neuro began to think I had a Parkinson’s plus syndrome instead of Parkinson’s disease. That MDS said he thought I probably had Multiple System Atrophy, but he didn’t want to put that label on me until we tried to rule everything out. After an MRI didn’t rule it out, he suggested, I raise my PD meds, because, although they were not working, I wasn’t on the maximum dosage. So, I am still waiting to see the Neuro for those results.

So, although I have seen over 8 doctors, I guess I was luckier than a lot of people, because my Neuro for the PD, just happened to be one of the co-directors for the MDA clinic and recognized my Muscular Dystrophy fairly quickly and is following my probable MSA.

I have never met another person with Myofibrillar Myopathy. The geneticist said she thought I was the only person in NE Ohio that had MFM. The largest study was done by the Mayo clinic between 1977-2003 that had 63 people. And I think I have only found around 8 people, including me, that have it on Inspire. Checking on the web, they say that the incidence can't be determined. So, I have been interested in finding or creating Database to find how many people have Myofibrillar Myopathy. I would also like to bring more awareness to MSA. A lot of people, including doctors, have never heard of it.

But, through it all, I have been pretty positive that is why my online user name is KeepsGoing.

"My personal motto is, 'Don't think about what you can't do, but think about what you CAN do.'" So, although I have pain stiffness, slowness, balance, mobility, double vision, bladder, insomnia, fatigue, writing, speech and swallowing problems, I can still take photographs, sing in my church choir, help watch over my nieces and hopefully, to help or inspire other people.

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# **“As long as I take my meds and watch what I eat, I believe I can lead a pretty full life.”**

**Clint, 49, Athens, GA**

I was feeling bad in December of 2000. Every joint in my body hurt, nausea, pains in my side. Thought I had a bad case of the flu. Went to see Dr. Steinbrook and had a physical. Bloodwork came back with high liver enzymes and high creatinine levels. Doctor ordered a sonogram and it showed a shrunken liver and enlarged spleen. I was referred to a Gastroenterologist (Dr. Medina), in March of 2001, I was diagnosed with Cirrhosis. In May of 2001 my Father passed away at 58 from a heart attack. In July of 2001, an endoscopy was performed which showed varices. My platelet count was too low to do a regular liver biopsy so they performed a transjugular liver biopsy. I was told I had End Stage Liver Disease (Cirrhosis) in Oklahoma at the age of 39. Went to St. Louis, Mo. to Barnes-Jewish Hospital (Dr. Crippin) & had more lab work. I was then referred to the Nazih Zuhdi Transplant Institute in OKC (Dr. Wright). First determined to be autoimmune hepatitis. I was given vitamin K shots and hepatitis A & B immunizations, treated with Prednisone, Aldactone, Lasix, Imuran, Neomycin, and Aciphex.

In early 2002, my wife hit me with the news she wanted a divorce and asked me to find another place to live. I didn't want the divorce, the last thing I wanted was to move out away from my kids and would have done anything to keep the marriage together after 17 years, but, she said there was no chance on working it out, and wanted me to move out. I guess the past year had taken its toll. Now on my own, slurred speech and neurological problems came hard and fast getting worse daily. Stumbling, falling down, couldn't tie my shoes or button my pants. People could no longer understand my speech, just walking to the bathroom was becoming a chore, couldn't hold a fork or write. Could no longer work.

So I contacted my mother in Florida. She and my Sister drove out to Oklahoma from Florida to get me and took me to my doctor's visits. By that time, I was very jaundiced and had ascites, very tired and constant pain, my liver was failing, my MELD score was high and I was told I had about 6 months, without a liver transplant, to live. Dr. Wright asked me if I had any family that I could stay with, said it would not be a good idea for me to live on my own and since my wife was divorcing me and my sons were minors, they took me back to Florida with them and we immediately started the application for a liver transplant. My hepatologist (Dr. Wright) said since I was going to Florida to stay with family he referred me to the University of Miami Jackson Memorial Transplant Center. With the help of my cousins, brother, sister and a good friend of mine, Alan, who loaded and drove my truck, I was in Florida.

