Celiac (SEE-lee-ak) disease is a digestive condition triggered by consumption of the protein gluten, which is primarily found in bread, pasta, cookies, pizza crust and many other foods containing wheat, barley or rye. People with celiac disease who eat foods containing gluten experience an immune reaction in their small intestines, causing damage to the inner surface of the small intestine and an inability to absorb certain nutrients.

Celiac disease can cause abdominal pain and diarrhea. Eventually, the decreased absorption of nutrients (malabsorption) that occurs with celiac disease can cause vitamin deficiencies that deprive your brain, peripheral nervous system, bones, liver and other organs of vital nourishment.

How is celiac disease diagnosed?

Celiac disease is suspected when individuals have signs or symptoms of malabsorption or malnutrition. Other diseases, however, can produce malabsorption and malnutrition, for example, pancreatic insufficiency (a pancreas that is not able to produce digestive enzymes), Crohn's disease of the small intestine, and small intestinal overgrowth of bacteria. It is important, therefore, to confirm suspected celiac disease with appropriate testing.

Small intestinal biopsy

Small intestinal biopsy is considered the most accurate test for celiac disease. Small intestinal biopsies can be obtained by performing an esophagogastroduodenoscopy (EGD). During an EGD, the doctor inserts a long, flexible viewing endoscope through the mouth and into the duodenum. A long, flexible biopsy instrument then can be passed through a small channel in the endoscope to obtain samples of the intestinal lining of the duodenum. Multiple samples usually are obtained to increase the accuracy of diagnosis. A pathologist then can examine the biopsies (under a microscope) for loss of villi and other characteristics of celiac disease such as increased numbers of lymphocytes.

Small intestinal biopsy does however, have some limitations. For example, acute viral gastroenteritis and allergy to cow's milk or soy protein can cause abnormal small intestinal biopsies that are indistinguishable from celiac disease. However, acute viral gastroenteritis is not easily confused with celiac disease because of the difference in the acuteness of symptoms. (Acute viral gastroenteritis has a sudden onset of symptoms and last only a few days.) It is however, easier to confuse cow's milk and soy protein allergies with celiac disease, but these allergic conditions are rare and primarily occur in young children. Despite these limitations, small intestinal biopsies are recommended even for individuals who have abnormal antibody tests for celiac disease. (See discussion that follows.)

Specific antibody tests for celiac disease

Antibodies are proteins that are produced by the immune system to fight viruses, bacteria, and other organisms that infect the body. Sometimes, however, the body produces antibodies against non-infectious substances in the environment (for example, in hay fever) and even against its own tissues (autoimmunity).

Blood tests that are specific for celiac disease include endomysial antibodies, anti-tissue transglutaminase antibodies, and anti-gliadin antibodies. In patients with celiac disease, anti-gliadin antibody is an antibody produced against gliadin in the diet and endomysial and anti-tissue transglutaminase antibodies are antibodies produced against the body's own tissues.

Endomysial antibodies and anti-tissue transglutaminase antibodies are highly reliable in diagnosing celiac disease. An individual with abnormally elevated endomysial and anti-tissue transglutaminase antibodies has a greater than 95% chance of having celiac disease. Anti-gliadin antibodies are less reliable and have a high false positive rate. Thus a person with an abnormally elevated anti-gliadin antibody level does not necessarily have celiac disease. Nevertheless, anti-gliadin antibody levels are useful in monitoring the response to treatment because anti-gliadin antibody levels usually begin to fall within several months of successful treatment of celiac disease with a gluten free diet.

Who should undergo antibody blood tests for celiac disease?

Some experts recommend that antibody blood tests be used to screen healthy persons with no signs or symptoms for celiac disease. In Italy, where celiac disease is common, all children are screened for celiac disease. Experts in the United States do not recommend screening healthy persons for celiac disease. Antibody blood tests are only recommended for individuals with a higher likelihood than normal of having celiac disease. These patients are:
1. Patients with chronic diarrhea (diarrhea that does not resolve in three weeks), increased amount of fat in the stool (steatorrhea), and weight loss
2. Patients with excess gas, bloating, and abdominal distension
3. First and second degree relatives of patients who have celiac disease
4. Children with growth retardation
5. Patients with unexplained iron deficiency anemia
6. Patients with skin rashes suggestive of dermatitis herpetiformis
7. Patients with recurrent painful mouth sores (aphthous stomatitis)

Patients with disease known to be associated with celiac disease. Examples of these diseases include insulin-dependent diabetes mellitus, autoimmune thyroid disease, rheumatoid arthritis, systemic lupus, ulcerative colitis, etc.

