How is Pernicious Anemia Diagnosed?

Your doctor will diagnose pernicious anemia based on your medical and family histories, a physical exam, and test results.

Your doctor will want to find out whether the condition is due to a lack of intrinsic factor or another cause. He or she also will want to find out the severity of the condition, so it can be properly treated.

Specialists Involved

Primary care doctors—such as family doctors, internists, and pediatricians (doctors who treat children)—often diagnose and treat pernicious anemia. Other kinds of doctors also may be involved, including:

- A neurologist (nervous system specialist)
- A cardiologist (heart specialist)
- A hematologist (blood disease specialist)
- A gastroenterologist (digestive tract specialist)

Medical and Family Histories

Your doctor may ask about your signs and symptoms. He or she also may ask:

- Whether you've had any stomach or intestinal surgeries
- Whether you have any digestive disorders, such as celiac disease or Crohn's disease
- About your diet and any medicines you take
- Whether you have a family history of anemia or pernicious anemia
- Whether you have a family history of autoimmune disorders (such as Addison's disease, type 1 diabetes, Graves' disease, or vitiligo). Research suggests a link may exist between these autoimmune disorders and pernicious anemia that's caused by an autoimmune response.

Physical Exam

During the physical exam, your doctor may check for pale or yellowish skin and an enlarged liver. He or she may listen to your heart for rapid or irregular heartbeats or a heart murmur.

Your doctor also may check for signs of nerve damage. He or she may want to see how well your muscles, eyes, senses, and reflexes work. Your doctor may ask questions or do tests to check your mental status, coordination, and ability to walk.

Diagnostic Tests and Procedures

Blood tests and procedures can help diagnose pernicious anemia and find out what's causing it.

Complete Blood Count

Often, the first test used to diagnose many types of anemia is a complete blood count (CBC). This test measures many parts of your blood. For this test, a small amount of blood is drawn from a vein (usually in your arm) using a needle.

A CBC checks your hemoglobin (HEE-muh-glow-bin) and hematocrit (hee-MAT-oh-crit) levels. Hemoglobin is an iron-rich protein that helps red blood cells carry oxygen from the lungs to the rest

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of the body. Hematocrit is a measure of how much space red blood cells take up in your blood. A low level of hemoglobin or hematocrit is a sign of anemia.

The normal range of these levels may be lower in certain racial and ethnic populations. Your doctor can explain your test results to you.

The CBC also checks the number of red blood cells, white blood cells, and platelets (PLATE-lets) in your blood. Abnormal results may be a sign of anemia, another blood disorder, an infection, or another condition.

Finally, the CBC looks at mean corpuscular (kor-PUS-kyu-lar) volume (MCV). MCV is a measure of the average size of your red blood cells. MCV can be a clue as to what's causing your anemia. In pernicious anemia, the red blood cells tend to be larger than normal.

Other Blood Tests

If the CBC results confirm that you have anemia, you may need other blood tests to find out what type of anemia you have.

A reticulocyte (re-TIK-u-lo-site) count measures the number of young red blood cells in your blood. The test shows whether your bone marrow is making red blood cells at the correct rate. People who have pernicious anemia have low reticulocyte counts.

Serum folate, iron, and iron-binding capacity tests also can help show whether you have pernicious anemia or another type of anemia.

Another common test, called the Combined Binding Luminescence Test, sometimes gives false results. Scientists are working to develop a more reliable test.

Your doctor may recommend other blood tests to check:

- Your vitamin B12 level. A low level of vitamin B12 in the blood indicates pernicious anemia. However, a falsely normal or high value of vitamin B12 in the blood may occur if antibodies interfere with the test.
- Your homocysteine and methylmalonic acid (MMA) levels. High levels of these substances in your body are a sign of pernicious anemia.
- For intrinsic factor antibodies and parietal cell antibodies. These antibodies also are a sign of pernicious anemia.

Bone Marrow Tests

Bone marrow tests can show whether your bone marrow is healthy and making enough red blood cells. The two bone marrow tests are aspiration (as-pi-RA-shun) and biopsy.

For aspiration, your doctor removes a small amount of fluid bone marrow through a needle. For a biopsy, your doctor removes a small amount of bone marrow tissue through a larger needle. The samples are then examined under a microscope.

In pernicious anemia, the bone marrow cells that turn into blood cells are larger than normal.

Source: National Heart, Lung, and Blood Institute, National Institutes of Health.

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