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'Fog of illness' never seemed to go away

Lack of antibodies makes it harder for sufferers of CVID to fight off infections

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For Judy Kozulak, 1998 was a rough year. Her marriage was failing and her mother had been diagnosed with Alzheimer's. So when she succumbed to bouts of pneumonia and bronchitis that winter, she chalked it up to stress and the usual winter maladies.

Two years later, though, she developed chronic thrush, a fungal infection that accumulates in the lining of the mouth and occurs most frequently in babies, older adults and people with weakened immune systems. Still, she recovered after a course of antibiotics and resumed her normal routine: Exercising each morning before work; ferrying her kids to and from school and activities; and organizing parties for friends on the weekends.

But by 2004, she was getting sinusitis three or four times a year. That fall, her primary care doctor referred her to an allergist. In addition to allergy tests, the doctor ordered blood work and a vaccine challenge to evaluate her immune system response. Tests revealed that one of her antibodies was in the low-normal range, but her reaction to the vaccine was good. To get her asthma under control, the doctor prescribed allergy shots.

Kozulak's health worsened during winter 2005. Despite getting a flu shot, she came down with the virus around Valentine's Day and was sick for two months with sinusitis and asthma. She began to feel better in April, but by the end of the month suffered yet another sinus infection. She was referred to an otolaryngologist who performed the first of what would ultimately be five surgical procedures on her sinuses.

A month later, she was prescribed steroids for another sinus flare-up. She also developed a cough that kept her up most of the night. "I was working in a fog of illness and sleep deprivation," she said.

In January 2006, Kozulak underwent a second sinus surgery and cultures revealed the presence of methicillin-resistant Staphylococcus aureus. She was referred to an infectious disease doctor. Another sinus culture taken that spring tested positive for yet another bacteria.

A couple of months later, she was placed on intravenous medications, requiring her to take a short-term medical leave from her job.

By the end of September 2006, she was doing somewhat better. She was taken off the antibiotics and antifungals and discharged by her allergist. But her instincts told her something was wrong. "My energy wasn't returning. I slept all the time," she said. "I would wake up in the morning and be too tired to move."

Believing her sinuses were the root of her problems, Kozulak sought a second opinion from an otolaryngologist at Johns Hopkins Hospital in Baltimore. The doctor referred her to an allergist who repeated the tests done two years earlier. This time her blood work revealed that her IgG level had dropped from low-normal to low. She was referred to an immunologist.

In spring 2007, Kozulak was sick for a month with another debilitating sinus infection. Her asthma had become so severe that she had to use a nebulizer. Vaccine challenges showed that her immune system was weak.

Finally, in August, Kozulak was diagnosed with common variable immune deficiency, or CVID.

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Men, women affected equally

CVID is a group of 20 to 30 primary immune deficiency disorders characterized by low levels of specific immunoglobulins or antibodies produced by the immune system to fight infections or disease. Its name derives from the fact that it is a relatively common form of immune disorder, but the degree and type of deficiency vary.

In some patients there is less IgG and IgA antibodies in the blood. In others all three types of antibodies – IgG, IgA and IgM – are deficient. In addition, about half of patients exhibit some degree of T-cell dysfunction. The causes of CVID are largely unknown, although research has identified the involvement of three genes in a small group of patients.

CVID affects males and females equally. The disorder typically affects people in their 30s and 40s, but about 20 percent of patients have symptoms of the disease or are found to be immunodeficient by age 16.

“This disease is in many ways like diabetes,” said Dr. Howard M. Lederman, professor of pediatrics and director of the Immunodeficiency Clinic at Johns Hopkins Children’s Center. “There are people who develop diabetes in infancy, there are people who develop diabetes in adolescence and there are people who don’t get it until they’re 60.

“People probably have some genetic susceptibility to develop CVID, but if you don’t run into the right infection or you don’t have some other change in your body, you don’t develop immunodeficiency,” he said.

In CVID, frequent bacterial infections of the ears, sinuses, bronchi and lungs are common. Patients may experience an enlarged spleen and lymph nodes. Gastrointestinal problems also may occur, due to the malabsorption of fat or certain sugars or a parasitic infection. Some patients develop autoantibodies that attack and destroy blood cells. Finally, patients may have an increased risk of cancer, especially those of the lymphatic system, skin and gastrointestinal tract.

Diagnosing CVID is fairly straightforward and involves measuring levels of antibodies in the bloodstream. Vaccine challenges are conducted to evaluate the patient’s ability to make protective amounts of antibodies. Certain cell functions may also be evaluated.

Still, a significant number of patients go undiagnosed for years. “The delay in diagnosis is not because it’s difficult to diagnose but because people don’t think about it,” Lederman said. “You have to have somebody who has a perspective to say, ‘This patient is getting one infection after the other, this is not what I usually see, I wonder if there’s a reason for that and run some tests.”

Common but not well-known

The standard treatment is immunoglobulin-replacement therapy, which can be administered at an infusion center or at home. Patients who suffer from chronic sinusitis or lung disease may need long-term treatment with broad-spectrum antibiotics. Physical therapy and draining of respiratory secretions also may be necessary.

Kozulak, who started treatments in September, has opted to self-administer her medication. “I had just started a new job, and I thought I’m going to need time off to do it around an infusion clinic’s schedule,” said the 51-year-old Hockessin resident who is now a nurse auditor at Thomas Jefferson University Hospital in Philadelphia.

There are about 250,000 diagnosed cases of primary immune deficiency disorders in the U.S., making it more common than a lot of well-known genetic diseases, including cystic fibrosis and Huntington’s. But primary immune deficiency disorders have a much lower profile.

“It’s something people haven’t heard about,” said Christine Belser, senior director of programs and education at the Immune Deficiency Foundation. “Everybody knows somebody who’s had a heart attack or somebody who has had cancer so they’re familiar with it.”

The foundation is working to raise awareness through patient and physician education, peer support and public policy initiatives, she said. It’s currently supporting legislation to increase Medicare reimbursements for intravenous immunoglobulin replacement therapy.
With good medical care, people with CVID usually have a normal life span. "They do as well or better as people in the same age group with normal immune systems," Lederman said.

Kozulak’s health has improved since beginning immunoglobulin therapy in September. "I am better, but I’m not who I was. I rest a lot," she said. "I still get infections, but they’re not as intense as they were pre-treatment."

She still gets regular CT scans to check for lung damage and must be proactive about any infection she might develop. Her medical “posse,” as she calls it, includes her primary care physician, an immunologist, an infectious disease specialist, an otolaryngologist and a pulmonologist.

Kozulak copes with the disorder through a combination of humor and acceptance. And she refuses to let it consume her. "I’m trying to live life as fully as I can because I don’t want to be CVID," she said. "I’m not CVID. It's just one part of my life."

FOR MORE INFORMATION

• Immune Deficiency Foundation – www.primaryimmune.org or (800) 296-4433

• Primary Immune Deficiency Disorder Support Group in Delaware – Judy Kozulak at DE-PIDDs@comcast.net

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