Myelodysplastic Syndrome Early Detection, Diagnosis, and Staging

Detection and Diagnosis

Catching cancer early often allows for more treatment options. Some early cancers may have signs and symptoms that can be noticed, but that is not always the case.

- Can Myelodysplastic Syndromes Be Found Early?
- Signs and Symptoms of Myelodysplastic Syndromes
- How Are Myelodysplastic Syndromes Diagnosed?

MDS Scores and Prognosis (Outlook)

Myelodysplastic syndrome scores provide important information about the anticipated response to treatment.

- How Are Myelodysplastic Syndromes Scored?
- Survival Rates for Myelodysplastic Syndromes

Questions to Ask About Myelodysplastic Syndromes

Here are some questions you can ask your cancer care team to help you better understand your diagnosis and treatment options.

- What Should You Ask Your Doctor About Myelodysplastic Syndromes?

Can Myelodysplastic Syndromes Be Found Early?
Currently, no special tests are recommended for early detection of myelodysplastic syndromes (MDS) in the general population.

Follow-up physical exams and blood tests may help find some cases of MDS in cancer survivors previously treated with certain chemotherapy drugs.

- References

See all references for Myelodysplastic Syndromes

 Signs and Symptoms of Myelodysplastic Syndromes

Myelodysplastic syndromes (MDS) cause low blood counts, which can be found on blood tests, sometimes even before symptoms appear. Shortages of one or more types of blood cells cause many of the symptoms of myelodysplastic syndrome (MDS):

- Shortage of red blood cells (*anemia*) can lead to excessive tiredness, shortness of breath, and pale skin.
- Not having enough normal white blood cells (*leukopenia*) can lead to frequent or severe infections; often the neutrophil is the type of white blood cell that is low - this condition is called *neutropenia*.
- Shortage of blood platelets (*thrombocytopenia*) can lead to easy bruising and bleeding. Some people notice frequent or severe nosebleeds or bleeding from the gums.

Other symptoms can include weight loss, fever, and loss of appetite. Of course, these problems not only occur with MDS but are more often caused by something other than cancer.

- References

See all references for Myelodysplastic Syndromes
How Are Myelodysplastic Syndromes Diagnosed?

If signs and symptoms suggest you may have MDS, the doctors will look at cells from your blood and bone marrow to confirm this diagnosis.

Blood cell counts and blood cell examination

The complete blood count (CBC) is a test that measures the different cells in the blood, such as the red blood cells, the white blood cells, and the platelets. The CBC is often done with a differential count (or “diff”), which is a count of the different types of white blood cells in the blood sample. In a blood smear, some of the blood is put on a slide to see how the cells look under the microscope.

Patients with MDS often have too few red blood cells. They may have shortages of white blood cells and blood platelets as well. Patients with RAEB (refractory anemia with excess blasts) may have a small number of myeloblasts in the blood. Blasts are very early cells that are produced by bone marrow stem cells and are normally only found in bone marrow. When blasts are present in the blood it is always abnormal and often signals a bone marrow problem. Blood cells from MDS patients may also have certain abnormalities in size, shape, or other features that can be seen under the microscope.

Blood abnormalities may suggest MDS, but the doctor cannot make an exact diagnosis without examining a sample of bone marrow cells.

Other blood tests

The doctor may also order tests to check for other possible causes of low blood counts, such as low levels of vitamin B12 and folate.
Bone marrow tests

Bone marrow samples are obtained from a bone marrow aspiration and biopsy, tests that are usually done at the same time. The samples are usually taken from the back of the pelvic (hip) bone. These tests are used first for diagnosis and classification and may be repeated later to tell if the MDS is responding to therapy or is transforming into an acute leukemia.

For a bone marrow aspiration, you lie on a table (either on your side or on your belly). After cleaning the area, the skin over the hip and the surface of the bone is numbed with local anesthetic, which may cause a brief stinging or burning sensation. A thin, hollow needle is then inserted into the bone and a syringe is used to suck out a small amount of liquid bone marrow (about 1 teaspoon). Even with the anesthetic, most patients still have some brief pain when the marrow is removed.

A bone marrow biopsy is usually done just after the aspiration. A small piece of bone and marrow (about 1/16 inch in diameter and 1/2 inch long) is removed with a needle that is twisted as it is pushed down into the bone. The biopsy may also cause some brief pain. Once the biopsy is done, pressure will be applied to the site to help prevent bleeding.

