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2022

Systemic Mastocytosis

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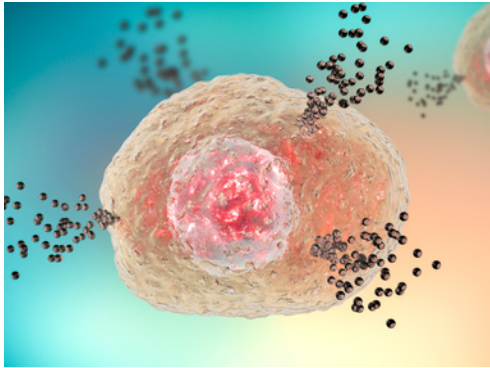
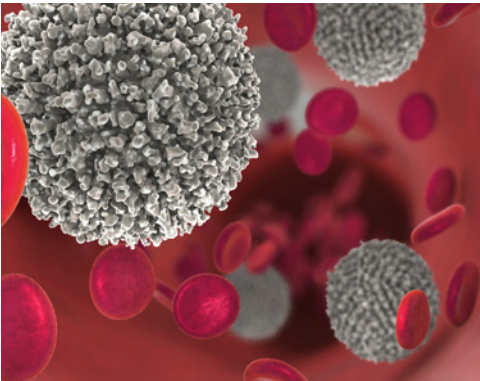
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Also supported by The Mast Cell Disease Society, Inc. (TMS)

The Mast Cell Disease Society, Inc. (TMS) is dedicated to providing multi-faceted support to patients, families, and medical professionals in our community and to leading the advancement of knowledge and research in mast cell diseases through education, advocacy and collaboration. We represent all those affected by Cutaneous and Systemic Mastocytosis and their variants, Mast Cell Activation Syndromes, and Hereditary Alpha-Tryptasemia. Email: info@tmsforacure.org, www.tmsforacure.org



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Basics

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Mastocytosis is a rare disorder that occurs when there are too many mast cells built up in your body. Mast cells are a type of white blood cell located all over your body. This chapter will provide an overview of the disease.

Blood

Blood is made up of red blood cells, platelets, plasma, and white blood cells. Plasma is the largest part of your blood. Plasma is made up of mostly water. Blood cells float in plasma. Blood takes oxygen and nutrients to your body's tissues, and carries away wastes.

There are 3 types of blood cells:

- Red blood cells (RBCs)
- White blood cells (WBCs)
- Platelets (PLTs)

Blood cells do not live forever. Normal red blood cells live for 3 months. Normal white blood cells live for 8 to 14 days. Normal platelets live for about 1 week (7 days). After cells reach these ages, they die off and are replaced by new cells. Your blood cells are being replaced in your body all the time.

Blood has many functions, they include:

- **Transportation** - Blood takes oxygen from the lungs to the cells of the body. It takes carbon dioxide from the body's cells to the lungs where it is breathed out. Blood also carries nutrients, hormones, and waste products around the body.

- **Regulation** - Blood helps to keep the acid-alkali balance of the body in check. It also plays a part in regulating body temperature. Increasing the amount of blood flowing close to the skin helps the body to lose heat.
- **Protection** - White blood cells attack and destroy invading germs (bacteria) and other pathogens. Blood clots form after an injury, which protects the body from losing too much blood.

Mast cells

Mast cells are a type of white blood cell found all over your body. They are part of your immune system. If bacteria or a virus is found, mast cells help the immune system to fight the infection (releasing a chemical called histamine). Histamine causes your blood vessels to expand and your skin to get itchy and swollen. It can also create a buildup of mucus, which makes the airways narrow. This response is used to protect your body from infections.

Mast cells also have other important functions. They help heal wounds, grow bone, and form new blood vessels. Mast cells are made in the bone marrow. Bone marrow is the spongy substance found in the middle of your bones.

Mastocytosis

Mastocytosis occurs when too many mast cells build up in your skin or organs such as the liver, spleen, bone marrow, and small intestines. When the mast cells are triggered they release substances similar to an allergic reaction. Sometimes, the reaction can cause severe inflammation that may result in organ damage.

There are two main forms of mastocytosis, they include:

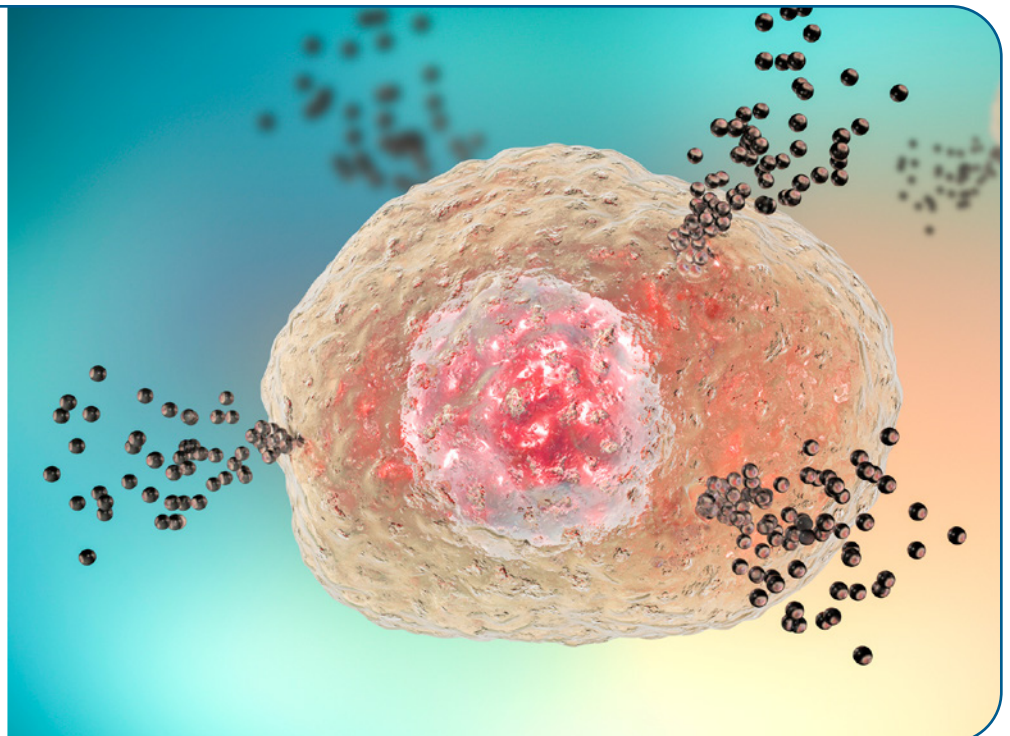
- **Cutaneous** - cutaneous mastocytosis affects the skin only. It occurs most often in children. During cutaneous mastocytosis, mast cells build up in the skin, causing red or brown lesions that itch. While cutaneous mastocytosis isn't life-threatening, it can cause a severe allergic reaction, which can be fatal.

Because cutaneous mastocytosis is limited to the skin, it is often treated with antihistamines and topical creams.

- **Systemic** - Systemic mastocytosis affects parts of the body other than the skin. It often occurs in adults. During systemic mastocytosis, mast cells accumulate in the bone marrow and organs (such as the intestines). If the systemic mastocytosis is aggressive (forms, grows, or spreads quickly), it can be life-threatening. Systemic mastocytosis includes two rare forms, mast cell leukemia and mast cell sarcoma. This book will focus on systemic mastocytosis.

Mast cell releasing histamine

Mast cells are a type of white blood cell. Histamine is released from mast cells in response to an allergen, causing an allergic reaction.



Subtypes

Systemic mastocytosis is broken up into subtypes.

- **Indolent systemic mastocytosis (ISM)** - A benign form of systemic mastocytosis. It refers to an abnormal accumulation of mast cells mainly in the bone marrow, but also in other organs or skin.
- **Smoldering systemic mastocytosis (SSM)** - A rare, slow to progress form of systemic mastocytosis (SM). It refers to a gradual buildup of mast cells in your internal organs.
- **Aggressive systemic mastocytosis (ASM)** - A loss of organ function (usually liver, gastrointestinal [GI] tract, bone, or bone marrow) due to mast cells getting in.
- **Systemic mastocytosis with an associated hematologic neoplasm (SM-AHN)** - Too many mast cells build up in certain tissues and organs, damaging them. Systemic mastocytosis with associated hematologic neoplasm refers to a mast cell buildup that occurs together with another blood disorder, usually a myelodysplastic syndrome, myeloproliferative disorder, or acute myeloid leukemia (AML).
- **Mast cell leukemia (MCL)** - An aggressive cancer described as a buildup of mast cells found in more than 20% of your body's tissues or organs. This subtype is very rare.
- **Mast cell sarcoma (MCS)** - A tumor made of abnormal mast cells has invaded your body's tissues. This condition is very rare, and is often not associated with additional skin issues.

Signs and symptoms

Symptoms of mastocytosis differ. They are based on the part of the body that is affected. Some people may experience severe symptoms that last anywhere from 15 to 30 minutes, others may not have any problems. Symptoms of mastocytosis are more likely to occur if “triggered”.

