LEMS Syndrome 101

Lambert-Eaton myasthenic syndrome (LEMS), a rare neuromuscular disorder, is difficult to diagnose and is frequently associated with cancer. Three people show how a positive attitude is what it takes to live with the disease.

By Cheryl L. Haggard

IS IT POSSIBLE for three people who have never met each other to cite chapter and verse of each other’s life stories? It is possible, if they share the diagnosis of Lambert-Eaton myasthenic syndrome (LEMS).

LEMS is an autoimmune disease that manifests as a neuromuscular (brain and muscle) disorder. LEMS is caused when the autoimmune response interferes with the release of acetylcholine (ACh), a neurotransmitter that activates muscles, regulating many bodily functions and helping carry messages from nerve cell to nerve cell in the brain.

The Mysterious Symptoms of LEMS

Before Sharon Southern was diagnosed with LEMS, she kept asking herself: “What’s wrong with me?” Sharon’s professional life as an employee of an Australian airline and her passion for swimming were interrupted by sudden onset of chronic dry mouth and muscle weakness in her extremities. “My mouth was so dry, I had to keep sipping water just so I could swallow,” Sharon says. “Then I started having bouts of diarrhea, then terrible constipation. When I noticed my left eye was beginning to droop and my body, especially my legs, became weak, I thought to myself, ‘You’re becoming a hypochondriac, and you’re only 38 years old!”

Bill Oehlke had a similar experience with LEMS. Climbing and lifting were all part of Bill’s daily routine as a fire chief. But after 11 years of progressively struggling with muscle weakness, even to the point of needing to push himself up with his arms from seated and prone positions, Bill’s wife of over 46 years knew something just wasn’t right. “When I couldn’t finish talking, chewing and swallowing, I thought I was next.”

Susan Harper, a social worker who lives with LEMS, can relate to Bill’s experience. “I was angry at myself, thinking my muscle weakness was due to laziness,” she explains. “When I was able to feel the bones in my legs, I was afraid that the next step was to plan my funeral.”

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The real next step for Susan, Bill and Sharon was to seek the best possible medical opinion for their mysterious and maddening symptoms, and get the proper diagnosis.

How LEMS Is Diagnosed

LEMS is easily misdiagnosed. After Susan was misdiagnosed with myriad diseases, including chronic inflammatory
demyelinating polyneuropathy (CIDP) and multifocal motor neuropathy, and then rediagnosed with Sjogren’s syndrome, lupus and connective tissue disorder, her third neurosurgeon concluded she might have been dealing with myasthenia gravis (MG), which has some symptoms similar to LEMS. She was then referred to a fourth neurosurgeon, who performed a series of diagnostic tests and scans that could distinguish between MG and LEMS.

The diagnostic tests included an electromyogram (EMG), which measures the electrical activity of muscles at rest and during a contraction, as well as nerve conductions studies (NCS), which measure how well and how fast the nerves can send electrical signals. With these tests, a physician can quickly and accurately begin to crack the code of a patient’s common complaints of weakening extremities, dry mouth and bladder dysfunction.

Dr. Robert Friedman, who consults on MG and LEMS cases for the Muscular Dystrophy Association, explains that there are clear clinical differences between the diseases. Proper diagnosis protocol is important to achieving an accurate diagnosis, particularly because LEMS has an underlying cancer risk, as much as 60 percent. “In myasthenia gravis, when you repeat the nerve stimulation, the muscle response declines rapidly, whereas in LEMS, it’s the complete opposite,” explains Dr. Friedman. “As LEMS patients exercise, they typically get stronger and stay stronger with proper medication.”

Along with EMG and NCS testing, blood work to detect antibodies to acetylcholine receptors is key to confirming LEMS. According to Dr. Friedman, this is where the rubber meets the road. Some 85 percent to 90 percent of people with LEMS test positive for antibodies against voltage gated calcium channel (VGCC), a protein that allows calcium entry into nerve cells, which is required for acetylcholine release. There is evidence that cancerous cells inappropriately make VGCC, triggering the immune system to make anti-VGCC antibodies.

