Emergency Protocol

Emergency treatment for full blown anaphylaxis always requires epinephrine. Patients with mast cell disorder should carry two self-injectable epinephrine units at all times. Units must be protected from heat and light. Patients should also have access to bottled water and at least one dose of their required medication. In addition, patients should wear medical identification jewelry and carry their medical history as well as documentation of relevant information including:

- Physician names and contact information
- Medications by indication including dosing
- Allergies, intolerances, and triggers
- Emergency contact information
- Emergency Protocol information
- A copy of this brochure
- Anesthesia article at www.tmsforacure.org

Serum tryptase level, and 24-hour urine for n-methyl histamine and prostaglandins should be started within 60 minutes of acute episode.

Organization and Support

The Mastocytosis Society, Inc. (TMS) is a non-profit 501c3 nonprofit organization dedicated to supporting patients affected by Mast Cell Disorders as well as their families, caregivers, and physicians through research, education and advocacy.

We are a patient-led organization guided by an expert medical advisory board.

TMS welcomes mast cell disorder patients of all ages. Anyone affected by or interested in learning about mast cell disorders is encouraged to join.

TMS sponsors online and regional support groups as well as a yearly joint patient and physician conference which includes a walk-a-thon, and CMEs when available. There are many volunteer opportunities. All proceeds are used to support the TMS mission. To learn more visit: www.tmsforacure.org

Become a Member

As a result of large gifts from our generous donors...

MEMBERSHIP IN TMS IS NOW FREE!

No payment is required starting February 1, 2017!

However, donations to support our mission of mast cell disorder research, education, support and advocacy are always welcome!

Please help us keep our membership free by donating now!

www.tmsforacure.org/donate-to-tms/

Benefits include: quarterly newsletter, discounted rate at the annual physician and patient conference, one vote at society elections, patient care coordination and actively supporting our mission statement.

To join TMS complete and submit the form below. You may join online at the TMS website:
www.tmsforacure.org/membership/

Name:________________________________
Address:_____________________________________________________________
City:________________ State:________________
Postal Code:_____ Country:________
Email:________________________________
Daytime Phone:___________________________

For Donations, visit:
www.tmsforacure.org/donate-to-tms/
or Mail check or money order to:

The Mastocytosis Society, Inc.
P.O. Box 416
Sterling, MA 01564

Copyright © 2016 The Mastocytosis Society, Inc.
Mastocytosis is a rare disease caused by an excess of mast cells in various tissues and organs of the body. The mast cells may be abnormal in both shape and function. In mast cell activation syndromes (MCAS) the number and shape of mast cells in the tissues may appear normal, but the mast cells are easily triggered to degranulate. In both mastocytosis and mast cell activation syndromes, mast cells degranulate or release their mediator contents. These mediators, which include histamine, tryptase, heparin, prostaglandins and leukotrienes, among others, are responsible for many symptoms seen in patients with mast cell disorders.

Symptoms may include: anaphylaxis, flushing, skin lesions/rashes, ie urticaria pigmentosa (UP) and telangiectasia macularis eruptiva perstans (TMEP), chest pain, nausea, vomiting, diarrhea, abdominal pain, bloating, GERD, fainting, blood pressure changes, itching, bone pain, osteoporosis, fatigue, weakness, anxiety/depression and cognitive difficulties.

The symptoms of mast cell disorders are highly variable and unpredictable. Mast cell disorders, including the skin rashes associated with them, are NOT CONTAGIOUS. These disorders may affect patients of all ages from newborn to adult. In pediatric and adult cutaneous disease, systemic symptoms may arise due to mediator release from skin lesions. These systemic symptoms by themselves are not an indication of systemic involvement.

Triggers for patients with mast cell disorders are highly individualized. The most common triggers include: heat, cold, temperature change, exercise, fatigue, friction, perfumes and odors, insect and other stings, certain foods, medications, alcohol, pressure, anesthetics, viral/bacterial/fungal infections, and stress of all kinds (environmental, physical and emotional). Some patients may experience reactions to certain medications, including but not limited to opiates, antibiotics, and NSAIDs. Use with caution.

For further information on administering anesthesia to a patient with a mast cell disorder please refer to the Anesthesia Article at: www.tmsforacure.org.

Currently there is no known cure for mastocytosis or MCAS. Patients require a careful balance of trigger avoidance and medication.

Diagnosis

The diagnostic criteria for mastocytosis was established in 2001. The criteria for diagnosing MCAS was proposed by international consensus in 2012. There are currently 7 categories of mastocytosis designated by the World Health Organization.

1. Cutaneous Mastocytosis: (CM) Benign disease in which the mast cell infiltration is confined to the skin.  
   A. Solitary Mastocytoma  
   B. Urticaria Pigmentosa  
   C. TMEP  
   D. Diffuse Cutaneous Mastocytosis

2. Indolent Systemic Mastocytosis: (ISM) Type most commonly seen in adult patients. Involve bone marrow and possibly other organs.

3. SM with assoc. clonal hematologic non-mast cell lineage disease: (SM-AHNMD)

4. Aggressive SM: (ASM)

5. Mast cell leukemia (malignant): very rare

6. Mast Cell Sarcoma (malignant): very rare

7. Extracutaneous Mastocytosis

In order to diagnose Systemic Mastocytosis a patient must fulfill one major criterion and at least one minor criterion. Alternately, a diagnosis may be made if a patient fulfills three minor criteria.

Major criterion: multifocal compact tissue infiltration by mast cells >15 in extracutaneous organ.

Minor criteria:

1. Prominent spindle shape of mast cells in >25% of mast cells
2. Atypical immunophenotype of mast cells with expression of CD2 and/or CD25
3. Presence of C-kit mutation (D816V)
4. Persistently elevated serum tryptase > 20ng/ml

The gold standard for diagnosis is an iliac crest bone marrow biopsy and aspirate. This includes flow cytometry and immunohistochemistry testing for normal and abnormal mast cells (CD117, tryptase, CD25). Common additional tests may include periodic serum tryptase levels, 24-hour urine collections for histamine metabolites and prostaglandins (mediator levels at baseline and within 60 minutes of degranulation or anaphylaxis), CBC with differential, serum chemistries, bone scan, bone density test, and skin biopsy. Additional tests may be required.

Treatment

Treatment of mast cell disorders depends both on the specific diagnosis and the presenting symptoms. For benign disease, treatment is directed at controlling the symptoms by trigger avoidance and medication. Typical medication used for these symptoms include:

- H1 antihistamines such as hydroxyzine, diphenhydramine, loratadine, fexofenadine, or cetirizine
- H2 antihistamines such as ranitidine, cimetidine, famotidine, or doxepin (H1,H2)
- Mast cell stabilizers: ketotifen or cromolyn sodium
- Leukotriene inhibitors such as montelukast, zileuton or zafirlukast
- Aspirin therapy-under doctor supervision
- Epinephrine
- Steroids (reserved for extreme conditions when a patient cannot be stabilized any other way)

For aggressive malignant disease, more powerful treatment including cytoreductive agents (chemotherapy) is indicated. Options may include interferon alpha/beta, imatinib (Gleevec) (if C-kit D816V negative), cladribine and other drugs now in clinical trials.

References