Common triggers include:

- Alcohol
- Surgery
- Spicy foods
- Exercise
- Insect stings
- Certain medications

Symptoms are associated with your type of mastocytosis:

- **Cutaneous mastocytosis** - The most common symptom is macules on the upper and lower extremities and abdomen. A macule is a flat, discolored (brown or tan) area of skin less than 1 centimeter (cm) wide.
- **Systemic mastocytosis** - You may experience symptoms found with cutaneous mastocytosis, as well as brain fog, low blood pressure (hypotension), abdominal pain, vomiting, diarrhea, fatigue, and frequent headaches. You may also experience an enlarged liver and spleen, anemia, or osteoporosis.

Speak to your health care provider about any symptoms you are experiencing.

Causes

Mastocytosis is most often caused by mutations (changes) in the *KIT* gene. The *KIT* gene tells the body to make a protein that helps to control cell growth, division, survival, and movement. Mutations in this gene is what causes an overproduction of mast cells. Most cases of systemic mastocytosis are not inherited. The cause of the mutation is because of a random change in the *KIT* gene. It is unknown why this mutation occurs.

It is important to tell your care team about all side effects so they can be managed.

Complications

A complication is an unwanted result of a disease or treatment. Complications may negatively affect your outcome of a disease.

Complications of systemic mastocytosis may include:

- Anaphylactic (allergic) reaction - rapid heartbeat, fainting, loss of consciousness and shock. A severe reaction may require a shot of epinephrine. An epinephrine injection is used to treat life-threatening allergic reactions.
- Blood disorders - anemia, cytopenia, and poor blood clotting
- Bone fractures (caused by disease)
- Peptic ulcer disease - ulcers and bleeding in your digestive tract
- Reduced bone density - you may be at risk of bone problems, such as osteoporosis
- Organ failure - inflammation and damage to the organs
- Liver problems - the liver can become swollen and may not work as well.
- Cancer - although rare, you may develop mast cell leukemia or mast cell sarcoma (cancer of the body's soft tissues)

Key points

- Mastocytosis is a rare disorder that occurs when there are too many mast cells built up in your body.
- Mast cells are a type of white blood cell found all over your body.
- Mastocytosis occurs when too many mast cells build up in your skin or organs such as the liver, spleen, bone marrow, and gastrointestinal tract.
- When the mast cells are triggered they release histamine which leads to an allergic reaction. Sometimes, the reaction can cause severe inflammation that may result in organ damage.
- Symptoms of mastocytosis differ. They are based on the part of the body that is affected.
- The most common symptom is macules on the upper and lower extremities and abdomen. A macule is a flat, discolored (brown or tan) area of skin less than 1 centimeter (cm) wide.
- Mastocytosis is most often caused by changes (mutations) in the *KIT* gene.

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Testing

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Treatment planning starts with testing. Accurate testing is needed to diagnose and treat systemic mastocytosis. This chapter presents an overview of the tests you might receive and what to expect.

Test results

Mastocytosis will be diagnosed based on a series of test results. Your diagnosis will determine your treatment plan. It is important you understand what these test results mean.

Keep these things in mind:

- Bring someone with you to doctor visits, if possible.
- Write down questions and take notes during appointments. Don't be afraid to ask your care team questions. Get to know your care team and help them get to know you.
- Get copies of blood tests, imaging results, and reports about your specific disease.
- Organize your papers. Create files for insurance forms, medical records, and test results. You can do the same on your computer.
- Keep a list of contact information for everyone on your care team. Add it to your phone. Hang the list on your refrigerator or keep it in a place where someone can access it in an emergency. Keep your primary care physician informed of changes to this list.



Create a medical binder

A medical binder or notebook is a great way to organize all of your records in one place.

- Make copies of blood tests, imaging results, and reports about your specific type of cancer. It will be helpful when getting a second opinion.
- Choose a binder that meets your needs. Consider a zipper pocket to include a pen, small calendar, and insurance cards.
- Create folders for insurance forms, medical records, and tests results. You can do the same on your computer.
- Use online patient portals to view your test results and other records. Download or print the records to add to your binder.
- Organize your binder in a way that works for you. Add a section for questions and to take notes.
- Bring your medical binder to appointments. You never know when you might need it!

General health tests

Medical history

A medical history is a record of all health issues and treatments you have had in your life. Be prepared to list any illness or injury and when it happened. Bring a list of old and new medicines and any over-the-counter medicines, herbals, or supplements you take. You may also want to bring in pictures of allergic reactions. Tell your doctor about any symptoms you have. A medical history will help determine which treatment is best for you. It is sometimes called a health history.

Family history

Some cancers and other diseases can run in families. Your doctor will ask about the health history of family members who are blood relatives. This information is called a family history. Ask family members about their health issues like heart disease, cancer, and diabetes, and at what age they were diagnosed.

Physical exam

During a physical exam, a health care provider may:

- ▶ Check your temperature, blood pressure, pulse, and breathing rate
- ▶ Check your weight
- ▶ Listen to your lungs and heart
- ▶ Look in your eyes, ears, nose, and throat
- ▶ Feel and apply pressure to parts of your body to see if organs are of normal size, are soft or hard, or cause pain when touched. Tell your doctor if you feel pain.

- ▶ Feel for enlarged lymph nodes in your neck, underarm, and groin. Tell your doctor if you have felt any lumps or have any pain.
- ▶ Conduct a complete skin exam

For a list of possible tests, [see Guide 1](#).

Testing takes time.
It might take days
or weeks for all test
results to come in.

Guide 1**Possible tests: Systemic mastocytosis**

Medical history and physical exam, including history of mast cell activation symptoms

Comprehensive metabolic panel with uric acid, LDH, and LFTs

Serum tryptase level

CBC with differential

Examination of blood smear

Bone marrow aspirate and biopsy with flow cytometry, immunohistochemistry, and cytogenetics

FISH test

Molecular testing

Myeloid mutation panel

CT/MRI or ultrasound of the abdomen/pelvis

DEXA scan

Skeletal survey

Organ-directed biopsy

24-hour urine studies (useful in certain circumstances)

HLA testing (useful in certain circumstances)

Assessment of symptom burden and quality of life (QOL) (useful in certain circumstances)

Blood tests

Blood tests check for signs of disease and how well organs are working. They require a sample of your blood, which is removed through a needle placed into your vein.

Comprehensive metabolic panel

A comprehensive metabolic panel (CMP) is a test that measures 14 different substances in your blood. A CMP provides important information about how well your kidneys and liver are working, among other things.

- **Uric acid** - A uric acid blood test is a medical test used to check the amount of a normal waste product in your blood. It is also known as a serum urate (SU) test or as a uric acid level.
- **Lactate dehydrogenase (LDH)** - LDH is a type of protein, known as an enzyme. LDH is found in almost all the body's tissues, including in the blood, heart, kidneys, brain, and lungs. When these tissues are damaged, they release LDH into the bloodstream or other body fluids. If your LDH blood or fluid levels are high, it may mean certain tissues in your body have been damaged by disease or injury.
- **Liver function tests (LFTs)** - LFTs look at the health of your liver by measuring chemicals that are made or processed by the liver. Levels that are too high or low signal that the liver is not working well or the bile ducts might be blocked.

Complete blood count with differential

A complete blood count (CBC) measures the levels of red blood cells (RBCs), white blood cells (WBCs), and platelets (PLTs) in your

blood. Your doctor will want to know if you have enough red blood cells to carry oxygen throughout your body, white blood cells to fight infection, and platelets to control bleeding. There are several types of white blood cells.

A differential counts the number of each type of white blood cell. It also checks if the counts are in balance with each other.

Serum tryptase level

Tryptase is an enzyme that is released with histamine and other chemicals, from mast cells when they are part of a normal immune response as well as an allergic reaction. This test measures the amount of tryptase in the blood.

Blood smear

A blood smear is often used as further testing if your CBC shows abnormal results. This test is used to evaluate the different types of blood cells. It may be used to help diagnose and/or monitor conditions that affect blood cell production.

Biopsies

A biopsy is the removal of a sample of tissue or group of cells for testing. It is an important part of an accurate diagnosis. Your sample should be reviewed by a pathologist who is an expert in the diagnosis of systemic mastocytosis. This review is often referred to as histology, histopathology, or hematopathology review. The pathologist will note the overall appearance and the size, shape, and type of your cells. Tests will be done on the biopsied cells.

