

DIVERSITY OF HEMATOLOGICAL DISORDERS UNCOVERED IN BONE MARROW EVALUATION

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ABSTRACT

Background: Bone marrow examination is a significant diagnostic tool for assessing a wide range of hematological and certain non-hematological disorders. The spectrum of disorders varies with patient population and referral patterns. This retrospective study was conducted to evaluate the distribution and trends of hematological and non-hematological disorders diagnosed on bone marrow examination.

Objectives: To highlight the relative frequencies of various conditions diagnosed on routine bone marrow examination.

Methods: This retrospective study was conducted in the hematology department of Chughtai Institute of Pathology between January 2022 and December 2023. The study comprised all bone marrow biopsies performed at Chughtai Institute of Pathology as well as biopsy specimens received for reporting from various centers. One thousand nine hundred and seventy-one bone marrow biopsies in all were documented. Relevant clinical data were obtained and statistically analyzed, including patient demographics, presenting symptoms, laboratory results, imaging tests, bone marrow morphology, immunophenotypic profiles, and diagnosis. The mean and standard deviation for age was determined. Qualitative variables such as gender and diagnosis were expressed as frequencies and percentages.

Results: Most prevalent hematological condition in the current study group, with 289 cases and a frequency of 14.7%, was precursor B-acute lymphoblastic leukemia. Acute myeloid leukemia (9.0%), Aplastic anemia (6.2%), Chronic myeloid leukemia (6.3%), Multiple myeloma (5.3%) and Non-Hodgkin lymphoma (5.2%) were the next most prevalent disorders. 4% patients had megaloblastic anemia, whereas 10.4% of the patients had normal morphology.

Conclusion: Bone marrow examination remains a vital diagnostic tool for a wide range of hematological and non-hematological conditions. This study determines the major conditions encountered and reveals trends in their relative frequencies, providing clinicians with valued insight into local disease patterns and supporting in targeted diagnostic evaluation.

Keywords: aplastic anemia, bone marrow examination, hematological disorders, leukemia, non-Hodgkin lymphoma

How to cite this article: Ali Z, Imram A, Malik N.A, Chughtai A.S, Aziz B Diversity Of Hematological Disorders Uncovered In Bone Marrow Evaluation. Pak Postgrad Med J 2026;37(3): 130-134

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Received: Feb 23,2026; Revised: Aug 26,2026

Accepted: Sep 07,2026

DOI: <https://doi.org/10.51642/ppmj.v37i02.883>

INTRODUCTION

Blood disorders encompass a wide spectrum of conditions, from simple ailments like iron deficiency anemia to more complex and cancerous conditions

including leukemias, myelomas, and lymphomas.¹ Globally, the prevalence of hematological malignancies varies, with lower rates in areas with less resources and higher rates in countries with greater resources. This is frequently the result of underdiagnosis and a lack of registration resources in developing countries.

Based on their cell lineage, hematological malignancies can be broadly divided into two groups: myeloid and lymphoid. Hodgkin and non-Hodgkin's lymphoma, multiple myeloma, acute lymphoblastic leukemia, and chronic lymphocytic leukemia are all prevalent subtypes of lymphoid neoplasms. Acute myeloid leukemia, myeloproliferative neoplasm, and myelodysplastic syndrome are the three primary forms of myeloid neoplasms.²

Hematological (blood-related) and non-hematological (non-blood-related) diseases can both be diagnosed with bone marrow biopsies.³ Patients usually manage this procedure well, even if it requires some invasion. It also offers quick insights into many cases, which helps with prompt diagnosis. In the field of hematology, this process is invaluable and cost-effective, making it easier to identify both malignant and non-malignant hematological illnesses.⁴ For diagnostic purposes, bone marrow aspiration and bone marrow biopsy are frequently carried out concurrently since they complement one another.⁵

Bone marrow aspirate smears offer cytological insights into hemopoietic cells, whereas bone marrow trephine biopsies assess cellularity, infiltration patterns, and fibrosis.⁴ Aplastic anemia, myelodysplastic syndromes, non-Hodgkin's lymphomas, granulomatous diseases, myeloproliferative disorders, and metastatic cancers that induce a fibrotic response are some of the conditions that are crucial for determining.^{6,7}

This study aimed to determine the spectrum and frequency of hematological disorders among patients presenting to a high-volume tertiary care referral center serving Punjab and the northern regions of Pakistan. The findings provide baseline data on the regional distribution of hematological disorders and may serve as a valuable reference for future clinical and epidemiological research

