



# The Mastocytosis Society

## Pediatric Mastocytosis Fact Sheet

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Pediatric Mastocytosis is a rare disease characterized by the presence of too many mast cells in the skin, and possibly other tissues. Mast cells are instrumental in mediating anaphylaxis, and children with mastocytosis are at risk to develop both provoked and unprovoked episodes of anaphylaxis. Pediatric mastocytosis may involve the bone marrow and the gastrointestinal tract, and symptoms in children can vary greatly from child to child. Basic treatment includes the avoidance of known triggers, having injectable epinephrine available at all times, H1 and H2 antihistamines to control itching and gastric acid hyper secretion, and a mast cell stabilizer. IV steroids may be necessary to treat progressive, severe bullae in infants. Many children may not complain of specific symptoms, may not be able to identify or localize a symptom, or may have every symptom while others may have very few. Below are a few quick points to remember when identifying a child with a potential mast cell disease:

### **Age of Onset:**

- The onset of pediatric mastocytosis usually occurs between birth and age two in over half of all cases.
- Disease is seen in both males and females, with a slight predilection for males.

### **Presentation:**

- In 90% of the cases the typical presentation involves cutaneous manifestations (skin lesions).  
These may include:

#### **Solitary or multiple Mastocytomas.**

- Lesions may be successfully surgically excised
- Serum tryptase may be slightly elevated over 20ng/ml
- There are no reports of systemic disease with solitary mastocytomas, but patients may still have systemic symptoms

#### **Urticaria Pigmentosa, (with bullae, plaques or blisters)**

- Usually seen without associated systemic disease, but patients may still have systemic symptoms
- Bullae and blistering lesions tend to fade after two years of age
- 50%-80% lesions fade by puberty
- Sun exposure decreases lesions in exposed areas of skin

#### **Diffuse Cutaneous Mastocytosis. (only occurring in infants under one year of age)**

- Skin thickened and diffusely infiltrated
- Blistering and dermatographism is seen
- Flushing is a common symptom
- Tryptase may be increased due to increased mast cell burden in the skin, and is indicative of systemic disease
- Transmembrane mutation can be found in family members of these children

### **Possible Symptoms/Occurances Rates:**

• Itching	80-90%
• Flushing	65-70%
• Bullae	50-55%
• Abdominal pain or gastrointestinal symptoms	40-45%
• Musculoskeletal lesions	18%
• Headache	12%



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### **Guidelines For Acquiring a Diagnosis:**

- Completion of a thorough patient history
- Careful skin examination and biopsy of lesions
- Acquisition of labs including CBC, peripheral smear, serum chemistry
- Testing for markers of systemic disease including CD2 and CD25, C-kit mutation D816V, serum tryptase levels.
- Exam of liver and spleen for hepatosplenomegaly
- Any other exam relevant to individual symptoms (endoscopy, colonoscopy, etc)

### **Treatment Guidelines:**

- H1 and H2 antihistamines
- Leukotriene antagonists
- Oral cromolyn sodium (Gastrocrom®)
- Systemic corticosteroids
- Injectable epinephrine
- Topical steroids
- No chemotherapy is indicated in cutaneous or indolent systemic disease in children.

### **Prognosis:**

- Benign course will be seen in eighty-percent of patients
- There is no known progression in children from indolent systemic to progressive, more aggressive disease.
- In a presentation by Dr. Melody Carter of the NIH/NIAID to TMS in 2006, she reported that all patients with systemic disease *initially presented* with systemic disease, and did not progress from cutaneous disease.
- 15-30% of pediatric mastocytosis cases persist into adulthood.
- Children with extensive bullous lesions appear to be at increased risk of shock or sudden death from anaphylaxis.

### **References:**

Information was also taken directly from presentations at TMS conferences by Dr. Melody Carter, Dr. Mariana Castells, Dr. Cem Akin, Dr. Luis Escribano, and Dr. Joseph Butterfield.

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