Bone marrow aspiration and biopsy

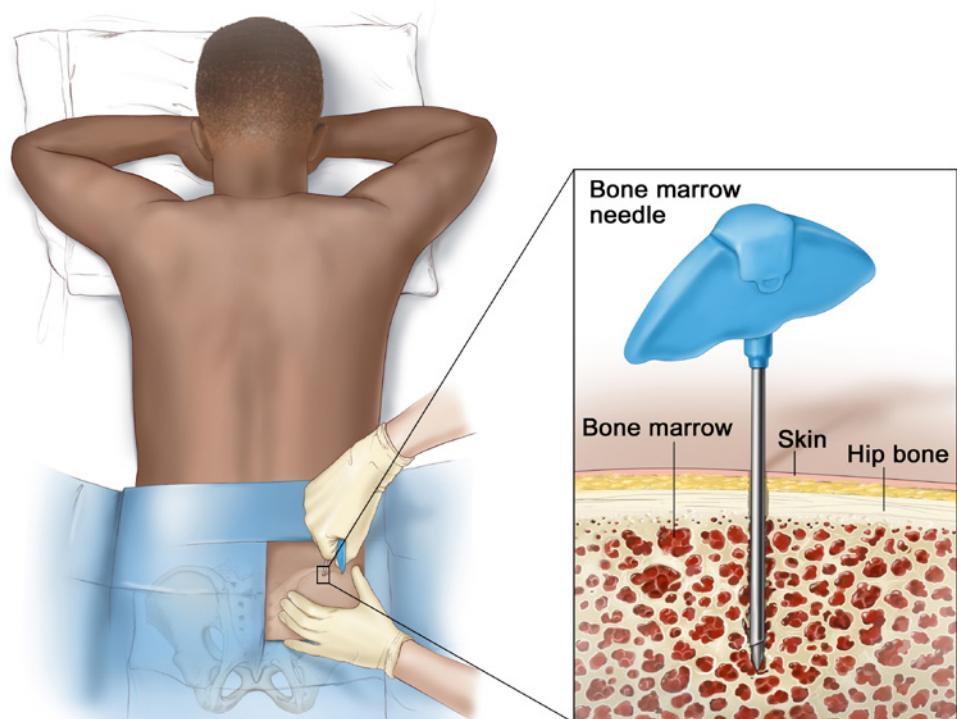
Bone marrow is like a sponge holding liquid. A bone marrow aspiration takes some of the liquid out of the sponge; a biopsy takes a piece of the sponge. For aspiration, a hollow needle

will be pushed through your skin and into the bone. Liquid bone marrow will then be drawn into a syringe. For the biopsy, a needle will be used to remove a core sample. The samples will be sent to a lab for testing. You may feel bone pain at your hip for a few days. Your skin may bruise.

Ask your provider about the type of bone marrow test you might have, where the sample will be taken, and if you will be given something to help relax.

Bone Marrow Aspiration and Biopsy

A bone marrow biopsy and aspiration is pictured. Here you see a hollow needle inserted through the bone to remove a sample.



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Organ-directed biopsies

For mastocytosis you can expect to receive one of the following organ-directed biopsies.

EGD or upper endoscopy

A gastrointestinal (GI) biopsy is often performed to confirm a diagnosis of mastocytosis. In an esophagogastroduodenoscopy (EGD), a device is guided down the throat into the esophagus, stomach, and upper parts of the small intestine (duodenum). An EGD is used to inspect the lining of these organs and to look for any signs of cancer or other abnormalities such as dilated blood vessels or ulcers. After the procedure, your throat may feel sore and you may feel some swelling.

Liver biopsy

In a liver biopsy, a needle may be inserted through the skin to remove a liver sample. It also may be inserted through a large vein in your neck (jugular), and threaded down to a vein (inferior vena cava) near your liver.

Immunophenotyping

Immunophenotyping uses antibodies to detect the presence or absence of white blood cell antigens. These antigens are proteins that can be found on the surface of or inside white blood cells. They are called biomarkers.

A complete blood test can count the number of white blood cells, but it cannot detect the subtle differences between different types of blood cancers. Immunophenotyping can detect these subtle differences.

There are 2 testing methods:

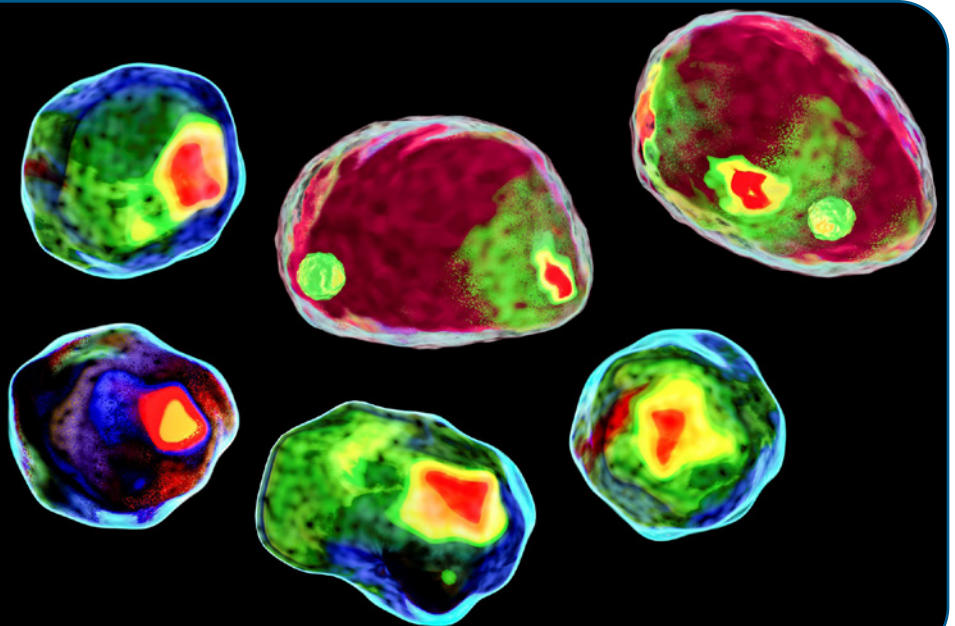
- Flow cytometry
- Immunohistochemistry

Flow cytometry

Flow cytometry is a laboratory method used to detect, identify, and count specific cells. Flow cytometry involves adding a light-sensitive dye

Cells studied with flow cytometry

Pictured are dyed cells that are passed through laser beams to help measure size and shape of cells.



to cells. The dyed cells are passed through a beam of light in a machine. The machine measures the number of cells, the size and shape of the cells, and other unique features of cells. Flow cytometry may be used on cells from circulating (peripheral) blood or from a bone marrow aspirate.

Immunohistochemistry

Immunohistochemistry (IHC) is a special staining process that involves adding a chemical marker to cells. The cells are then studied using a microscope.

Genetic tests

Your doctor may suggest tests for gene mutations. Genes tell cells what to become and what to do. In a process called mutation, something goes wrong in the genetic code. This can cause cells to grow and divide out of control.

Genetic testing looks for the presence or absence of certain gene mutations. Test results will help your treatment team learn more about your disease and make a treatment plan.

Since this test doesn't need growing cells, it can be performed on either a bone marrow or blood sample. A bone marrow sample is needed to get all the information your doctor needs to help plan your care.

Cytogenetics

Cytogenetics is a type of genetic test that uses samples of tissue, blood, or bone marrow to look for changes in chromosomes. A chromosome is a long DNA (deoxyribonucleic

acid) molecule with genetic material of an organism. The test specifically looks for broken, missing, rearranged, or extra chromosomes.

Myeloid mutation panel

A myeloid mutation panel is a blood test used to identify mutated genes. This test helps to diagnose and manage diseases such as acute myeloid leukemia (AML), myelodysplastic syndrome (MDS), myeloproliferative neoplasm (MPN), and MDS/MPN.

Biomarker tests

Biomarker tests also referred to as molecular tests are used to look for changes in one or more genes, and to determine if any abnormal genes or proteins are present.

FISH test

Fluorescence in situ hybridization (FISH) is a method that involves special dyes called probes that attach to pieces of DNA. FISH can look for translocations (switching parts between 2 chromosomes) and inversions (switching parts within one chromosome) that are too small to be seen with other methods. However, it can only be used for known changes. It cannot detect all the possible changes found within your chromosomes (karyotype).

***KIT* gene mutations**

A mutation in the *KIT* gene is the most common genetic change found in systemic mastocytosis. The *KIT* gene provides instructions for making a protein. It is important for the development and role of certain cell types. This includes reproductive cells (germ cells), early blood cells (hematopoietic stem cells), white blood cells (mast cells), cells in the gastrointestinal tract called interstitial cells of Cajal, and cells called melanocytes. *KIT D816V* is the gene specific to systemic mastocytosis.

Imaging

Imaging tests take pictures of the inside of your body. A radiologist, an expert in interpreting test images, will write a report and send this report to your doctor. Your test results will be discussed with you.

CT scan

A computed tomography (CT or CAT) scan uses x-rays and computer technology to take pictures of the inside of the body. It takes many x-rays of the same body part from different angles. All the images are combined to make one detailed three-dimensional (3D) picture.

In most cases, contrast will be used. Contrast material is used to improve the pictures of the inside of the body. Contrast materials are not dyes, but substances that help enhance and improve the images of several organs and structures in the body. It is used to make the pictures clearer. Contrast might be taken by mouth (oral) or given through a vein (intravenous [IV]). The contrast is not

permanent and will leave the body in your urine immediately after the test.

Tell your doctors if you have had allergic reactions to contrast in the past. This is important. You might be given medicines, such as diphenhydramine (Benadryl®) and prednisone (steroids), to avoid the effects of those allergies. Contrast might not be used if you have a serious allergy or if your kidneys are not working well.

MRI scan

A magnetic resonance imaging (MRI) scan uses radio waves and powerful magnets to take pictures of the inside of the body. It does not use x-rays. Contrast might be used.

Ultrasound

An ultrasound (US) uses high-energy sound waves to form pictures of the inside of the body. A probe will be pressed onto your abdomen. This is similar to the sonogram used for pregnancy. Ultrasound is painless and does not use x-rays, so it can be repeated as needed. It can show small areas of cancer that are near the surface of the body. Sometimes, an ultrasound or CT is used to guide a biopsy.

DEXA scan

A DEXA scan is an imaging test that measures bone strength (density). DEXA scans are used to determine your risk for osteoporosis (bone loss).