A pathologist (a doctor specializing in the diagnosis of diseases using laboratory tests) examines the bone marrow samples under a microscope. A hematologist (a doctor specializing in medical treatment of diseases of the blood and blood-forming tissues) or an oncologist (a doctor specializing in medical treatment of cancer) usually reviews these as well.

The doctors will look at the size and shape of the cells and see whether the red cells contain iron particles or whether the other cells contain granules (microscopic packets of enzymes and other chemicals that help white blood cells fight infections). The percentage of marrow cells that are blasts is particularly important. Blasts are very early cells that are produced by bone marrow stem cells. Blasts eventually mature into normal blood cells. In MDS, the blasts do not mature properly, so there may be too many blasts and not enough mature cells. For a diagnosis of MDS, a patient must have less than 20% blasts in the bone marrow. A patient who has more than 20% blasts in the bone marrow is considered to have acute leukemia.

Different types of tests that are done on the bone marrow help the doctor diagnose MDS:

Immunocytochemistry
Cells from the bone marrow sample are treated with special antibodies that cause certain types of cells change color. The color change can be seen only under a microscope. This testing is helpful in distinguishing different types of MDS or leukemia from one another and from other diseases.

**Flow cytometry**

This technique is sometimes used to examine the cells from bone marrow and blood samples. It is very helpful in diagnosing and classifying the type of MDS. It is also used in diagnosing leukemia and lymphoma. A sample of cells is treated with special antibodies and passed in front of a laser beam. Each antibody sticks only to certain types of cells. If the sample contains those cells, the laser will cause them to give off light. The instrument detects the light, and a computer counts the cells. This test may not be needed for all patients.

**Cytogenetics**

This test looks at the chromosomes inside the cells. DNA in human cells is packed into chromosomes. Each cell should have 46 chromosomes (23 pairs). Abnormal chromosomes are common in MDS. Sometimes parts of chromosomes or even whole chromosomes are missing. MDS cells may also have extra copies of all or part of some chromosomes. Chromosome translocations (portions of chromosomes may trade places with each other) may also be seen.

Cytogenetic testing can take several weeks because the bone marrow cells need time to grow in laboratory dishes before their chromosomes can be viewed under the microscope. The results of cytogenetic testing are written in a shorthand form that describes which chromosome changes are present. For example:

- A minus sign (-) or the abbreviation “del” is used to mean a deletion. For example, if a copy of chromosome 7 is missing, it can be written as -7 or del(7). Often, only a part of the chromosome is lost. There are 2 parts to a chromosome, called p and q. Thus the loss of the q part of chromosome 5 is written 5q- or del(5q).
- A plus sign is used when there is an extra copy of all or part of a chromosome. +8, for example, means that chromosome 8 has been duplicated, and too many copies of it are found within the cell.
- The letter t is used to indicate a translocation

Chromosome changes commonly seen in MDS include deletions in chromosomes 5 and 7 or an extra chromosome 8. Certain chromosome changes, such as del(5q) (a deletion of a part of chromosome 5), can predict a better outcome (as long as there are
no other chromosome changes). Other changes, such as deletions of chromosome 7 or changes in 3 or more chromosomes, have a poorer outlook.

**Molecular genetic studies**

These tests are another way to find chromosome and gene abnormalities. An example of this is *fluorescent in situ hybridization* – more commonly called FISH. In FISH, specific gene sequences are tagged with a fluorescent dye. These may correspond to a certain area of a chromosome or even a certain translocation. An advantage of FISH is that it doesn’t require actively dividing cells. This allows the testing to go a bit faster than cytogenetic testing. FISH is very good for finding translocations – it can even find some that may be too small to be seen with usual cytogenetic testing.

Polymerase chain reaction (PCR) is another molecular genetic test that can be used to look for specific gene abnormalities.

Molecular genetic studies are not needed to make a diagnosis in most cases of MDS, but they can be useful in determining a person’s outlook.

- **References**
  See all references for Myelodysplastic Syndromes

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**How Are Myelodysplastic Syndromes Scored?**

Doctors often group cancers into different stages based on the size of the tumor and how far the cancer has spread from the original site in the body. The stage of a cancer can help predict the outlook for a cancer. Often, the cancer’s stage is used to decide which treatment is needed.

However, myelodysplastic syndromes (MDS) are diseases of the bone marrow. They cannot be staged by looking at the size of a tumor like some other cancers. Other
factors are used to predict outlook and determine the need for treatment. These factors include the patient’s blood counts, the appearance of their bone marrow, their age, and certain chromosome changes.

