**Case 1:**Carole

**Age:** 24

**Ethnic Background:** Caucasian/White

**Country:** United Kingdom

**Symptoms Began:**8 months ago

**Symptoms detail:**\*Pre September 2013\* Significant abdominal bloating - worse after eating but ever present and sometimes random. Incredibly fatigued and lethargic. \*Early September 2013\* Regurgitating acid into mouth constantly. Constantly nauseous. Band of pain under ribs - Tight crushing feeling. Pain scale 5 all the time - worse when laying down and after eating. \*Early November (Seemingly immediately following a Gastroscopy)\* Feels as though hit hard in very centre of top of stomach - Pain scale 5 all the time, up to 8 seemingly randomly as well as after eating. Sometimes hitting 9.5(!) on the pain scale. \*Mid November\* Started vomiting at least once daily - worse after eating. Constipation and diarrhoea. \*Beginning of December\* Pain that comes in waves and feels like being squeezed hard in lower right hand side of stomach just above pelvis - Pain scale 5 to 8 seemingly randomly. \*Mid January 2014\* Severe and continuous joint pain particularly in hips and knees. I believe visible swelling on knees and bruising around pelvis and elbows.

**Note to previous symptoms:**\*December 2013\* Was in hospital for 5 weeks from 12th November to 20th December. Increased pain in upper, centre of stomach and pelvic area. Very heavy period. Increased pain has continued since. \*February 2014\* Readmitted to hospital with even further increased pain to upper stomach. Shortly after admittance experienced an extraordinarily heavy period that alarmed a couple of nurses. Magnesium levels low again and brought up with infusions. Prescribed 6 Magnesium Glycerophosphate 4mmol/tablet per day to attempt to maintain. \*March 2014\* All pains continue. Magnesium levels have fallen despite supplements and as many magnesium rich foods as possible. No explanations offered for cause of continuing magnesium deficiency.

**Current medications:**

* Insulin (Novorapid / Lantus), as required / 20 units, daily
* Ursodeoxycholic acid, 500mg, 3 times a day
* Cetirizine, 10mg, once a day
* Chlorophenamine, 4mg, once a day
* Hydroxychloroquine, 200mg, twice a day
* Lisinopril, 20mg, once a day
* Bisacodyl, 5mg, once a day
* Cyclizine, 50mg, 3 times a day
* Domperidone, 20mg, 4 times a day
* Gabapentin, 300mg, 3 times a day
* Lanzoprazole, 30mg, twice a day
* Oramorph, 2.5ml, as required up to 4 times a day (using about twice on average)

**Problems with specific body parts or systems:**

* Abdominal or digestion
* Abnormal bleeding or bruising
* Joint or muscular

**Personal Medical History:** Constant ear infections as child - grommets when 14. Tonsillitis many times through childhood. Used to suffer asthma as child. Subsided late teens. Appendix removed when 17. (2007) Admitted to hospital with suspected pneumonia. (2008) Routine blood tests flagged abnormal Liver Function. (May 2011) Admitted to hospital with undiagnosed abdominal pains. Upper right hand AND general. Eventually subsided after months of pain control. (Aug 2011 to March 2012) Primary Sclerosing Cholangitis (PSC) diagnosed to fairly high confidence. (March 2012) Admitted to hospital again with upper right hand abdominal pain. Gall Bladder removed. (March 2012) Pain subsided completely and painkillers stopped by July 2012 July 2012 to end August 2013 - very few problems except for three blood-clots in right calf / knee. (November 2012)

**Family Medical History:**Both parents Type 2 Diabetic.

**Personal lifestyle:** Student (but drank very little!) Non smoker.

**Diagnostic or imaging test abnormalities:** Gastroscopy - small hiatus hernia and mild gastritis. MRCP of abdomen - normal bar slight dilation of bile ducts - discounted as "likely caused by opiate pain medications." CT Scan of head and abdomen - Possible torsioned ovary - discounted by gyno doctors as symptoms "not typical". X-Ray's of abdomen and chest - Normal bar constipation. Barium swallow - Normal. Transvaginal ultrasound - Normal. Endoscopic ultrasound - Normal. Numerous blood / stool / urine samples - possible Porphyria markers (conclusive blood test believed to be negative (not been told otherwise yet.)) \*Liver function awry \*Magnesium very low \*Potassium very low \*Other bloods "slightly atypical"Gastroscopy and colonoscopy repeated in February 2014. Gastroscopy again showed nothing significant. Colonoscopy was not fully clear but doctors were confident nothing significantly untoward was present.