METHODS

It was a retrospective study conducted in the hematology department of Chughtai Institute of Pathology between January 2022 and December 2023. The study proposal was approved by Institutional Review Board of Chughtai Institute of Pathology, Lahore (CIP/IRB/1257). The study included all bone marrow biopsies performed at Chughtai Institute of Pathology as well as biopsy specimens received for reporting from various centers during the above-mentioned time period. Biopsies with inadequate/suboptimal reporting material were excluded from the study. One thousand nine hundred and seventy-one bone marrow biopsies in all were documented. Relevant clinical data were obtained and statistically analyzed, including patient demographics, presenting symptoms, laboratory results, imaging tests, bone marrow morphology, immunophenotypic profiles, and diagnosis. 2% xylocaine was used as local anesthetic during the bone marrow procedure. Giemsa and specialty stains such as Perl's stain, PAS, Sudan black B, and others were used to stain the peripheral blood sample and the bone marrow aspirate smears. Hematopoiesis, iron reserves, abnormal cells, storage disorders, infiltration with non-hematopoietic malignancies, etc. were all evaluated in bone marrow smears. After decalcification trephines were embedded in paraffin. Hematoxylin and eosin staining was applied to the slides

following standard tissue processing. In other cases, immunohistochemistry was also used to diagnose the condition. If a BMA has well-preserved marrow particles for cytological analysis, including differential counting and microscopic identification of aberrant cells, it is deemed sufficient. The trephine biopsy is considered adequate for histopathological evaluation, if the core length is 1.5–2.0 cm and containing at least 5 - 6 evaluable hematopoietic intertrabecular spaces/pockets. Statistical analysis was performed using SPSS version 25.0. The mean $\pm$ SD for age was determined. Among the qualitative variables for which frequency and percentage were calculated were gender and diagnosis.

RESULTS

The age distribution showed that 676 cases (34.3%) were between the ages of 1 and 18, 603 cases (31%) were between the ages of 19 and 45, 350 cases (17.7%) were between the ages of 46 and 60, and 342 cases (17%) were older than 60 (Table 1). Of the 1971 patients, 820 cases (41.6%) were female and 1151 cases (58.4%) were male (Table 2).

Table 1: Age distribution (n=1971)

Age (years)	No. of patients	%
1-18	676	34.3
19-45	603	31.0
46-60	350	17.7
>60	342	17.0
Total	1971	100.0

Table 3 shows that the most prevalent hematological condition in the current study group, with 289 cases and a frequency of 14.7%, is precursor B-acute lymphoblastic leukemia. Acute myeloid leukemia (9.0%), Aplastic anemia (6.2%), Chronic myeloid leukemia (6.3%), Multiple myeloma (5.3%) and 'Non-Hodgkin lymphoma' (5.2%) were the next most prevalent disorders. 4% of patients had megaloblastic anemia, whereas 10.4% of patients had normal morphology.

Table 2: Gender distribution (n=1971)

Gender	No. of patients	%
Male	1151	58.4
Female	820	41.6
Total	1971	100.0

DISCUSSION

The study of bone marrow is essential for the detection of various hematological and non-hematological illnesses. Aspirations are mostly used for cytological details; however, trephine biopsies are useful for cellularity, fibrosis, architectural pattern, metastatic deposits, and are suitable for immunohistochemistry.

These can also be used to monitor the effectiveness of chemotherapy in patients receiving it.⁸

Table 3: Diagnosis on bone marrow examination (n=1971)

Diagnosis	No of patients	%age
Normal morphology	206	10.4%
Megaloblastic anemia	79	4.0%
Mixed deficiency anemia	63	3.2%
Aplastic anemia	122	6.2%
Immune thrombocytopenia	71	3.6%
Acute lymphoblastic leukemia	289	14.7%
Acute myeloid leukemia	178	9.0%
Chronic myeloid leukemia	125	6.3%
Chronic lymphocytic leukemia	40	2.0%
Multiple myeloma	104	5.3%
Myelodysplastic neoplasm	58	2.9%
Myelofibrosis	34	1.7%
Essential thrombocythemia	14	0.7%
Polycythemia vera	10	0.5%
MDS/MPN	10	0.5%
Metastatic bone marrow	26	1.3%
Storage disorders	7	0.4%
Hodgkin lymphoma	26	1.3%
Non-Hodgkin lymphoma	102	5.2%
Congenital dyserythropoietic anemia	8	0.4%
Erythroid hyperplasia	51	2.6%
PRCA	13	0.7%
Reactive bone marrow	68	3.4%
Hypersplenism	23	1.2%
Myeloid hyperplasia	17	0.9%
Iron deficiency anemia	21	1.1%
Chronic eosinophilic leukemia/HES/eosinophilia	4	0.2%
BI phenotypic acute leukemia	4	0.2%
HLH/ increased macrophage activity	16	0.8%
Granulomatous disease	7	0.4%
Evans syndrome	5	0.3%
Post chemotherapy bone marrow in remission	147	7.5%
Anemia of chronic disease	7	0.4%
Bone marrow infestation with Malarial parasite	4	0.2%
Therapy related AML	3	0.1%
Amegakaryocytic thrombocytopenia/ selective megakaryocytic hypoplasia	3	0.1%
Osteopetrosis	2	0.1%
Leishmaniasis	2	0.1%
Inherited platelet disorder	2	0.1%
Total cases	1971	100%