**International Prognostic Scoring System**

The International Prognostic Scoring System (IPSS) is a system developed for staging MDS. It was intended for use with the FAB classification system. It rates 3 factors:

- The percentage of blasts in the bone marrow (scored on a scale from zero to 2)
- Chromosome abnormalities (scored from zero to 1)
- The patient’s blood counts. (scored as zero or 0.5)

Each factor is given a score, with the lowest scores having the best outlook. Then the scores for the factors are added together to make the IPSS score. The IPSS puts people with MDS into 4 groups:

- Low risk
- Intermediate - 1 risk (Int-1)
- Intermediate - 2 risk (Int-2)
- High risk

**WHO Prognostic Scoring System (WPSS)**

More recently, a scoring system was developed based on 3 factors:

- The type of MDS based on the WHO classification
- Chromosome abnormalities
- Whether or not the patient requires blood transfusions

<table>
<thead>
<tr>
<th>WHO type</th>
<th>Chromosomes</th>
<th>Needs Transfusions</th>
</tr>
</thead>
<tbody>
<tr>
<td>RA, RARS, (del)5q</td>
<td>Good</td>
<td>No</td>
</tr>
<tr>
<td>RCMD, RCMD-RS</td>
<td>Intermediate</td>
<td>Yes</td>
</tr>
<tr>
<td>RAEB-1</td>
<td>Poor</td>
<td></td>
</tr>
<tr>
<td>RAEB-2</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

This system puts patients with MDS into 5 groups
Survival Rates for Myelodysplastic Syndromes

Survival rates are often used by doctors as a standard way of discussing a person's prognosis (outlook). Some patients with cancer may want to know the survival statistics for people in similar situations, while others may not find the numbers helpful, or may even not want to know them.

Median survival is one way to look at outcomes and measures the amount of time for half the patients in a certain group to die. This is a middle value – half the patients live longer than this, and half do not live this long. These numbers are based on patients diagnosed some time ago. Improvements in treatment since these numbers were gathered may result in a more favorable outlook for people now being diagnosed with a myelodysplastic syndrome.

These survival rates are based on previous outcomes of large numbers of people who had the disease, but they cannot predict what will happen in any particular person's case. Many factors may affect a person’s outlook, such as the patient’s age and health, the treatment received, and how well the disease responded to treatment. Your doctor can tell you how the numbers below apply to your particular situation.
The following survival statistics are based on the International Prognostic Scoring System (IPSS) risk groups. These were published in 1997 and do not include patients treated with intensive chemotherapy.

<table>
<thead>
<tr>
<th>IPSS risk group</th>
<th>Median survival</th>
</tr>
</thead>
<tbody>
<tr>
<td>Low</td>
<td>5.7 years</td>
</tr>
<tr>
<td>Int-1</td>
<td>3.5 years</td>
</tr>
<tr>
<td>Int-2</td>
<td>1.2 years</td>
</tr>
<tr>
<td>High</td>
<td>5 months</td>
</tr>
</tbody>
</table>

The WHO Prognostic Scoring System (WPSS) risk groups can also be used to predict outcome. These statistics were published in 2007 based on patients diagnosed between 1982 and 2004.

<table>
<thead>
<tr>
<th>WPSS Risk Group</th>
<th>Median Survival</th>
<th>Risk of Leukemia (within 5 years)+</th>
</tr>
</thead>
<tbody>
<tr>
<td>Very low</td>
<td>12 years</td>
<td>3%</td>
</tr>
<tr>
<td>Low</td>
<td>5.5 years</td>
<td>14%</td>
</tr>
<tr>
<td>Intermediate</td>
<td>4 years</td>
<td>33%</td>
</tr>
<tr>
<td>High</td>
<td>2 years</td>
<td>54%</td>
</tr>
<tr>
<td>Very high</td>
<td>9 months</td>
<td>84%</td>
</tr>
</tbody>
</table>

+ The percentage of people who will develop leukemia within 5 years of being put into this risk group.

- References
  See all references for Myelodysplastic Syndromes

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What Should You Ask Your Doctor About Myelodysplastic Syndromes?
It is important to have frank, open, and honest discussions with your doctor about your condition. Your doctor and the rest of the health care team want to answer all of your questions. For instance, consider these questions:

- What type of myelodysplastic syndrome do I have?
- What is my prognostic score?
- What treatment choices do I have?
- Which treatment, if any, do you recommend, and why?
- What are the side effects of the treatments that you recommend?
- How can I help reduce the side effects I may have from the treatment?
- What is the outlook for my survival?
- Should I get a second opinion, and can you recommend an expert in this field?

References

See all references for Myelodysplastic Syndromes

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