ORIGINAL ARTICLE

Biomedica - Official Journal of University of Health Sciences, Lahore, Pakistan
Volume 41(2):73-77
https://doi.org/10.24911/BioMedica/5-1235

This is an open access article distributed in accordance with the Creative Commons Attribution (CC BY 4.0) license: https://creativecommons.org/licenses/by/4.0/) which permits any use, Share — copy and redistribute the material in any medium or format, Adapt — remix, transform, and build upon the material for any purpose, as long as the authors and the original source are properly cited. © The Author(s) 2025

The spectrum of bone marrow examination findings at a tertiary care centre in Lahore: a retrospective analysis

Rabia Ahmad[1*], Ayesha Khalid[2], Hira Arshad[3], Ahmad Saleem[4]

ABSTRACT

Background and Objective: Bone marrow examinations are typically safe procedures. Even though complications are uncommon, they can occur and may include significant bleeding in individuals with low platelet counts and infections, usually at the skin puncture site, particularly in immunocompromised patients. The conditions examined include bone disorders, as well as hematologic and non-hematologic malignancies in the bone marrow.

Methods: This study analyzed the retrospective data from Department of Pathology, Allama Iqbal Medical College/Jinnah Hospital, Lahore, including 373 patients who underwent bone marrow trephine biopsy between January 2023 and June 2024. Patient records were reviewed for demographic and diagnostic data. Statistical analysis was performed using the chi-square test, with $p < 0.05$ considered significant.

Results: Of the 373 patients, 206 (55.2%) were male, and 167 (44.8%) were female, yielding a male-to-female ratio of 1.2:1. The most common age group was 21-30 years, comprising 68 cases.

Bone marrow aspiration (BMA) was most frequently performed for suspected acute leukemia, followed by hepatosplenomegaly. Pancytopenia was observed in 59 cases (15.8%), while immune thrombocytopenia was noted in 3 cases. Normal trilineage hematopoiesis was reported in 65 cases. Aplastic anemia and hypocellular marrow were identified in 12 (3.2%) and 29 (7.7%) cases, respectively.

Acute leukemia was diagnosed in 77 cases (21%), with acute myeloid leukemia (AML) accounting for 21 cases (5.6%). A statistically significant association was found between age, gender, and AML, with a higher frequency in older male patients ($p = 0.04$). Multiple myeloma and other plasma cell disorders were diagnosed in 3 cases (0.8%), while myelofibrosis was identified in another 3 cases (0.8%). Hemophagocytic lymphohistiocytosis, myelodysplastic syndrome, and lymphoma were seen in 3, 11, and 8 cases, respectively.

Conclusion: Acute leukemia, particularly AML, was the most frequent diagnosis on BMA, followed by hepatosplenomegaly and pancytopenia. Significant association of AML with older male patients underscores the need for early evaluation in this group. Bone marrow examination remains a valuable diagnostic tool for diverse hematological disorders in our setting.

Keywords: Bone marrow, trephine biopsy, leukemia, aplastic anemia, myelodysplastic syndrome.

Received: 14 January 2025 **Revised date:** 21 April 2025 **Accepted:** 26 May 2025

Correspondence to: Rabia Ahmad
*Associate Professor, Department of Pathology, Allama Iqbal Medical College, Jinnah Hospital, Lahore, Pakistan.
Email: rabia1908@yahoo.com
Full list of author information is available at the end of the article.

Introduction

Bone marrow examination is a key diagnostic procedure utilized in the evaluation of a wide range of hematological disorders, malignancies, and systemic diseases affecting the bone marrow. It provides essential information regarding the functional status of the marrow, including its ability to produce an adequate number of blood cells. A bone marrow biopsy and aspiration together help determine whether the marrow is normocellular, hypocellular, or hypercellular, and whether abnormal cellular infiltration is present. Although generally considered safe, bone marrow examination may rarely be associated with complications such as bleeding, particularly in thrombocytopenic patients, and infection, especially at the puncture site in immunocompromised individuals.[1]