In the present study, a total of 1,971 bone marrow biopsies were evaluated for hematological abnormalities. Of these, 676 cases (34.3%) involved patients aged 1–18 years, 603 cases (31%) were aged 19–45 years, 350 cases (17.7%) were aged 46–60 years, and 342 cases (17%) were aged over 60 years. These results were highly comparable to those of Gilotra et al⁹ and Shah R et al.¹⁰ In contrast, Atchyuta et al., reported the highest proportion of cases among patients aged 31–40 years (19%), followed by those aged 51–60 years.¹¹ Erythroid hyperplasia (34.2%) and megaloblastic anemia (10.9%) were the most common non-malignant disorders, while chronic leukemias (10.4%) were more frequent than acute leukemias (6.4%).¹¹ These findings differ from our study, in which acute leukemias were the most common hematological malignancies and aplastic anemia was the predominant non-malignant disorder. The difference in age distribution may be attributable to the relatively higher number of pediatric referrals for bone marrow examination received by our Pathology Department. Similar to our findings, a male predominance was observed, with males accounting for 1,151 (58.4%) cases compared with 820 (41.6%) females among the 1,971 patients studied.

The most prevalent hematological condition in the current study population is precursor B-acute lymphoblastic leukemia, which accounts for 289 cases with a frequency of 14.7%. Acute myeloid leukemia (9.0%), Aplastic anemia (6.2%), chronic myeloid leukemia (6.3%), multiple myeloma (5.3%), non-Hodgkin lymphoma (5.2%) were the next most prevalent disorders. 4% of patients had megaloblastic anemia, while 10.4% had normal morphology. Similar research was also conducted by Pudasaini S¹², Qahtani AS¹³, and Chowdhury MRK et al.¹⁴ In a study conducted by Shastri et al., nutritional anemia was reported as the most common etiology, followed by aplastic marrow.¹⁵ In contrast, our findings differed, with acute lymphoblastic leukemia being the most frequently observed hematological disorder.

However, these results are consistent with a number of international research carried out in other Asian nations including Nepal¹⁶, as well as in nearby nations like Iran.¹⁷ A study conducted in Iran showed acute leukemias being the highest in number.¹⁷ Studies from the United States, Canada, and Africa have also shown similar patterns.^{18,19} Our study is in line with Tshabalala et al's study, which revealed that leukemias were the most common.¹⁸

Our results are in contrast to research by Iqbal et al., that found that acute myelogenous leukemia and acute lymphoblastic leukemia were equal in number (23.08%).²⁰ This discrepancy may be explained by differences in the age distribution, with AML more common in adults and ALL more common in youngsters. There were more pediatric patients in our study. On the other hand, disorders including chronic granulomatous disease, pure red cell aplasia, storage diseases and

visceral leishmaniasis were less common. Malignancies are more common than benign illnesses in the distribution of bone marrow diagnosis, highlighting the diagnostic relevance of bone marrow biopsy procedure. Our findings are in contrast to Omer et al.'s findings, which showed that aplastic anemia and Chronic Myeloid Leukemia (CML) were the most prevalent disorders.²¹ This study has some limitations. As a retrospective, single-center study conducted at a tertiary care referral center, its results may not be generalizable to the broader population. Referral bias, particularly among pediatric patients, may have influenced the observed spectrum of hematological conditions. Acute lymphoblastic leukemia was the most frequently observed hematological illness in our study, likely reflecting the higher percentage of pediatric patients referred to our center. In addition, operator-related factors affecting specimen adequacy could not be objectively evaluated because of the retrospective study design. Finally, ancillary investigations, including flow cytometry, cytogenetics, and molecular studies, were not included in the analysis, as they were frequently performed later in the diagnostic workup and were not consistently documented in the original bone marrow reports.

CONCLUSION

Bone marrow examination continues to play a central role in the diagnosis of hematological and selected non-hematological disorders. In this retrospective analysis, malignant disorders were more frequent, with acute leukemias particularly precursor B-acute lymphoblastic leukemia predominating, especially in children. Benign conditions, including megaloblastic and aplastic anemia, were also commonly observed. Overall, the diagnostic distribution aligns with regional and international literature, reaffirming the value of bone marrow examination in patients presenting with unexplained hematological abnormalities. These local frequency patterns can help clinicians prioritize early suspicion of acute lymphoblastic leukemia, particularly precursor B-ALL, in pediatric patients presenting with unexplained cytopenias, prompting timely bone marrow evaluation and definitive diagnosis.

ETHICAL APPROVAL

Ethical approval of article was granted by the Institutional Review Board of Chughtai Institute of Pathology, Lahore vide CIP/IRB/1257.

AUTHOR'S CONTRIBUTIONS

ZA: Conceived idea, design, manuscript writing

AI: Data analysis, statistical analysis

NAM: Data collection, data analysis

ASC: Critical review

All Authors: Approval of the final version of the manuscript to be published

CONFLICT OF INTEREST

Authors declare no conflict of interest.

FUNDING SOURCE: None

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