



The Mastocytosis Society

What is Mastocytosis or Mast Cell-related disorder?

According to Merriam-Webster Online, *mastocytosis* (pronounced mas•to•cy•to•sis), means an "excessive proliferation of mast cells in the tissues." (Retrieved 4/6/09 from <http://www.merriam-webster.com/medical/mastocytosis>.)

Mast cells are something that are produced naturally in every body, necessary to assist the body in fighting possible foreign threats to the system. Individuals with mastocytosis have an abundance of mast cells and the mast cells do not work properly in reaction to a [trigger](#), sometimes unknown.

In the early to mid-20th century, all forms of mast cell disease were undifferentiated and were grouped under the name *mastocytosis*. Mastocytosis specifically means "an abnormal increase in the number of mast cells," but we now know that definition, taken from the root words, relates to some very specific mast cell disorders, and may not apply to others. Some of the research done in the latter part of the 20th century laid the groundwork for much of the work done today. Over the last 30 years, there has been an explosion of interest in, and research into, the various mast cell diseases, resulting in many different categories being defined, and the definitions are still evolving.

So, for the purposes of this section, we will refer to the general term mast cell diseases which encompasses the following very general subcategories*:

Cutaneous Mastocytosis, referring to the skin, including

Urticaria Pigmentosa, referred to as **UP**, relating to hives and skin lesions

Telangiectasia Macularis Eruptiva Perstans, referred to as **TMEP**, relating to a rare form of the skin disease, most often occurring in adults, and consisting of generally smaller lesions than are typically seen in UP

Systemic Mastocytosis, involving more than one (1) organ (skin, gastro-intestinal, liver, etc.), with or without cutaneous manifestations outlined above, including

Indolent Mastocytosis, relating to slowly developing

Aggressive Mastocytosis, as it suggests, more aggressively developing Mastocytosis with associated hematologic disorder
Mast Cell Leukemia

Mast Cell Activation Disorder or Syndrome, referred to as **MCAD**

Pediatric mast cell disorders typically include the following

Solitary Mastocytoma, as it suggests, a solitary or single "clump" of mast cells or lesions

Urticaria Pigmentosa (explained above)

Diffuse Cutaneous Mastocytosis, as it suggests, diffuse skin involvement of hives and lesions

It is less common for children to suffer from systemic symptoms, but there are a number of cases.

In 2000, at a meeting in Vienna, Austria, a consensus was reached about what criteria must be fulfilled for a diagnosis of Mastocytosis (see our Research article entitled [A Consensus Document](#) for more information). Many people met the new criteria. However, many patients who had been formerly diagnosed with [Systemic Mastocytosis](#) did not seem to fit into the agreed-upon criteria, possibly because their diagnostic work-up was done incorrectly, or was not conclusive, or because they were not tested for all the criteria. Over the last few decades, some researchers began differentiating between the different forms of mast cell diseases. A few began individually defining new categories, one of which is called [Mast Cell Activation Syndrome or Disorder](#) (MCAS/MCAD). Although the various forms of mast cell disease may present with some of the same symptoms, and may be treated with the same medications and avoidance of known triggers, the cause of the [symptoms](#) is what makes them separate, but related, entities. Indeed, mastocytosis and other mast cell disorders are heterogeneous, meaning they can present in many different ways. Ultimately, the cause of each different form of mast cell disease may dictate how they are [treated](#).

What are the Symptoms of Mast Cell Diseases?

What we know about Systemic Mastocytosis is that in many cases, it is a neoplastic disease, meaning that it involves new or abnormal cell growth. (Please note - this may not apply to most cases of pediatric and/or familial Mastocytosis.) In this case the cells involved are [mast cells](#), which are normally contained in body tissues. Mast cells release certain mediators, or chemicals, of which one is histamine, into the body in response to certain events. People with [Systemic Mastocytosis](#) develop an increase in the number of mast cells, or they develop abnormally shaped mast cells, which may not function properly. In addition, the mast cells fail to die off when they are supposed to, further increasing the total mast cell burden. This die off is called [apoptosis](#). Apoptosis is programmed into normal cells, but in people with mast cell disorders, the mast cells may fail to die off, resulting in an increased number of mast cells in the body. When these mast cells are [triggered](#), they can [degranulate](#), and release their contents all at once, or they can slowly leak their contents in response to a trigger. This can cause many acute and potentially serious symptoms, which include, but are not limited to, the following:

