



Mastocytosis



WHAT YOU NEED TO KNOW

You or your loved one has been diagnosed with mastocytosis.
What does it mean and how will it affect you?

This fact sheet will help you:

Learn about
mastocytosis and
how it is diagnosed

Get an overview
of treatment
options

Understand
what happens
next

What is mastocytosis?

Mastocytosis is a blood cancer where too many mast cells (white blood cells that help fight infection) build up in the body's tissues and organs. The two main types of mastocytosis are:

- Cutaneous mastocytosis (affects the skin)
- Systemic mastocytosis (affects internal areas such as bone marrow, liver, spleen, and gastrointestinal tract)

Mast cells

Mast cells are white blood cells that help the body fight infection. They are part of the immune system. They also help bones grow, blood vessels form, and wounds heal.

About mastocytosis

- Mastocytosis is a rare disorder where the body produces too many mast cells.
- Around 1 in 10,000 to 20,000 people worldwide have mastocytosis.
- It is often linked to an acquired (not inherited) mutation of the *KIT* D816V gene, which causes mast cells to grow nonstop and become activated.
- The activated mast cells lead to naturally occurring chemicals such as histamine being released by your immune system.
- The cause is unknown. In most cases, the genetic change is random.
- It affects men and women equally.
- It may present with one or both forms: cutaneous or systemic.
- In children, it may be diagnosed in their first year. It involves the skin and often resolves during puberty.

Symptoms of mastocytosis

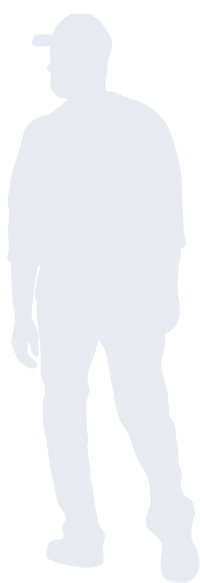
Mastocytosis symptoms are many and varied. They are often caused by too many mast cells. It's important to avoid anything that triggers an allergic reaction.

You may experience:

- A sudden drop in blood pressure and airways becoming narrow, blocking your breathing
 - When you're in anaphylaxis (severe and potentially life-threatening allergic reaction)
- Itching, flushing, hives, swelling, red or brown skin blotches
 - A response to an allergic reaction
- Wheezing, shortness of breath, blood pressure changes, dizziness, fainting
 - A response to an allergic reaction
- Nausea, diarrhea, vomiting, and stomach pain
 - When you are having an allergic reaction
- Anemia or bleeding disorders
 - When you have a buildup of mast cells in organs such as the bone marrow and/or an enlarged liver or spleen
- Muscle and joint pain, osteoporosis
 - When you have ongoing systemic mastocytosis
- Fatigue, lack of energy, mood changes, headache, brain fog
 - When you have ongoing systemic mastocytosis

Histamine

Histamine is a chemical released by your immune system as part of an allergic reaction. It causes symptoms like itching, sneezing, and watery eyes.



Your diagnosis

With a diagnosis, your healthcare team can determine the right treatment for you. Your healthcare team includes an oncologist (the primary cancer doctor), who works with a range of professionals such as your family doctor, nurse practitioners, nurses, pharmacist, and social worker to manage your treatment and provide support. Depending on the type of cancer, other specialists like a pathologist, radiologist, or surgeon may also be involved in your care.

Your test results help them predict how mastocytosis will likely progress and how you may respond to treatment.

Here are some possible tests you may undergo:

Name of test	Description
Medical history and physical exam	Your healthcare provider will review past illnesses, injuries, and symptoms. They will look at your lungs, heart, and other organs, and carefully examine your skin.
Blood tests	Blood tests help determine the need for treatment and the extent of the disease. You will be tested for tryptase, a protein that mast cells release.
Biopsy	Biopsy tests may include collection of bone marrow, skin samples, and/or tissue from the gastrointestinal tract. These tests will help determine the benefits of specific therapies.
Imaging tests	A computed tomography (CT) scan uses a computer linked to an X-ray machine to make a series of detailed pictures of areas inside the body. Magnetic resonance imaging (MRI) uses magnetic fields and radio waves to create images of the body's organs, including the brain and tissues.
Ultrasound	This test uses high-frequency sound waves to create pictures of internal organs, tissues, and blood flow.
Skeletal or bone survey	This test takes X-rays of the bones to look for any holes or lesions.
Bone density test	This test checks how dense your bones are. It is done to assess bone strength and the risk of osteoporosis and fractures.
Liver, adrenal, and thyroid function tests	These tests measure the levels of various substances in the body.
Genetic tests	These tests look for an acquired (not inherited) mutation of the <i>KIT</i> D816V gene.

Classification of mastocytosis

Classifying mastocytosis is based on:

- The location of mast cells
- Signs and symptoms
- Clinical presentation (how the disease appears to your healthcare team)
- Test results

Identifying the type and subtype of mastocytosis is an important step in planning your treatment. Subtypes vary depending on the organs involved and the severity of the disease. Types and subtypes include:

Cutaneous mastocytosis (CM)

- Affects the skin and often causes itchy reddish-brown lesions
- More common in children
- Subtypes:
 - Urticaria pigmentosa (also called maculopapular CM)
 - Telangiectasia macularis eruptiva perstans (TMEP)
 - Diffuse CM (a rare form in infants)
 - Mastocytoma (infant and child form)

Systemic mastocytosis (SM)

- Affects internal organs like the lymph nodes and liver
- More common in adults
- Has significant symptoms and abnormalities in the blood
- Skin lesions are common
- Subtypes:
 - Indolent (mild or no symptoms)
 - Smouldering (slow to progress)
 - Aggressive (fast growing)
 - Systemic mastocytosis with an associated blood cancer
 - Mast cell leukemia (rare and aggressive)

Mast cell sarcoma (MCS)

- Very rare
- Shows up as a solid tumour of atypical mast cells in bones or soft tissues





Clinical trials are research studies that aim to improve the care and treatment of people living with cancer.