Skeletal survey

A skeletal survey, also referred to as a bone survey, is a series of x-rays taken to look at all the bones in your body. This test is used to determine if bone disease is present.

Other tests

You may receive additional tests such as urine testing, HLA typing, or a quality of life (QOL) assessment. These tests will help to determine the extent of your disease and treatment for symptoms.

Urine test

Instead of a blood test, urine tests are often used to help determine the presence of histamine in your body. The test consists of providing urine over a 24-hour period to evaluate histamine production over a longer time frame.

HLA typing

Human leukocyte antigens (HLAs) are proteins found on the surface of most cells. They play an important role in your body's immune response. HLAs are unique to each person. They mark your body's cells. Your body detects these markers to tell which cells are yours. Each person's set of HLAs is called the HLA type or tissue type.

HLA typing is a blood test that detects a person's HLA type. This test is done before a donor (allogeneic) blood stem cell transplant. To find a donor match, your proteins will be compared to the donor's proteins to see how many proteins are the same.

Quality of life assessment

You may be asked to complete a quality of life (QOL) assessment called an MC-QoL. It is a disease-specific tool that looks to identify your physical and mental wellness, as well your ability to function in your daily activities.

Hereditary alpha-tryptasemia

Hereditary alpha-tryptasemia (HαT) refers to a biochemical trait. A trait is a change in your DNA. DNA stands for deoxyribonucleic acid. It is in every cell of every living thing.

Those with HαT have inherited extra copies of the *alpha tryptase* gene (*TPSAB1*). Extra copies of this gene cause increased levels of the trypase protein in blood tests. Trypase is a protein made by mast cells. Trypase is often used to test for a mast cell activation. A mast cell activation results in an allergic reaction.

Symptoms of HαT may include:

- Itchy skin, flushing, hives, and anaphylaxis
- Gastrointestinal (GI) symptoms such as bloating, abdominal pain, diarrhea and/or constipation, heartburn, and difficulty swallowing
- Connective tissue symptoms such as hypermobile joints and scoliosis
- Cardiac symptoms such as a racing or pounding heartbeat or blood pressure swings with/without fainting
- Anxiety, depression, and panic attacks

Key points

- Mastocytosis will be diagnosed based on a series of test results. Your diagnosis will determine your treatment plan. It is important you understand what these tests mean.
- Blood tests check for signs of disease and how well organs are working. They require a sample of your blood, which is removed through a needle placed into your vein.
- A biopsy is the removal of a sample of tissue or group of cells for testing. It is an important part of an accurate diagnosis.
- Immunophenotyping uses antibodies to detect the presence or absence of white blood cell antigens. These antigens are proteins that can be found on the surface of or inside white blood cells. They are called biomarkers.
- Genetic testing looks for the presence or absence of certain gene mutations. Test results will help your treatment team learn more about your disease and make a treatment plan.
- Imaging tests take pictures of the inside of your body.
- You may receive additional tests such as urine testing, HLA typing, or a quality of life assessment. These tests will help to determine the extent of your disease and treatment for symptoms.

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Treatment

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This chapter presents an overview of the different treatment types for systemic mastocytosis. Together, you and your doctor will choose a treatment plan that is best for you.

Treatment team

Treatment decisions should involve a multidisciplinary team (MDT). An MDT is a team of doctors, health care workers, and social care professionals from different professional backgrounds who have knowledge (expertise) and experience with systemic mastocytosis. This team is united in the planning and implementation of your treatment.

Ask who will coordinate your care.

You know your body better than anyone. Help other team members understand:

- How you feel
- What you need
- What is working and what is not

Keep a list of names and contact information for each member of your team. This will make it easier for you and anyone involved in your care to know whom to contact with questions or concerns.



Your treatment team

Treating systemic mastocytosis takes a team of doctors and other experts. Your treatment team may include:

Dermatologist: specializes in the diagnosis and treatment of skin diseases.

Hematologist: an expert in blood diseases and cancers.

Pathologist: an expert in evaluating and testing tissue to diagnose and classify disease

Gastroenterologist: specializes in diagnosing and treating diseases that occur in the gastrointestinal system.

Allergist: specializes in the diagnosis and treatment of asthma and other allergic diseases.

Antihistamines

Antihistamines, also referred to as allergy medicine, block histamines to stop allergy symptoms. A histamine is a chemical made by the body that is released by blood basophils (white blood cells) into the bloodstream when your immune system is defending against a potential allergen. Antihistamines can control itchy skin, watery eyes, as well as other allergic symptoms of mastocytosis.

Epinephrine

Epinephrine is an drug used in emergencies to treat an allergic reaction. Epinephrine helps to quickly improve breathing, stimulate the heart, raise a dropping blood pressure, reverse hives, and reduce swelling of the face, lips, and throat. The drug is injected through your skin, often in the thigh.

A list of antihistamines most often used for the treatment of systemic mastocytosis can be found in [Guide 2](#).

Guide 2

Antihistamine treatment options: Systemic mastocytosis

Brompheniramine maleate

Cetirizine hydrochloride (Zyrtec®)

Chlorpheniramine maleate (Chlor-Trimeton®)

Cimetidine (Tagamet®)

Desloratadine (Clarinetx®)

Dimenhydrinate (Dramamine®)

Diphenhydramine hydrochloride

Doxylamine (Unisom®)

Epinephrine (EpiPen®)

Famotidine (Pepcid®)

Fexofenadine hydrochloride (Allegra®)

Hydroxyzine hydrochloride

Hydroxyzine pamoate (Vistaril®)

Levocetirizine dihydrochloride (Xyzal®)

Loratadine (Claritin®)

Meclizine hydrochloride

Targeted therapy

Targeted therapy is a form of systemic therapy that works throughout your body. It is a drug therapy that focuses on specific or unique features of cancer cells. Targeted therapies seek out how cancer cells grow, divide, and move in the body. These drugs stop the action of molecules that help cancer cells grow and/or survive.

Tyrosine kinase inhibitor

A tyrosine kinase inhibitor (TKI) is a type of targeted therapy that blocks the signals that cause cancer to grow and spread. TKIs might be used alone or in combination with other systemic therapies like chemotherapy.

TKIs used to treat systemic mastocytosis include:

- PDGFR - directed TKI - Avapritinib (Ayvakit™)
- BCR-ABL - directed TKI - Imatinib mesylate (Gleevec®)
- FLT3 - directed TKI - Midostaurin (Rydapt®)

Chemotherapy

Chemotherapy kills fast-growing cells throughout the body, including cancer cells and normal cells. Chemotherapy drugs used for the treatment of systemic mastocytosis affect the instructions (genes) that tell cells how and when to grow and divide.

Cladribine (Leustatin®) is a chemotherapy drug used to treat systemic mastocytosis.

Immunosuppressants

Immunosuppressive therapy (IST) uses materials made either by the body or in a laboratory to improve, target, or restore immune system function. IST is a type of drug therapy that lowers the body's immune response to allow bone marrow stem cells to grow and make new blood cells. Immunosuppressants can be topical or systemic.

ISTs used to treat systemic mastocytosis include:

- Biologic response modifier
- Prednisone
- Steroids

Biologic response modifiers

Biologic response modifiers (BRM), is a type of treatment that activates the body's immune system to fight cancer. BRM is used to help the immune system do its job. BRM is also referred to as immunotherapy. Peginterferon alfa-2a (Pegasys®), is a specific BRM used to treat systemic mastocytosis.

Prednisone

Prednisone is used to decrease the number of mast cells and block the release of histamine in systemic mastocytosis.

Steroids

Corticosteroids or steroids are lab-made immunosuppressants used to reduce inflammation. Steroids can be placed on the skin as an ointment or gel, inhaled through the mouth, used as a mouth (oral) rinse, taken as a pill, or given as an infusion through an intravenous (IV) needle.

Steroids can cause short-term and long-term side effects. Ask your care team about possible side effects.

Steroids used to treat systemic mastocytosis:

- ▶ Beclomethasone dipropionate (Diprolene®)
- ▶ Dexamethasone/dexamethasone sodium phosphate
- ▶ Hydrocortisone (Cortef®)
- ▶ Methylprednisolone (Solu-Medrol®)

Get to know your care team and let them get to know you.

Steroids

In systemic mastocytosis, steroids are used to help reduce allergies and inflammation. The most common types of steroids are tablets, liquid, or intravenous (injection into a vein).



Clinical trials

A clinical trial is a type of medical research study. After being developed and tested in a laboratory, potential new ways of fighting diseases need to be studied in people. If found to be safe and effective in a clinical trial, a drug, device, or treatment approach may be approved by the U.S. Food and Drug Administration (FDA).

Everyone with cancer should carefully consider all of the treatment options available for their cancer type, including standard treatments and clinical trials. Talk to your doctor about whether a clinical trial may make sense for you.

Phases

Most cancer clinical trials focus on treatment. Treatment trials are done in phases.

- Phase I trials study the dose, safety, and side effects of an investigational drug or treatment approach. They also look for early signs that the drug or approach is helpful.
- Phase II trials study how well the drug or approach works against a specific type of cancer.
- Phase III trials test the drug or approach against a standard treatment. If the results are good, it may be approved by the FDA.
- Phase IV trials study the long-term safety and benefit of an FDA-approved treatment.



