Transcript Details

This is a transcript of a continuing medical education (CME) activity. Additional media formats for the activity and full activity details (including sponsor and supporter, disclosures, and instructions for claiming credit) are available by visiting:
https://reachmd.com/programs/cme/differential-diagnosis-of-indolent-systemic-mastocytosis/32713/

Released: 02/28/2025
Valid until: 02/28/2026
Time needed to complete: 38m

ReachMD
www.reachmd.com
info@reachmd.com
(866) 423-7849

Differential Diagnosis of Indolent Systemic Mastocytosis

Announcer:
Welcome to CME on ReachMD. This episode is part of our MinuteCE curriculum. Prior to beginning the activity, please be sure to review the faculty and commercial support disclosure statements as well as the learning objectives.

Dr. Boggs:
This is CME on ReachMD. I'm Dr. Nathan Boggs. Joining me today is Dr. Tracy George.

Let's start our discussion with a case.

A 37-year-old female state park ranger and mother of 2 presented to an allergist for evaluation of several months of diffuse itching. She relayed to the allergist that for 8 months she had noticed intense pruritus when her uniform rubs against her flanks and upper legs, including during trail clearing at work and getting dressed in the morning. She noted that the trigger that brought her in now, as opposed to earlier, was that she more recently started waking up multiple times at night when the same areas of her skin brush against the covers.

She noted additional problems of flushing and raised welts when taking warm showers, necessitating lukewarm or cold water. She has taken diphenhydramine to treat the pruritus but has limited its use to bedtime due to side effects. She noted to her physician that her spouse and children have commented that she's more irritable.

Review of systems revealed that she missed several days of work due to new fatigue and increased frequency of migraine headaches. The allergist performed a physical exam that demonstrated too-numerous-to-count reddish-brown 0.5- to 1-cm macules on sun-protected regions, including flanks, chest, mid and lower back, and upper legs. The spleen and liver were not palpable. Gentle irritation of one macule resulted in a wheal-and-flare reaction.

Dr. George, what testing would you recommend for this patient?

Dr. George:
I think the first thing that we need to do is to biopsy the skin lesions and to rule out or rule in cutaneous mastocytosis. So when you send the skin lesion, make sure you let your pathologist know you're trying to rule out mastocytosis so they can do the appropriate stains.

So then, thinking along those lines, getting a basal serum tryptase level will be important.

Also, in terms of systemic mastocytosis, I think just getting the baseline CBC and a metabolic panel. I know you said the liver isn't enlarged on physical exam, but maybe doing LFTs and then getting an alkaline phosphatase as well, just thinking about mastocytosis. And then finally, I would do a peripheral blood KIT for D816V, and I would use a highly sensitive assay when you're sending that.

What additional testing would you do in terms of allergy immunology?

Dr. Boggs:
Many times we might check a total IGE as part of the evaluation for SM. You mentioned several of the tests that we might order in the clinic. And then, for this patient, I probably would go straight to hematopathology for this patient just because of the positive Darier's sign

on the skin exam.

What bone marrow testing would you recommend at this point?

Dr. George:
There's a number of different tests that should be performed on the bone marrow. The first is a flow cytometry for mast cells, and this should be performed on the bone marrow aspirate. I would also do the highly sensitive KIT testing on the bone marrow as well, and usually that's done on the bone marrow aspirate as well. You'll also want to do chromosome analysis. It's a good opportunity to do that, and so I would do that on the bone marrow aspirate.

A KIT immunohistochemical stain or CD117 nicely will highlight mast cells. It's super bright in the mast cells. It might be a little dim in erythroid or myeloid precursors. And then the most specific stain is a mast cell tryptase stain for mast cells, so you're going to want to do both. Then, you're going to look for aberrant expression of CD25 on mast cells, as well as CD30. And those are also done via immunohistochemical stains. You could do CD2, honestly, but I prefer that being done by flow cytometry. I find it's harder to interpret on the bone marrow biopsy.

And finally, I would do a CD34 stain, because mast cells should be 34 negative and it also is nice so you can look for increased blasts. You'll want to do reticulin and trichrome stains. Those are special stains that you can do to look for increased fibrosis.

If this patient meets criteria for indolent systemic mastocytosis, my next question is what treatment options would you consider for this patient?

Dr. Boggs:
I think, historically, the treatment options have revolved around symptom-directed medication, so if itching and urtication of the macules is one of their biggest problems, then I think antihistamines historically have been what allergists have used to treat the symptoms.

Other treatment options might be omalizumab, which there's a long history of omalizumab use in chronic spontaneous urticaria that's extrapolated to be beneficial for patients with SM. And then there's additional medications, including cromolyn antihistamines targeting H2 as opposed to H1. In this patient's case, headache and fatigue were some of the symptoms. In cases where there's multiple different symptoms, chronic daily symptoms, tyrosine kinase inhibitors are definitely now becoming more of the standard of care. So I think probably for this person, that's where I would go.

Dr. George:
That sounds like a reasonable approach.

Dr. Boggs:
Yeah. And it's an evolving landscape for sure,

Unfortunately, our time is up. Thank you for watching.

Announcer:
You have been listening to CME on ReachMD. This activity is provided by Total CME, LLC and is part of our MinuteCE curriculum. To receive your free CME credit, or to download this activity, go to ReachMD.com/CME. Thank you for listening.