**Personal struggle:**\*Unable to work due to pain. (Or indeed concentrate on hobbies or even books.) \*Difficulty eating due to pain and nausea has caused significant weight-loss (13kg since November 2013). \*Unable to sleep and awoken from sleep from pain. \*Starting to feel pretty 'down'.

**POSSIBLE DIAGNOSIS:** Post Cholecystectomy Syndrome.

= The patient may be experiencing Post Cholecystectomy Syndrome which is described as the presence of symptoms after cholecystectomy. Two types of problems may arise. The first problem is continuously increased bile flow into the upper GI tract which may contribute to gastritis and esophagitis. The second consequence is related to the lower GI tract where diarrhea and colicky lower abdominal pain may arise.

= Certain medications like lanzoprazole or proton-pump inhibitors which the patient is taking cause hypomagnesemia which could result in lethargy/fatigue that the patient is experiencing. Magnesium helps relaxes the muscles and if there is insufficient amount of magnesium in the body, muscle cramps occur. I believe this also causes the heavy period.

**Case 2:** Severe swelling, increased painful sensation throughout body, hand joints dislocate

Johna

48 years old

Canada

**Ethnic Background:** Caucasian/White

**Symptoms Began:** 1 years 7 months ago

**Symptoms detail:** Severe swelling of all body parts including extremities and internal organs. Severe sharp and aching pain. Limited mobility. Sleeping in a recliner due to shoulders being compromised if I roll around and severe pins and needles sensation causing sharp pain enough to wake me. Heart palpitations. Compromised joint integrity. All symptoms increase in severity with precipitation.

**Current medications:**

* Actonel 35mg 1/week
* Folic acid 5 mg daily 6/wk
* Leflunomide 10 mg 1/diem 7/wk
* Temazepam 30 mg 1/diem 7/wk
* Mirtazapine 30 mg 1/diem 7/wk
* Glumetza 500mg 1/diem 7/wk
* Chromium 500mcg 1/diem 7/wk
* Cinnamon 3000 mg 1/diem 7/wk
* Multivitamin 1/diem 7/wk
* B12 1000mg 1/diem 7/wk
* Vitamin C 1500 mg 1/diem 7/wk
* OxyNeo 10mg 2/diem 14/wk
* Calcium 1200mg 1/diem 7/wk
* Pregabalin 25 mg 2/diem 14/wk
* Raberazole 20 mg 1/diem 7/wk
* Methotrexate .9ml 1/wk
* Oxycocetacet 5/325 prn

**Problems with specific body parts or systems:**

***Heart or cardiovascular***

Heart Palpitations. The heart issues happened several times in July and August. Increasing in severity and duration each time. The heart issues did not re-appear until December and have been once or twice a month with no notable increase in pain.

***Abdominal or digestion***

Abdominal swelling. The abdomen swells all over up to 42 inches approximately 48 hrs. before snow or rain. We know its organ swelling due to results of an ultrasound. It can stay swollen until the weather passes or reduce slightly after 24 hrs. My waist is usually 32 to 34 inches. My generalized swelling changes constantly causing me to wear athletic clothing that expands and reduces with it.

***Joint or muscular***

Compromised joint integrity. The joint and muscular issues are constant but the areas effected seem to vary. For example last March it was my feet ankles and hands. Lately its been my hands, thumbs, shoulders and back. It can be worse or more limited in the morning and at night.

***Whole body***

Increased pain sensation and severe swelling in whole body. When the swelling goes down the pain usually reduces BUT I have had swelling without pain and conversely I’ve had pain without swelling. These occurrances are rare. This has been ongoing for 18 months.

