

How is Myelodysplastic Syndromes (MDS) Diagnosed?

Proper diagnosis of Myelodysplastic Syndromes (MDS) is crucial and the disease is often not diagnosed until the patient is seen by a hematologist.

Bone marrow tests can diagnose MDS. Samples of bone marrow are important to make the diagnosis of MDS. Two types of samples are taken. One is called a bone marrow biopsy, which is obtained by removing a small piece of the bone along with the marrow inside the bone. The second is called a bone marrow aspirate, which is obtained by drawing out a sample of liquid from the bone marrow space.

Many tests are performed on the bone marrow biopsy and aspirate. A hematologist will also look at your sample under the microscope to look for abnormal cell shapes and sizes, which can also indicate that someone has MDS.

Other tests that are performed on bone marrow samples include:

- **Chromosome analysis** to determine chromosome changes in bone marrow cells. Each cell in the body contains chromosomes (46 tightly coiled strands of DNA). Chromosomes contain all the information that cells need to function normally. In about half of patients with MDS, one or more chromosomal changes can be identified. The most common abnormalities are seen in chromosomes 5, 7, 8, and 20.
- **Flow cytometry** to see if the bone marrow cells are developing normally and to obtain an estimate of the blast count.
- **Molecular genetic studies** including some only done at Albert Einstein are performed to determine the specific genetic traits of the bone marrow cells. Analysis of mutations and other epigenetic changes in bone marrow cells are also done in selected cases.