For some people with a blood cancer, a clinical trial may be the best treatment choice. Talk to your healthcare team for more information.

Treatment of mastocytosis

New treatment approaches will help manage mastocytosis symptoms and complications, including infections and fatigue. The types of treatment can vary widely and may include clinical trials.

You may experience mild to severe side effects during treatment, depending on your age, overall health, and treatment plan. If so, let your healthcare team know.

Side effects can affect people in different ways. Most side effects improve or go away after treatment ends. New drugs and therapies can help control most side effects.

Types of treatment

There are many steps you can take to minimize the symptoms and avoid an allergic reaction. Common mastocytosis treatments and possible side effects include:

Medications to help you manage mastocytosis and side effects

- Antihistamines, moisturizers, and corticosteroid creams for itchy skin lesions
- Injectable epinephrine (EpiPen) in case of a severe, potentially life-threatening allergic reaction (anaphylactic shock)
- Cromolyn sodium to reduce mast cell activation
- Bisphosphonates to strengthen weak bones
- Leukotriene inhibitor to reduce flushing (when the skin gets red and warm due to increase in blood flow)
- Non-steroidal anti-inflammatory to relieve bone pain
- Vitamin C to reduce blood histamine levels

**Treatments
for advanced
systemic
mastocytosis
and side
effects**

Chemotherapy uses medicine (chemicals) to kill cancer cells. It also affects the body's healthy, fast-growing cells (such as hair, nails, and blood cells). Chemotherapy is often given in combination with immunotherapy.

- **Potential side effects:** low blood cell counts (white, red, and platelets), infection, bleeding, anemia, nausea, diarrhea, vomiting, loss of appetite, brain fog (chemo brain), fatigue, shortness of breath, diseases or disorders affecting the heart (cardiopathy), temporary hair loss, mouth sores, rashes, secondary cancers, and nerve damage (neuropathy)

Immunotherapy uses an intravenous drug that can either boost or pause your immune system to help your body fight cancer. Immunotherapy is done in addition to chemotherapy. It is available in Canada to treat mastocytosis only through a clinical trial.

- **Potential side effects:** rashes, fatigue, diarrhea, nausea, vomiting, and decreased thyroid hormone levels

Targeted therapies are a type of drug therapy that targets specific substances on cancer cells. These drugs are often given in pill form and may be helpful if the cancer has come back after treatment. Some targeted therapies are available in Canada for some types of systemic mastocytosis, while others are only available through a clinical trial.

- **Potential side effects:** low blood cell counts (white, red, and platelets), infection, bleeding, anemia, skin problems, high blood pressure, fatigue, diarrhea, neuropathy, and slower healing time for wounds

A **stem cell transplant** gives you healthy stem cells to replace those damaged by cancer or intense chemotherapy and radiation treatments. Your body relies on stem cells to produce blood cells.

Two main types of stem cell transplants are used to treat blood cancers:

- **Autologous:** The stem cells come from your body. This allows you to receive high doses of chemotherapy (sometimes with radiation). Using your own stem cells helps your bone marrow produce new blood cells and reset your immune system.
- **Allogeneic:** The stem cells come from a donor. These donor cells replace the damaged ones in your bone marrow, potentially offering a long-term cure.
 - **Potential side effects:** low white blood cell count (increased risk of infection), low platelet count (increased risk of bleeding or bruising), low red blood cell count (causes fatigue, dizziness, and shortness of breath), pain and issues with your digestive system, skin and hair problems, issues with your organs or central nervous system, and in the case of allogeneic stem cell transplants, possible graft-versus-host disease (GvHD) or veno-occlusive disease (affecting the small vessels leading to your liver)



Factors that affect treatment

Discuss your treatment options with your healthcare team to make sure you understand the benefits and risks of each approach. Your treatment plan is based on:

- Your age and overall health
- Your symptoms and test results
- The subtype of mastocytosis
- Disease status (first diagnosis or relapse, which is when the disease returns after initial treatment)
- Any other medical problems you may have
- Your lifestyle and preferences

Long-term or late effects of treatment

Medical follow-up is important after treatment for mastocytosis. You may need blood tests, bone marrow tests, or imaging tests to determine if you need further treatment. Your healthcare team will provide a care plan listing follow-up visits and the tests you will have at those visits.

You may experience long-term or late effects of your treatment:

- **Long-term side effects** can last for months or years after treatment ends. Examples include fatigue, fertility issues (the ability to conceive and have children), secondary cancers, and nerve damage.
- **Late effects** are medical problems that do not show up until years after treatment ends. See your healthcare team to get follow-up care for possible early detection of heart disease, secondary cancers, fertility issues, thyroid problems, trouble concentrating, or chronic fatigue.



Living with mastocytosis can be overwhelming. Seek medical help if you feel “down” or “blue” or don’t want to do anything and your mood does not improve over time. These could be signs of depression, an illness that should be treated even when you’re undergoing treatment for mastocytosis. Treatment for depression has important benefits for people living with cancer. Remember, you are not alone.

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