

Myelodysplastic Syndromes - MDS - Diagnosis [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board](#) [2], 10/2014

ON THIS PAGE: You will find a list of the common tests, procedures, and scans that doctors can use to find out what's wrong and identify the cause of the problem. To see other pages, use the menu on the side of your screen.

Doctors use many tests to diagnose MDS. Some tests may also determine which treatments may be the most effective. This list describes options for diagnosing MDS, 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 MDS:

Blood tests [3]. A complete blood count, or CBC, test measures the numbers of red blood cells, white blood cells, and platelets. Blood tests may also be done to rule out other conditions that can cause symptoms similar to MDS, such as low levels of vitamin B12, folate, copper, and thyroid problems.

Peripheral (circulating) blood smears. A drop of blood is placed on a slide, smeared into a thin film, and placed under a microscope for examination. The percentages of the different types of cells are counted, and the appearance of cells under the microscope, called cell morphology, is looked at to find out if or how the cells are different from healthy cells.

Bone marrow aspiration and biopsy [4]. These two procedures are similar and often done at the same time to examine the bone marrow. Bone marrow has both a solid and liquid part. A bone marrow aspiration removes a sample of the fluid with a needle. A bone marrow biopsy is the removal of a small amount of solid tissue using a needle. The samples are then analyzed by a pathologist to determine the percentage of red blood cells, white blood cells, platelets, and blasts. A pathologist is a doctor who specializes in interpreting laboratory tests and evaluating cells, tissues, and organs to diagnose disease. A common site for the bone marrow aspiration and biopsy 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, which is medication to block the awareness of pain, may be used. The appearance of the bone marrow tissue, along with blood cell counts and chromosomal analysis (see below), is needed to confirm a diagnosis of MDS.

Molecular testing. Your doctor may recommend running laboratory tests on a bone marrow sample to identify specific genes, proteins, and other factors unique to MDS. Results of these tests will help decide your doctor plan treatment, find out how treatment is working, and the likelihood of recovery after a stem cell transplant (see [Treatment Options](#) [5]).

Cytogenetic (chromosomal) analysis. Looking at the chromosomes of the cells in the blood and bone marrow shows specific abnormalities that help doctors tell the difference between MDS and other blood disorders. About 50% of people with MDS have one or more chromosomal abnormalities, regardless of the [subtype](#) [6]. Primary MDS often has one chromosomal abnormality. Secondary MDS often has many or complex chromosomal changes. The most common abnormalities affect chromosomes 5, 7, 8, 11, 12, and 20.

Immunophenotyping. Immunophenotyping is the examination of antigens, a specific type of protein, on the surface of the MDS cells. Immunophenotyping can help identify the type of MDS.

After diagnostic tests are done, your doctor will review all of the results with you. If the diagnosis is MDS, these results also help the doctor describe the disease.

The next section helps explain the different subtypes and classification for MDS. Use the menu on the side of your screen to select MDS Subtypes and Classification or you can select another section, to continue reading this guide.

Links:

[1] <http://www.cancer.net/cancer-types/myelodysplastic-syndromes-mds/diagnosis>

[2] <http://www.cancer.net/about-us>

[3] <http://www.cancer.net/node/24716>

[4] <http://www.cancer.net/node/24409>

[5] <http://www.cancer.net/node/19387>

[6] <http://www.cancer.net/node/19386>