Bone marrow evaluation comprises two complementary techniques: bone marrow aspiration (BMA) and trephine biopsy. Aspiration primarily facilitates the assessment of cellular morphology, whereas trephine biopsy provides detailed information regarding marrow architecture,

cellularity, fibrosis, and patterns of infiltration by abnormal cells.[2] The most common indication for bone marrow examination is the presence of unexplained cytopenias involving one or more hematopoietic cell lines.[2] Its diagnostic importance becomes particularly evident when routine laboratory investigations fail to establish a definitive diagnosis.[3] Bone marrow examination is also valuable in identifying hematological malignancies, metastatic involvement, and various non-hematological conditions affecting the marrow.[4]

Trephine bone marrow biopsy plays a pivotal role in the diagnosis, staging, and follow-up of both hematological and non-hematological disorders. It is especially important in monitoring patients undergoing chemotherapy or bone marrow transplantation.[5] The procedure provides comprehensive insight into marrow cellularity, hematopoietic activity, and structural organization.[6] While bleeding remains the most frequently reported complication, issues related to pain assessment and management are often under-recognized.[5]

Given the diagnostic value of bone marrow examination, the present study was conducted to evaluate the spectrum of hematological disorders, including acute and chronic leukemias, lymphomas, and selected benign conditions, based on findings from BMA and biopsy at a tertiary care hospital in Lahore.

Methods

This retrospective cross-sectional study was conducted at the Department of Pathology, Allama Iqbal Medical College/ Jinnah Hospital, Lahore, after obtaining approval from the Institutional Ethical Review Board. A total of 373 patients who underwent bone marrow examination during the study period, from January 2023 to June 2024, were included.

BMA and trephine biopsy were performed under strict aseptic conditions from the posterior superior iliac crest using a sterile disposable bone marrow biopsy needle, following standard procedural protocols. Bone marrow aspirate smears were prepared immediately and stained with Giemsa stain for cytomorphological evaluation. Trephine biopsy specimens were fixed in 10% neutral buffered formalin, processed routinely, embedded in paraffin wax, sectioned, and stained with hematoxylin and eosin for histopathological examination. All specimens were evaluated by experienced pathologists to assess marrow cellularity, architecture, hematopoietic elements, and the presence of abnormal infiltrates in accordance with established diagnostic criteria.

Patients of all age groups and both genders who underwent BMA and/or trephine biopsy for the evaluation of hematological or suspected systemic disorders during the study period were included. Only those cases with adequate bone marrow samples and complete clinical and laboratory data were considered eligible for analysis.

Cases were excluded if they represented repeat bone marrow examinations of the same patient during the study period, to avoid duplication of data. Additionally, cases with inadequate or poorly preserved bone marrow samples, including insufficient aspirate or non-representative trephine biopsy specimens, were excluded. Patients with incomplete clinical information, missing laboratory parameters, or inconclusive diagnostic records were also excluded to ensure the reliability of data analysis.

The study aimed to evaluate the spectrum and frequency of hematological and non-hematological disorders diagnosed through bone marrow examination at a tertiary care center. Aplastic anemia was defined as the presence of bicytopenia or pancytopenia in peripheral blood (hemoglobin <10 g/dl, absolute neutrophil count $<1.5 \times 10^9$/l, and platelet count $<50 \times 10^9$/l) in association with bone marrow cellularity of less than 50%. Hypocellular bone marrow was defined as marrow cellularity of less than 50%, with or without accompanying peripheral cytopenias.

Statistical analysis

Data were entered and analyzed using the Statistical Package for Social Sciences, version 20. Categorical variables were expressed as frequencies and percentages. The chi-square test was applied to assess associations between variables, and a *p*-value <0.05 was considered statistically significant.

Results

A total of 373 patients were included in this study, with an age range of 0-85 years. There were 206 (55.2%) males and 167 (44.8%) females, with a male-to-female ratio of 1.2:1. The majority of patients belonged to the 21-30 years age group (*n* = 68; 18%), followed by the 11-20 years group (*n* = 71; 19%) (Table 1).

