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# Myelodysplastic Syndrome Early Detection, Diagnosis, and Staging

Know the signs and symptoms of myelodysplastic syndrome. Find out how MDS is tested for, diagnosed, and staged.

## 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](#)
- [Tests for Myelodysplastic Syndromes](#)

## MDS Scores and Prognosis (Outlook)

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

- [Myelodysplastic Syndrome Prognostic Scores](#)
- [Survival Statistics 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.

- [Questions to Ask Your Doctor About Myelodysplastic Syndromes](#)

# Can Myelodysplastic Syndromes Be Found Early?

At this time, there are no widely recommended tests to screen for myelodysplastic syndromes (MDS). (Screening is testing for cancer in people without any symptoms.)

MDS is sometimes found when a person sees a doctor because of [signs or symptoms](#) they are having. These signs and symptoms often do not show up in the early stages of MDS. But sometimes MDS is found before it causes symptoms because of an abnormal result on a [blood test](#) that was done as part of a routine exam or for some other health reason. MDS that is found early does not always need to be treated right away, but it should be watched closely for signs that it's progressing.

For some people who are known to be at [increased risk](#)<sup>1</sup>, such as people with certain inherited syndromes or people who have received certain chemotherapy drugs, doctors might recommend close follow-up with blood tests or other exams or tests to look for possible early signs of MDS.

## Hyperlinks

1. [www.cancer.org/cancer/types/myelodysplastic-syndrome/causes-risks-prevention/risk-factors.html](http://www.cancer.org/cancer/types/myelodysplastic-syndrome/causes-risks-prevention/risk-factors.html)

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# Signs and Symptoms of Myelodysplastic Syndromes

A main feature of myelodysplastic syndromes (MDS) is that they cause low blood cell counts. Sometimes this is found on [blood tests](#), even before symptoms appear. In other cases, symptoms related to shortages of one or more types of blood cells (cytopenias) are the first sign of MDS:

- Having too few red blood cells (*anemia*) can lead to feeling tired, dizzy, or weak, as well as shortness of breath and pale skin.
- Not having enough normal white blood cells (*leukopenia*), especially cells called neutrophils (*neutropenia*), can lead to frequent or severe infections.
- Having too few blood platelets (*thrombocytopenia*) can lead to easy bruising and bleeding. Some people have frequent or severe nosebleeds or bleeding from the gums.

Other symptoms can include:

- Weight loss
- Fever
- Bone pain
- Loss of appetite

These symptoms are more likely to be caused by something other than MDS. Still, if you have any of these symptoms, especially if they don't go away or get worse over time, see your doctor so that the cause can be found and treated, if needed.

## References

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## Tests for Myelodysplastic Syndromes

- [Blood cell counts and blood cell examination](#)
- [Other blood tests](#)
- [Bone marrow tests](#)
- [Lab tests of bone marrow or blood samples](#)

Some people have [signs or symptoms](#) that suggest they might have a myelodysplastic syndrome (MDS). If you have symptoms, your health care provider will get a complete medical history, focusing on your symptoms and when they began. You will also be examined for possible causes of your symptoms.

For other people, MDS might be suspected based on the results of blood tests that are done for another reason.

In either case, if MDS is suspected, you will likely need tests to look at your blood and bone marrow cells to see if you have MDS or some other health condition.

### Blood cell counts and blood cell examination

The **complete blood count (CBC)** is a test that measures the levels of red blood cells, white blood cells, and platelets in your blood. 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 (anemia). They may have shortages of white blood cells and blood platelets as well. Patients with some types of MDS might also have myeloblasts ("blasts") in the blood. These are very early forms of blood cells that are normally only found in bone marrow. Blasts in the blood are not normal and are often a sign of 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. For example, low levels of iron, vitamin B12, or folate can cause anemia. If one of these is found to be abnormal, a diagnosis of MDS is much less likely.

## 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 they may be repeated later to tell if the MDS is responding to treatment or is transforming into an acute leukemia.

For a **bone marrow aspiration**, 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. 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 is removed with a slightly larger needle that is put 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.

## Lab tests of bone marrow or blood samples

A pathologist (a doctor specializing in the diagnosis of diseases using laboratory tests) examines the bone marrow and blood samples under a microscope. Other doctors,

such as a hematologist (a doctor specializing in medical treatment of diseases of the blood and blood-forming tissues), might review these as well.