**Personal Medical History:** 3 kidneys tonsils removed age 4 fertility issues: 3 miscarriages 4 term pregnaciesoverectomy left ovary 1993 bowel adhesions requiring surgery twice appendectomy due to endometrium in appendix yet no other areas in body including uterus abcesses post-op on multiple occasions i.e. after appendectomy, ectopic, breast reduction and partial adomnioplasty Sepsis in armpit after multiple breast infections Infection specialist investigated possible Lupus 3 times removal of sweat glands in groin due to severe infection Motor Vehicle Accident July 2001 mild ABI, severe deep soft tissue damage fibromyalgia arthritis bowel scope and upper GI (Crohn’s disease scopes and biopsy’s are clear.) flu twice and then staph infection Oct 2012 took medical leave March 2013 as I was no longer able to use my hands well enough to dress never mind look after my family lumps on tendons in feet late June 2013 Treatment for Rheumatoid arthritis despite neg blood work Complete loss of sex drive early onset puberty age 9 early onset menopause age 45

**Family Medical History:** Heart disease Diabetes Arthritis (all forms) Fibromyalgia Rheumatoid arthritis Autism

**Personal lifestyle:** Educational assistant in a public school for high behaviour autistic males PSW Smoke 1/2 pk to pk/day for 30 yrs. and continue to do so Social drinking only 3-4 times to 8-10 times per year Hyperflexible until car accident (2001) 10,000 steps per day plus gym class support until Oct. 2012

**Diagnostic or imaging test abnormalities:** As long as I can remember blood work shows severe white cell and inflammatory cell activity Neg to lupus, rheumatoid, or any other blood work for autoimmune inflammatory disorder bone density and x-rays show excellent bone condition

**Secondary or partial diagnoses:**

* Sjogren's Syndrome
* Prediabetic

Personal struggle: Sick of being sick and sick of being in pain Had to hire a full time PSW to manage household needs and personal support 4 children with high functioning Asperger's 2 children with serious scoliosis condition requiring surgeries Husband is 11 yrs my senior and had knee replaced last summer plus he works full time My hands are so bad that my thumb dislocates at will I had a normal to high sex drive prior to all of this Sleep is not quality due to pain levels serious reduction in mobility and i can barely wash and dress myself (this process can take hrs to achieve) My children do not remember me well as I have been unwell most of their childhood Depressed on a regular basis but I keep trying I think I probably have lupus, sjogrens, and rhuematiod on top of my arthritis and fibromyalgia I felt as though the intensity of the swelling was so bad to my organs that had the lumps in my feet not occurred I wouldn't have had the methotrexate treatment and I would've been dead by Christmas The methotrexate has prevented it from progressing but it isn't my cure

