Mastocytosis

Overview

Mastocytosis is a disorder that can occur in both children and adults. It is caused by the presence of too many mast cells in your body. You can find mast cells in skin, lymph nodes, internal organs (such as the liver and spleen), and the linings of the lung, stomach, and intestine. Mast cells play an important role in helping your immune system defend these tissues from disease. Mast cells attract other key players of the immune defense system to areas of your body where they are needed by releasing chemical “alarms” such as histamine and cytokines.

Mast cells seem to have other roles as well. Found to gather around wounds, they may play a part in wound healing. For example, the typical itching you feel around a healing scab may be caused by histamine released by mast cells. Researchers also think mast cells may have a role in the growth of blood vessels. No one with too few or no mast cells has ever been found. This fact indicates to some scientists that having too few mast cells may be incompatible with life.

The presence of too many mast cells, or mastocytosis, can occur in two forms—cutaneous and systemic. The most common cutaneous (skin) form is also called urticaria pigmentosa, which occurs when mast cells infiltrate the skin. Systemic mastocytosis is caused by mast cells accumulating in the tissues and can affect organs such as the liver, spleen, bone marrow, and small intestine.

Researchers first described urticaria pigmentosa in 1869. Systemic mastocytosis was first reported in the scientific literature in 1949. The true number of cases of either type of mastocytosis remains unknown, but mastocytosis generally is considered to be an “orphan disease.” (Orphan diseases affect approximately 200,000 or fewer people in the United States.)

Symptoms

Chemicals released by mast cells cause changes in your body’s functioning that lead to typical allergic responses such as flushing, itching, abdominal cramping, and even shock. When too many mast cells are in your body, the additional chemicals can cause

- Musculoskeletal pain
- Abdominal discomfort
- Nausea and vomiting
• Ulcers
• Diarrhea
• Skin lesions

They can also cause episodes of hypotension (very low blood pressure and faintness) or anaphylaxis (shock).

**Diagnosis**

Your doctor can diagnose cutaneous mastocytosis by the appearance of your skin and confirm it by finding an abnormally high number of mast cells on a skin biopsy. The diagnosis of systemic mastocytosis is made when an increased number of abnormal mast cells is found during an examination of your bone marrow.

Other tests that are important in evaluating a suspected case of mastocytosis include measurement of a protein (tryptase) from mast cells in your blood and a search for specific genetic mutations that health experts associate with this disease.

**Treatment**

Doctors use several medicines to treat mastocytosis symptoms, including antihistamines (to prevent the effect of mast cell histamine) and anticholinergics (to relieve intestinal cramping). A number of medicines treat specific symptoms of mastocytosis.

• Antihistamines frequently treat itching and other skin complaints.
• Certain antihistamines work specifically against ulcers; proton pump inhibitors also relieve ulcer-like symptoms.
• Epinephrine is used to treat symptom flares which occur with shock, referred to as “anaphylaxis.” Two types of antihistamines treat severe flushing and low blood pressure before symptoms appear.
• Topical steroids temporarily reduce skin lesions that are cosmetically disturbing.
• Steroids treat malabsorption, or impaired ability to take in nutrients.

In cases in which mastocytosis is malignant, cancerous, or associated with a blood disorder, steroids and/or chemotherapy may be necessary.

**Research**

National Institute of Allergy and Infectious Diseases (NIAID) scientists have studied and treated patients with mast cell diseases for more than three decades at the National Institutes of Health (NIH) Clinical Center.
Some of the most important research advances for this rare disorder include improved diagnosis of mast cell disease, identification of growth factors that are responsible for increased mast cell production, and improved treatment. For example, researchers have developed drugs that help block the division of mast cells and the action of chemicals released from mast cells.

Scientists are also focusing on identifying gene mutations associated with the disease. Several such mutations have been identified at NIH in a cell receptor for a mast cell growth factor and in key molecules within mast cells that control cell activation. Understanding such mutations helps researchers understand the causes of mastocytosis, improve diagnosis, and develop better treatment methods.

More information

National Institutes of Health
Office of Rare Diseases Research
Genetic and Rare Diseases Information Center
P.O. Box 8126
Gaithersburg, MD 20898-8126
888-205-2311 or 301-251-4925
rarediseases.info.nih.gov/gard/

National Library of Medicine
MedlinePlus
8600 Rockville Pike
Bethesda, MD 20894
888–FIND–NLM (888–346–3656) or 301–594–5983
www.medlineplus.gov

The Mastocytosis Society
P.O. Box 731
Brenham, TX 77834
909-20MASTO (909-206-2786)
www.tmsforacure.org

National Organization for Rare Disorders
55 Kenosia Avenue
P.O. Box 1968
Danbury, CT 06813–1968
800–999–6673 or 203–744–0100
www.rarediseases.org