

Learn the symptoms and become your own health advocate

by Andrea Atkins

diseases doctors miss



Some diseases are difficult to diagnose because they masquerade as something else. Others elude doctors because they haven't been trained to recognize them. And some can have so many symptoms that doctors may be confounded for months—or years. All the more reason to learn about these “medical mysteries.”

multiple sclerosis

Ellen Sue Stern was 26 when she began to have a searing pain across the right side of her face. Sometimes it was accompanied by debilitating fatigue and muscle weakness. Often it was so bad that she'd wind up at the emergency room on a morphine drip for the pain. Astoundingly, this went on for 25 years.

Several doctors were stymied, says Ellen Sue, a mother of two. “They told me, ‘Basically, you’re just going to have to live with this.’”

She began to wonder whether she was crazy. Finally, out of des-

peration, Ellen Sue, who lives in Minneapolis, called a neurologist she'd seen almost 10 years earlier.

The doctor told her the pain she was describing was called trigeminal neuralgia, a condition usually experienced by people with brain tumors. He never mentioned any other possible causes, so a terrified Ellen Sue, who was then 48, explored her symptoms online. Her search led her to multiple sclerosis, a disease of the central nervous system that attacks the fatty tissue that surrounds nerve fibers. Scar tissue develops around the nerves and impairs their ability to conduct

information from the brain to other body parts. Ellen Sue insisted on an MRI, which sealed the diagnosis.

It's been four years since her MS diagnosis, and while Ellen Sue still has pain, she now knows what she needs: steroids to relieve the discomfort; 12 hours of sleep a night, which seems to help stem the fatigue and muscle exhaustion; and other drugs that slow the course of MS's degenerative nature.

WHAT TO WATCH FOR MS is difficult to diagnose because its symptoms can be so varied. The most common signs are blurred vision, loss of balance, (Please turn to 62)

DISEASES DOCTORS MISS

Continued

poor coordination, numbness and extreme fatigue, according to Patricia O'Looney, Ph.D., associate vice president of biomedical research programs at the National MS Society. Since the brain controls every function of the body, patients may also experience bladder and bowel difficulties and cognitive problems.

HOW IT'S DIAGNOSED There is no one diagnostic tool, Dr. O'Looney says. A neurologist will conduct a thorough examination, which may include a test in which electrodes are attached to the body at various places to measure the speed of the nervous system's response to stimuli. An MRI (which can show brain scarring) may help confirm the diagnosis.

TREATMENT There are six FDA-approved medications that can help lessen the frequency and severity of MS attacks, Dr. O'Looney says. They also reduce the number of sclerotic lesions in the brain and slow the progression of the disability. Most are therapies aimed at regulating the immune system. Tysabri is the most recently approved drug.

THE GOOD NEWS "As devastating as a diagnosis of MS is, it is so much better than it was 20 years ago," Dr. O'Looney says. "There are very effective therapies that are slowing the disease. No one should go to a doctor's office, hear she has MS, and think there's nothing to be done."

TO LEARN MORE Visit nmss.org or call 800-FIGHT-MS.

celiac disease

From the time she was 15, Jana Simone of Los Angeles was anemic. But it wasn't until she was 27 and lost her first pregnancy at eight months that she knew something was terribly wrong.

During her second pregnancy, doctors monitored her closely. Her labor was induced at eight months due to fetal distress. She delivered a healthy baby girl, but Jana was anything but healthy.

"Two back-to-back pregnancies, and my system was completely depleted," recalls Jana, now 44.

Not only was she iron-deficient, but Jana was constantly fighting unbearable fatigue. She had frequent sore throats, fevers, sinus infections and flu-like achiness. Her doctors diagnosed her with lupus (see page 66) once they found actual lupus antibodies in her body. But medications never seemed to help, nor did any other treatment. Then her rheumatologist suggested testing her for celiac disease, an autoimmune disease in which the small intestine is damaged by gluten, a substance found in wheat products and many other foods.

She had never heard of the condition, but the test was positive, and Jana, then 40, switched to a gluten-free diet. The achiness, sore throats and fevers quickly disappeared, as did the signs of lupus in her blood. Gone also were her constant fatigue and anemia.

Studies show that most celiac patients go untreated for an average of 11 years and see at least four doctors before diagnosis, says Peter

H.R. Green, M.D., director of the Celiac Disease Center at Columbia University in New York. Left untreated, celiac can develop into anemia, osteoporosis and rheumatoid arthritis. It can also lead to a secondary autoimmune problem.

WHAT TO WATCH FOR The most common symptoms are diarrhea, gas, bloating, pain, cramps and weight loss. But fatigue, rashes, headaches and joint pain can also be signs.

HOW IT'S DIAGNOSED A blood test can reveal indicators for celiac disease, but a small bowel biopsy, done by endoscopy (it takes about 10 to 20 minutes), will confirm the diagnosis.

TREATMENT Completely eliminating gluten from the diet.

THE GOOD NEWS Once patients begin a gluten-free diet, symptoms usually disappear, which is why prompt diagnosis is extremely important—before any long-term damage is done to the body. And more doctors are becoming aware of the disease.

TO LEARN MORE Visit celiac.org or call the Celiac Foundation at 818-990-2354. *(Please turn to 66)*

get the right diagnosis

If you've been struggling with baffling symptoms, follow these steps before your next doctor's visit.

Share your records with the physician Send them ahead of time so the doctor can look over them before meeting you, says Susan Manzi, M.D., director of the Lupus Center of Excellence at the University of Pittsburgh Schools of Health Sciences. "Sometimes physicians like to review records before the actual visit."

Give a complete health history Know your background and your family's history, says Eric Esrailian, M.D., M.P.H., clinical instructor of medicine, Division of Digestive Diseases, David Geffen School of Medicine at UCLA.

Know your medications Know how long you took them, at what dosage and how often, Dr. Manzi says.

Accept that there may be a mental health component Many of the hard-to-diagnose diseases can bring on depression or anxiety, Dr. Manzi says. It doesn't mean you're crazy, but psychological intervention may, in fact, help you.

Follow the doctor's instructions Some treatments focus on reducing the body's reaction to symptoms, says Scott Stuart, M.D., professor of psychiatry at the University of Iowa. "If you experience pain but quit the treatment, don't be surprised if your pain continues," he counsels.