“As long as I take my meds and watch what I eat, I believe I can lead a pretty full life. There was a time we wondered if I would make 42. I'll be 50 this year.”

Day by day the speech was getting worse, and motor skills even more so. I then had to use a wheelchair, I could no longer feed myself and had a hard time swallowing. Then the tremors started. They couldn't figure out what was happening to me. I could not communicate with them to tell them what was happening all I could do was grunt. The Doctors kept saying End Stage Liver Disease isn't causing this. I was tested for Parkinson's disease, ALS, etc. I had MRI's, CAT scans, Sonograms, EKG's, ECG's and bloodwork, lots and lots of bloodwork. My platelet count had fallen to 52,000 (140,000 is normal). If I just bumped something my skin would tear and I would bleed for an hour. Was admitted to North Collier Hospital diagnosed with hepatic encephalopathy. Was there about a week. My Mother had to fly back out to Oklahoma to finalize my divorce for me. I took Lactulose 4x a day for about a month to get rid of Ammonia in my blood. During one of the test appointments at UMJMMC, I had a spell, fell down couldn't get back up and was admitted to the hospital for about a week. More tests.

By then I could no longer stand, walk, talk, use my arms or hands, had dysphagia, drooling, and severe muscle spasms, Finally a physician, Dr. Jau at UMJMTC pre transplant clinic listened to what my mother was telling him, "I am noticing changes every day for the worse." He then remembered a study of Wilson's disease in Medical School and noticed the Kayser-Fleischer rings around my eyes without the slit lamp. Ordered a 24-hour urine and ceruloplasmin test. And found I was loaded with copper—it was everywhere, blood, eyes, and brain. Diagnosed with Wilson's disease, he started treatment with Zinc and Penicillamine (Cuprimine, Depen). I went home and on about the third day I broke out in hives and became lethargic and non responsive. The EMT's took me to Cleveland Clinic Hospital in Naples, Fla. the town where I was living. I was admitted. Almost catatonic, by this time I had no motor skills, no speech, couldn't make a sound or press a button to call for assistance. Couldn't move any part of my body. For the most part I was quadriplegic. The Gastroenterologist at Cleveland Clinic immediately changed the Penicillamine (Cuprimine, Depen) to Trientine (Syprine) and started more rounds of tests. The staff there was great and saw to my every need. I was in the hospital for a month or more. I started showing signs of improvement. I then saw Cleveland Clinic's Dr. Hanson (neurologist), and Dr. Avalos (gastroenterologist), and they told me the Wilson's disease was caught too late and the neurological damage was probably irreversible. Totally non-ambulatory, I then had to be admitted to a convalescent home to be cared for until my mother was able to retire from her employment to take care of me. I was bedridden for nearly a year, confined to a wheelchair for at least another.

Had they caught mine earlier  
I would not have endured  
about 8 years of hell.

In the meantime, I had an appointment with LifeLink at Tampa General Hospital for a transplant and they ran tests and I was told my liver was improving (the only organ in the human body that can regenerate itself) and I might not have to have a transplant. Best news yet! Over the next couple of years I spent going

to Physical Rehabilitation, Speech Therapy, and endless Doctors appointments. Had to learn to walk, speak, write, eat on my own, retake my drivers test and motorcycle test. Had a lot of muscle atrophy from the past couple of years, muscle cramps & tremors for a long while. The only way I could communicate was with a Letter Board, Spelling Ace, or laptop. Made it to both of my sons' graduations 2004 (in a wheelchair) and 2006 (walking, light tremors) and was able to drive out and attend my 30-year class reunion in 2009.

