

Polycythemia Vera with Eosinophilia Associated with FIP1L1-PDGFR A: A Case Report

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BACKGROUND: Myeloproliferative neoplasms associated with Platelet Derived Growth Factor Alpha (PDGFRA) rearrangement typically present as chronic eosinophilic leukemia or acute leukemia. An association between polycythemia vera and these disease entities has not previously been documented. We have a case report of a 54 year old Hispanic male with clinically diagnosed polycythemia vera who upon further examination was found to have FIP1L1-PDGFR A mutation by fluorescence in situ hybridization (FISH)

MATERIALS AND METHODS: Diagnostic specimens obtained in order to assess the patient's lack of response to hydroxyurea included peripheral blood smear, complete blood count, bone marrow aspirate, and bone marrow biopsy. Wright-stained slides of the peripheral blood smear and bone marrow aspirate, hematoxylin-eosin stained slides of the bone marrow biopsy were evaluated by pathology. Chromosome analysis, FIP1L1-PDGFR A (FISH) and ETV6-PDGFR B (FISH) were also performed on the aspirate specimens.

RESULTS: Peripheral blood showed marked leukocytosis (White blood cell count of 24,800 cells/uL) with marked eosinophilia (51%), erythrocytosis (Hemoglobin of 17.6g/dL while receiving 500 mg hydroxyurea daily and therapeutic phlebotomy as needed). Bone marrow biopsy slides showed a marked increase in eosinophils (28% on manual differential), an increase in mast cells, marked reticulin fibrosis (reticulin staining), and absent iron stores. FISH testing of the marrow aspirate showed the FIP1L1-PDGFR A mutation via loss of CHIC-2. Complete chromosomal analysis and ETV6-PDGFR B tests were normal.