Finding a clinical trial

In the United States

NCCN Cancer Centers

[NCCN.org/cancercenters](https://www.nccn.org/cancercenters)

The National Cancer Institute (NCI)

[cancer.gov/about-cancer/treatment/clinical-trials/search](https://www.cancer.gov/about-cancer/treatment/clinical-trials/search)

Worldwide

The U.S. National Library of Medicine (NLM)

clinicaltrials.gov/

Need help finding a clinical trial?

NCI's Cancer Information Service (CIS)

1.800.4.CANCER (1.800.422.6237)

[cancer.gov/contact](https://www.cancer.gov/contact)

Who can enroll?

Every clinical trial has rules for joining, called eligibility criteria. The rules may include age, cancer type and stage, treatment history, or general health. These requirements ensure that participants are alike in specific ways and that the trial is as safe as possible for the participants.

Informed consent

Clinical trials are managed by a group of experts called a research team. The research team will review the study with you in detail, including its purpose and the risks and benefits of joining. All of this information is also provided in an informed consent form. Read the form carefully and ask questions before signing it. Take time to discuss with family, friends, or others you trust. Keep in mind that you can leave and seek treatment outside of the clinical trial at any time.

Start the conversation

Don't wait for your doctor to bring up clinical trials. Start the conversation and learn about all of your treatment options. If you find a study that you may be eligible for, ask your treatment team if you meet the requirements. If you have already started standard treatment you may not be eligible for certain clinical trials. Try not to be discouraged if you cannot join. New clinical trials are always becoming available.

Frequently asked questions

There are many myths and misconceptions surrounding clinical trials. The possible benefits and risks are not well understood by many with cancer.

Will I get a placebo?

Placebos (inactive versions of real medicines) are almost never used alone in cancer clinical trials. It is common to receive either a placebo with a standard treatment, or a new drug with a standard treatment. You will be informed, verbally and in writing, if a placebo is part of a clinical trial before you enroll.

Are clinical trials free?

There is no fee to enroll in a clinical trial. The study sponsor pays for research-related costs, including the study drug. You may, however, have costs indirectly related to the trial, such as the cost of transportation or child care due to extra appointments. During the trial, you will continue to receive standard cancer care. This care is billed to—and often covered by—insurance. You are responsible for copays and any costs for this care that are not covered by your insurance.

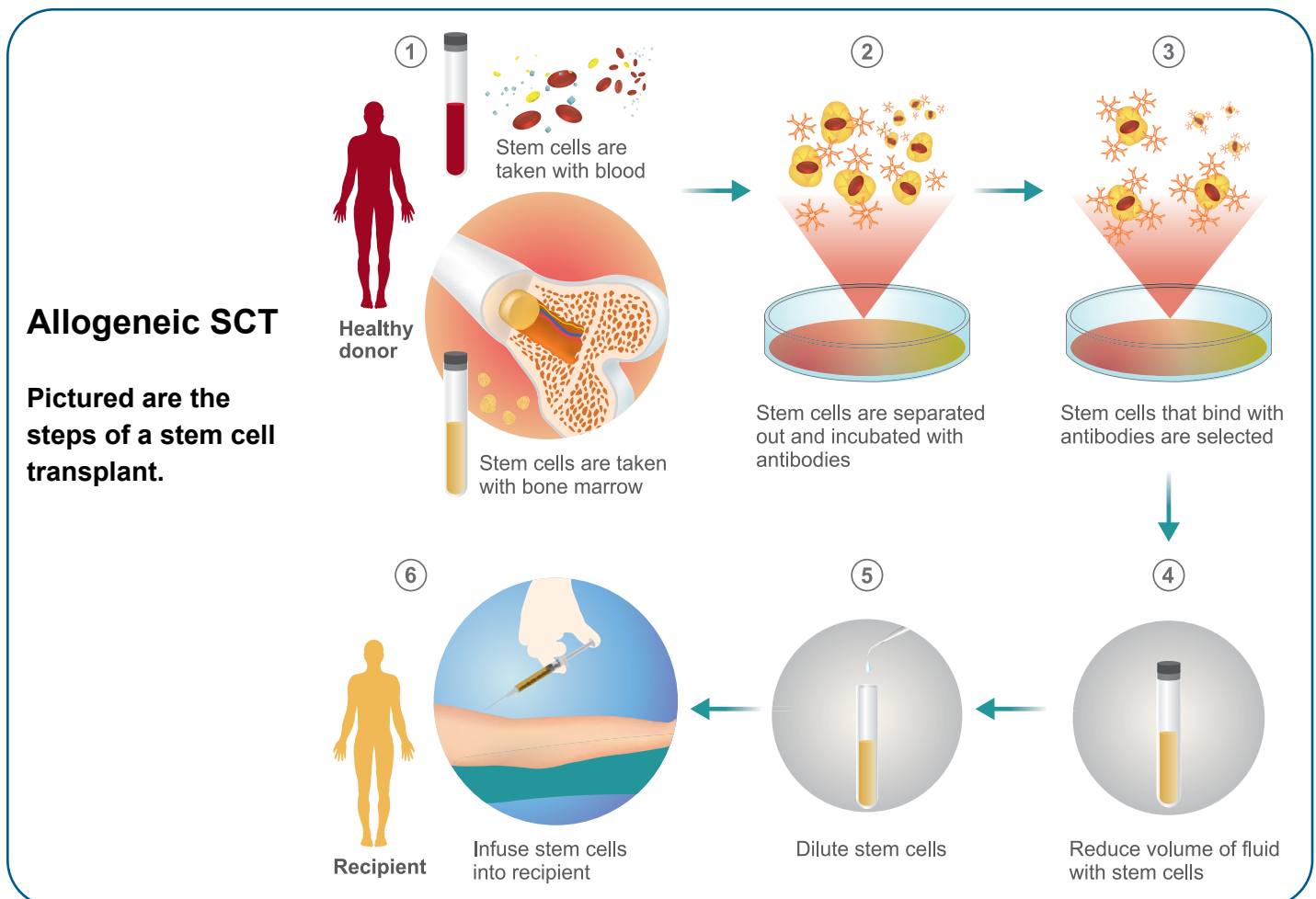
Allogeneic HCT

Hematopoietic cell transplant (HCT) (also referred to as bone marrow transplant [BMT] or stem cell transplant [SCT]) destroys cells in the bone marrow and replaces them with new, healthy blood cells. An allogeneic transplant uses healthy stem cells from a donor. The donor may or may not be related to you.

Before an SCT, you will receive a procedure called conditioning. Conditioning uses chemotherapy (and/or radiation therapy) to destroy bone marrow cells. This procedure creates room for the healthy stem cells and weakens the immune system so your body does not kill the transplanted cells.

After conditioning, you will receive the healthy stem cells through a transfusion. A transfusion is a slow injection of blood products into a vein. This can take several hours. New, healthy blood cells will form. This is called engraftment. It usually occurs about 2 to 4 weeks after the transplant.

Until the new, healthy blood cells form, you will have little or no immune defense. You may need to stay in a very clean room at the hospital or be given antibiotics to prevent or treat infection. While waiting for the cells to engraft, you will likely feel tired and weak.



Allogeneic HCT may help cure mast cell diseases, however it is rarely used because of risks and other effective treatment options.

Observation

Observation is a plan that closely watches your condition. Observation may also be referred to as surveillance or watch-and-wait. During this time, you will have tests on a regular basis to look for any new or changing symptoms. You will not have any treatment during observation.

General supportive care

Supportive care is health care that relieves symptoms and improves quality of life. It might include pain relief (palliative care), emotional or spiritual support, financial aid, or family counseling. Tell your care team how you are feeling and about any side effects. Best supportive care, supportive care, and palliative care are often used interchangeably.

Diarrhea

Diarrhea is frequent and watery bowel movements. Your care team will tell you how to manage diarrhea and may recommend medicines to stop the diarrhea. It is important to drink lots of fluids. Changes to your diet might help.

Distress

Distress is an unpleasant experience of a mental, physical, social, or spiritual nature. It can affect how you feel, think, and act. Distress might include feelings of sadness, fear, helplessness, worry, anger, and guilt.

Depression, anxiety, and sleeping problems are common in cancer. Talk to your doctor and with those whom you feel most comfortable about how you are feeling. There are services and people who can help you. Support and counseling services are available.

For more information, see *NCCN Guidelines for Patients: Distress During Cancer Care*, available at [NCCN.org/patientguidelines](https://www.nccn.org/patientguidelines).

Fatigue

Fatigue is extreme tiredness and inability to function due to lack of energy. There are treatments for fatigue. Let your care team know how you are feeling and if fatigue is getting in the way of doing the things you enjoy. Eating a balanced diet, exercise, yoga, and massage therapy can help. You might be referred to a nutritionist or dietitian (an expert in nutrition and food) to help with fatigue.

Fertility

Fertility is the ability to have children. In order to preserve one's fertility, action may be needed before starting treatment. However, this is not always possible. Those who want to have children in the future should be referred to a fertility specialist to discuss the options.

Nausea and vomiting

Nausea and vomiting might be a sign of systemic mastocytosis. You will be given medicine to treat nausea and vomiting.

