INDICATIONS FOR TESTING
- Symptoms consistent with mast cell activation
- Flushing, loose stools or diarrhea, anaphylaxis
- No identifiable secondary cause of mast cell activation

Characteristic skin lesions (mastocytosis in the skin)

Yes

Adult
Proceed to bone marrow aspirate and biopsy to rule out systemic mastocytosis

Child
No further testing, unless symptoms suggest systemic disease

No

ORDER
Serum tryptase AND testing for the KIT D816V mutation

Tryptase ≥15 ng/mL and KIT D816V positive
Tryptase <15 ng/mL and KIT D816V positive
Tryptase ≥15 ng/mL and KIT D816V negative
Tryptase <15 ng/mL and KIT D816V negative

Monitor tryptase
Consider alternative diagnosis besides systemic mastocytosis (eg, mast cell activation syndrome)

Patients may be negative for KIT D816V mutation and still have systemic mastocytosis; consider full KIT gene sequencing.

High-sensitivity assay for detection of KIT D816V mutation (eg, allele-specific polymerase chain reaction [PCR]) recommended.