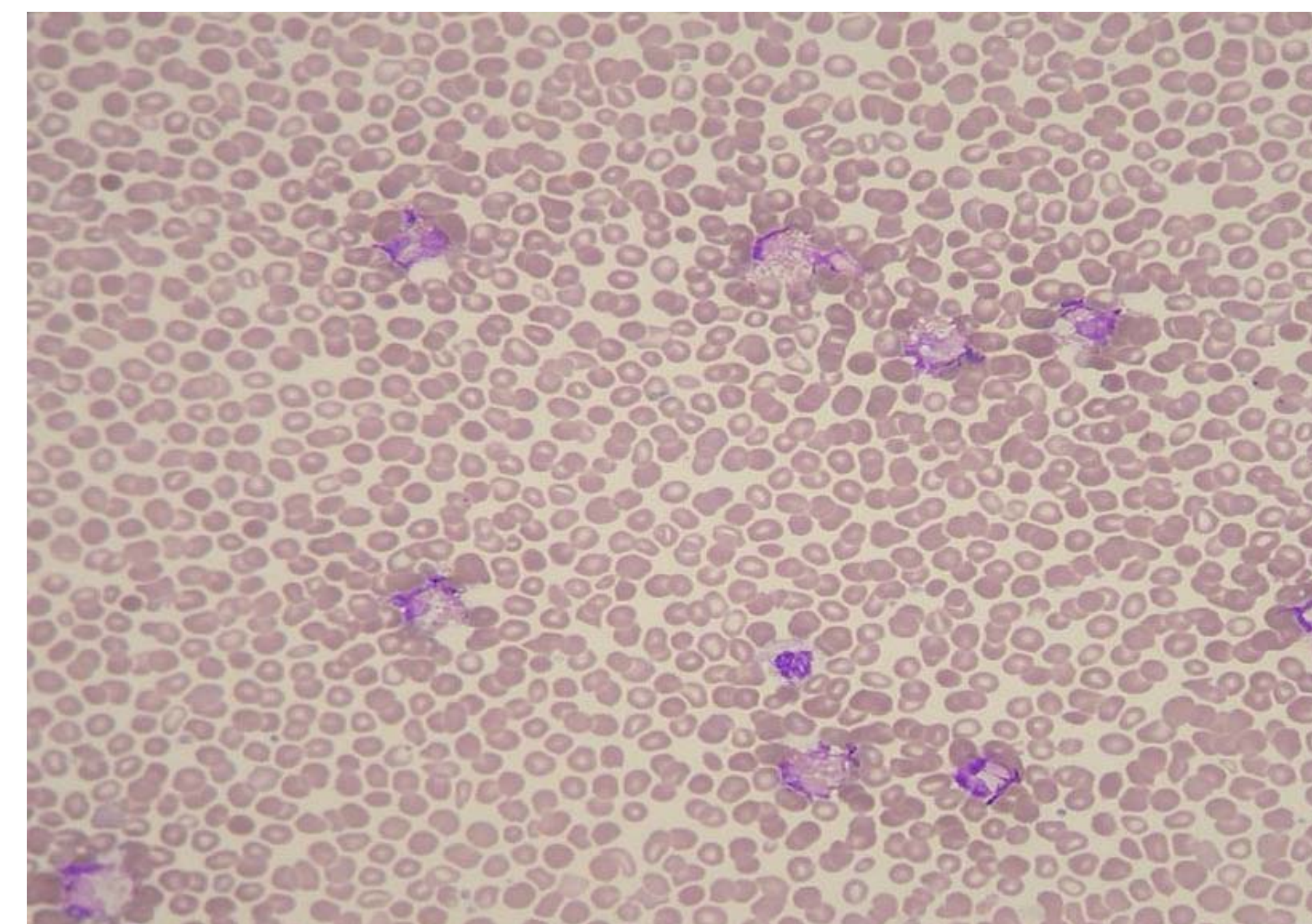


Fig 1. Peripheral Smear Wright Stain (20x)