The doctors will look at the size, shape, and other features of the cells. The percentage of cells in the bone marrow or blood that are *blasts* (very early forms of blood cells) is particularly important. 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 and blood. A patient who has more than 20% blasts is considered to have [acute myeloid leukemia \(AML\)](#)<sup>1</sup>.

Other types of lab tests can also be done on the bone marrow or blood samples to help diagnose MDS:

### **Flow cytometry and immunocytochemistry**

For both flow cytometry and immunocytochemistry, samples of cells are treated with antibodies, which are proteins that stick only to certain other proteins on cells. For immunocytochemistry, the cells are then looked at under a microscope to see if the antibodies stuck to them (meaning they have these proteins), while for flow cytometry a special machine is used.

These tests can be helpful in distinguishing different types of MDS or leukemia from one another and from other diseases.

### **Chromosome tests**

These tests look at the chromosomes (long strands of DNA) inside the cells. Each cell should have 46 chromosomes (23 pairs). Abnormal chromosomes are common in MDS (see below).

**Cytogenetics:** In this test, the cells are looked at under a microscope to see if the chromosomes have any abnormalities. A drawback of this test is that it usually takes about 2 to 3 weeks because the cells must grow in lab dishes for a couple of weeks before their chromosomes can be viewed.

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. The loss of the q part of chromosome 5 is written 5q- or del(5q).

- A plus sign is used when there is an **addition** (an extra copy of all or part of a chromosome). +8, for example, means that chromosome 8 has been duplicated, and there are too many copies of it within the cell.
- The letter *t* is used to indicate a **translocation**, in which parts of two chromosomes have traded places with each other. For example, if chromosomes 8 and 21 have swapped pieces, it would be written as t(8;21)

Certain chromosome changes in MDS cells can help predict the likely course of MDS. For example, a deletion of a part of chromosome 5, or del(5q), usually predicts a better outcome (as long as there is no more than one other chromosome change, and it isn't a loss of part of chromosome 7). Changes in 3 or more chromosomes or the deletion of chromosome 7 tend to have a poorer outlook.

**Fluorescent in situ hybridization (FISH):** This test looks more closely at cell DNA using fluorescent dyes that only attach to specific gene or chromosome changes. An advantage of FISH is that it doesn't require actively dividing cells, so it can usually provide results within a couple of days. 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):** This is a very sensitive DNA test that can also find some chromosome changes too small to be seen under a microscope, even if there are very few abnormal cells in a sample.

### Other molecular and genetic tests

Other types of lab tests can also be done on the samples to look for specific gene or other changes in the MDS cells. This is sometimes referred to as [biomarker testing](#)<sup>2</sup>.

For example, the MDS cells are tested for changes (mutations) in genes such as IDH1. People whose MDS cells have changes in this gene are more likely to be helped by treatment with certain [targeted therapy drugs](#)<sup>3</sup>.

## Hyperlinks

1. [www.cancer.org/cancer/types/acute-myeloid-leukemia.html](http://www.cancer.org/cancer/types/acute-myeloid-leukemia.html)
2. [www.cancer.org/cancer/diagnosis-staging/tests/biomarker-tests.html](http://www.cancer.org/cancer/diagnosis-staging/tests/biomarker-tests.html)
3. [www.cancer.org/cancer/types/myelodysplastic-syndrome/treating/chemotherapy.html](http://www.cancer.org/cancer/types/myelodysplastic-syndrome/treating/chemotherapy.html)

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# Myelodysplastic Syndrome Prognostic Scores

- [Revised International Prognostic Scoring System \(IPSS-R\)](#)
- [WHO Prognostic Scoring System \(WPSS\)](#)
- [Other prognostic factors](#)

For most types of cancer, the **stage** of the cancer – a measure of how far it has spread – is one of the most important factors in selecting treatment options and in determining a person's outlook (prognosis).



But myelodysplastic syndromes (MDS) are diseases of the bone marrow. The outlook for these cancers isn't based on the size of a tumor or whether the cancer has spread. Because of this, doctors use other factors to predict outlook and decide when to treat. Some of these factors have been combined to develop scoring systems.

