SOHO ANNUAL MEETING 2024

Systemic Mastocytosis in a 70 year old Filipino, Female With Cutaneous, Bone Marrow and Gastrointestinal Manifestations: A Case Report

R. Recio-Aquino[1]
1. The Medical City, Manila, Philippines

THE MEDICAL CITY
Where Patients are Partners

OBJECTIVE

This report highlights the importance of a high index of clinical suspicion in patients presenting with atypical skin lesions and vague systemic symptoms. Systemic mastocytosis (SM) may be indolent at times, appropriate work up and prompt treatment is important because transformation to an aggressive form imposes increased risk of morbidity and mortality.

CASE

A 70-year-old Filipino woman with no history of allergy presented with an erythematous-to-hyperpigmented, flat, pruritic rash on her extremities extending to the trunk and face. She developed significant weight loss, recurrent epigastric pain, and hematochezia. A work up showed anemia and thrombocytopenia with elevated eosinophil count. Colonic nodule and bone marrow biopsies showed eosinophilia and mast cell infiltration positive for CD117 and CD25 and negative for PDGFR. She was given symptomatic relief and started on Imatinib for advanced SM with skin, gastrointestinal, and bone marrow involvement. Stem cell transplantation will be considered depending on treatment response. Regular follow up and monitoring is being done in the outpatient setting.

IMAGES

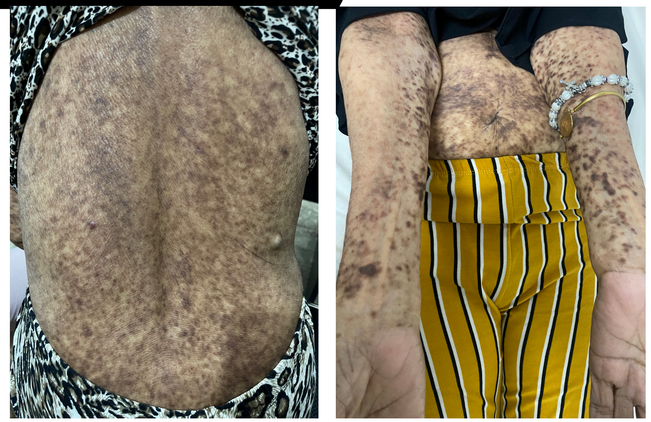


Figure 1. Skin lesions. Generalized Multiple flat, well defined, hyperpigmentd coalescing macules

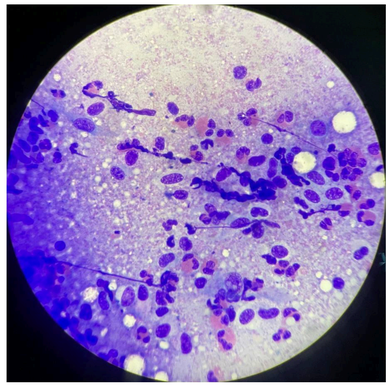


Figure 2. Bone Marrow aspirate smears. Hypercellular marrow with megakaryocytic pleomorphism, erythroid and granulopoietic dysplasia with predominance of eosinophilic cells with irregularly shaped nuclear outlines, dense chromatin and granular cytoplasm

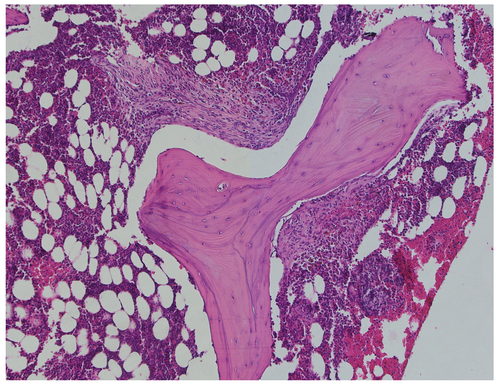


Figure 3. Bone Marrow core bone biopsy. Considering reactive marrow with some focal areas of spindle shaped cells (probably mast cells), admixed with eosinophils and small lymphocytes suggesting Mastocytosis.

DISCUSSION

Systemic mastocytosis is a rare neoplasm from KIT-driven expansion and accumulation of neoplastic mast cells in various tissues. Mast cell activation plays a significant role in disease progression mostly in cutaneous disease. Other subtypes that present as hematologic neoplasms with organ damage denote advanced or aggressive disease with increased risk of morbidity and mortality. Presently, there is still insufficient data on the incidence and prevalence of cutaneous mastocytosis and SM or a clear gender predominance. It is considered a nonhereditary, somatic disorder and regarded as a persistent disease without spontaneous remission.

Clinical symptoms result from the release of proinflammatory and vasoactive mediators from mast cells. Symptoms may be indolent. Patients may or may not have skin manifestations with nonspecific constitutional symptoms. Some with more aggressive disease may exhibit vague gastrointestinal symptoms, hematologic findings, unexplained lymphadenopathy or splenomegaly, and osteoporosis. Delay in diagnosis impairs quality of life and may lead to life-threatening consequences.

ACKNOWLEDGEMENTS

Special appreciation to The Medical CIty, Section of Hematology mentors and co-fellows.

CONTACT INFORMATION

rocenrecio@gmail.com
(+63)9176325016