Trouble eating

Sometimes side effects from systemic mastocytosis might cause you to feel not hungry or sick to your stomach (nauseated). You might have a sore mouth. Healthy eating is important during treatment. It includes eating a balanced diet, eating the right amount of food, and drinking enough fluids. A registered dietitian can help. Speak to your care team if you have trouble eating or maintaining your weight.

Pain

Pain is common in systemic mastocytosis. It might be caused by nausea and vomiting or abdominal cramps from diarrhea. Tell your care team about any pain or discomfort. You might meet with a palliative care specialist or with a pain specialist to manage pain.

Keep a pain diary

A pain diary is a written record that helps you keep track of when you have pain, how bad it is, what causes it, and what makes it better or worse. Use a pain diary to discuss your pain with your care team. You might be referred to a specialist for pain management.

Include in your pain diary:

- The time and dose of all medicines
- When pain starts and ends or lessens
- Where you feel pain
- Describe your pain. Is it throbbing, sharp, tingling, shooting, or burning? Is it constant, or does it come and go?
- Does the pain change at different times of day? When?
- Does the pain get worse before or after meals? Does certain food or drink make it better?
- Does the pain get better or worse with activity? What kind of activity?
- Does the pain keep you from falling asleep at night? Does pain wake you up in the night?
- Rate your pain from 0 (no pain) to 10 (worst pain you have ever felt)
- Does pain get in the way of doing the things you enjoy?

Key points

- ▶ Treatment decisions should involve a multidisciplinary team (MDT). An MDT is a team of doctors, health care workers, and social care professionals from different professional backgrounds who have knowledge (expertise) and experience with systemic mastocytosis.
- ▶ Antihistamines, also referred to as allergy medicine, reduce or block histamines to stop allergy symptoms. Antihistamines can control itchy skin, watery eyes, as well as other allergic symptoms of mastocytosis.
- ▶ Chemotherapy kills fast-growing cells throughout the body, including cancer cells and normal cells.
- ▶ Immunosuppressive therapy (IST) uses materials made either by the body or in a laboratory to improve, target, or restore immune system function.
- ▶ A clinical trial is a type of medical research study.
- ▶ A hematopoietic cell transplant (HCT) destroys cells in the bone marrow and replaces them with new, healthy blood cells. An allogeneic transplant uses healthy stem cells from a donor. The donor may or may not be related to you.
- ▶ Observation is a plan that closely watches your condition. You will not have any treatment during observation.
- ▶ Supportive care is health care that relieves symptoms and improves quality of life. It might include pain relief (palliative care), emotional or spiritual support, financial aid, or family counseling.



We want your feedback!

Our goal is to provide helpful and easy-to-understand information on cancer.

Take our survey to let us know what we got right and what we could do better:

[NCCN.org/patients/feedback](https://www.nccn.org/patients/feedback)

4

Systemic mastocytosis

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36 ISM and SSM

37 Advanced

38 Mast cell sarcoma

39 Key points



Systemic mastocytosis is a rare disease that is caused by too many mast cells in your body. When mast cells are triggered they release substances similar to an allergic reaction. This chapter reviews the different types of systemic mastocytosis as well as testing and treatment options.

Diagnosis

Systemic mastocytosis is a rare disorder defined by an abnormal buildup of mast cells in the skin, bone marrow, and internal organs (such as the liver, spleen, gastrointestinal tract, and lymph nodes).

Systemic mastocytosis is often diagnosed due to signs or symptoms. For example, a symptom of skin lesions may be confirmed by

a skin biopsy. During a skin biopsy, a sample of skin tissue is taken and looked at under a microscope to look for a buildup of mast cells. If there are no cutaneous lesions or if the skin biopsy reveals uncertain results, a bone marrow biopsy may be performed.

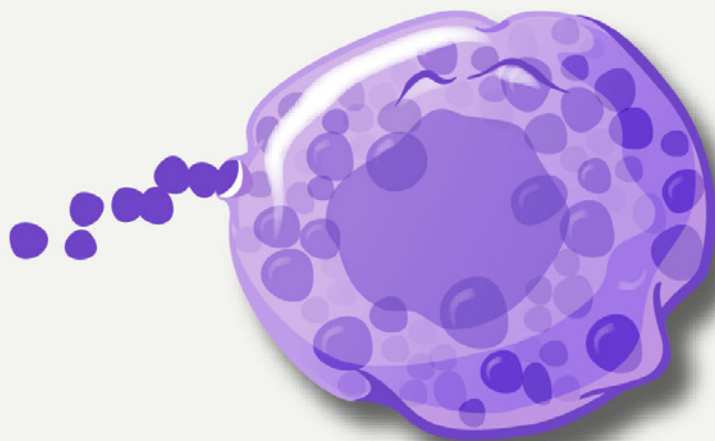
Skin is not the only organ affected by systemic mastocytosis. You may receive blood and/or urine tests to measure the levels of specific chemicals or substances related to mast cells. If tests show high levels of these substances, you will be diagnosed with systemic mastocytosis. Other tests you may receive include a bone scan, gastrointestinal workup, or genetic testing to confirm there is a mutation in the *KIT* gene.

If the systemic mastocytosis is aggressive (forms, grows, or spreads quickly), it can be life-threatening. Systemic mastocytosis includes two rare forms, mast cell leukemia and mast cell sarcoma.

Mast Cell

When triggered, mast cells release granules filled with chemicals (such as histamine), which sets off an allergic reaction.

Image: [https://commons.wikimedia.org/wiki/File:Mast_Cell_\(30107399584\).jpg](https://commons.wikimedia.org/wiki/File:Mast_Cell_(30107399584).jpg)



ISM and SSM

Indolent systemic mastocytosis

Indolent systemic mastocytosis (ISM) is a benign (not harmful) form of systemic mastocytosis. It refers to an abnormal buildup of mast cells. Mast cells often build up in the bone marrow, as well as other organs or skin. ISM has a low mast cell buildup compared to other types of systemic mastocytosis. Some people may experience maculopapular (flat and raised) skin lesions, an enlarged liver or spleen, or pain and discomfort within the gastrointestinal tract. ISM has a low risk of moving into a more aggressive disease.

Smoldering systemic mastocytosis

Smoldering systemic mastocytosis (SSM) is a form of systemic mastocytosis that is slow to progress into a more advanced disease. It refers to a gradual buildup of mast cells in your internal organs. Symptoms of SSM typically include an enlarged spleen, an increase in bone marrow (as seen in anemia or certain cancers), and skin lesions such as urticaria pigmentosa. People can remain stable for years or may develop a more aggressive type of systemic mastocytosis.

Treatment options

Primary treatment options for indolent or smoldering systemic mastocytosis include:

- Anti-mediator drug therapy
- Clinical trial

Systemic mastocytosis

Systemic mastocytosis is a rare disorder caused by too many mast cells.



Further testing

After primary treatment, you are likely to receive regular testing to look for any new or changing signs or symptoms.

Tests may include:

- Physical exam and/or blood tests - expect every 6 to 12 months
- DEXA scan - every 1 to 3 years for those with osteopenia/osteoporosis
- Quality of life assessment

If testing shows your disease has progressed, you will be restaged and treated based on the new stage.

Advanced

Aggressive systemic mastocytosis

Aggressive systemic mastocytosis (ASM) is described as a loss of organ function (usually liver, gut, bone, or bone marrow) due to mast cells getting in and taking over. Examples of organ function loss include low numbers of white blood cells, anemia, low platelets, liver dysfunction, and bone fractures due to bone lesions.

Treatment options

Primary treatment options for ASM include:

- Clinical trial
- Avapritinib (Ayvakit™)
- Midostaurin (Rydapt®)

Other treatment options may include:

- Cladribine (Leustatin®)
- Peginterferon alfa-2a with or without prednisone
- Imatinib mesylate (Gleevec®) (in certain circumstances)

If you respond to primary treatment, you may be asked to continue it and/or consider a test for an allogeneic HCT.

If you have little or no response to primary treatment, you will be restaged and treated based on your new stage of disease.

Systemic mastocytosis with AHN

Systemic mastocytosis with an associated hematologic neoplasm (SM-AHN) refers to a large number of mast cells that build up in certain tissues and organs, damaging them. Affected tissues or organs may include the bone marrow, lymph nodes, bone, liver, spleen, and small intestine. Systemic mastocytosis with associated hematologic neoplasm refers to a mast cell buildup that occurs together with another blood disorder, usually a myelodysplastic syndrome, myeloproliferative disorder, or acute myeloid leukemia (AML). Myeloproliferative and myelodysplastic disorders are the most common diseases associated with SM-AHN.

For more information, see *NCCN Guidelines for Patients: Myelodysplastic syndromes or Myeloproliferative neoplasms*, available at [NCCN.org/patientguidelines](https://www.nccn.org/patientguidelines).

Treatment options

Primary treatment options for systemic mastocytosis with an associated hematologic neoplasm (SM-AHN) include:

- Clinical trial
- Avapritinib (Ayvakit™)
- Midostaurin (Rydapt®)

Other treatment options may include:

- Cladribine (Leustatin®)
- Peginterferon alfa-2a with or without prednisone

If you respond to primary treatment, you may be asked to continue it and/or consider a test for an allogeneic HCT.

If your disease gets worse, you may be given AHN-directed therapy. AHN-directed therapy is based on your specific blood disorder. It may include consideration of an allogeneic HCT.

If you have no response to treatment, you will undergo more testing to restage your disease.

