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Memorial Sloan Kettering
Cancer Center

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— aplastic anemia

certain leukemias, including acute leukemia and large granular lymphocytic leukemia

HIV infection

an overactive immune system

Proper diagnosis is crucial so that you receive the most-effective treatment for MDS. Often, you will be referred to a hematologist (a doctor who specializes in blood disorders). A definitive diagnosis can be provided only after a specialist called a hematopathologist examines a sample of your bone marrow.

Blood Tests to Diagnose MDS

A very basic blood test called a CBC can determine whether the numbers of various types of blood cells are within normal ranges. CBC stands for complete blood count. It is a routine test that is usually done as part of your regular medical checkup or before you have surgery.

Abnormalities in this test provide the first sign of the disease. In MDS, red blood cell levels may be low, which causes anemia. Platelet levels may also be low, which can cause bleeding and bruising. Low white blood cell levels may lead to infections.

A hematologist will also look at the blood sample under a microscope in a test called a blood smear. This can identify any abnormal cell shapes and sizes, which can also indicate MDS.

In addition, tests will be done to look for other causes of low blood counts, including thyroid disease, low vitamin levels, and iron deficiency.

Bone Marrow Tests to Diagnose MDS

Myelodysplastic Syndrome (MDS) Doctors & Other Experts

Select from the list below to learn more about MSK MDS experts, their education, training, board certifications, current publications, and specific areas of expertise.

[Learn more](#) →

If blood tests do not show another reason for the abnormal blood counts, samples of your bone marrow will be examined to look for MDS. Two types of samples are taken. One is called a bone marrow biopsy. It is obtained by removing a small piece of the bone along with the marrow inside the bone. The second sample is called a bone marrow aspirate. It is obtained by drawing out liquid from the bone marrow space.

Many tests are performed on the bone marrow biopsy and aspirate samples. These tests can help confirm a diagnosis of MDS. For example, a test might show that the cells look abnormal. The tests can also determine the subtype of MDS and help doctors determine the most-effective treatment and prognosis.

The following tests are performed on bone marrow samples:

Cytogenetic studies help determine chromosome changes in bone marrow cells. Each cell in the body contains chromosomes. These are 46 tightly coiled strands of DNA. Chromosomes contain all the information that cells need to function normally. About half of people with MDS have one or more chromosomal changes. The most common abnormalities are seen in chromosomes 5, 7, 8, and 20.

Histochemistry studies allow doctors to look at bone marrow cells to determine whether the cells appear abnormal. With these tests, the bone marrow blast count can also be determined. A pathologist can look for other causes of bone marrow malfunction.

Flow cytometry passes cells through a laser beam to see whether the bone marrow cells are developing normally and to estimate the blast count.

Molecular genetic studies are highly sensitive DNA and RNA tests. These tests determine the specific genetic traits of bone marrow cells.

Research Studies

New diagnostic tests and procedures are emerging as a result of research performed at Memorial Sloan Kettering and at other institutions. Your doctor may ask whether you are willing to have additional blood or bone marrow samples taken for this type of research.

These tests are not necessary to make a diagnosis of MDS, and they are not required tests. However, research on samples taken from people with MDS is vital to our ongoing efforts to learn more about MDS and to develop better treatments. Talk with your Memorial Sloan Kettering doctor to learn more about research studies.

Request an Appointment

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Available Monday through Friday, 8 a.m. to 6 p.m. (Eastern time)

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