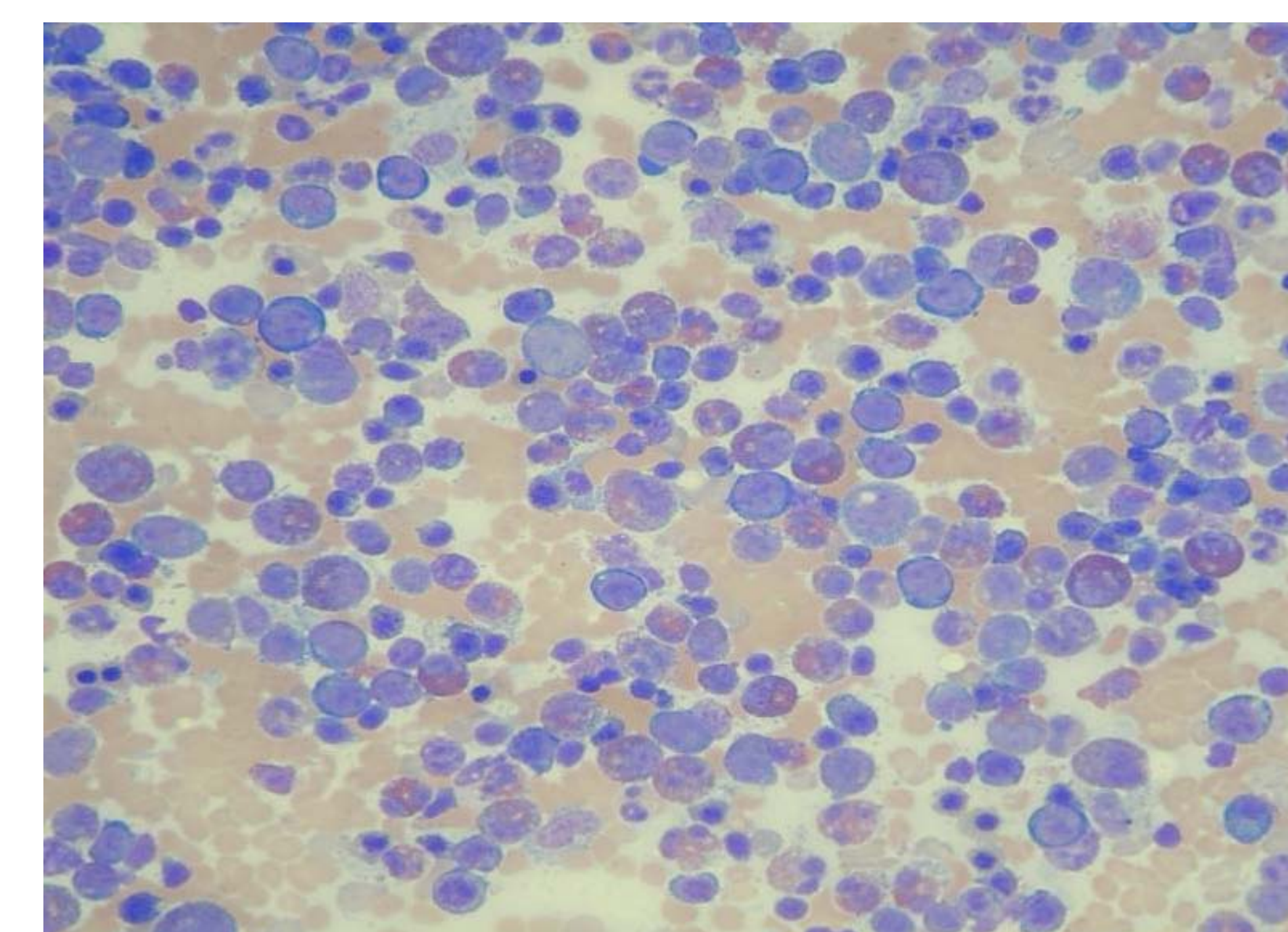


Fig 2. Marrow Aspirate Wright Stain (40X)

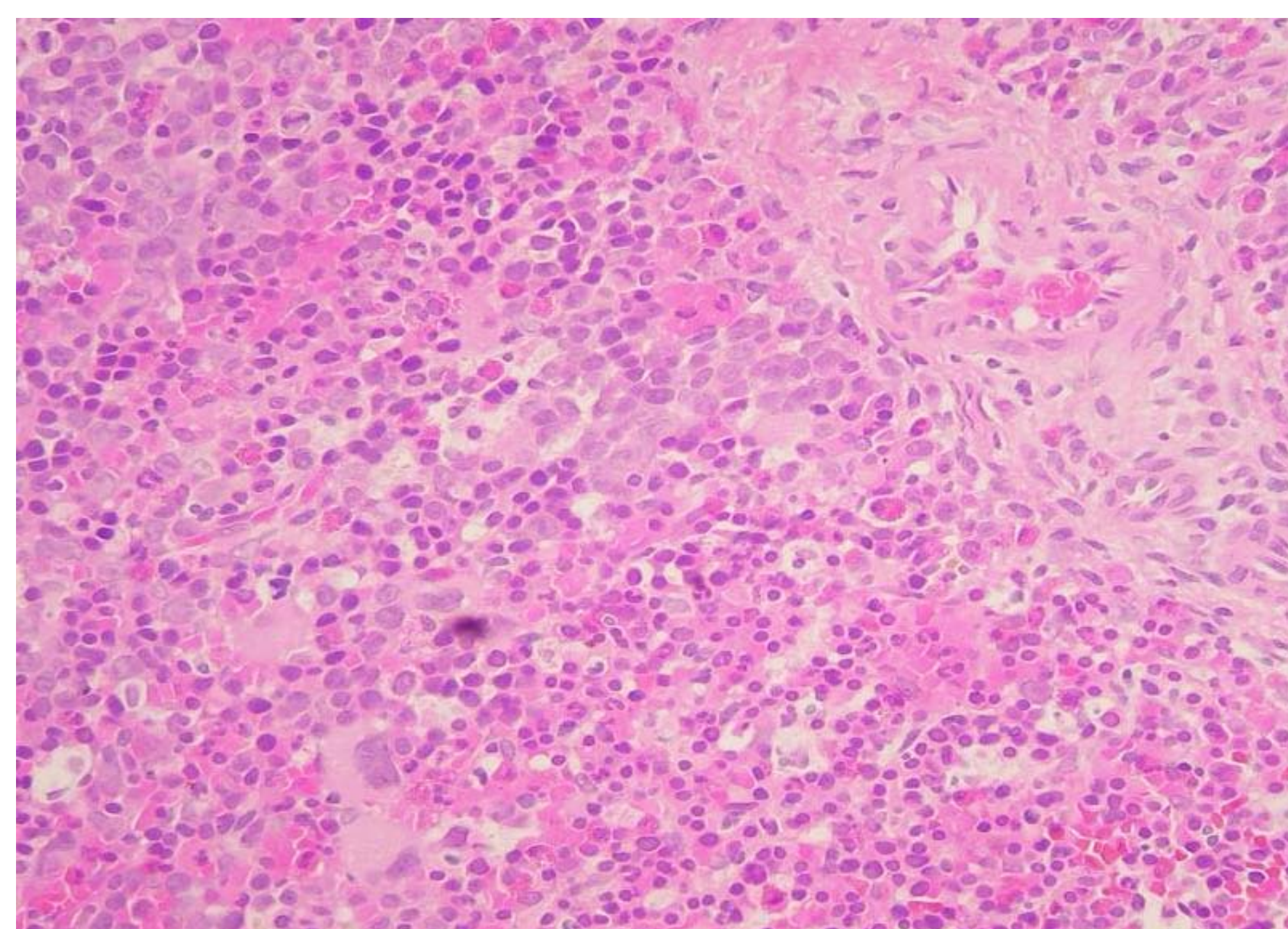


Fig 3. H and E Marrow Biopsy (40X)