**POSSIBLE DIAGNSOSIS:** Lyme Disease. Lyme Disease often mocks autoimmune disorders and even causes them undetected.

**Case 3:**Alphonse

31 years old

Kentucky, United States

**Ethnic Background:** Caucasian/White

**Symptoms Began:** 5 months ago

**Symptoms detail:** This is the log I've been keeping since symptoms started so I could review with my doctor. 12/9/13 \* Early signs of cold / sinus infection. Common for me this time of year (typically diagnosed with Sinus Infection either in Dec or Jan the last several years). 12/10 - 12/13 \* Felt miserable, with fairly consistent symptoms (green mucus, tired, achey, congestion, etc.) 12/14 \* First noticed a mild pain, near the Pelvis / Hip Joint on the front side. General groin area, on RIGHT side only. Thought that perhaps this was some Lymph Node swelling, which prompted me to think I had a Sinus Infection and not a common cold. \* Went to Kroger Little Clinic. Received Prescription for Augmentin and Refill of Flonase. \* Began meds, and took for ten days (full length) 12/15 - 12/23 \* Cold / Sinus symptoms slowly improved over the first 3-5 days of taking Augmentin \* In this same period, I stopped noticing the dull groin area pain 12/24 - 1/1/14 \* Felt pretty fine, overall, until the night of 1/1 where I started to feel like a cold was coming on again 1/2/14 \* Woke up feeling miserable, same as in early December. Tired, Achey, Slightly Nauseous (nothing severe), Green mucus, etc. \* Note: was a big barometric pressure drop around 1/1 - 1/2 if that affects sinuses? \* Self treated, rest, water, neti-pot, fruits, veggies, hot drinks, etc. \* The cold / sinus symptoms gradually improved between 1/3 - 1/5. Only mild symptoms exist. \* \*\*\*However, during this same time frame, I started to notice the dull groin pain again. Primarily on the right side where leg meets pelvis, where I believe the Inguinal Lymph Nodes to be located. The pain would come and go. Was mostly a dull ache without a defined exact location. Primarily Right Side. 1/5 \* Drove to Nashville for work, noticed the discomfort much more while driving. Seems to be somewhat aggravated by sitting. Still mild, dull ache 1/6 \* In and out of meetings, walking around. Noticed slightly, but not too much. 1/7 \* Drove home from Nashville, noticed the pain more. Slightly stinging on occasion. Called Dr. office for appointment. Between 1/7 - 1/9 \* The dull pain occasionally occurred on the LEFT side hip/pelvis joint. General lower abdomen discomfort, especially when sitting. Heavy feeling. The pain seems to move around somewhat now, with even occasional mild pain on inner right thigh, very briefly. \* Somewhat more sharp pain around/beneath pubic bone \* Generally achy in back and joints 1/10 \* Slight pain, occasionally on inner right thigh. Did blood work at dr office. Appt moved to Monday the 13th. \* Later in day started to feel congestion type symptoms with LEFT side of head/face. Sore throat on left when swallowing, ear ache, face and scalp sensitive. All on left side. Continued overnight with ear ache being more intense. \* 120mg Sudafed "12hr" \* @ 10:30pm 1/11 \* AM: Congestion symptoms persist. All on left side. Not too achy compared to recent days. Don't currently notice the groin / abdominal pain. Ear pain not as constant as it was overnight. Notice mostly when swallowing. \* 400mg Guaifenesin \* 500mg Acetamenaphin \* @ 9:15am \* For entire day, left scalp, face, inner ear and left throat in specific spot was moderate to severe pain. Pain level 8/9. \* PM took another 120mg Sudafed and 500mg Acetamenaphin \* Groin pain was virtually non-existent through entire day. 1/12 \* Left face etc pain present when woke. Pain level 5/6. \* Took 120mg Sudafed and 2 extra strength excedrin. Which took edge off and brought pain level down to 1/2, and eventually 0. \* Dull lower ab groin pain very minor. Barely noticeable. \* Scalp / Fain pain returned around 7pm and overnight 1/13 \* Mild scalp, fain and ear pain (left side) and some mild left side nasal congestion. \* Took 120mg Sudafed \* Mild low middle groin pain, comes and goes \* Saw Dr. Said blood work and urinalysis all looked normal. Suggested possible infection, and possibly prostatitis. Prescribed 10 day supply of Levaquin antibiotic. 1/14 - 1/19 \* Took meds at lunch every day. More or less felt the same, with slightly improving sinus symptoms. Groin discomfort was mild but noticeable and was mostly consistent. Seem to notice more in the morning but also occurs throughout the day. 1/20 \* Both groin and sinus symptoms were the best they've been as of today. 1/21 \* Woke feeling similar to 1/20. Overall pretty good. Brief noticeable pain, lasted a couple minutes. Right of public bone. Specific location. Aside from that felt pretty good all day. 1/22 \* Dull discomfort, mild, returned. Lower groin area. Modestly noticeable throughout day. More so noticed when sitting. \* Last day of Levaquin prescription \* Called Dr about refill or plans. Called back and said wait until Friday and call back to see about refill or next steps. 1/23 \* Groin discomfort more noticeable than yesterday when woke up. Not severe, but uncomfortable and noticeable. 1/24 \* Same as prior day in the AM. Felt pretty good through mid afternoon. Didn't notice much. More discomfort overnight. \* Called Dr again. Wouldn't refill my prescription and wanted me to come back on Tuesday 1/25 \* Similar discomfort when woke. Mid to lower groin. Somewhat like a mild muscle ache or strain. Dull pain. Mid-low abdominal. Took Motrin but didn't seem to do much.

**Current medications:**

* Levaquin, 500mg, Daily - Just Finished a 10 day supply. None currently

**Problems with specific body parts or systems:**

Head or neck

Abdominal or digestion

Joint or muscular

**Personal Medical History:** Overall healthy and in good condition. Basic annual physical and blood work shows things in normal range. I do have seasonal and environmental allergies (cat, mold, pollen, etc.) and a related (small) nasal polyp. I take flonase seasonally. No known medical allergies.