In 2006, I moved to Georgia and was referred to Dr. Norman Gitlin, my hepatologist whom I now see every six months, him and PA-C Cathey Williams at Emory University Hospital in Atlanta. I was feeling so good I thought I could help a friend who was working, building a balcony, lost my balance took about a 12' head-dive off a retaining wall onto the asphalt broke my right wrist in three places and broke my jaw and had 15 stitches in my chin. But here it is 2011 the speech is not the best but I am understandable. I'm walking, talking, still have fatigue, insomnia, and bouts where I lose balance (I have 3 ruptured disks from falls taken learning to walk again) but other than that I'm fully functional. I now take zinc to block the Copper from getting in and Syprine (1500mg/day) to pull it out and will have to for the rest of my life (I can deal with that). It's been tough but the worst is behind me. As long as I take my meds and watch what I eat, I believe I can lead a pretty full life. There was a time we wondered if I would make 42. I'll be 50 this year. The best course of action is to be diagnosed early, if they catch it before any damage is done they can treat it with medication. But like me, if it's caught too late you can still come back. Stay positive and never give up hope. It's been a long road. A lot of things happened in between. Had it not had been for the prayers, support of family, friends I would not have made it to 43. So doctors if you have any patients with liver trouble having any neurological symptoms such as balance, slurred speech, altered gait, tremors, order a 24 hour urine test, a slit lamp exam (ophthalmologist), and check ceruloplasmin levels. If they are not looking for it it's hard to detect until it's too late. Had they caught mine earlier I would not have endured about 8 years of hell. For 39 years I had no symptoms or issues at all. If you have any siblings or children have them tested also. Though it is genetic, I hope mine never get it.

# “I have asked his three docs to work together on his symptoms.”

Arlene Johnson, 55, Carson, WA

My husband Steve, 60, began to have severe headaches on Tuesday and on Thursday afternoon, May 25, 2010, he was sent home from work but his boss because he was confused and disoriented. He had a seizure, rolled an F250 truck and went by ambulance to the hospital. He missed hitting anyone, missed a gas line by 6 inches and we think he had angels on his shoulders. The docs said he had fallen asleep, or had a brain injury from the wreck. His CT scan was clear. We kept telling them it had begun a few days before. Finally I refused to let them take him upstairs until they did an MRI.

The ER doc did the MRI during the night and said it could be three things, a brain bruise, a tumor or an infection. They said they would treat all three. Because it was a holiday weekend it took six days to get the results but they treated him the entire time. They did follow up MRIs and spinal tap and the infectious disease doc was convinced it was Herpes Simplex Encephalitis (HSE), which it was. Steve went home with 21 days of antibiotics.

We relish every good day and suffer through the bad with the help and love of our family and friends.

He recovered enough in two months to be able to drive, and go back to work part time—he is retired from his other job. His neurologist said he has made a remarkable recovery. However, he still has bouts of severe nausea, and fatigue. I have asked his three docs to work together on his symptoms—thank heavens for Kaiser’s email system. We relish every good day and suffer through the bad with the help and love of our family and friends.

# “We can’t give up. We are not done here yet.”

Kathy, 54, Hughesville, Pennsylvania

I am not happy with the thought of complete blindness but am thankful to still be here and feel the sun on my face and wind in my hair.

I have serpigenuous choroiditis and I only have very limited vision left in my right eye. (My left eye is all scars and no vision left. I was diagnosed in 1998 and it took six months and >6 MDs to diagnose. I have been to major medical centers like Will’s eye and Wilmer eye Institute (were I continue to go now) before they were sure of my diagnosis. I have tried all the so called recommended treatments (meds, chemo, more meds) and nothing will stop it. Little is really known about this as to why it happened, to who, how to stop—but I won’t give up. Everyday new ideas and things are being done to prevent blindness.

I am not happy with the thought of complete blindness but am thankful to still be here and feel the sun on my face and wind in my hair. We can’t give up. We are not done here yet. My motto is never give in and keep moving forward.

# **“When I was younger, I never thought I’d meet another single person outside of my family with this disease.”**

**“kkdk,” Maplewood, NJ**

My story is a bit unusual for Fabry’s disease, I think, because I’ve known about it since I was little. I’m now in my 40’s. When I was younger, I never thought I’d meet another single person outside of my family with this disease. Although the population is still small, the network and support, treatment, etc., is more than I had imagined. My father had a kidney transplant in the early 70’s and he was diagnosed when my mother was pregnant with me. Although he passed away in 1989 related to Fabry’s, his story is much more typical of the delayed diagnosis.

