Chronic Myeloid Leukemia (CML): Diagnosis



How is chronic myeloid leukemia (CML) diagnosed?

If your healthcare provider thinks you might have CML, you'll need certain exams and tests to be sure. You'll be asked about your health history, your symptoms, risk factors, and family history of disease. A physical exam will be done.

What tests might I need?

You may have one or more of these tests:

- **Blood tests.** CML is often found with blood tests before you have any symptoms. Blood is taken from your arm or hand with a small needle. These tests can look at the numbers of different types of blood cells. People with CML often have too many white blood cells.
- Bone marrow aspiration and biopsy. This involves taking out small amounts of bone marrow fluid
 (aspiration), solid bone marrow tissue (core biopsy), or both. Bone marrow samples are often taken
 from the back of the hip (pelvic) bone.
 - Bone marrow aspiration. For bone marrow aspiration, the skin over the hip is numbed. A thin, hollow needle is then put into the pelvic bone. A syringe is used to pull out a small amount of liquid bone marrow. You may have some brief pain when the marrow is removed.
 - Bone marrow biopsy. This is often done just after the aspiration. A slightly bigger needle is used to take out a small core of bone and marrow. The biopsy may also cause some brief pain. The fluid and bone marrow are sent to a lab and checked for the number, size, and maturity of blood cells and abnormal cells. Other tests will also be done on these cells.

How blood or bone marrow is tested

Nearly all people with CML have a genetic change called the Philadelphia chromosome, an abnormal gene called BCR-ABL, or both. People with CML also have too many white blood cells. Tests are used to look for these changes. These are some of the tests often done on blood or bone marrow samples to confirm a CML diagnosis and learn more about the CML cells:

- Cytogenetics. For these tests, cells are grown in a lab for a week or more. The chromosomes inside the cells are then stained with special dyes and looked at with a microscope. Major problems in the chromosomes can often be seen. But smaller changes may not be visible. These tests often take a few weeks because the cells need time to be grown in the lab.
- Fluorescence in situ hybridization (FISH). This test is another way to look for changes in chromosomes. The cells in the sample are stained with fluorescent dyes that will attach only to certain parts of chromosomes. The cells are then viewed with a special light. This test can find some chromosome changes that can't be seen with standard cytogenetic testing. It's also a quicker test.
- Polymerase chain reaction (PCR). This is a very sensitive test that can detect very low levels of BCR-ABL in a test sample. It works by increasing the amount of genetic material in a sample so that it can be found. This test can find very small levels of the BCR-ABL gene that other tests may not find.

Getting your test results

Your healthcare provider will contact you with the results. Your provider will talk with you about other tests you may need if CML is found. Make sure you understand the results and what follow-up you need.

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