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/the-many-clinical-presentations-of-non-advanced-systemic-mastocytosis/14542/

Released: 12/21/2022
Valid until: 12/21/2023
Time needed to complete: 1h 22m

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

The Many Clinical Presentations of Non-Advanced Systemic Mastocytosis

Announcer:
Welcome to CME on ReachMD. This episode is part of our MinuteCME 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. Castells:
Hello everybody. I'm Dr. Mariana Castells. I am the director of the Mastocytosis Center at the Brigham and Women's Hospital, and today I will be talking to you about the clinical presentations of indolent systemic mastocytosis in smoldering systemic mastocytosis. I will do that by illustrating clinical vignettes. Mastocytosis is a rare disease and the true incidence and prevalence is not clearly known.

We have seen that to be more frequent in Caucasian populations, but the true evaluation of mastocytosis in other races and populations has not been undertaken yet. So one in a hundred thousand cases is potentially the incidence of the disease and we are looking into future studies that will allow us to see its prevalence and incidence in other races.

The first vignette is an acute presentation in a young woman, 32-year-old female who presents with her daughter to a mall and losses consciousness suddenly and is found in a pool of blood after feeling hot and dizzy and having a fluttering heart. She tells the ER doctors once she arrives there that her medical problems have been panic attacks, flushing, and a rash, and that she has had the rash for over 10 years and was told that the rash was freckles where in fact the rash was a typical rash of mastocytosis. The second clinical presentation would be also acute presentation for an adult who presents with a drug reaction.

So this is a 33 year old male who's a runner runs the Boston Marathon. He at the end uses 800 milligrams of ibuprofen presents with sudden onset of flushing. He becomes hypotensive. He loses consciousness, he was found to be very short of breath before being intubated and whose tryptase is found to be over 2000 nanograms per milliliter when he's found in the intensive care unit And his symptoms when further questioning are that the patient has had flushing has had chronic fatigue, depression, anxiety, bone pain, chest pain where he has had multiple rule outs for cardiac disease and he has had few lesions in his chest that nobody has paid much attention to that where upon bone marrow biopsy he's found to have mast cell aggregates. He is positive for KIT mutation and his baseline triptase is 32 and in fact the patient has systemic mastocytosis.

Third presentation here for cutaneous mastocytosis is an acute pediatric presentation. This is a four month old boy who presents with fever, viral disease, skin blistering, and then he's seen in the hospital given vancomycin and presents a seizure develops acute respiratory distress syndrome and cardiac arrest. His triptase is elevated as you can see here over hundreds and a biopsy of his skin shows an increase in triptase positive mast cells that indicates that the patient has diffuse cutaneous mastocytosis and a novel KIT mutation is found in this child who survives and with treatment is able to get out of the intensive care unit and recover from the episode.

Here we have another presentation of systemic mastocytosis as hymenoptera anaphylaxis and in this particular patient a 47-year-old man presents with loss of consciousness five minutes after being stung by a yellow jacket in his backyard. He's evaluated, resuscitated, and his allergy evaluation indicates that he's truly allergic with IgE antibodies to yellow jacket and honeybee, he is placed on hymenoptera immunotherapy, and he is found to have more of those events with triptase level during those episodes being 29 and he

has no skin lesions and his baseline level is 25. Again, the patient is evaluated for mastocytosis and find to have aggregates in the bone marrow and hymenoptera induced anaphylaxis is one of the presentations of systemic mastocytosis.

Finally, a 44-year-old woman with allergies who actually presents an episode with burning heart chest pressure, lightheadedness, is given epinephrine and receives multiple of epinephrine. Has a triptase that is not elevated but presents peripheral blood mutation KIT D816V positive presents what is known as monoclonal muscle syndrome where there is no typical aggregates but abnormal mast cells. Finally, a 30-year-old male with osteoporosis and unprovoked fractures of hips with bilateral prosthesis is found to have intermittent episodes of diarrhea, inability to concentrate, mental fogginess, flushing, and again unprovoked anaphylaxis with elevated tryptase and his hip biopsy shows that he has aggregates of mast cells that are CD25 positive.

So, the presentation of mastocytosis mainly stems for a cutaneous rash that can be monomorphic in adults and polymorphic in children and diffusing some children and can present with cutaneous mastocytoma and aggregate of mast cells in the skin that present upon scratching a very positive Darier's sign and that is highly specific and for cutaneous mastocytosis and in adults it's associated with systemic mastocytosis. So the presentation of mastocytosis can have symptoms that go from five to 55 years. There is no increase in allergies or asthma and there is very few familial association with that. Cutaneous are symptoms are as you have seen prominent abdominal and gastrointestinal symptoms, psychological symptoms, and adverse reactions to medications including non-steroidal anti-inflammatory medications, opioids, and sometimes abnormal blood counts.

So the patients are referred from different specialists, dermatologists internist, gastroenterologist, allergist, neurologists, endocrinologists, and even hematologists and they have different diagnosis that can mimic mastocytosis, that is why the duration of the symptoms can be so long. And if you see here that for adults on systemic mastocytosis is the most common presentation. In children, is cutaneous mastocytosis and is a temporary phenomenon.

So we have classified the muscle disorders as primary with the clonal muscle activation disorders secondary and idiopathic, and when we see a patient we have to make sure that when the patient has mast cell activation symptoms we have to make sure that we know how to categorize based on the history, based on the presentation, the symptoms, KIT mutation, and tryptase levels. And we have here the different categories. So essentially at the end of the day what is important for patients who come to see us mastocytosis is associated with cutaneous, with gastrointestinal, the systemic symptoms including anaphylaxis.

So essentially the presentation of mastocytosis requires that we look for multi-organ involvement and particularly the organs that we have to look the most commonly are the cutaneous like the skin, the gastrointestinal, neuropsychiatric symptoms and also the presence of anaphylaxis. Thank you so much for your participation today. We hope that this educational program will help you raise awareness of a rare disease such as mastocytosis that you will be able to help your patients with an early diagnosis and potentially a better treatment and management outcome.

Announcer:
You have been listening to CME on ReachMD. This activity is jointly provided by Global Learning Collaborative (GLC) and TotalCME, Inc. and is part of our MinuteCME curriculum.

To receive your free CME credit, or to download this activity, go to ReachMD.com/CME. Thank you for listening.