Figure 3. Differential Diagnosis of Systemic Mastocytosis

Suspect Systemic Mastocytosis (SM)  
(arthralgia, bone pain, flushing, gastrointestinal distress, headache, myalgia, pruritus, and/or urticaria)

Bone Marrow Morphology

Mast cell aggregates (≥15 per aggregate)  
>25% of mast cells are immature or atypical

SM diagnosed; consider KIT mutation analysis to select therapy

Mast cell aggregates (≥15 per aggregate)  
<25% of mast cells are immature or atypical

Positive for major diagnostic criterion

Mast cell aggregates (<15 per aggregate)  
<25% of mast cells are immature or atypical

Negative for major and minor diagnostic criteria

Mast cell aggregates (<15 per aggregate)  
>25% of mast cells are immature or atypical

Positive for minor diagnostic criterion

KIT Mutation Analysis
Immunophenotyping
Serum Tryptase

Major diagnostic criterion:  
Mast cell aggregates ≥15 per aggregate

Minor diagnostic criteria:
>25% of mast cells are immature or atypical  
KIT D816V detected  
Positive for CD2 and/or CD25  
Serum tryptase >20 ng/mL

SM diagnosed if 1 major and 1 minor or 3 minor criteria are met

This algorithm is intended as a guide for using Quest Diagnostics laboratory tests to diagnose systemic mastocytosis. It is based on reference 7. The algorithm is provided for informational purposes only and is not intended as medical advice. A physician’s test selection and interpretation, diagnosis, and patient management decisions should be based on his/her education, clinical expertise, and assessment of the patient.