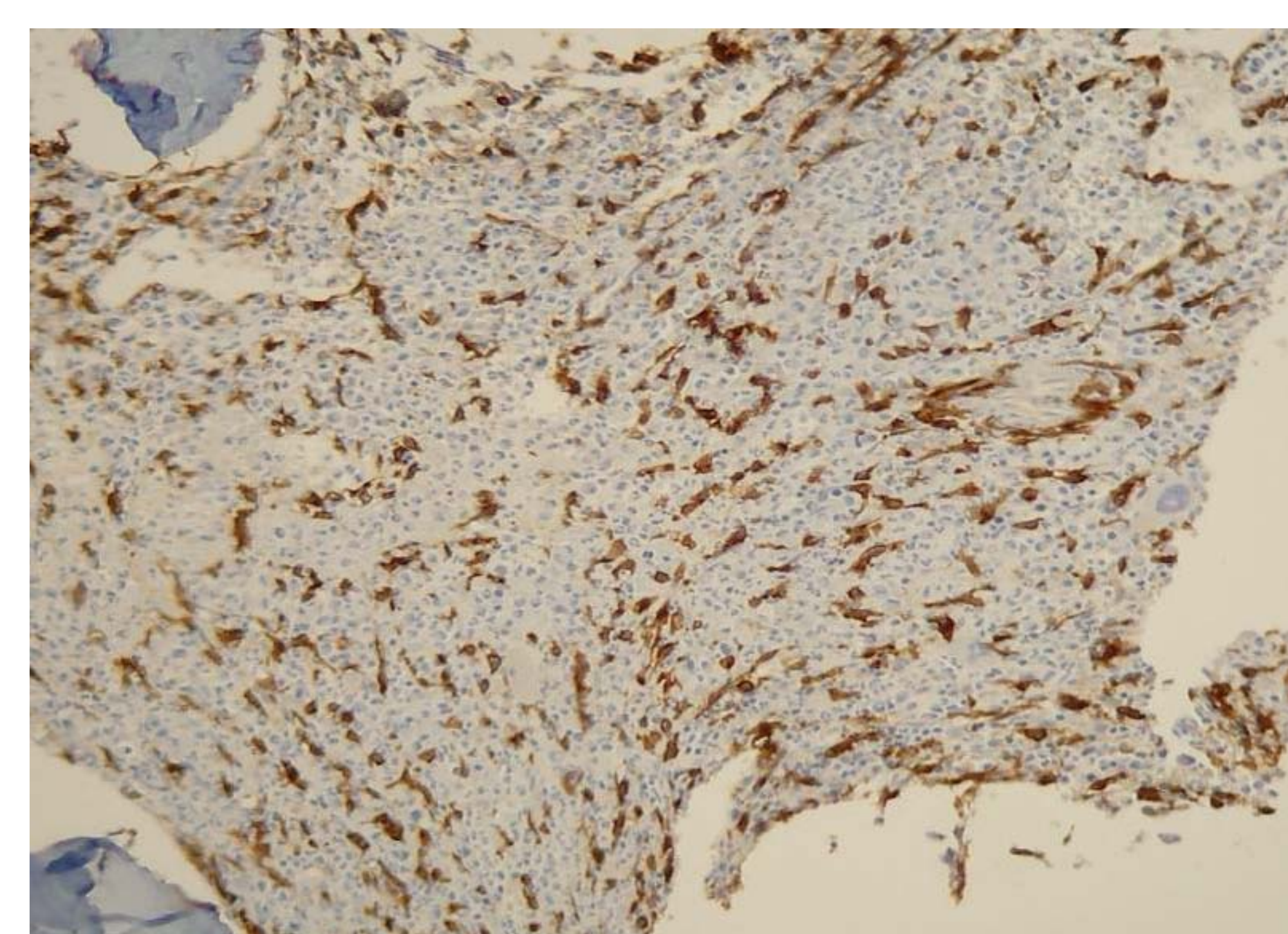


Fig 4. Tryptase Immunohistochemical Stain (20X)