Table 1. *Age and gender distribution of study participants (n = 373).*

Age group (years)	Male (*n*)	Female (*n*)	Total (*n*)	Percentage (%)
0-10	11	2	13	3.5
11-20	36	35	71	19.0
21-30	31	37	68	18.2
31-40	35	27	62	16.6
41-50	37	24	61	16.4
51-60	34	19	53	14.2
61-70	12	20	32	8.6
>70	10	3	13	3.5
Total	206	167	373	100

Table 2. *Indications for bone marrow examination (n = 373).*

Indication	Frequency (*n*)	Percentage (%)
Acute leukemia	101	27.1
Hepatosplenomegaly	87	23.3
Lymphadenopathy	69	18.5
Pancytopenia	59	15.8
Pyrexia of unknown origin/ Lymphoma staging	51	13.7
Suspected metastasis	6	1.6
Total	373	100

Table 3. *Spectrum of bone marrow diagnoses (n = 373).*

Diagnosis	Frequency (*n*)	Percentage (%)
Acute leukemia	77	20.6
CML	54	14.5
Normal marrow morphology	65	17.4
Hypocellular marrow	29	7.8
Aplastic anemia	12	3.2
AML	21	5.6
Acute promyelocytic leukemia	10	2.7
MDS	11	3.0
Chronic lymphocytic leukemia	7	1.9
Lymphoma	8	2.1
Plasma cell disorders	3	0.8
HLH	3	0.8
Myelofibrosis	3	0.8
Essential thrombocythemia	2	0.5
Hairy cell leukemia	2	0.5
Polycythemia vera	3	0.8
Immune thrombocytopenia	6	1.6
Sideroblastic anemia	1	0.3
Eosinophilia	1	0.3
Miscellaneous	54	14.5
Total	373	100

Bone marrow examination was most commonly performed for the evaluation of suspected acute leukemia (*n* = 101; 27%), followed by hepatosplenomegaly (*n* = 87; 23.3%) and lymphadenopathy (*n* = 69; 18.4%). Other indications included pancytopenia (*n* = 59; 15.8%), pyrexia of unknown origin/lymphoma staging (*n* = 51; 13.6%), and suspected metastasis (*n* = 6; 1.6%) (Table 2).

On bone marrow examination, acute leukemia was the most frequent diagnosis, observed in 77 (21%) cases. Among these, acute myeloid leukemia (AML) constituted 21 (5.6%) cases, confirmed by Sudan Black B positivity. Chronic myeloid leukemia (CML) was identified in 54 (14.4%) cases, while normal marrow morphology with trilineage hematopoiesis was seen in 65 (17.4%) cases.

Table 4. *Distribution of major diagnoses according to age and gender.*

Diagnosis	Male (*n*)	Female (*n*)	Total (*n*)	*p*-value*
Acute leukemia	44	33	77	0.40
AML	16	9	25	0.04
Aplastic anemia	6	5	11	0.09
APML	4	6	10	0.40
CML	28	26	54	0.50
Hypocellular marrow	13	16	29	0.40
Lymphoma	5	3	8	0.45
MDS	5	6	11	0.50

*Chi-square test.

Non-malignant conditions included hypocellular marrow (*n* = 29; 7.7%) and aplastic anemia (*n* = 12; 3.2%). Other hematological disorders observed were myelodysplastic syndrome (MDS) (*n* = 11; 2.9%), lymphoma infiltration (*n* = 8; 2.1%), chronic lymphocytic leukemia (*n* = 7; 1.8%), and plasma cell disorders (*n* = 3; 0.8%). Rare findings included hemophagocytic lymphohistiocytosis (HLH) (*n* = 3; 0.8%), myelofibrosis (*n* = 3; 0.8%), essential thrombocythemia (*n* = 2; 0.5%), hairy cell leukemia (*n* = 2; 0.5%), and sideroblastic anemia (*n* = 1; 0.3%) (Table 3).

A statistically significant association was observed between age, gender, and AML, with higher frequency noted in older age groups and males ($p = 0.04$). No significant associations were observed for most other conditions due to smaller sample sizes (Table 4).

Discussion

This retrospective study highlights the diagnostic significance of bone marrow examination in evaluating a broad spectrum of hematological disorders and marrow involvement in systemic diseases. The procedure was generally well tolerated, with only mild pain and discomfort reported in a small number of cases. Although minor bleeding was observed in some patients, it was effectively managed without major complications, supporting the overall safety profile of bone marrow biopsy.

