CT-118/Systemic Mastocytosis in Armenia: A Rare Disease with Two Documented Cases

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Introduction

Systemic mastocytosis (SM) is a rare myeloid disorder characterized by the accumulation of neoplastic mast cells in various tissues. It frequently involves activating mutations in the KIT gene, particularly D816V. The estimated incidence is approximately 1.21 per 100,000 individuals. Due to its rarity and symptom overlap with more common conditions, SM is often misdiagnosed or diagnosed late.

Aim

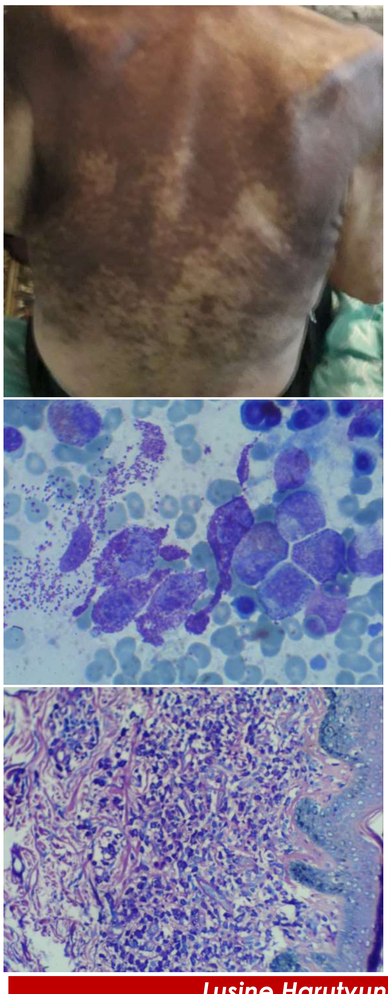
The purpose of this abstract is to present two cases of SM diagnosed in Armenia, outlining diagnostic

and treatment barriers and emphasizing the challenges in managing this rare condition in a low-resource healthcare setting.

Methods

Clinical data were extracted from patient records archived at Yeolyan Hematology and Oncology Center.
Diagnostic confirmation involved bone marrow aspiration, histopathology, immunohistochemistry, and external molecular testing for KIT D816V mutations.

Figure 1. Patient 1. Skin involvement and Histology



Results

Patient 1: A 69-year-old male presented with fatigue, weight loss, abdominal discomfort, and skin lesions. Workup showed mild anemia, hepatosplenomegaly, lymphadenopathy, and ascites. Bone marrow revealed hypercellularity and 28% atypical mast cells. Skin and lymph node biopsies confirmed mast cell infiltration with extensive immunophenotyping (CD25+, CD117+, Ki67 ~10%, EBV/CD34/TdT-negative). KIT D816V mutation (15%) was detected in Basel. He was started on midostaurin (self-funded), which stabilized symptoms for 2 months but was discontinued due to cost. Imatinib was ineffective, and he died 6 months post-diagnosis due to complications from exudative pleuritis.
Patient 2: A 74-year-old male diagnosed with systemic mastocytosis in 2022 based on hypercellular bone marrow with dense paratrabecular spindled mast cells, fibrosis, and osteosclerosis. CBC showed anemia (Hb 88 g/L) and mild splenomegaly. Further diagnostics (CD117, tryptase, KIT mutation) were planned, but the patient emigrated, taking the paraffin block and was lost to follow-up.

Summary

Diagnosing SM is challenging due to its varied presentation and the need for specialized tools. In resource-limited settings like Armenia, the absence of molecular testing (e.g., KIT D816V) and immunophenotyping further complicates diagnosis. In Patient 1, confirmation relied on international collaboration; Patient 2 emigrated in search of diagnostic and treatment access.
Treatment barriers are equally concerning. Midostaurin, the standard for advanced SM, is unaffordable for most patients, with no governmental reimbursement. As seen in Patient 1, lack of access led to treatment discontinuation and poor outcomes. Imatinib, ineffective for D816V-positive cases, was the only available option.
Systemic gaps, including the lack of national registries, reference centers, and trained specialists, limit research and care. Addressing these challenges through international partnerships, better diagnostics, and access to essential drugs is crucial to improving outcomes in underserved regions.

References

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