CONDITION

Myelofibrosis

Myelofibrosis is a blood cancer that disrupts blood cell production by causing scarring in the bone marrow.

WHAT IT IS
Blood cancer causing bone marrow scarring

BODY SYSTEM
Blood and bone marrow

MAIN SYMPTOMS
Fatigue, breathlessness, abdominal fullness, night sweats

WHAT CAUSES IT
Acquired blood-cell mutations, often JAK2, CALR, or MPL

HOW IT IS DIAGNOSED
Blood tests, mutation testing, and bone marrow biopsy

MAIN TREATMENT
Monitoring, symptom control, targeted medicines, or stem cell transplant

CAN IT BE CURED?
Sometimes - donor stem cell transplant can cure

IS IT AN EMERGENCY?
Sometimes - heavy bleeding, stroke symptoms, or severe breathlessness

LONG-TERM OUTLOOK
Often long-term, shaped by blood counts and genetic changes

Overview

Myelofibrosis is a type of blood cancer called a myeloproliferative neoplasm. It begins when an abnormal blood-forming stem cell makes altered copies of itself. These cells release signals that cause scar tissue, called fibrosis, to build up in the bone marrow.

Bone marrow is the soft tissue inside bones where blood cells are normally made. As scarring increases, the marrow may struggle to make healthy red blood cells, white blood cells, and platelets. Red blood cells carry oxygen, white blood cells help fight infection, and platelets help stop bleeding.

The spleen and liver may begin making blood cells to compensate. This can enlarge the spleen, causing discomfort or fullness under the left ribs.

Primary myelofibrosis develops on its own. Secondary myelofibrosis develops in someone who previously had another blood condition, usually polycythaemia vera or essential thrombocythaemia.

The condition can behave very differently from one person to another. Some people remain stable without treatment for years. Others develop worsening blood counts, troublesome symptoms, or a faster-moving form of the condition.

Symptoms

Myelofibrosis may be found on a routine blood test before it causes symptoms. When symptoms occur, they may come from low blood counts, an enlarged spleen, or inflammation caused by abnormal blood-cell signals.

Possible symptoms include:

Anaemia means there are too few healthy red blood cells. It is common in myelofibrosis, but tiredness and breathlessness can have many other causes.

Symptoms can change gradually. New abdominal swelling, greater fatigue, worsening night sweats, or more frequent bleeding should be discussed at routine follow-up rather than simply accepted as part of the condition.

- Tiredness, weakness, dizziness, or breathlessness from anaemia
- Fullness, discomfort, or pain under the left ribs from an enlarged spleen
- Feeling full after eating only a small amount
- Night sweats, fever, itching, or unplanned weight loss
- Easy bruising, nosebleeds, or bleeding gums
- Frequent or slow-to-clear infections
- Bone or joint pain
- Gout, caused by a build-up of uric acid
- Reduced concentration or difficulty keeping up with usual activities

Causes and risk factors

Myelofibrosis usually results from genetic changes acquired during a person's lifetime. These changes affect how blood-forming cells grow and communicate. They are not changes that a

person caused through everyday behaviour.

Many people have a change in one of three genes: JAK2, CALR, or MPL. These genes are involved in signals that tell blood cells when to grow. Other genetic changes may also influence how the condition behaves.

These mutations are usually present only in the abnormal blood cells. Myelofibrosis is therefore not generally inherited directly by children. Some families do have a higher tendency to develop myeloproliferative conditions, but this does not mean that relatives will necessarily develop one.

Risk rises with age, although younger adults can be affected. Having polycythaemia vera or essential thrombocythaemia increases the chance of developing secondary myelofibrosis.

Exposure to high levels of ionising radiation or certain industrial chemicals has been linked with myeloproliferative cancers, but most people with myelofibrosis have no clear environmental trigger.

Diagnosis

No symptom or blood test result can diagnose myelofibrosis by itself. Assessment usually combines blood tests, examination, genetic testing, and a bone marrow biopsy.

A full blood count measures red cells, white cells, and platelets. The results may show anaemia or unusually high or low platelet and white-cell counts. A blood film allows the cells to be examined under a microscope. Myelofibrosis may cause immature cells and unusually shaped, teardrop-like red cells to enter the bloodstream.

Blood tests may also check liver and kidney function, iron, vitamin levels, uric acid, and lactate dehydrogenase, or LDH. LDH may rise when blood cells are being produced or broken down quickly.

Mutation testing looks for JAK2, CALR, MPL, and sometimes other genetic changes. Finding a mutation supports the diagnosis and can help estimate future risk, but a mutation alone does not confirm myelofibrosis.

During a bone marrow biopsy, a small sample of bone and marrow is usually taken from the back of the hip using local anaesthetic. A specialist examines its cells and the amount of scar tissue. Sometimes it is difficult to draw out liquid marrow because of the fibrosis, but a solid biopsy sample can still provide important information.

Tests are also used to rule out other causes of marrow scarring and related blood cancers. An ultrasound or other scan may measure the spleen and liver.

Treatment and management

Treatment is based on symptoms, blood counts, spleen size, general health, genetic findings, and the estimated risk of the condition progressing. Age matters, but it is not considered on its own.

If the condition is stable and causing few or no symptoms, regular monitoring may be appropriate. This is active observation rather than a lack of care. Follow-up generally includes symptom review, physical examination, and blood tests.

JAK inhibitors are targeted medicines that reduce abnormal growth signals inside blood cells. Examples used for myelofibrosis include ruxolitinib, fedratinib, pacritinib, and momelotinib. The most suitable option is influenced by symptoms and blood counts. These medicines can reduce spleen size and symptoms such as night sweats or itching. Some may also help anaemia in particular circumstances.