The study population showed a male predominance (55.2%), with a male-to-female ratio of 1.2:1. The most frequently represented age group was 21-30 years (18%), indicating a higher burden of hematological evaluations in young adults. In contrast, a previous study reported a nearly equal gender distribution (male-to-female ratio 1.02:1) with the majority of cases occurring in the 0-10 years age group, suggesting possible regional or referral-based differences in patient demographics.[7]

In the present study, the most common indication for bone marrow examination was suspected acute leukemia, followed by hepatosplenomegaly. These findings are consistent with those reported by Nadda et al.[5], who also identified acute leukemia as the leading indication. Pancytopenia was observed in 15.8% of cases, whereas other studies have reported anemia as the most frequent indication.[7] This variation may reflect differences in referral patterns, particularly the higher number of patients referred for anemia evaluation in certain centers.[7-9]

A subset of patients (14.4%) underwent bone marrow examination for pyrexia of unknown origin, lymphoma staging, and miscellaneous conditions, while lymphadenopathy was present in 18.4% of cases. The frequency of immune thrombocytopenia (1.6%) in this study was lower compared to previously reported rates (13%-15.7%).[1,2,10] This discrepancy may be attributed to the fact that bone marrow biopsy was primarily performed in treatment-resistant cases, rather than as a routine diagnostic step.

The spectrum of hematological disorders observed in this study included both malignant and benign conditions. Acute leukemia was the most common diagnosis (21%), whereas studies from other regions, including Karachi and Peshawar, have reported CML as the most prevalent malignancy.[8,9] Among acute leukemias, a substantial proportion comprised AML, with many patients presenting with fever and bleeding manifestations. The inability to determine the frequency of acute lymphoblastic leukemia due to the lack of flow cytometry is a limitation; however, the relatively higher frequency of AML may reflect the presence of a specialized oncology referral setup at our institution.

Hypocellular marrow and aplastic anemia collectively accounted for 11% of cases in this study. In contrast, a significantly higher frequency (76%) has been reported in a study from Peshawar, possibly due to regional environmental exposures or genetic predispositions.[8]

Myelofibrosis was observed in 0.8% of cases, which is much lower than the frequency (43.5%) reported by Ali Khan et al.[11] from Rawalpindi Pakistan. Plasma cell disorders were also identified in 0.8% of cases, whereas similar or modestly higher frequency (0.5%-3%) has been reported in other studies.[12,13] These differences may be influenced by population characteristics, diagnostic facilities, and referral bias.

The retrospective nature of the study represents a key limitation, as complete clinical and follow-up data were not available in all cases. Despite this, the study provides valuable insight into the local disease spectrum and diagnostic utility of bone marrow examination in a tertiary care setting.

Limitations of the Study

The study has several limitations. Its retrospective design limited the availability of complete clinical and follow-up information. Being conducted at a single center, the findings may not be fully generalizable to other populations. Additionally, the lack of advanced diagnostic techniques, such as flow cytometry and molecular studies, restricted the detailed sub-classification of certain hematological disorders. There is also a possibility of selection bias, as only patients undergoing bone marrow examination were included, and treatment outcomes or prognostic correlations were not evaluated.

Conclusion

Bone marrow examination is a vital diagnostic tool for a wide range of hematological and selected non-hematological disorders. Acute leukemia was the most commonly identified condition, while sideroblastic anemia was the least frequent in this study. The findings reflect the local disease pattern and highlight the diagnostic utility of bone marrow evaluation. Further prospective and multicenter studies are recommended to validate and expand upon these results.

Acknowledgement

The authors would like to acknowledge the administration of Allama Iqbal Medical College/Jinnah Hospital Lahore, Pakistan, for providing support during the execution of research work.

List of Abbreviations

BM	Bone marrow
AML	Acute myeloid leukemia

Conflict of interest

None to declare.

Grant support and financial disclosure

None to disclose.

Ethical approval

The ethical approval of the study was granted by the Institutional Ethics Committee of Allama Iqbal Medical College, Lahore, Pakistan vide Letter no: ERB159/1/06-02-2024/S1ERB.

Authors' contributions

RA, HA: Conception and design of study, acquisition and analysis of data, drafting of manuscript, and critical intellectual input.
AK, AS: Drafting of manuscript, critical intellectual input.
ALL AUTHORS: Approval and responsibility of the final version of the manuscript to be published.