**Family Medical History:** Glaucoma on Father's side. Nothing else major which I am aware of.

**Personal lifestyle:** I'm self employed and work at a computer mostly. Though I'm up and down throughout the day. Busy running around with young kids. Not much in the way of "dedicated exercise" Non smoker. Social Drinker (a few beers). No drug use, etc.

**Diagnostic or imaging test abnormalities:** I don't have a copy of the results, but, Doctor did: Physical Exam, Blood work, and urinalysis. The doctor said that all looked normal - nothing unusual.

**Personal struggle:** I never get worried about much. I don't generally seek medical help or expertise for anything (one of those many guys who never want to call the doctor, etc.) However, this has me extremely paranoid. We have a 3 yr old, a 1 yr old, and my wife is pregnant with our third which is due in a couple of months. I'm afraid something bizarre is wring with me. I try not to bring it up too much around my wife so as not to worry her but she knows something is not right and is concerned. Hoping someone can provide some ideas or insights, thank you!

**Other Notes:** As mentioned, I have allergies and every winter end up with a bad sinus infection (and one year even a bad allergic reaction as a result of the infection. The doctor thought possibly this issue was "Prostatitis" but aside from dull groin and abdominal pain the symptoms don't really match up. I have no "burning when peeing" symptom, no ejaculation pain, no sensation of having to pee more often, no blood in stool or urine. I've been married nearly 8 years, and together for 10. 100% faithful so no random contraction of some std. Hope that helps, let me know if I can answer more questions.

POSSIBLE DIAGNOSIS: Sinusitis and inguinal hernia.

**Case 4:**Lester

54 years old

Massachusetts, United States

**Ethnic Background:** Jewish/Ashkenazi

**Symptoms Began:** 4 months ago

**Symptoms detail:** I'm a healthy male, and am very physically active. I swim a mile most days, play tennis, and cycle about 1000 miles a year. I was noted at a medical appointment to have a pulse of 40. Since then, my pulse is often in the low 30s, and doesn't go up beyond 60 even with serious exertion. I feel fine - no dizziness and no fainting. So - essentially no symptoms.

**Current medications:**

* simvastatin 10mg/day
* aspirin 81 mg/day

**Problems with specific body parts or systems:**

Heart or cardiovascular

See above. Low heart rate without symptoms

**Personal Medical History:** Healthy. Low HDL with borderline high cholesterol - 10 year cardiac risk by current AHA algorithm is around 3.2% with or without cholesterol therapy.

**Family Medical History:** Dad MI at 65

**Personal lifestyle:** Vigorous daily exercise Lacto ova pesca vegetarian

**Diagnostic or imaging test abnormalities:** Not applicable

**Personal struggle:** No struggle at all

POSSIBLE DIAGNOSIS: A well man.

= Your resting heart rate is the heart pumping the lowest amount of blood you need because you’re not exercising. If you’re sitting or lying and you’re calm, relaxed and aren’t ill, your heart rate is normally between 60 (beats per minute) and 100 (beats per minute), Stein said.

But a heart rate lower than 60 doesn’t usually signal a medical problem. It could be the result of taking a drug such as a beta blocker. A lower heart rate is also common for people who get a lot of physical activity or are very athletic, Stein said. Active people often have lower heart rates because their heart muscle is in better condition and doesn’t need to work as hard to maintain a steady beat.

“Moderate physical activity doesn’t usually change the resting pulse much,” Stein said. “If you’re very fit, it could change to 40. A less active person might have a heart rate between 60 and 100,” he added. That’s because the heart muscle has to work harder to maintain bodily functions, making it higher.