## Revised International Prognostic Scoring System (IPSS-R)

The revised International Prognostic Scoring System (IPSS-R) is based on 5 factors:

- The percentage of blasts (very early forms of blood cells) in the bone marrow
- The type and number of chromosome abnormalities in the cells
- The level of red blood cells (measured as hemoglobin) in the patient's blood
- The level of platelets in the patient's blood
- The level of neutrophils (a type of white blood cell) in the patient's blood

Each factor is given a score, with the lowest scores having the best outlook. Then the scores for the factors are added up to put people with MDS into 5 risk groups:

- Very low risk
- Low risk
- Intermediate risk
- High risk
- Very high risk

These risk groups can be used to help predict a [person's outlook](#). This can be helpful when trying to determine the best [treatment](#)<sup>1</sup> options.

This system has some important limitations. For example, it was developed before many of the current treatments for MDS were available, so it only took into account people who were not treated for their MDS. It also did not include people who have MDS as a result of getting chemotherapy (secondary MDS). But this system can still be helpful and is still widely used.

## WHO Prognostic Scoring System (WPSS)

The World Health Organization (WHO) scoring system is based on 3 factors:

- The [type of MDS based on the WHO classification](#)<sup>2</sup> (For example, certain types of MDS-SLD and MDS-del(5q) tend to have the best outlook, whereas MDS-EB tends

to have the worst.)

- Chromosome abnormalities (grouped as good, intermediate, or poor)
- Whether or not the patient needs regular blood transfusions

Each factor is given a score, with the lowest scores having the best outlook. Then the scores are added up to put people with MDS into 5 risk groups:

- Very low risk
- Low risk
- Intermediate risk
- High risk
- Very high risk

These risk groups can be used to help predict a [person's outlook](#), as well as how likely the MDS is to transform into [acute myeloid leukemia \(AML\)](#)<sup>3</sup>. This can be helpful when trying to determine the best treatment options. But as with the IPSS-R, this system has some important limitations. For example, it does not include people who have MDS as a result of getting chemotherapy (known as *secondary MDS*).

Both the IPSS-R and the WPSS can be complex, and different doctors might use different systems. If you have MDS, talk to your doctor about which system they use, which risk group you are in, and what it might mean for your treatment and outlook.

## Other prognostic factors

Along with the factors used in these scoring systems, doctors have found other factors that can also help predict a person's outlook. These include:

- A person's age
- A person's performance status (how well they're able to do normal daily activities)
- The severity of low blood cell counts
- The results of certain blood tests, such as the serum ferritin level
- Certain gene or chromosome changes that are not accounted for in the scoring systems

## Hyperlinks

1. [www.cancer.org/cancer/types/myelodysplastic-syndrome/treating.html](http://www.cancer.org/cancer/types/myelodysplastic-syndrome/treating.html)
2. [www.cancer.org/cancer/types/myelodysplastic-syndrome/about/mds-types.html](http://www.cancer.org/cancer/types/myelodysplastic-syndrome/about/mds-types.html)
3. [www.cancer.org/cancer/types/acute-myeloid-leukemia.html](http://www.cancer.org/cancer/types/acute-myeloid-leukemia.html)

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# Survival Statistics for Myelodysplastic

# Syndromes

- [Survival statistics don't tell the whole story](#)
- [Survival statistics for MDS](#)

Survival statistics are a way for doctors and patients to get a general idea of the outlook (prognosis) for people with a certain type of cancer. They can't tell you how long you will live, but they may help give you a better understanding about how likely it is that your treatment will be successful. Some people will want to know the survival statistics for their cancer, and some people won't. If you don't want to know, you don't have to.

**Median survival** is one way to look at outcomes. It is the time after diagnosis at which half the patients in a certain group are still alive, and half have died. This is a middle value – half the patients live longer than this, and half do not live this long.

## Survival statistics don't tell the whole story

Survival stats are often based on previous outcomes of large numbers of people who had the disease, but they can't predict what will happen in any particular person's case. There are some limitations to remember:

- The numbers below are based on patients diagnosed with a myelodysplastic syndrome (MDS) some time ago. Improvements in treatment since these numbers were gathered may result in a better outlook for people now being diagnosed with MDS.
- These numbers are based on [prognostic scores](#) that take into account certain factors, such as the type of MDS, the results of certain blood tests, and whether the abnormal cells have certain chromosome changes. But other factors might also affect a person's outlook, such as the patient's age and health, and how well the disease responds to treatment.

Your doctor can tell you how the numbers below apply to you.

## Survival statistics for MDS

The following survival statistics are based on the [revised International Prognostic Scoring System \(IPSS-R\) risk groups](#). It's important to note that this system is based largely on people who were diagnosed many years ago and who *did not get treatments*

such as chemotherapy for their MDS.