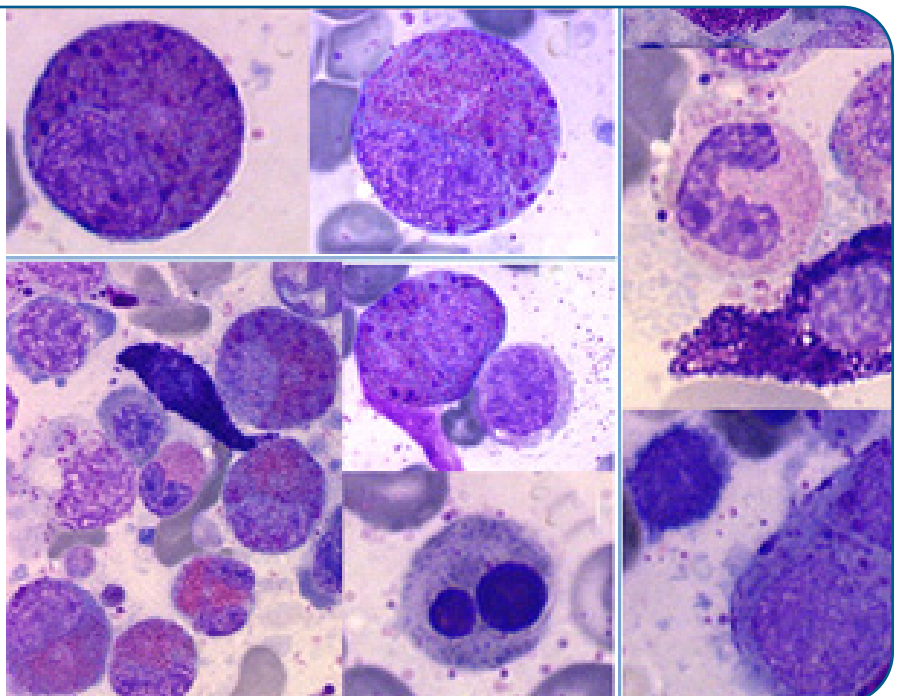
Mast cell sarcoma

Mast cell sarcoma, is an extremely aggressive form of sarcoma. A sarcoma is a tumor made of cells from connective tissue. Mast cell sarcoma is extremely rare.

For more information, see *NCCN Guidelines for Patients: Soft tissue sarcoma*, available at [NCCN.org/patientguidelines](https://www.nccn.org/patientguidelines).

Sarcoma cells

Sarcoma is an uncommon group of cancers found in bones and connective tissue such as fat and muscle.



Key points

- Systemic mastocytosis is often diagnosed due to signs or symptoms.
- Indolent systemic mastocytosis (ISM) is a benign (non-cancerous) form of systemic mastocytosis. It refers to an abnormal buildup of mast cells. Mast cells often build up in the bone marrow, as well as other organs or skin.
- Smoldering systemic mastocytosis (SSM) is a form of systemic mastocytosis that is slow to progress into a more advanced disease. It refers to a gradual buildup of mast cells in your internal organs.
- Aggressive systemic mastocytosis (ASM) is described as a loss of organ function (usually liver, gut, bone, or bone marrow) due to mast cells getting in and taking over.
- Systemic mastocytosis with an associated hematologic neoplasm (SM-AHN) refers to a large number of mast cells that build up in certain tissues and organs, damaging them.
- Mast cell sarcoma is an extremely aggressive form of sarcoma. Mast cell sarcoma is extremely rare.

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Mast cell leukemia

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Mast cell leukemia (MCL) is a very rare and aggressive type of systemic mastocytosis. MCL does not have positive outcomes.

Diagnosis

Leukemia is a group of cancers that's caused by cells in your bone marrow that produce abnormal or underdeveloped blood cells. Leukemias are classified based on the type of cells that are affected. Mast cell leukemia (MCL), is a type of leukemia that is caused by an abnormal growth of cells from myeloid progenitor cells.

Myeloid progenitor cells (stem cells) can become mast cells, platelets, red blood cells, and white blood cells.

There is no clear reason why MCL develops, but several gene mutations in the *KIT* gene have been linked to it.

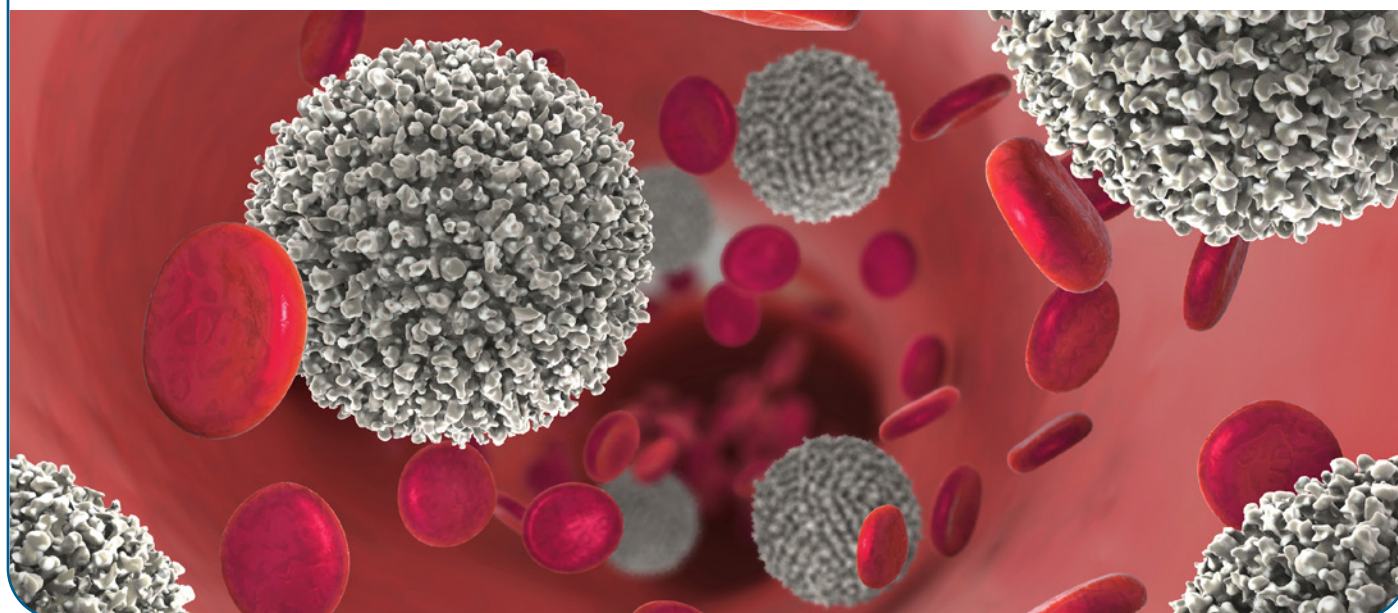
Mast cell leukemia (MCL) is an aggressive condition that leads to the buildup of mast cells in your bone marrow and other tissues.

Symptoms of MCL may include:

- Low blood pressure
- Fatigue
- Gastrointestinal distress

Mast cell leukemia

MCL originates in the bone marrow (myeloid tissue). It involves an overgrowth of mast cells. This crowds out production of red blood cells and platelets as pictured.



Treatment options

Primary treatment options for mast cell leukemia (MCL) include:

- Clinical trial
- Avapritinib (Ayvakit™)
- Midostaurin (Rydapt®)
- Cladribine (Leustatin®)

If you respond to primary treatment, you may be asked to continue it and/or consider a test for an allogeneic HCT.

If your disease gets worse (progresses to AHN [associated hematologic neoplasm] or AML [acute myeloid leukemia]), you may be given AHN-directed therapy. AHN-directed therapy is based on your specific blood disorder. It may include a multiagent chemotherapy and/or consideration of an allogeneic HCT.

If you have no response to treatment, you will undergo more testing to restage your disease.

No response to treatment refers to any of the following:

- Return or progression of organ damage
- Enlarged spleen or liver
- Progression of systemic mastocytosis symptoms
- Resistance to drug therapy

MCL is very serious and life-threatening. Treatment will focus on supportive care. This includes symptom relief, quality of life, and longer survival. It is important to ask your health care provider about your prognosis.

Advance care planning

Advance care planning refers to making decisions now about the care you would want to receive if you become unable to speak for yourself.

Advance care planning is for everyone, not just for those who are very sick. It is a way to ensure your wishes are understood and respected. Your plan can be changed at any time.

Advance care planning starts with an honest conversation with your doctor. Ask your doctor about the course your cancer will take, called a prognosis. Find out what you might expect if your cancer spreads. Discuss the medicines or therapies that will give you the best quality of life.

Include family and friends in your advance care planning. Make your wishes clear. It is important that everyone understands what you want.

Key points

- Mast cell leukemia (MCL) is a very rare and aggressive type of systemic mastocytosis.
- Mast cell leukemia (MCL) is a type of leukemia that is caused by abnormal growth of cells from myeloid progenitor cells.
- There is no clear reason why MCL develops, but several gene mutations in the *KIT* gene have been linked to it.
- Symptoms of MCL include low blood pressure, rashes, and itchy skin.
- MCL is very serious and life-threatening. Treatment will focus on supportive care.