I thought until about six years ago that I was ‘just a carrier’ with some symptoms until more research on females has suggested otherwise so that was a big change in how I have approached my treatment. I have two children as well. My six-year-old was diagnosed in utero and has been receiving treatment since he was three. My younger son I had through PGD/IVF to ensure that he would not have the disease.

Thanks for all the continued efforts to increase awareness of rare diseases like Fabry’s.

# “I’m just learning as I go.”

“mlofferosky,” 34, Milwaukee, WI

I was born with Tuberous Sclerosis. I was diagnosed in infancy. I had a tumor growing behind my right eye at about 2 weeks old. They removed my eye and I have had a prosthetic eye since. I am still learning about my disease as I go. I still feel like I don’t know as much as I should. I remember taking a lot of tests. I had EEGs, MRIs and more from an early age. I had seizures since I was little until about age 12.

I had started feeling auras again a few years back and had been put back on seizure medication which I am still on (Carbatrol 2x daily at 300 MG each) and also with my TS comes delayed fine and gross motor skills. I have sleep issues. I have what they call angiofibromas on my face which are bumps on my nose and elsewhere on my face. I have depression and Seasonal Affective Disorder, anxiety disorder, possibly more.

My disability is one of the reasons I lost my last job. I am currently unemployed and looking and also trying to get Social Security Disability. I have a lack of concentration. I used to love to read but now I can’t really concentrate on it. I have sleep issues which is also related to the TS. It’s frustrating because I’m just learning as I go. The clinicians I have seen are many. I was studied growing up at the University of Wisconsin, Madison. They liked me but I don’t have much memory of it. Since I was diagnosed at birth and because I was off my medication at about 12 when my seizures initially stopped (also correlating with my parents’ divorce) I really don’t have much knowledge of TS, only what I learn talking to others and I just find as I go that I’m learning daily. Now that I’m unemployed and have no insurance I find it harder. I see a doctor at a free clinic and am able to get some meds for free or at least generic. If someone finds out they have just been diagnosed, I would recommend that if they have the medical resources to find out as much as they are able to.

# “I pray for a cure every day.”

“Joan45,” 65, Tennessee

Familial Amyloidosis Polyneuropathy: That is the disease my husband has. Unfortunately, it has taken about two years before he was diagnosed. He started with electrical pains in his feet. Our family doctor at the time brushed it off and told him to get inserts for his shoes.

Finally, as things got worse, he went to a neurologist and also changed family doctors. Suggesting more than one opinion, we proceeded to go to number two neurologist who wasted six months just watching him walk. Finally, number three said you have CIDP. Nope. Ten months with this and wrong diagnosis: By now, my husband was falling daily, losing feeling in his feet and going up his legs. Sepsis from Prednisone and Imuran, then IVIG infusion did not help nor did plasma pheresis. He was in so much pain that he could not even stand having a sheet on him at night. We went to another hospital and he was diagnosed correctly. A second facility did many tests but said he was too old for a liver transplant at age 63. Also, as we left, they thought they had a trial for him but another disappointment as it was not his mutation. I would have thought they would have checked that first. We had driven 1,150 miles with nothing for him.

One good thing came out of this as the genetic doctor told us to keep looking at trials in case one opened up. He also disagreed with the one doctor saying you need to be under 50 for a transplant. We went home very depressed and I really did not want to look on the computer for more info but I knew I had to. My poor husband could not bear to read about his disease, so I began again. I did find one trial that was closed at one time and opened up again. My husband became the last one to be accepted. Although it is supposed to slow the progression there still needs to be something to STOP THE PROGRESSION AND CLEAR ALL AMYLOID THAT IS IN HIS BODY WHICH WILL DESTROY HIS ORGANS. Sad thing is his uncle died of this dreadful disease 25 years ago and because it is so rare it still is misdiagnosed and still, no known cure. We

You must  
advocate for  
yourself and be  
strong.

wrote down amyloidosis on one of the papers we were given to fill out from the third neurologist even though we never knew originally that this disease his uncle died of could involve my husband. The neurologist never read it or acknowledged it.