### IPSS-R risk group Median survival

|              |           |
|--------------|-----------|
| Very low     | 8.8 years |
| Low          | 5.3 years |
| Intermediate | 3 years   |
| High         | 1.6 years |
| Very high    | 0.8 years |

The [WHO Prognostic Scoring System \(WPSS\) risk groups](#) can also be used to predict outcome – both median survival and the chance that the MDS will transform into [acute myeloid leukemia \(AML\)](#)<sup>1</sup> within 5 years. These statistics were published in 2007 based on patients diagnosed between 1982 and 2004.

### WPSS Risk Group Median Survival Risk of AML (within 5 years)

|              |            |     |
|--------------|------------|-----|
| Very low     | 11.8 years | 3%  |
| Low          | 5.5 years  | 14% |
| Intermediate | 4 years    | 33% |
| High         | 2.2 years  | 54% |
| Very high    | 9 months   | 84% |

Remember, these survival statistics are only estimates – they can't predict what will happen to any individual person. Many other factors can also affect a person's outlook. We understand that these statistics can be confusing and may lead you to have more questions. Talk to your doctor to better understand your specific situation.

## Hyperlinks

1. [www.cancer.org/cancer/types/acute-myeloid-leukemia.html](http://www.cancer.org/cancer/types/acute-myeloid-leukemia.html)

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## Questions to Ask Your Doctor About Myelodysplastic Syndromes

- [When you're told you have MDS](#)
- [When deciding on a treatment plan](#)
- [During and after treatment](#)

It is important to have open and honest discussions with your cancer care team about your myelodysplastic syndrome (MDS). You should feel free to ask any question, no matter how minor it might seem. For instance, consider these questions:

### When you're told you have MDS

- How sure are you about the diagnosis of MDS?
- Can you explain what MDS is? How is it different from leukemia?
- Do I need any other [tests](#) before we can decide on treatment?

- Do I need to see any other types of doctors?
- What [type of myelodysplastic syndrome](#)<sup>1</sup> do I have?
- Which [risk group](#) does my MDS fall into? How might this affect my prognosis and [treatment options](#)<sup>2</sup>?
- Are there other factors that could affect my outlook or treatment options?
- If I'm concerned about the costs and insurance coverage for my diagnosis and treatment, who can help me?

## When deciding on a treatment plan

- How much experience do you have treating MDS?
- What [treatment choices](#)<sup>3</sup> do I have? Do we need to treat the MDS right away?
- Which treatment, if any, do you recommend, and why?
- Should I get a [second opinion](#)<sup>4</sup> before starting treatment? Can you suggest a doctor or cancer center?
- What should I do to be ready for treatment?
- How long will treatment last? What will it be like? Where will it be done?
- What are the risks or side effects of the treatments that you recommend? How long are they likely to last?
- Will treatment affect my daily activities?
- What is the outlook for my survival?

## During and after treatment

Once treatment begins, you'll need to know what to expect and what to look for. Not all of these questions may apply to you, but getting answers to the ones that do may be helpful.

- How will we know if the treatment is working?
- What type of [follow-up](#)<sup>5</sup> will I need during and after treatment?
- Is there anything I can do to help manage side effects?
- What symptoms or side effects should I tell you about right away?
- How can I reach you on nights, holidays, or weekends?
- Do I need to change what I eat during treatment?
- Are there any limits on what I can do?
- Should I exercise? What should I do, and how often?

- Can you suggest a mental health professional I can see if I start to feel overwhelmed, depressed, or distressed?
- What would my options be if the treatment isn't working?
- Where can I find more information and support?

Along with these sample questions, be sure to write down any others you want to ask. For instance, you might want information about recovery times so that you can plan your work or activity schedule. Or you might want to ask about [clinical trials](#)<sup>6</sup> that might be right for you.

Keep in mind that doctors aren't the only ones who can give you information. Other health care professionals, such as nurses and social workers, can answer some of your questions. To learn more about speaking with your health care team, see [Talking With Your Doctor](#)<sup>7</sup>.

## Hyperlinks

1. [www.cancer.org/cancer/types/myelodysplastic-syndrome/about/mds-types.html](http://www.cancer.org/cancer/types/myelodysplastic-syndrome/about/mds-types.html)
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## Written by

The American Cancer Society medical and editorial content team



<https://www.cancer.org/cancer/acs-medical-content-and-news-staff.html>

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