6

Making treatment decisions

45 It's your choice

45 Questions to ask your doctors

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It's important to be comfortable with the cancer treatment you choose. This choice starts with having an open and honest conversation with your doctor.

It's your choice

In shared decision-making, you and your doctors share information, discuss the options, and agree on a treatment plan. It starts with an open and honest conversation between you and your doctor.

Treatment decisions are very personal. What is important to you may not be important to someone else.

Some things that may play a role in your decision-making:

- What you want and how that might differ from what others want
- Your religious and spiritual beliefs
- Your feelings about certain treatments like surgery or chemotherapy
- Your feelings about pain or side effects such as nausea and vomiting
- Cost of treatment, travel to treatment centers, and time away from school or work
- Quality of life and length of life
- How active you are and the activities that are important to you

Think about what you want from treatment. Discuss openly the risks and benefits of specific treatments and procedures. Weigh options and share concerns with your doctor. If you take the time to build a relationship with

your doctor, it will help you feel supported when considering options and making treatment decisions.

Second opinion

It is normal to want to start treatment as soon as possible. While cancer can't be ignored, there is time to have another doctor review your test results and suggest a treatment plan. This is called getting a second opinion, and it's a normal part of cancer care. Even doctors get second opinions!

Things you can do to prepare:

- Check with your insurance company about its rules on second opinions. There may be out-of-pocket costs to see doctors who are not part of your insurance plan.
- Make plans to have copies of all your records sent to the doctor you will see for your second opinion.

Support groups

Many people diagnosed with cancer find support groups to be helpful. Support groups often include people at different stages of treatment. Some people may be newly diagnosed, while others may be finished with treatment. If your hospital or community doesn't have support groups for people with cancer, check out the websites listed in this book.

Questions to ask your doctors

Possible questions to ask your doctors are listed on the following pages. Feel free to use these questions or come up with your own. Be clear about your goals for treatment and find out what to expect from treatment.

Questions to ask about diagnosis and testing

1. What type of mastocytosis do I have?
2. Is there a cancer center or hospital nearby that specializes in mastocytosis?
3. What tests are needed? What other tests do you recommend?
4. What will you do to make me comfortable during testing?
5. How do I prepare for testing?
6. How and where will the test be done?
7. How soon will I know the results and who will explain them to me?
8. Would you give me a copy of the pathology report and other test results?
9. Will you explain my pathology report to me?
10. Who will talk with me about the next steps? When?
11. Should my mast cells be tested for the *c-KIT* mutation?

Questions to ask about treatment options

1. How can I manage the symptoms of mastocytosis?
2. Will my age, health, and other factors affect my options?
3. Is there a better treatment option based on my age and other risk factors?
4. What is the goal of each treatment?
5. Is there an option that is less expensive?
6. Can I stop treatment at any time?
7. What will happen if I stop treatment?
8. What support services are available to me?
9. Could the treatment affect my fertility? If so, should I speak to a fertility specialist before treatment?
10. How often will I need follow-up visits after I finish treatment?
11. Who should I call with questions or concerns?

Questions to ask about clinical trials

1. What clinical trials are available for me?
2. Has the treatment been used before?
3. What are the risks and benefits of this treatment?
4. What side effects should I expect? How will the side effects be controlled?
5. How long will I be in the clinical trial?
6. Will I be able to get other treatment if this doesn't work?
7. How will I know if the treatment is working?
8. Will the clinical trial cost me anything? If so, how much?
9. What type of long-term follow-up care will I have?

Questions to ask about follow-up care

1. What is the chance that the mastocytosis will come back? What are potential signs and symptoms?
2. What long-term side effects or late effects are possible based on the treatment I received?
3. What follow-up tests will I need, and how often will I need them?
4. How do I get a treatment summary and follow-up care plan to keep in my personal records?
5. Who will be leading my follow-up care?
6. What support services are available to me after treatment? To my family?

Questions to ask your doctors about their experience

1. Who will be part of my health care team, and what does each member do?
2. Who will be leading my overall treatment?
3. What is your experience in treating people with mastocytosis?
4. Who else will be on my treatment team?
5. What other diagnostic tests or procedures will I need?
6. I would like to get a second opinion. Is there someone you recommend?
7. How many patients like me (of the same age, gender, race) have you treated?
8. Will you be consulting with experts to discuss my care? Whom will you consult?
9. How many procedures like the one you're suggesting have you done?
10. Is this treatment a major part of your practice?
11. How many of your patients have had complications? What were the complications?
12. Who will manage my day-to-day care?

Resources

Blood & Marrow Transplant Information Network

bmtinfonet.org

Chemocare

chemocare.com

Global Genes®

globalgenes.org

Lab Tests Online

labtestsonline.org

The Mast Cell Disease Society

tmsforacure.org

MedlinePlus

medlineplus.gov/genetics/condition/systemic-mastocytosis

National Bone Marrow Transplant Link

nbmtlink.org

National Coalition for Cancer Survivorship

Canceradvocacy.org/toolbox

National Hospice and Palliative Care Organization

nhpco.org/patients-and-caregivers

NCCN Reimbursement Virtual Resource

NCCN.org/reimbursement

National Organization for Rare Disorders (NORD)

rarediseases.org/rare-diseases/mastocytosis

OncoLink

oncolink.org

Patient Access Network Foundation

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Target Systemic Mastocytosis

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UK Mastocytosis Support Group

ukmasto.org



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Words to know

acute myeloid leukemia (AML)

A fast-growing cancer that starts in the bone marrow and causes too many young white blood cells to be made.

allogeneic stem cell transplant (alloSCT)

A cancer treatment that replaces abnormal blood stem cells with healthy donor cells.

antihistamines

Reduce or block histamines to stop allergy symptoms. Can control itchy skin, watery eyes, as well as other allergic symptoms of mastocytosis.

anemia

A condition where the number of red blood cells are low.

best supportive care

Treatment given to prevent, control, or relieve side effects and improve comfort and quality of life.

biopsy

Removal of small amounts of tissue from your body to test for disease.

blood stem cell

A blood-forming cell from which all other types of blood cells are formed. Also called hematopoietic stem cell.

bone marrow

The sponge-like tissue in the center of most bones.

bone marrow aspiration

The removal of a small amount of liquid bone marrow to test for disease.

chemotherapy

Drugs that kill fast-growing cells, including normal cells and cancer cells.

chromosome

A threadlike structure of nucleic acids and protein found in the nucleus of most living cells, carrying genetic information in the form of genes.

clinical trial

A study of how safe and helpful tests and treatments are for people.

complete blood count (CBC)

A lab test that includes the number of blood cells.

contrast

A substance put into your body to make clearer pictures during tests that take pictures of the inside of the body.

cytogenetic testing

A test that uses a microscope to examine a cell's chromosomes.

dermatologist

A doctor who specializes in the diagnosis and treatment of skin diseases.

deoxyribonucleic acid (DNA)

A chain of chemicals in cells that contain coded instructions for making and controlling cells.

diagnosis

To identify a disease.

differential

A measure of the different types of white blood cells present in a blood sample.

fatigue

Severe tiredness that limits the ability to function.

fluorescence in situ hybridization (FISH)

A lab test that uses special dyes to look for abnormal changes in a cell's genes and chromosomes.

gene

A set of coded instructions in cells for making and controlling cells.

hematopoietic cell transplant (HCT)

A type of treatment that destroys cells in the bone marrow, then replaces them with new, healthy blood-forming cells from another person. Also called stem cell transplant (SCT) or bone marrow transplant (BMT).

human leukocyte antigen (HLA)

A cell protein by which your body knows its own cells from foreign cells.

imaging test

A test that makes pictures (images) of the insides of the body.

immune system

The body's natural defense against infection and disease.

immunotherapy

A medicine that increases the activity of your body's disease-fighting system.

karyotype

A test that uses a microscope to examine a cell's chromosomes.

liver function tests

Tests that measure chemicals made or processed by the liver.

myelodysplastic syndromes (MDS)

Myelodysplastic syndromes are a rare group of bone marrow disorders.

myeloproliferative neoplasm (MPN)

A cancer in which the bone marrow makes too many red blood cells, white blood cells, or platelets.

mutation

A mutation occurs when a DNA gene is damaged or changed in such a way as to alter the genetic message carried by that gene.

palliative care

Specialized medical care aimed at increasing quality of life and reducing pain and discomfort for people with serious, complex illness.

pathologist

A doctor who is an expert in testing cells and tissue to find disease.

platelet (PLT)

A type of blood cell that helps control bleeding. Also called thrombocyte.

red blood cell (RBC)

A type of blood cell that carries oxygen from the lungs to the rest of the body. Also called an erythrocyte.

side effect

An unhealthy or unpleasant physical or emotional response to treatment.

stem cell transplant (SCT)

A type of treatment that replaces abnormal blood stem cells with healthy cells. Also called hematopoietic cell transplant (HCT) or bone marrow transplant (BMT).

supportive care

Health care that includes symptom relief but not cancer treatment. Also called palliative care or best supportive care.

systemic therapy

Treatment that works throughout the body.

targeted therapy

A drug treatment that targets and attacks specific cancer cells.

white blood cell (WBC)

A type of blood cell that helps fight infections in the body. Also called a leukocyte.

NCCN Contributors

This patient guide is based on the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Systemic Mastocytosis, Version 3.2021. It was adapted, reviewed, and published with help from the following people:

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