**Case 5:**Kylie

17 years old

Nebraska, United States

**Ethnic Background:** Caucasian/White

**Symptoms Began:** 1 years ears 8 months ago

**Symptoms detail:** In summer of 2012, Kylie started being extremely exhausted, even after getting 10-12 hours of sleep a night. She then began to have episodes where she experienced the following symptoms: - Eyes would go blank - Difficulty walking - Could answer questions correctly - Could slowly retrieve information. She wanted to sleep. She recovered after hour and a half with no liquids or meds. These episodes increased in frequency and occurred at any time of day. Then in Feb 2013, her episodes progressed and she began experiencing the following symptoms: - Total body collapse - She can hear us, and respond with a yes/no groan - Cannot move muscles - Eyes move back in her sockets and then move back and twitch right to left rapidly. (The eye wiggling hurts the muscles in her eyelids and behind her eyes - it is fast, and continuous. The neurologist told us no one could do that on purpose and believes it to be neurological) - Heavy sweating in left armpit occurs prior to the episode. - Rash on neck - Facial twitching on left side of her face (sometimes right) - Headache - If episode occurs while eating, can push food out of the mouth with her tongue - Can usually answer questions if we physically move her lips to speak - She does have reflex action during the episode in her limbs. Recently, limbs have begun to stiffen The episode lasts from a few seconds to usually a minute or two up to as long as 35 minutes. The number of episodes have increased in frequency to as much as 40-50 per day. They come in clusters often - the other day, 20 minutes worth of episodes, a few seconds or a minute each. Sometimes the episodes are isolated. Update 1/4/2014: The neurologist has put her back on the Xyrem, even though the spinal tap ruled out narcolepsy with cataplexy. She went from 50 episodes one day to 5 the next. Since then, it's been about a week of 0-8 episodes a day. We tried this once before, but it was unsuccessful in helping at all when she was on other meds, such as clomipramine.We found the term 'oculogyric crisis' online- the youtube videos of this eye problem strongly resemble Kylie's eye movements.She has been going stiff more and more during episodes. Her legs, trunk, and sometimes arms. She has been stiff for as long as an hour and a half.Current episodes - a few seconds to an hour and a half... (though had 3 to 4 hour recurring ones in hospital)... muscles go lax, eyelids close, eyeballs go back in sockets then back and forth rapidly (4 Hz per sec), she remains loose or body now stiffens - sometimes just legs, sometimes legs and trunk, sometimes arms, too. She can hear us, respond through clenched teeth, can feel. There is usually some facial twitching. Sometimes if can't verbalize, can respond with eyebrows or quick smile or partial smile or frown.

**Current medications:**

* Xyrem
* Allergy shots, every 2 weeks

Kylie is no longer taking Xyrem, due to it not being narcolepsy. She also has discontinued allergy shots. Her neuro has placed her on Baclofen, which seems to be helping - she is sleeping better and 0-3 episodes a day the last week! She is also on the rescue medicine of Valium for episodes with stiffening lasting 30 minutes or longer.Currently NOT on Xyrem or allergy shots Now on Baclofen, which seems to be helping - down to 0 - 7 episodes a day instead of 50-70 Has valium as rescue med if stiffening lasts over 20 minutes

**Problems with specific body parts or systems:**

***Eyes or vision***

Eyes move at 4 Hz/sec during episodes under lids - they move back towards forehead then side to side She sometimes experiences dizziness, and once in a while double vision

***Neurological***

Body collapses while staying conscious, lids close but eyeballs move, can respond through clenched teeth or facial muscle movement Sometimes body stiffens or part of it, sometimes just limp

***Whole body***

total body collapse as explained above

**Personal Medical History:** Adenoidectomy @ age 7 Tonsillectomy @ age 8 Worms (don't know what kind) @ age 14 Seasonal allergies - has taken Zyrtec in past for it and now takes allergy shots every two weeks Diagnosed with dyslexia as a child, but has done very well with Orton-Gillingham program. She did have a long bout with diarrhea one year ago- December till March. Possibly due to minocycline, which she was taking for acne. After she stopped taking minocycline, she seemed better in a couple of weeks, but noticably lost weight, became lethargic. She also had mild acid reflux. We think Kylie may have experienced her first lethargic episode in April 2012, when she was away on a trip. (This is before she became extremely exhausted in the summer and after diarrhea.)

**Family Medical History:** Maternal grandmother - Extra mammory Paget's Disease of the Breast, diagnosed @ 57 Maternal grandfather - Diabetes, diagnosed @ 75 Paternal grandfather - high blood pressure Mother - sleep walks Paternal uncle - GuillianBarre Syndrome - 3 times @ 33-34 Father - high triglycerides high blood pressure, gout @ 50'sPaternal grandmother - Rheumatoid arthritus Father – gout

**Personal lifestyle:** Kylie was a high school sophomore when she first started experiencing her episodes. She was a starter on the volleyball team, practiced French horn regularly and was preparing to try out for the All-State band. She was on the honor roll at school, had several friends. She exercised regularly and was in great physical shape. She does not smoke or drink.

