



Case of the Quarter: Q1 2015

ANSWER

I. Immunohistochemistry (200x)

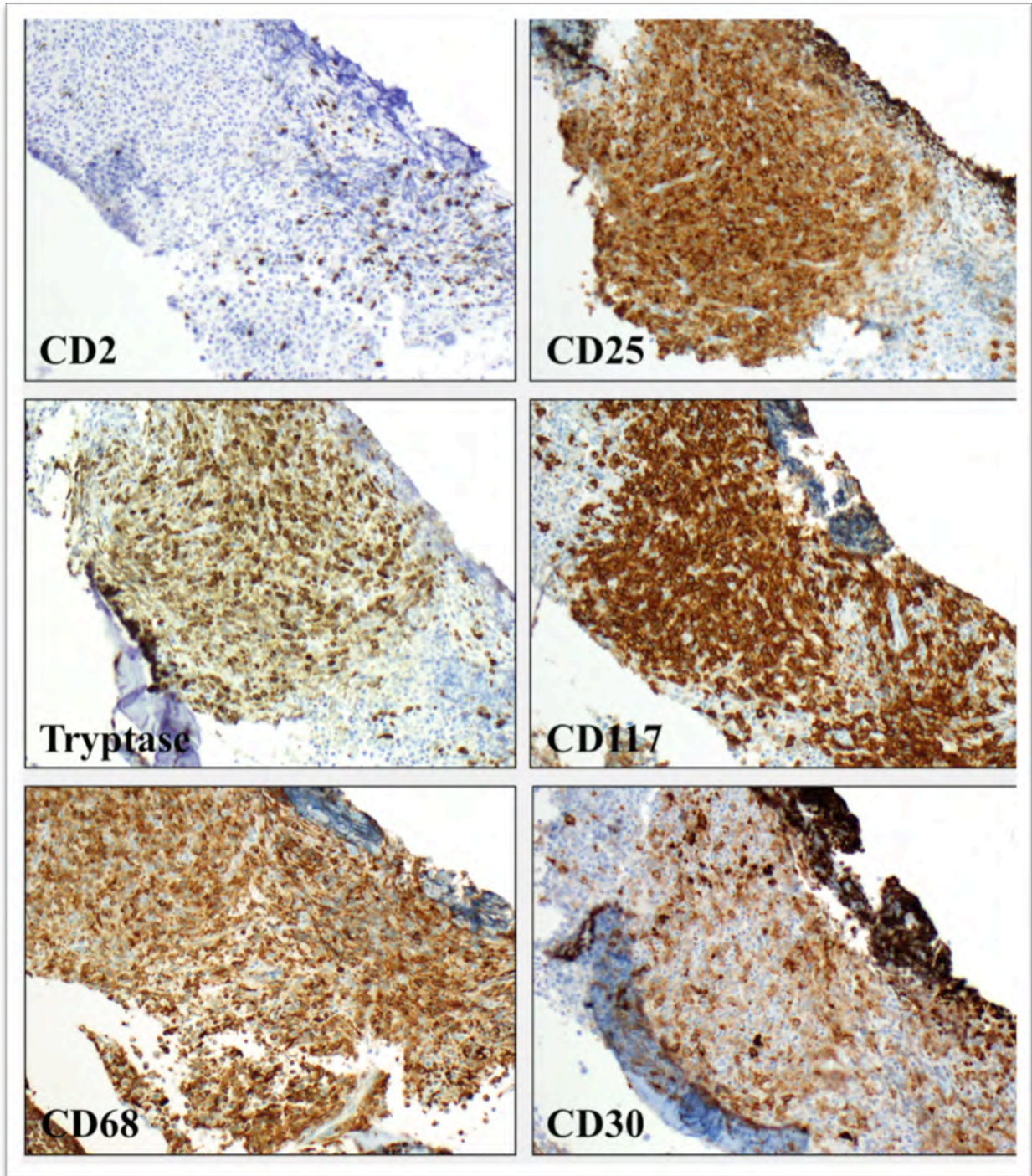


Figure 6



II. Findings

1. The aspirate differential count revealed 5% mast cells.
2. The differential count demonstrated no increase in the blast population to suggest transformation to acute myeloid leukemia. This was confirmed by flow cytometry which shows approximately 1% CD34+ blasts.
3. The marrow shows erythroid precursors with megaloblastic changes. Additionally, there are dysplastic unilobated megakaryocytes present (Figure 4). These findings are consistent with the patient's history of myelodysplastic syndrome.

III. Diagnosis

Systemic mastocytosis with associated clonal hematological non-mast-cell lineage disease (SN-AHNMD)

IV. Discussion

Systemic mastocytosis has an incidence of approximately 5-10 new cases per one million population per year.

Diagnostic criteria:

A diagnosis of systemic mastocytosis can be made when the major criteria plus one minor criterion or at least three minor criteria are present.

Major criterion:

Multifocal, dense infiltrates of mast cells (≥ 15 mast cells in aggregates) in the bone marrow and/or extracutaneous organs.

Minor criteria:

1. In bone marrow biopsy, extracutaneous organ biopsy, or bone marrow aspirate, $>25\%$ of the mast cells have atypical morphology
2. Detection of KIT mutation at codon 816
3. Aberrant expression of CD2 and/or CD25
4. Serum total tryptase persistently exceeding 20 ng/mL

SN-AHNMD can be diagnosed when the criteria for both systemic mastocytosis and a clonal hematological neoplasm are met.

Interesting note:

CD30, which is a member of tumor necrosis factor receptor superfamily, is normally restricted to expression in lymphoid cells. Aberrant expression of CD30 has been observed in systemic mastocytosis, especially in more advanced disease. As a diagnostic marker, CD30 is not as sensitive as CD25. However, its utility is useful in cases of well-differentiated systemic mastocytosis. Unlike CD25, CD30 is aberrantly expressed in the majority of well-differentiated systemic mastocytosis cases.