“Once a patient tests positive for voltage gated calcium channel antibodies, we confirm LEMS and go looking for a tumor,” Dr. Friedman continues. “LEMS suddenly takes a back seat, and we go after cancer.”

And timing is everything. “After a positive VGCC antibody test, we begin with everything available to us to find the neoplasm, including PET [positron emission tomography], CT [computed tomography], MRI [magnetic resonance imaging] and chest X-rays,” Dr. Friedman explains, although sometimes it may take up to five years before a tumor is detected. And, “because of the high risk of cancer in LEMS patients, we must routinely check for carcinoma. It’s always looming in the back of our minds.”

Physicians and patients are particularly concerned about a LEMS diagnosis because small cell lung cancer (SCLC) is most frequently associated with LEMS. What is interesting is that 3 percent of patients with SCLC have LEMS, and almost all SCLC sufferers with LEMS have a smoking history. The good news is this: If cancer is not found within two years, the chances of a LEMS patient developing cancerous tumors decrease.

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Treating LEMS

According to Dr. Friedman, successful treatments depend on the patient. Ultimately, a good marriage of medical therapy and support systems can create positive quality-of-life outcomes for patients with LEMS.

Prior to cancer treatment, or in LEMS patients without cancer, immunosuppressant drugs, intravenous
immune globulin (IVIG) and/or plasmapheresis (a plasma exchange where antibodies are removed from the blood) are quite helpful. According to the Muscular Dystrophy Association, IVIG is essentially an infusion of antibodies that might work by dialing down the immune system’s production of its own antibodies, much as warm air tells a thermostat to stop pumping out heat.

For many LEMS patients, symptom relief is achieved with Mestinon, which allows more ACh to accumulate, improving the transmission of electrical impulses, and/or 3, 4-diaminopyridine (DAP), an FDA-designated orphan drug for the treatment of LEMS. “When I arrived home after my first dose of DAP, I sat down and went to cross my legs,” Susan recalls. “In the past, I needed to use my hands to lift my legs. I was able to lift my leg with just my leg! In fact, I almost kicked myself in the head as this was such a dramatic change.”

Living with LEMS

For Bill, Susan and Sharon, a positive attitude is the common denominator for living with such an uncommon disease.

“They made me a permanent greeter at church because I’m so well-known,” Bill joked. “Not known because of my LEMS, but because I’m such a likable guy!” After almost 16 years living with LEMS, Bill defied the cancer statistics. He published two books about the fire department and was working on a third when, in February 2008, he passed away due to complications from LEMS.

Within months of Susan’s LEMS diagnosis, she was found to have a 0.5 millimeter spot in one lung that has been carefully monitored. This past December, she had her five-year CT scan, and since there is no change in the spot, she is considered to be cancer-free, which, says Susan, “is an incredible boost to my morale!” To keep her mind and her body sharp, Susan took to the art of cross-stitch and sends her finished products “to people sicker than I am,” she says. “I was tired of existing as a human being instead of a human doing.” Some of her work is being used by an online group called Love Quilts, which creates and gives cross-stitched quilts to ill children. Her work also has been sold to support a children’s hospice and an air ambulance in England. In addition to cross-stitch, Susan started an online support group for people with LEMS.

Nine months after being diagnosed with LEMS, Sharon was diagnosed with breast cancer and had a mastectomy “that almost took my life,” she explains. She suffered an internal bleed and had to have several blood transfusions. “Lying in my hospital bed after my mastectomy, I had to make a choice about my illness,” Sharon recalls. “I made the decision: ‘I don’t just plan to survive, I plan to thrive.’” Now, five years later, she is cancer-free. And, whenever she feels temporary emotional twinges related to her LEMS, she will mentally rehearse her father’s favorite saying: “I complained I had no shoes, until I saw a man with no feet.”

Related Organizations

Muscular Dystrophy Association: www.mdusa.org
Myasthenia Gravis Foundation of America: www.myasthenia.org
National Organization of Rare Disorders (NORD): www.rarediseases.org
American Autoimmune Related Diseases Association: www.aarda.org
Autoimmune Information Network: www.aininc.org
LEMS Yahoo Group: groups.yahoo.com/group/Lambert-Eaton/join

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