ABOUT JANET

The unusual muscle spasms began when I was about 15. I called them "charlie horses" that occurred in my feet and calf muscles, sometimes in my forearms. They were painful, and made it difficult or impossible to walk or use those muscles. If I ignored the spasms and continued the activity they did progress until my body involuntarily was spasm-ed into a fetal position. The fatigue after these spasms occurred was sometimes brutal. I learned very quickly to avoid that kind of involvement. I also learned that these occurrences happened when I was involved in exercise, usually extreme exercise, ultimately, the more I did the less I could do.

The first muscle biopsy done in 1980 is the only one of four total that I have done that has survived through time. All four biopsies say about the same thing, and described the sample in the same way as this first one. (see report) The last three all stated that this was an abnormal muscle biopsy indicating "a slowly degenerating muscle disease, type unknown". These were all done between 1980 and 1995. The medical work ups through those years was extensive, thorough and provided information that I did not have MS, any known (at the time) form of Mitochondrial disorder, nor did I have Lupus, or a myriad of other disease including Glycogen Storage diseases.

During that time period I saw two different neuromuscular specialist who just happen to "see" in office what happens when my muscles fail, they simply quit. Both diagnosed me with some form of Muscular Dystrophy, type unknown, nothing I could do about it, so learn to work within the parameters my body was dictating. (5/2015 possibly defects in dysferlin protein, chromosome 5, Distal Muscular dystrophy, diagnosis not yet made)

In 1987 I was diagnosed with Chronic Fatigue Syndrome and that diagnoses coupled with a unknown neuromuscular disease is what my Social Security Disability was based on.

I have been diagnosed with or treated for the following:

Pre-diabetes 1972
Muscular Dystrophy 1978
Hysterectomy 1979 (both ovaries retained) from Dalkon Shield damage
Probable MS -- 1979
Lypomas on back removal 1984
IBS 1985 routine colonoscopy, IBS is what doctor told me
Chronic Fatigue Syndrome 1986-87
Neuropsych Evaluation 1986, intelligent, normal psych, abnormal testing show unusual form of dementia, fatigue evident in results, CFS
MS genetic testing of five alleles involved w/MS 1994, I had 4, sister had all
Trigeminal Neuralgia 1990
Gallbladder removed 1996
Vocal Dysphonia 1996 or 7 treated by botox injections, still receiving
Lyme Disease 1997, treated, improved 2007, treated, improved
High blood pressure 2003 continue treatment today
Neurocardiogenicsyncopy failed test about1998
Torticulus (head & neck) 1999 treated by botox injections, still receiving
Heart Attack 2003, MI right coronary artery, clot recovered and stint placed
Breast biopsy 2004 and 2006
Genetic testing for breast cancer gene 2006, results negative
surgical correction of deviated septum 2007
B12 very low, 2007 began B12 self injections, continue today, 1ml 3 X wk
Lyme Disease 2008, treated, improved
Inflammation around heart 2008 treated with Celebrex
Rheumatoid Arthritis 2009, A-typical, little joint damage, RA positive factors
Insulin Resistance rather than diabetes or hypoglycemia 2010 to now
Pneumonia, atypical COPD 2010, treated, improved
Higher blood pressure 2012
Cataract removal surgery 2013, both eyes
Pneumonia from accidental acid/base explosion 2013
Angioplasty 2013, 2 new stints placed, 1st stint remains good
Became involved at Scripts medical in San Diego, told genetic testing for heart disease is positive, heart is very slowly failing, but not yet into heart failure. 2013 thru 2014 These records are available.
Low B6 2014 taking 50mg B6
Finally saw Neuromuscular specialist, Dr. Romine, supports genetic testing because no clear direction for specific testing

THESE PROBLEMS HAVE OCCURRED IN MY MATERNAL FAMILY
(from my memory, Grandparents forward)

Juvenile Pagent's Disease -- Grandfather's sister's son
Ovarian (Grandmother's mother died in her 30's) and Breast Cancer and Inflammatory Breast Cancer (my sister died at 50)
Lung Cancer -- my aunt and mother (Adenoma Carcinoma w/ unknown primary, suspected breast cancer as the origin of the metastasis) mother died at 62, aunt at 68
Non-Hodgkin's Lymphoma -- aunt treated and recovered
Heart Attack -- uncle died when he was about 66, cousin at 50.
Alzheimer's -- grandmother, occurred at 95 died at 102
Thyroid Disease -- aunt and sister and sister's daughter
Multiple Sclerosis -- daughter of sister w/IBC has primary progressive
sister w/5 alleles positive for MS also has MS
Trigeminal Neuralgia -- sister, brother, myself
RA -- mother, myself
Monostatic Fibrous Dysplasia -- aunt's daughter, multiple surgeries until she was 13, then stopped all growth and did final surgery.

PATERNAL

Little is known of father's family, only my childhood observations, like my Grandfather who worked as rural mail carrier,retired early, owned his own filling station, then suddenly quit, retired to his chair, had difficulty walking, walked with a cane and died in his mid-sixties. This was not an uncommon occurrence in Dad's family. Dad's paternal family was large, a majority lived in the Chicago area, last name Simpson, connected to the Tillman Tyler Simpson clan. My Grandmother was O'Brien, family from Cork County Ireland, settled in Illinois.