Authors' Details

Rabia Ahmad[1], Ayesha Khalid[2], Hira Arshad[3], Ahmad Saleem[4]

1. Associate Professor, Department of Pathology, Allama Iqbal Medical College, Jinnah Hospital, Lahore, Pakistan
2. Associate Professor, Department of Pathology, Ameer Ud Din Medical College, Lahore, Pakistan

3. Director Clinical Governance, Strategic Management and Internal Policy Unit, Specialized Health Care and Medical Education Department, Government of Punjab, Lahore, Pakistan
4. Additional Principal Medical Officer, Department of Physiology, Allama Iqbal Medical College, Lahore, Pakistan

References

1. Mayo Clinic. Bone marrow biopsy and aspiration. 2024 [cited 2025 May]. Available from: https://www.mayoclinic.org/tests-procedures/bone-marrow-biopsy/about/pac-20393117/
2. Raskin RE, Messick JB. Bone marrow cytologic and histologic biopsies: indications, technique, and evaluation. Vet Clin North Am Small Anim Pract. 2012;42(1):23–42. https://doi.org/10.1016/j.cvsm.2011.10.001
3. Agrawal S, Bhandari R, Gowda VN, Gupta A, Singh N, Chowdhury N, et al. Hematological and biochemical predictors of bone marrow metastases in non-hematological malignancies: a clinico-pathological analysis. Preprint. medRxiv. 2020. https://doi.org/10.1101/2020.12.16.20248306
4. Claveau JS, Murray DL, Dispenzieri A, Kapoor P, Binder M, Buadi F, et al. Value of bone marrow examination in determining response to therapy in patients with multiple myeloma in the context of mass spectrometry-based M-protein assessment. Leukemia. 2023;37(1):1–4. https://doi.org/10.1038/s41375-022-01779-8
5. Nadda R, Repaka R, Mallik N, Sahani AK. A prospective survey on trephine biopsy of bone and bone marrow: an experience with 274 Indian patients' biopsies. Eur J Med Res. 2023;28(1):193. https://doi.org/10.1186/s40001-023-01167-7
6. Goyal S, Singh UR, Rusia U. Comparative evaluation of bone marrow aspirate with trephine biopsy in hematological disorders and determination of optimum trephine length in lymphoma infiltration. Mediterr J Hematol Infect Dis. 2014;6(1):e2014002. https://doi.org/10.4084/MJHID.2014.002
7. Bashir S, Fahim Akhtar, Misbah Salim, Maham Arshad, Zahra Tasleem Khan, Farhan Akhtar. Diagnostic significance of bone marrow examination amongst the patients with abnormal haematological parameters. J Ayub Med Coll Abbottabad. 2023;35(4):619–22. https://doi.org/10.55519/JAMC-04-12357
8. Ullah I, Khan D, Qureshi V. Etiological spectrum of bone marrow biopsy in patients with pancytopenia. Cureus. 2024;16(11):e73138. https://doi.org/10.7759/cureus.73138
9. Fatima N, Ashfaq M, Hussain W, Raza M, Anwar A. Clinical spectrum, laboratory profile and outcome of aplastic anemia in children presenting at tertiary care hospital. Prof Med J. 2025;32(11):1543–9. https://doi.org/10.29309/TPMJ/2025.32.11.9917
10. Bilal H, Ali S, Kiani MT. Hematological disorders on bone marrow examination in a tertiary care Centre. Pak J Pathol. 2024;35(1):44–8. https://doi.org/10.55629/pakjpathol.v35i1.791
11. Ali Khan M, Suhaib Ahmed, Muhammad Arif Siddique, Maryam Khan, Memoona Khan, Zainab Akram. Landscape of somatic mutations in myeloproliferative neoplasm in Pakistani population. Pak J Med Sci. 2025;41(7):2003–9. https://doi.org/10.12669/pjms.41.7.12129
12. Tschautscher MA, Jevremovic D, Buadi FK, Lacy MQ, Gertz MA, Dispenzieri A, et al. Utility of repeating bone marrow biopsy for confirmation of complete response in multiple myeloma. Blood Cancer J. 2020;10:95. https://doi.org/10.1038/s41408-020-00363-6
13. Fend F, Dogan A, Cook JR. Plasma cell neoplasms and related entities-evolution in diagnosis and classification. Virchows Arch. 2023;482(1):163–77. https://doi.org/10.1007/s00428-022-03431-3