Why is it important to accurately diagnose celiac disease?
Diagnosis of celiac disease should be firmly established before commencing treatment with a gluten free diet for several reasons.

How are malabsorption and malnutrition evaluated in celiac?
1. The gluten free diet is a life-long and tedious commitment that should not be taken lightly. It is more costly than a normal diet and has significant social implications for dining out.
2. Patients with irritable bowel syndrome (IBS) may experience improvements in bloating, abdominal pain, and diarrhea with a gluten free diet. These patients may be misdiagnosed as having celiac disease. Without confirmation of celiac disease by small intestinal biopsy, they may be unnecessarily committed to life-long gluten restriction.
3. A gluten free diet can lower blood antibody levels and allow the microscopic appearance of the small intestine to lose the typical appearance of celiac disease, complicating subsequent efforts at making a firm diagnosis of celiac disease.

Celiac disease causes malabsorption of nutrients and leads to malnutrition. Tests are available that help in the evaluation of malabsorption and malnutrition; however, because other diseases can cause both malabsorption and malnutrition, these tests cannot be used to diagnose celiac disease.

Stool examination for malabsorption
Fat in a sample of stool placed on a glass slide can be stained with a dye (Sudan stain) to make the fat visible under the microscope as globules. Stool from patients with celiac disease often contains many stained globules of fat, and Sudan staining is a quick and easy screening test for increased amounts of fat in the stool (steatorrhea). To conclusively diagnose steatorrhea, however, stool is collected over a 72-hour period, and the fat in the stool is chemically measured and quantified. Steatorrheic stools have abnormally high quantities of fat. Since malabsorption and steatorrhea can occur with other intestinal diseases (such as small intestinal bacteria overgrowth, prior small intestinal resection, extensive Crohn's disease of the small intestine, and chronic pancreatitis), stools with large amounts of fat only raise the suspicion of celiac disease but cannot be used to diagnose celiac disease.

Blood tests for malnutrition and vitamin deficiencies
Malabsorption reduces the absorption of protein and causes a reduction in blood protein levels. This can be seen commonly as a reduced blood level of albumen, the most concentrated protein in blood. Other proteins in blood, for example, pre-albumen and transferrin also may be reduced.

Intestinal malabsorption can lead to deficiencies and low blood levels of iron, calcium, vitamin B12, folate, Vitamin D and vitamin K. These deficiencies, in turn, can lead to other blood test abnormalities such as:

Iron deficiency anemia: Iron is an important component of hemoglobin in red blood cells. When iron is deficient, production of red blood cells is impaired, and anemia develops. Iron deficiency anemia can occur either through loss of blood (with its iron-containing red blood cells) or lack of intestinal iron absorption. Heavy menstrual bleeding and cancer of the colon that bleeds into the intestine are two common causes of iron deficiency anemia due to blood loss. Celiac disease causes iron deficiency anemia by reducing intestinal iron absorption. In fact, iron deficiency anemia can be an important clue to the presence of celiac disease.

Abnormally prolonged prothrombin time (ProTime): ProTime is a blood test that measures how quickly blood clots. Clotting of blood requires special proteins or clotting factors, many of which are made by the liver. Formation of clotting factors by the liver requires vitamin K. When vitamin K absorption from the intestine is reduced, as in celiac disease, the production of clotting factors by the liver is inadequate, and blood clotting is delayed. Delayed clotting is reflected in an abnormal ProTime, and individuals with an abnormal ProTime have a higher risk of abnormal or excessive bleeding.

Iron deficiency anemia, abnormal ProTime, steatorrhea, and low iron and vitamin levels can occur in diseases other than celiac disease. Therefore the presence of these abnormalities only raises the suspicion of celiac disease but does not specifically diagnose celiac disease.