A Case of Systemic Mastocytosis Diagnosed Endoscopically

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Abstract

Mastocytosis, or mast cell proliferation is very rare. 60% of patients with systemic mastocytosis (SM) will have gastrointestinal involvement, with the colon being the most prevalent site affected in these cases. However, most patients are diagnosed through bone marrow biopsy. Indolent SM, which is characterized by both gastrointestinal and cutaneous symptoms in the absence of bone marrow suppression is extremely rare and often missed due to the complexity of the diagnosis. We present a patient with abdominal pain, flushing and nausea who was diagnosed endoscopically with systemic mastocytosis, likely indolent type.

Case Presentations

A 39 YO F with no significant PMH presented to the GI clinic with dull and non-radiating RLQ pain associated with nausea and flushing of her face, lips, and ears. The patient appeared well-nourished and her examination, including perianal and digital rectal exams were unremarkable. She had a normocytic anemia (Hbg 11.7 g/dL, MCV 90.6 fl), but otherwise complements, CRP and ESR were normal. A CT abdomen/pelvis demonstrated a soft tissue nodule near the ileocecal region measuring 1.5x1.2 cm, concerning for a carcinoid tumor. Colonoscopy revealed numerous 1-5 mm yellow-white mucosal nodules with a central hyperpigmentation visualized in the entire colon. There were discontinuous areas of non-bleeding and ulcerated mucosa in the transverse colon, ascending colon and cecum. Cold forceps biopsies from the ileal and colonic mucosa revealed sheets of eosinophils mixed with clusters of mast cells, showing atypical morphology, including oval to short spindled nuclei and focal clustering. IHC stains were positive for CD117, CD25, and Tryptase. OnkoSight KIT Sequencing detected Tier 1 genomic alterations in KIT p.Asp816Val, strongly supporting the diagnosis of systemic mastocytosis. She was started on Loratadine and Famotidine.

Discussion

Only 10% of all mastocytosis is considered systemic, with the most common site of involvement being the bone marrow. However, of those patients with SM approximately 60% will also have gastrointestinal symptoms. A case series observing the clinicopathologic features in 5 patients determined the colon to be the most prevalent site of involvement. 4 out of 5 patients had cutaneous symptoms, gastrointestinal symptoms and bone marrow involvement, however up to 50% of patients with SM often lack cutaneous symptoms at time of diagnosis. Endoscopically, patients were found to have mucosal nodularity, loss of normal architecture and friability with pathologically positive dense mast cell infiltration of the lamina propria. This is similar to our patient who had colonic involvement characterized by mucosal nodularity, friability and ulceration, however our patient likely had indolent SM given the absence of bone marrow suppression. Our patient was also unique in the way in which she was diagnosed, endoscopically.

Conclusions

Mastocytosis is a rare disease characterized by heterogeneous clinicopathological features and variable treatment and prognosis. In the absence of classical cutaneous lesions, bone marrow suppression and/or serum tryptase elevations, the diagnosis of indolent systemic mastocytosis can be easily missed. As many patients lack cutaneous symptoms at time of diagnosis, the clinical suspicion should remain high if other more common diseases can be excluded. This is a rare case of indolent systemic mastocytosis diagnosed endoscopically.

References