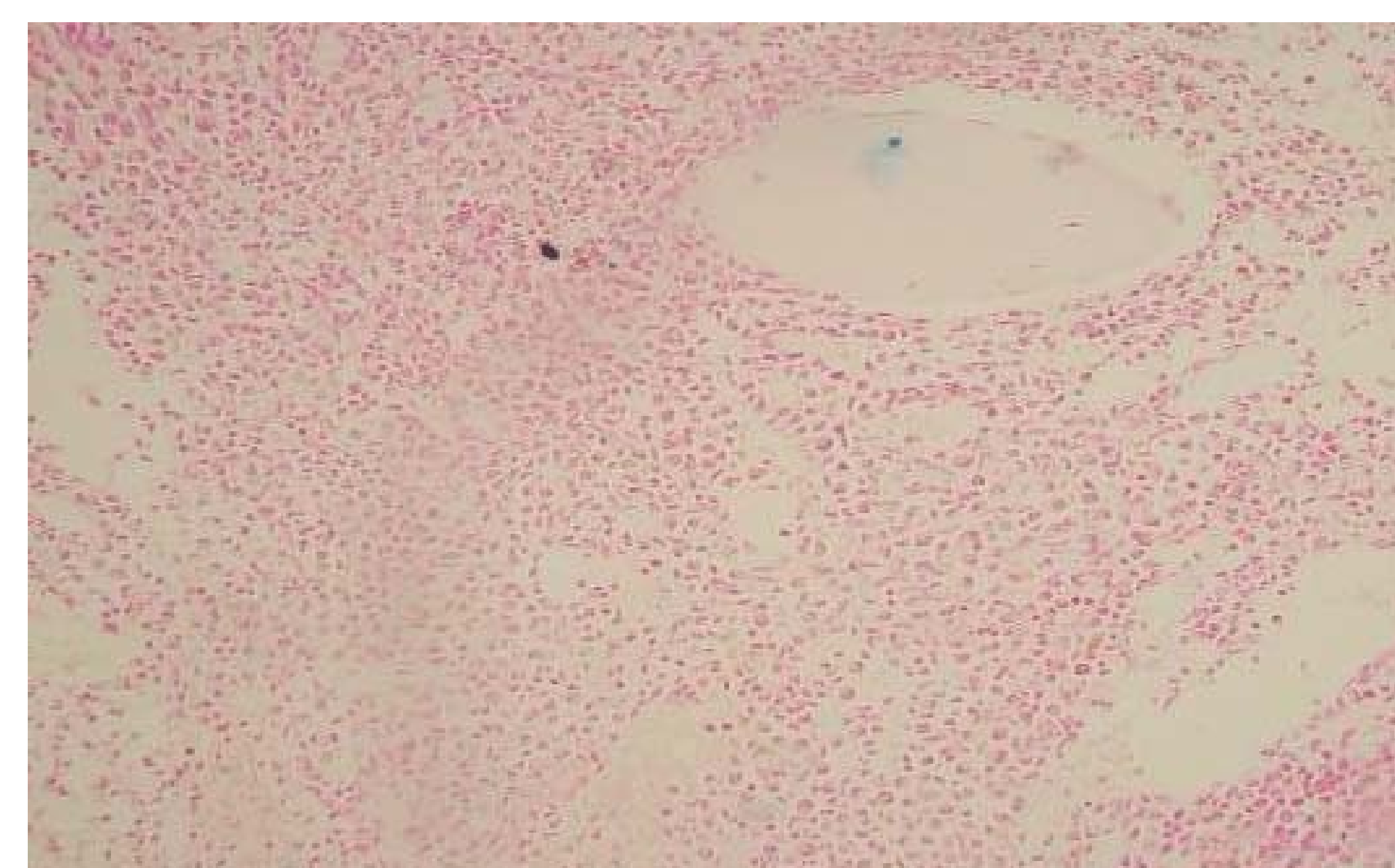


Fig 5. Iron Stain (40X)

