

Mast Cell Disorders Testing Algorithm

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INDICATIONS FOR TESTING
 Findings consistent with mast cell disorder and/or mast cell activation in 2 organ systems, eg:

- Hematologic (abnormal CBC)
- Allergic/immunologic (recurrent anaphylaxis)
- Dermatologic (skin lesions)
- Gastroenterologic (diarrhea, abdominal pain)
- Neurologic

AND
 No identifiable secondary cause of mast cell activation

Abbreviations	
CM	Cutaneous mastocytosis
HαT	Hereditary alpha-tryptasemia
REMA	Spanish Network on Mastocytosis
SM	Systemic mastocytosis

Characteristic skin lesions (urticaria pigmentosa) present (mastocytosis of the skin)

Yes

No

Child

Adult

ORDER
 Serum tryptase
 AND
 Calculate REMA score

Anaphylaxis
 AND/OR
 REMA score ≥ 2

Yes

No

Serum tryptase ≥ 8 ng/mL

Serum tryptase < 8 ng/mL

Consider alternative diagnoses

ORDER
 Tryptase genotyping (testing for HαT)^a

Determine corrected^c serum tryptase based on genotype

Elevated

Not elevated

ORDER
 KIT D816V testing
 AND
 Bone marrow biopsy and aspirate as indicated^b

Monitor and consider alternative diagnoses

ORDER
 KIT D816V testing
 AND
 Perform bone marrow biopsy and aspirate as indicated^b
 AND CONSIDER
 Tryptase genotyping (testing for HαT)^a if tryptase ≥ 8 ng/mL

Monomorphic or other lesions

ORDER
 Serum tryptase
 AND
 KIT D816V testing

Polymorphic lesions

ORDER
 Serum tryptase
 AND
 CBC with differential

Serum tryptase > 11.4 ng/mL

ORDER
 KIT D816V testing

References

- National Comprehensive Cancer Network. [NCCN Clinical Practice Guidelines in Oncology: systemic mastocytosis](#). Version 2.2024. Published Mar 2024; accessed Mar 2024.
- Akin C. [How to evaluate the patient with a suspected mast cell disorder and how/when to manage symptoms](#). *Hematology Am Soc Hematol Educ Program*. 2022;2022(1):55-63.
- Rossignol J, Arock M. [Diagnosis of mastocytosis: emerging iceberg?](#) *Blood*. 2024;144(4):350-352.
- McMurray JC, Pacheco CS, Schornack BJ, et al. [Standardized indolent systemic mastocytosis evaluations across a health care system: implications for screening accuracy](#). *Blood*. 2024;144(4):408-419.
- Lange M, Hartmann K, Carter MC, et al. [Molecular background, clinical features and management of pediatric mastocytosis: status 2021](#). *Int J Mol Sci*. 2021;22(5):2586.
- Navarro-Navarro P, Álvarez-Twose I, Pérez-Pons A, et al. [KITD816V mutation in blood for the diagnostic screening of systemic mastocytosis and mast cell activation syndromes](#). *Allergy*. 2023;78(5):1347-1359.

Refer to the ARUP Consult [Mast Cell Disorders](#) topic for diagnostic criteria and classification information

^aA positive test confirms HαT. HαT may coexist with SM.
^bBone marrow biopsy and aspirate should only be performed in children if there is high suspicion for SM. In adults, bone marrow biopsy should be performed if mastocytosis of the skin is present (confirmed by skin biopsy). In the absence of mastocytosis of the skin, bone marrow biopsy should be performed if there is anaphylaxis and/or a REMA score ≥ 2 .
^cThere are multiple options to determine corrected serum tryptase; refer to the ARUP Consult [Mast Cell Disorders](#) topic.