There are some wonderful doctors out there but we also found out that you have to fight to get copies of medical information as we were turned down not once but twice on the biopsy so there are some not so great doctors too. You must advocate for yourself and be strong. We are now going to two doctors who are researchers and also using the support system for familial amyloidosis polyneuropathy better known as FAP. We have never heard again from the doctor where we were told, "You're too old." As it turns out maybe he did us a favor as liver transplants are harder on older patients and my husband is still around and still in the trial. I pray for a cure every day.

# “The only advice I can give is to trust in yourself, your love and your beliefs.”

Melissa Valdez, 35, San Antonio, TX

My son Benjamin has TSC. About two weeks before he was born in November 2009, I requested one final ultrasound because I wanted to know how much he weighed. Up until this day, everything was perfect throughout my pregnancy. As soon as we saw his heart (we had seen it so many times before!) we noticed white spots. I asked the technician what they were and she told me she didn't know but the doctor would talk to me (of course she didn't want to give bad news). She looked at the spots several times, and from different angles to take measurements. The doctor explained that we needed to go see a fetal cardiologist (I didn't even know that title existed!) They rushed us off for a more detail sonogram. At that sonogram, I was told it was likely that he had TSC.

Once the cardiologist confirmed that there was no restriction in blood flow, that made me feel better but as I read online about TSC it was very stressful wondering what was going to happen. They delivered him at 38 weeks, via C-Section to lessen the stress on his body. At the delivery, the NICU, and cardiologist was standing by waiting to care for him from the beginning. He had nine rhabdomyomas at birth but was otherwise very healthy. When he was three weeks old he had an MRI that revealed several cortical tubers. The neurologist prepared us for the absolute worst and explained that he would most likely

We are doing all the research possible to ensure we have as much information possible to help our son through any symptoms he may endure.

be severely mentally retarded. At about six weeks, his ash leaf marks appeared. By the time he was five months old he had already seen the cardiologist (multiple times), neurologist, ophthalmologist, geneticist, had MRI's, CT scans, EEG's. . .He was diagnosed quickly and we received the best care I can imagine. My only complaint is the hospital drew blood for the genetic test right after he was born but they lost it. When the geneticist went to ask them about it, they told him it was never drawn, though I have paperwork saying it was. When he was 13 months old I went to have it drawn again, but since the Athena lab (the only one to run the TSC test) is in Massachusetts, and the weather was bad, the blood didn't make it there in time due to weather delays and was un-testable. So, we still have not found out those results. Luckily for us, my son has no experienced any seizures, does not have any tumors in his kidneys, lungs, or optic nerve.

Had it not been for that final ultrasound, we wouldn't even know he has TSC, we would just think he had white birthmarks :-). I sometimes wish I hadn't asked for another ultrasound because I would have been able to enjoy more time with him without all of the stress. But, on the other hand, I am very grateful. I know this is a tricky one to diagnose and a lot of people have to go through a lot of testing prior to receiving a diagnosis. Now, if/when my son starts having problems that I know are related, we will not have to "discover" what is wrong. We are doing all the research possible to ensure we have as much information possible to help our son through any symptoms he may endure.

The only advice I can give is to trust in yourself, your love and your beliefs. Do all that it takes to support your loved one. Being "different" while growing up is very difficult. . .all we, as parents, can do is to learn all that is possible about our children's rare disease so we can help them understand when they begin to ask questions.

# About Inspire



Princeton, N.J.-based Inspire creates safe, moderated online communities, organized by medical condition, for patients to share information and to support one another.

Inspire builds and manages these communities in partnership with more than 70 national patient advocacy and support organizations. For more information, go to [www.Inspire.com](http://www.Inspire.com), or email [team@inspire.com](mailto:team@inspire.com). Follow Inspire on Twitter at [www.twitter.com/teaminspire](http://www.twitter.com/teaminspire), and on Facebook at [www.facebook.com/Inspiredotcom](http://www.facebook.com/Inspiredotcom).

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