JAK inhibitors do not usually remove every abnormal cell or cure the condition. They can also lower blood counts and may affect infection risk, so blood tests and safety monitoring are needed. Treatment should not be stopped or changed without medical guidance because symptoms can return quickly.

Anaemia may be managed with red blood cell transfusions or medicines that support blood-cell production. The best approach depends on its cause and severity. Some people gradually need transfusions more often, while others do not.

An allogeneic stem cell transplant replaces abnormal marrow-forming cells with cells from a compatible donor. It is the only treatment with the potential to cure myelofibrosis. However, it can cause serious complications, including severe infections and graft-versus-host disease, in which donor immune cells attack the recipient's tissues. It is generally considered for selected people whose expected benefit outweighs these risks.

Radiotherapy to the spleen or surgical removal of the spleen is now used only in particular situations because both can cause significant complications. Clinical trials may provide access to newer treatments or combinations.

Supportive care can include treatment for pain, itching, gout, infection, or nutritional problems. Emotional support and practical help with work, fatigue, or travel for appointments can also be important.

Complications

Myelofibrosis can affect more than blood counts. An enlarged spleen may cause ongoing pain,

early fullness after meals, or pressure on nearby organs. Less often, blood-cell production outside the marrow forms growths in other parts of the body.

Low platelets or poorly functioning platelets can increase bleeding. In other people, the condition increases the chance of a blood clot. A clot may affect a leg, the lungs, the heart, the brain, or blood vessels around the liver and digestive organs.

Low white-cell counts and some treatments can make infections more likely. Anaemia can become severe enough to limit activity or require transfusions.

Increased blood flow and pressure around the liver may cause portal hypertension. This can lead to enlarged veins in the digestive tract, fluid in the abdomen, or bleeding.

A minority of cases change into acute myeloid leukaemia, a faster-growing blood cancer. Changes in blood counts, a rise in immature cells called blasts, or rapidly worsening symptoms may lead to further testing. Progression is not inevitable.

Prevention

There is no proven way to prevent most cases of myelofibrosis. The mutations usually arise without a clear or controllable cause.

If you have polycythaemia vera or essential thrombocythaemia, regular follow-up can identify signs of progression earlier, but it cannot guarantee that myelofibrosis will be prevented.

Avoiding tobacco and unnecessary exposure to harmful chemicals or radiation supports general health but has not been shown to prevent myelofibrosis specifically. Because the condition is not usually inherited directly, routine testing of healthy relatives is not generally useful unless a specialist identifies an unusual family pattern.

Living with it and what to expect

Myelofibrosis is usually a long-term condition. Its course is influenced by symptoms, blood counts, chromosome and gene findings, age, and overall health. Risk scores combine several of these features to help guide monitoring and treatment decisions.

Some people have few symptoms and stable blood tests for years. Others develop increasing anaemia, spleen enlargement, weight loss, or a need for more treatment. A person's risk category can change, so it may be reassessed over time.

Regular follow-up helps track changes before they cause major problems. Keeping a brief record of fatigue, bleeding, infections, night sweats, weight, and abdominal discomfort can

make gradual changes easier to describe.

Fatigue may continue even when treatment improves blood tests or reduces spleen size. Pacing activities, planning rest, and addressing sleep, pain, nutrition, and mood may help. Exercise that matches your ability can support strength, but activity may need to be adjusted if anaemia, an enlarged spleen, or bleeding risk is significant.

Travel, work, and family life often remain possible, although appointments, transfusions, or treatment effects may require planning. People being considered for stem cell transplant usually need a detailed assessment and substantial support during recovery.

When to get medical help

Contact your haematology team promptly if you develop a clear increase in tiredness, abdominal swelling, night sweats, weight loss, bruising, bleeding, or infections. These changes usually need assessment, but they are not automatically an emergency.

Seek urgent medical advice for a fever or signs of infection, particularly if you have been told that your white-cell count is low or you are taking treatment that affects immunity. Follow any specific fever plan given by your treatment centre.

Get emergency help for:

These symptoms can have causes other than myelofibrosis, but they need immediate assessment.

- Severe difficulty breathing, chest pain, fainting, or blue-grey lips
- Sudden facial drooping, arm weakness, speech trouble, or confusion
- Heavy bleeding that does not stop
- Vomiting blood or passing black, tar-like stools
- Sudden severe abdominal pain, especially under the left ribs
- A painful swollen leg with sudden breathlessness

Common questions

Is myelofibrosis cancer?

Yes. It is a blood cancer within a group called myeloproliferative neoplasms. It often develops more slowly than acute leukaemia, although its behaviour differs between people.

Can myelofibrosis be cured?

A donor stem cell transplant can cure some people. Because the treatment has substantial risks, it is not suitable for everyone. Other treatments aim to control symptoms, improve blood counts, reduce spleen size, and slow complications.

How quickly does myelofibrosis progress?

There is no single timeline. Some cases remain stable for years, while others change more quickly. Blood counts, symptoms, genetic findings, and the number of immature blood cells help estimate risk more accurately than time since diagnosis alone.

Is myelofibrosis inherited?

Usually not. The main mutations are generally acquired in blood-forming cells during life rather than inherited from a parent. Rare family patterns can occur, but inheriting a tendency does not mean someone will definitely develop the condition.

Why does myelofibrosis make the spleen bigger?

When scarred bone marrow cannot make enough blood cells, the spleen may begin producing them. This extra work can enlarge it, causing fullness, discomfort under the left ribs, or feeling full soon after starting a meal.

Can myelofibrosis turn into leukaemia?

It can progress to acute myeloid leukaemia, but this does not happen to everyone. Regular blood tests and symptom reviews help identify changes that need further investigation.

Sources

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