Abdominal pain	Anaphylaxis
Blood pressure changes & shock	Bone pain (mild to severe/debilitating)
Chest pain	Cognitive difficulties/brain fog
Degenerative disc disease	Diarrhea
Dizziness/vertigo/lightheadedness	Faintness
Fatigue	Flushing
Gastroesophageal reflux	Hematological abnormalities
Hives & other rashes	Inflammation of the esophagus
Intestinal cramping and bloating	Itching, with and without rashes
Irritable bowel	Liver, spleen and other organ involvement
Malabsorption	Migraine headaches
Muscle pain	Nausea
Osteoporosis /Osteopenia	Peripheral neuropathy and paresthesias
Rapid heart rate	Vomiting

People who have been told they have *Mast Cell Activation Syndrome or Disorder* (MCAS/MCAD) may have a normal, or nearly normal, number of mast cells. However, their mast cells "behave badly" - that is, they are easily triggered to release their contents, which results in many of the same symptoms that people with Mastocytosis experience. The danger of *anaphylaxis* and shock is present with MCAD/MCAS, but unlike Mastocytosis, this syndrome may not have the potential to progress to a more aggressive or malignant stage. Nevertheless, people with either Mastocytosis and MCAS/MCAD can be either very stable or extraordinarily ill on a day-to-day basis, and managing the unpredictability of the mast cell diseases and their symptoms can be quite challenging.

What are Triggers?

Triggers are stimuli that can set off a mast cell response, potentially leading to a mast cell attack. Avoidance of various triggers (things that can set off a mast cell attack) can do much to improve quality of life and reduce the need for medication, but that is often easier said than done, as the triggers can be almost anything, including, but not limited to, the following:

Alcohol	Anesthetic agents
Antibiotics	Bacteria or fungi
Certain foods	Cold (temperature)
Coloring & flavoring in foods	Coloring & flavoring in medication
Emotional upset	Environmental toxins
Exercise or exertion	Fatigue
Fever	Friction
Friction	Heat (temperature)
Infection with viruses, bacteria or fungi	Mold
MSG	Narcotics
Perfumes	Pesticides
Plasma expanders (i.e. dextran)	Preservatives
Room freshener sprays	Smells
Spices	Stress
Sunlight	

There is great variation from person to person in what is a trigger, and even within the same person. The triggers may change day-to-day - that is, heat may set off an attack on one day, but not on other days. The above list is not complete, but is meant to show the wide range of triggers that affect mast cells.

Some people with the indolent form of Mastocytosis, and/or people with Mast Cell Activation Disorder or Syndrome, have been told by some physicians that they can expect a nearly normal life expectancy, as long as they keep themselves as stable as possible by avoiding triggers and taking medications as prescribed. However, since it is not clear what is causing mast cell activation in patients with MCAD, this may not apply. Mast cell diseases are extremely unpredictable, and some people can very quickly develop acute symptoms that may require immediate medical attention. That is why it is advisable to stay within range of a medical facility, and to carry a written protocol from your mast cell disease specialist for emergency care.

How Are Mast Cell Diseases Diagnosed?

Mast cell diseases can be diagnosed by:

- Skin biopsies
- Blood tests
- Bone marrow biopsy with aspirate flow cytometry
- Bone density and bone scan
- Radiologic, CT scan
- Careful evaluation of response to treatment

For more information on how mast cell diseases are diagnosed, please review our [consensus](#) document.

How Are Mast Cell Diseases Treated?

While a few people manage to remain stable and healthy by avoiding dietary and environmental triggers, many people with mast cell disease take a medication protocol that involves some or all of the following:

- H1 blockers - antihistamines like hydroxyzine (Atarax®), diphenhydramine (Benadryl®), Doxepin®, loratadine (Claritin®), and cetirizine (Zyrtec®)
- H2 blockers - antihistamines like ranitidine (Zantac®) or famotidine (Pepcid®)
- Leukotriene inhibitors like Singulair®, Accolate®, or Zflo®
- Mast cell stabilizers like oral cromolyn sodium (Gastrocrom®), cromolyn sodium nasal solution (NasalCrom®) or Ketotifen (Apo®-Ketotifen, Zaditen®)

In addition, many people require:

- Proton pump inhibitors like omeprazole (Prilosec®), pantoprazole (Protonix®), lansoprazole (Prevacid®)
- Inhaled bronchodilators such as albuterol (Ventolin®)
- Corticosteroids

More aggressive forms of the disease may require the use of chemotherapeutic agents and/or cytoreductive therapies. Further information about the use of these agents in treating mast cell diseases can be found at cancer treatment centers.

*This is a very general overview of the most common Mast Cell Disorders, written by patients/caregivers and TMS Board Members. (Please note: We are not discussing all Mast Cell Related Disorders, such as Asthma, on this site, but rather are focusing on some of the disorders that were previously collectively called Mastocytosis.) For specific medical information, please consult your physician. We will be adding more specific information about these diseases to this web site in the near future.