Finally Understood

A Family's Journey with Fabry Disease

By Jean Swanson

"What she needs is some tough love.", the doctor said to Grandpa, "I find nothing wrong with your wife. I believe it is all in her head. Carry her out to the barn or mailbox daily and make her walk back for exercise. Perhaps shock treatment would help". The year was about 1930.

Once a vibrant mother of six, Jennie Johnson had become riddled with aches and pains, with bouts of depression and anxiety in her early thirties.

Grandma's response was to give up, to shut down physically and emotionally. Grandpa Melvin did indeed carry Jennie out for daily exercise, but he would not be able to take care of her long for he too became disabled by heart disease. My father, Orville, would sit by his bedside while the older children tended to the barn chores. Jennie lost her husband in 1931 and moved in with her mother, unable to care for the children yet at home. 19 year old Robert ran the home farm with 10 year old Orville helping before and after school each day, eating a type of gruel they made called "mush". Their 16 year old sister Sarah moved to Cameron to work as a housemaid for a family, earning her room and board in order to attend High School. Three years later, when Jennie's mother passed away, she showed up on the doorstep of her oldest daughter's house, with suitcase in hand. Newlyweds, Una and Otis took her in. In her quiet, odd ways, she carried with her the stigma of an imagined illness, and yet her children loved her devotedly.

Each Christmas Eve, Una hosted a large feast in which the entire family gathered, bearing dishes of traditional Norwegian and Swedish foods. Savory smells from Swedish meatballs filled the air, rumagrut was on the stove, while lefse, krumkake and rosettes lay in wait on the table, ready to be devoured. But Grandma was not a part of the festivities, she would spend most of the evening in her bedroom. I hardly knew her, but I have a vivid memory of someone braiding her long grey hair in the bedroom and wrapping it in a bun before she joined us in the living room.

"Sometimes she wouldn't join us at all," said my sister Dianne, remembering the Johnson Christmas Eve gatherings. "Dad would say it was a good Christmas Eve if Grandma came out of the bedroom. I always wanted her to come out because it made Dad happy."

When she did join us, she would sit quietly in a corner chair or stand watching from the hallway door. We cousins would perform music and readings for Grandma, the aunts and uncles. I remember my sister Dianne playing "The Little Drummer Boy" on the piano with my sister Donna and I singing. I thought it was magical, but Grandma sat in still silence. The Christmas story was read, presents distributed and then began the cousins' long cold walk to the church for a Christmas Eve service. Though her children would have driven her, Grandma stayed behind.

"That woman was nuts!" exclaimed cousin Albert.

“She wasn’t a cozy grandma. She lived with us, and she was crabby and anxious. We couldn’t leave her alone, ever. When she paid her little bit of rent to my dad she would slap it on the table and walk away,” said Linda.

“She was better in her later years,” said Tom to Linda. “I would go and stay with her when your family was gone. We had some nice talks.” (Tom now lives in the two story farmhouse that Jennie and her husband Melvin had once made a home).

“Sometimes she stayed at our house. We connected,” Sandra said. “Grandma and I were both introverts. We understood each other.”

In 2023, Jennie's grandchildren gathered together at my cousin Susan's lakeside cabin. Susan told us about a genetic disease called Fabry which was discovered through DNA tests in her family, and her sister's family. They explained that is passed down from a defective X chromosome, meaning it cannot be transferred from father to son, but father to daughter is 100% and mother to daughter is a 50% chance. It creates an enzyme deficiency that causes tissues in the body to retain waste, often bringing about an array of possible physical symptoms including chronic muscle and nerve pain, as well as insomnia, depression and anxiety. They wondered if the source was Grandma Johnson.

At the time, my nephew Luke was experiencing severe hand and feet tingling, confounding the doctors. When told about our family discussion, his doctor called it the “smoking gun.” Luke’s DNA test revealed Fabry disease. Galafold, a medicine by Amicus Therapeutics, was ordered just when Luke’s pain had flared at its worst. His symptoms subsided. After more testing, 22 of Jennie’s descendants were found to be Fabry positive, including myself and two of my children. Grandma’s symptoms were real. It had taken 88 years to find the cause of her physical pain and emotional problems. She was finally understood.

Jennie's parents immigrated from Norway, near Bergen. The University of Bergen is a center of research for Fabry today and there we find a small symptom graph in a study from one family with the same Fabry strain. We are likely related to the family in the study, whose onset of symptoms is later in life and less severe than other Fabry strains. We can now trace the root cause of unexplained heart, muscle and nerve tissue problems for our family back through generations: father and uncles, cousins and sisters, children and grandchildren.

As for me, in the year 2005, I experienced constant tingling in my hands and feet as well as extreme muscle tightness aggravated by repetitive motion. I was 47 years old. This eventually prevented me from playing piano and guitar for my Music Teacher job and I could not do even simple repetitive tasks, like wrapping presents, without pain.

“What shall we call this?” my doctor said after scratching his head. Unlike Grandma’s doctor, he simply believed me. It was labeled “Chronic Myofacial Pain” and “Fibromyalgia”. I was prescribed muscle relaxers, physical therapy, massage therapy and stretching exercises while I took a medical leave from teaching. I had tools for relief. My family and my faith were a constant well to draw support. Three months later I returned to the job I loved and continued 10 more years, thankful for every moment of music making, and time connecting with students. In retirement I am still able to direct church choir, play a limited amount of piano and give lessons. Symptoms still flare, but it is manageable. I notice a definite improvement of symptoms since taking Galafold in the past two years.

Naming the condition two years ago finally connected the pieces of an 18 year old puzzle, bringing the whole family new medicine and specialists who not only believe us, but are studying us, using our data for research to help defeat Fabry disease. Galafold, the prescription medicine, does not ease symptoms for everyone, but it helps prevent further progression of the disease for our strain. I am thankful to TAF (The Assistance Fund) for providing financial assistance to make it possible for me to afford the medication. Without this assistance I could not afford this life saving drug. I am thankful for Amicus Assist who take an active role in monitoring and educating us along the path.

“Mom, It’s a bad Fabry day,” said my daughter one day about her muscle aches. Her years of migraines have also been attributed to Fabry disease.

“For me too Eliza,” I replied, “I played too much piano the last few days.”

“It’s a handicap, isn’t it?” she replied.

Yes, it is often difficult, but then I remember a Grandma who had worse than nothing, even her sanity was questioned. Pain may have prevented her from helping out around the house or doing simple crafts like knitting or sewing. No doubt she was misunderstood, perhaps viewed as lazy, and her symptoms created anxiety that altered her personality. I think of her now with fondness and a sweet, tender love. I feel as though I understand her, even though in reality I hardly knew her. When I look at the contented smile on her last photo, I have hope that she found inner peace. But this I know, that the world is a better place because Jennie Johnson lived. Her story ends with joy, through the lives of her many amazing descendants.

Jennie Boyum Johnson (1888–1965)

This essay began as an exercise for a writing class at the University of Superior entitled, “Finding Your Inner Narrative”.

Family Photographs

Images shared by Jean Swanson as part of her family’s story.









