Mastocytosis - Diagnosis

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Diagnosis

ON THIS PAGE: You will find a list of the common tests and procedures that doctors can use to find out what?s wrong and identify the cause of the problem. To see other pages in this guide, use the colored boxes on the right side of your screen, or click ?Next? at the bottom.

Doctors use many tests to diagnose a disease and find out its extent. Some tests may also determine which treatments may be the most effective. A biopsy is the only way to make a definitive diagnosis of mastocytosis. If a biopsy is not possible, the doctor may suggest other tests that will help make a diagnosis. This list describes options for diagnosing this condition, and not all tests listed will be used for every person. Your doctor may consider these factors when choosing a diagnostic test:

- Age and medical condition
- Type of disease suspected
- Signs and symptoms
- Previous test results

In addition to a physical examination, the following tests may be used to diagnose mastocytosis:

Blood/urine tests [2]. Tests are done on samples of the patient?s blood and urine that may be able to find high levels of mast cells and histamine. Blood tests can also show how well the liver, kidneys, and other vital organs are working. The blood?s level of tryptase (an enzyme) may be higher for people with systemic mastocytosis, which may help distinguish systemic mastocytosis from similar disorders.

Biopsy [3]. A biopsy is the removal of a small amount of tissue for examination under a microscope. Other tests can suggest that mastocytosis is present, but only a biopsy can make a definite diagnosis. The sample removed during the biopsy is analyzed by a pathologist (a doctor who specializes in interpreting laboratory tests and evaluating cells, tissues, and organs to diagnose disease). Skin biopsies are often used to diagnose mastocytosis.

Bone marrow aspiration and biopsy [4]. A sample of the bone marrow helps diagnose systemic mastocytosis. Bone marrow aspiration and biopsy are similar procedures and are often done at the same time. Bone marrow has both a solid and a liquid part. A bone marrow biopsy is the removal of a small amount of solid tissue using a needle. An aspiration removes a sample of fluid with a needle. The sample(s) are then analyzed by a pathologist. A common site for a bone marrow biopsy and aspiration is the pelvic bone, which is located in the lower back by the hip. The skin in that area is numbed with medication beforehand, and other types of anesthesia (medication to block the awareness of pain) may be used.

Molecular testing. Your doctor may recommend running laboratory tests on the mast cells to identify specific genes, proteins, and other factors unique to the mastocytosis. Results of these tests will help decide whether your treatment options include a type of treatment called targeted therapy (see the Treatment [5] section).

After these diagnostic tests are done, your doctor will review all of the results with you. If the diagnosis is mastocytosis, these results will help the doctor identify the type and help plan treatment.

Choose ?Next? (below, right) to continue reading this guide to learn about the different stages of this condition. Or, use the colored boxes located on the right side of your screen to visit any section.

Links:
[1] http://www.cancer.net/about-us
[2] http://www.cancer.net/node/24716
[3] http://www.cancer.net/all-about-cancer/cancernet-feature-articles/cancer-screening-and-prevention/biopsy-what-expect
[4] http://www.cancer.net/node/24409
[5] http://www.cancer.net/node/19230