

Celiac Disease: Finally on the Map

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by Christine McLaughlin

A patient enters the primary care center complaining of symptoms including those indicative of anemia, chronic fatigue, learning disabilities, depression and osteoporosis. Maybe she does have each of these disorders, but something you've learned recently tells you otherwise. You suspect she might have celiac disease (CD) and suggest getting her tested. Your very astute hunch is likely a result of all the work done at the University of Maryland Center for Celiac Research in Baltimore in the past 14 years.

A decade ago, diarrhea and weight loss were the top symptoms related to CD and are still most commonly associated with it in the medical community. "People are surprised to learn you can have symptoms that have nothing to do with the GI tract," said Alessio Fasano, MD, professor of pediatrics, medicine and physiology and director of the Center for Celiac Research. "As a matter of fact, out-of-the-intestine celiac disease is more common than within the intestine, so to speak."

The reason, explains Fasano, is because tests have been developed in recent years that provide a better understanding of the disease earlier before the intestinal symptoms can be present.

In fact, CD is not nearly as rare as once believed. Previous projections had about one in 10,000 people with CD. However, the Center for Celiac Research found the numbers to be much more dramatic: more like one in 133.

Celiac Disease Basics

To understand the advances made with the disease, it's important to understand its fundamentals. CD is unlike a food allergy because it is a genetically based immune response to gluten, a protein found in wheat and other grains. An allergy to wheat is a reaction people can outgrow. But in people who have CD, gluten sets off an autoimmune reaction that causes the destruction of the villi in the small intestine - the body's own antibodies attack the intestine causing damage and illness. An allergy does not attack the body.

Despite the damage that can occur from CD, it's not uncommon to develop the disease without any symptoms at all. Fasano explains the target organ of the autoimmune insult is the small intestine. To develop gastrointestinal symptoms like diarrhea, a certain amount of the intestine must be destroyed. If there's only a small percentage destroyed, the rest of the intestine can subsidize the job and there may be no GI symptoms. However, depending on the location of the destruction, other symptoms like weakness from malabsorption of iron or weakened bones from malabsorption of calcium and vitamin D may be present. GI symptoms could still appear down the line.

A blood test is available to screen for the antibodies to CD, but an endoscopy/biopsy of the intestine must be performed to confirm the diagnosis. There's also a genetic test that can look at the genes necessary to develop CD - with two specific HLA genes being necessary (DQ2 and DQ8). Actually, about 30 percent of the population carry the gene associated with CD. A major finding about susceptibility to CD, according to Fasano, is that it must include three things or "a holy trinity," as he put it. A person must be genetically predisposed to genes that put that person at risk for the disease, have an environmental trigger (gluten) that is mismanaged by the immune system because of the genetic makeup, and have a "leaky gut" so the trigger can enter the body.

Research Happenings

Since opening in 1996, the Center for Celiac Research has made countless other discoveries. For example, it was traditionally believed CD only developed in newborns with voluminous stool and failure to thrive. Through studies involving all age groups including infants, children and adults, researchers have learned the disease can occur at any point in one's life.

Researchers and patients at the center are currently involved in several studies. For example, in an effort to improve the ability of individuals with CD or food allergies to find foods that are healthy for them, center researchers are working on assisting the FDA with a study examining how patients interpret food labels.

Another study involves infants at risk of developing CD because they have a parent or a sibling with the disease. In this study, the researchers are trying to find out whether changing the timing of introducing gluten in solid food influences if CD can be delayed or even prevented.

"We know at least some of the markers for genetic susceptibility that reside in the genes. We know gluten somehow crosses the intestinal barrier and causes the body to make antibodies to itself. And we know the gluten protein is a big molecule and should not get through the barrier," said Elaine Puppa, MEd, MSN, RN, nurse research coordinator at the center. "What we are trying to learn more about is how it gets there and why the body responds by attacking itself." According to Fasano, the researchers are fortunate to have a wealth of research in the center's portfolio from the very basic, such as how an immune system with CD acts differently than a normal immune system, all the way to hands-on clinical practice studies that examine the distribution of symptoms, why some people develop two symptoms and others develop many more, and everything in between.

"This is the autoimmune disease where we know all the ingredients of the recipe. We know the genes involved and it is the only autoimmune disease for which we know the trigger is gluten. And that it's treatable. . . At the same time, it's the only one we can turn on and off [expose or detract gluten] at will and that will give us a tremendous tool to study the basics of autoimmunity," he explained. In other words, because autoimmune diseases work basically the same way, this kind of information can be extrapolated for other conditions like diabetes and MS.

Nurses Are Key

All of the research findings would not be possible without the center's expert nurses. They're invaluable, according to Fasano, who said that, because of the strong nurse/patient relationship, patients are more comfortable asking questions. At the same time, the expert nurses know exactly what to ask the patients to glean the most accurate information and are then able to be their best advocates.

Maggie Martin, BSN, RN, clinical nurse, is one of those nurses who has a variety of responsibilities. She works directly with the patients in the outpatient clinic and is charged with taking medical history and assessing their specific needs, facilitating their care with insurance and prescriptions, coordinating test results, conducting patient education and assisting their care with any specialists. She also helps recruit patients for research studies.

"Once they're diagnosed, it's especially rewarding to see symptoms resolve once patients are on the gluten-free diet," Martin said.

Puppa works on the research side of the center as a patient advocate. She helps design research to ensure it's patient-friendly and translates the research language so patients can understand it. She's also responsible for recruiting patients for studies and ensuring they understand what they're agreeing to, and then arranging the research for them in the minimum amount of visits. Once they're in the study, she monitors for any complications and side effects, and analyzes the data looking for trends. She also manages regulatory research requirements to acquire Institutional Review Board scientists and physicians, and ensures the research is safe for the patient, has potential benefits and is scientifically sound.

"I love this job because it's never boring. You get to work with patients and treat them as a whole being. And you get [to] work [with patients] through the entire lifespan," Puppa said.

Humble Beginnings

Fasano moved to the U.S. from Salerno, Italy, in 1993 to study diarrheal diseases and micro-organisms. He realized there was very little research here on CD and decided to develop the center, opening it in 1996. But, getting it established was an uphill battle. Most of his colleagues

didn't understand CD and were dismissive about his mission to open a facility to help improve the quality of life for people with CD.

"There were no labs doing the testing for CD, so we had to set up our own labs. There were no tests to screen for CD so we had to develop new tests. There was no awareness whatsoever, so we had to start a campaign of awareness of doing hundreds and hundreds of grand rounds a year to educate colleagues on CD," Fasano said.

Now, you can't go into a grocery store without seeing shelves dedicated to gluten-free foods, and that's largely a result of the awareness spread by the center since its inception.

"If you told me in 1996 we would be where we are today with celiac disease, I'd say you must be nuts," Fasano laughed. "It's not possible."

But thankfully, for millions of people, it has been.

Christine McLaughlin is a frequent contributor to ADVANCE.

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