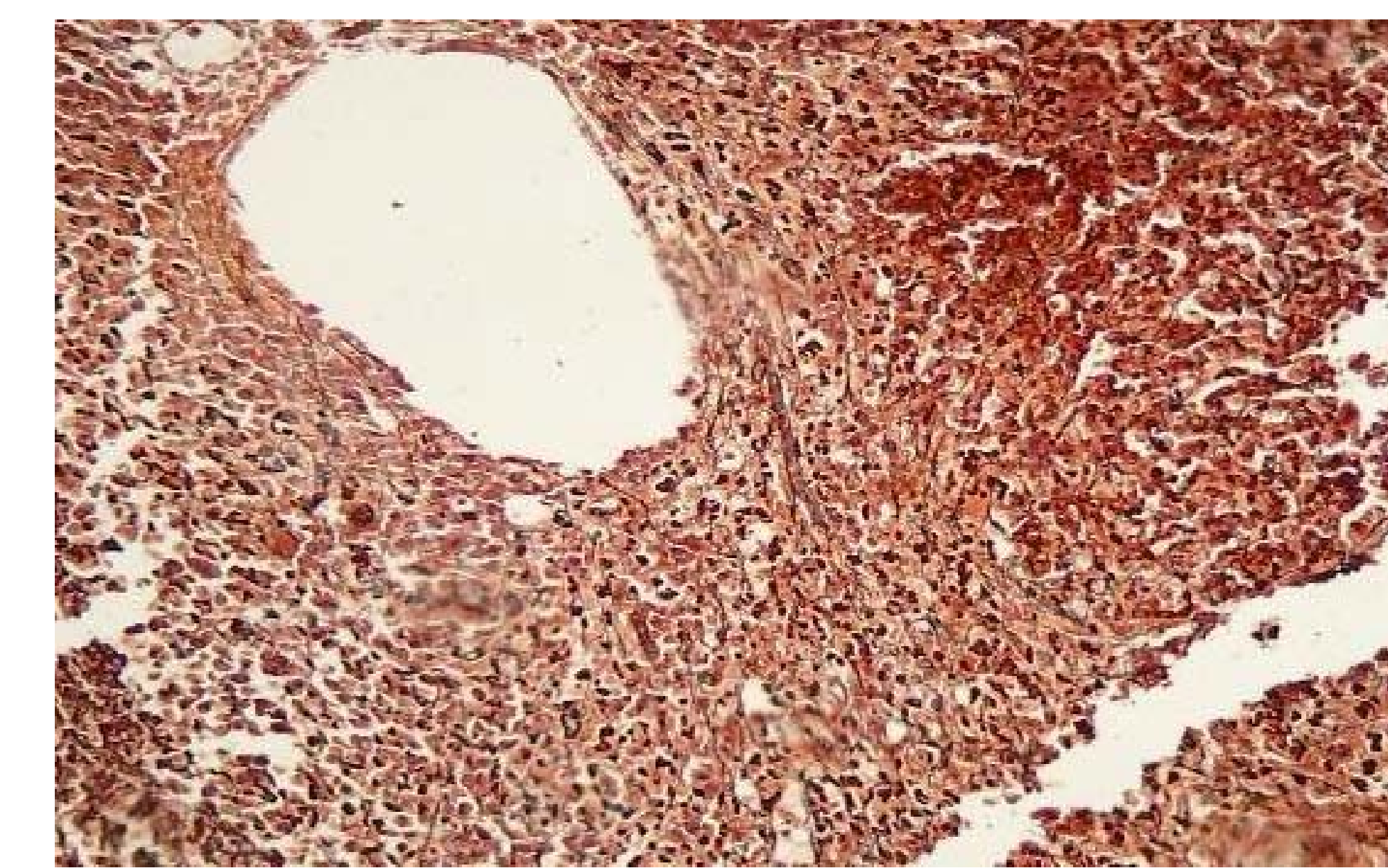


Fig 6. Reticulin Stain (40X)

CLINICAL COURSE:

- Induction of 200mg imatinib mesylate therapy (trade name Gleevec) followed by 100mg daily maintenance given to treat eosinophilia associated with FIP1L1-PDGFR A
- On imatinib, the patient's leukocytosis (White blood count 7400 cells/uL), peripheral eosinophilia (6%) and polycythemia (Hemoglobin 12.0 g/dL) improved without continuation of hydroxyurea or therapeutic phlebotomy.

CONCLUSIONS:

- To the best knowledge of the authors, there has been no case report of myeloproliferative neoplasm with PDGFR A rearrangement in the setting of clinical polycythemia vera.
- Further investigation of the relationship between polycythemia vera and eosinophilia with PDGFR A rearrangement may be warranted.

REFERENCES

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