**Diagnostic or imaging test abnormalities:** CAT - clear Drug tests - clear EEG's - clear EKG - clear MRI's. MSLT sleep test - fell asleep in 2 of 5 naps, one nap - fell asleep in 17 minutes, but went into REM quickly Polysomnograms - clear - mild sleep apnea HLA-DQB1\*06:02 test - positive (checking for possibility of narcolepsy) Spinal tap - checking hypocretin levels - 250.19, so not narcolepsy with cataplexy BUT had been diagnosed with this earlier and episodes disappeared for 4 months on Xyrem... tiredness did not disappear Porthuria test - clear Psychiatric evaluations - cleared Thyroid - we think they checked this and it was clearneuropsych testing. Kylie was hospitalized for almost 2 weeks for drug trials related to disease the dr suspected. Phenobarbital did take her episodes completely away - but left her extremely drunk acting. Valium ended up being more effective than Ativan for a rescue med with the stiffening episodes. Baclofan seems to help. He tested her blood for antibodies related to Stiff Person Sndrome, but they were within normal. He has scheduled other tests for next week - a spinal tap, blood work, and ultrasounds of her abdomen and ovaries. We have also had tests that were negative for Lyme's Disease, West Nile virus, toxicara, and toxiplasmosis.Ultrasound of abdomen areas - clear except for cyst with some calcium on left side - dr not concerned with it

**Excluded diagnoses:**

* Narcolepsy with cataplexy
* Epilepsy
* Migraines
* Non-epileptic seizure disorder
* Lyme's Disease West Nile Virus ToxicaraToxiplasmosis

**Secondary or partial diagnoses:**

* Articulation Perception Disorder

**Personal struggle:** She has gone from being an active vibrant teenage girl to someone who can't get to school on time because she is so tired. She also can't attend classes regularly because she is tired and having so many episodes. She either has to stay in the special ed room or go to class in a wheelchair with a para. She can no longer drive, no longer participate in the sports she loves, and can no longer be by herself. She is embarrassed to go out in public. Even in quiet settings, she is just sitting and not engaged mentally. Our family life has deteriorated - we can't just 'be' --we have to consider what she can and can't do. We have to be home very early so she can get her rest. Her sister (two years younger and one year behind in school) feels like her caretaker instead of just a sister. Good things happen, too - we are tighter in many ways, and we appreciate the small things in life more. In general, our life has come apart - she can't even consider colleges right now, which is what she should be doing. Kylie's episodes are decreasing in frequency since she has been put back on Xyrem. Although this is great, it doesn't give us a diagnosis, and even one episode is too many - she can't drive, can't do much by herself, and is way too tired. When school starts, she is afraid she will be worn out from the day, and more episodes will appear.

**Other Notes: Medicines Tried:**TrileptolKeppra: she had horrible reactions to these - began acting like a small child Topomax: made left hand drop and she couldn't move it for a minute or so at a time Amitriptylene: did seem to help headaches and sleep some, but not nearly enough and as she titrated off of it is when the current type episodes began Xyrem: episodes were better the next day, and disappeared completely for 4 months, except for sleepiness, then had a cold and episodes began coming back Adderall, Adderall ER: made heart race too fast Provigil: helps get her going in AM Effexor ER: seemed to have more problems Clomipramine - evened her moods, but didn't seem to help anything. Also - she doesn't remember A THING from Nov - March while she was on the epilepsy drugs through the Amitriptylene Episodes get worse when she is sick. She has been a tired child since about halfway through kindergarten, but not as tired as the beginning of this. If she can concentrate, she can hang on for a while and avoid experiencing an episode (i.e. a marching band event) Normally her pulse is 60-70 BPM, but we've measured her pulse at 110 BPM in the morning, while she is in bed. Psychiatrists have confirmed that Kylie's episodes are not psychogenic.

POSSIBLE DIAGNOSIS: Multiple Sclerosis.