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Diagnostic Utility of Bone Marrow Aspiration and Biopsy in Various Hematological Disorders: A Study of 3 Years

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KEYWORDS

Bone marrow aspiration, Bone marrow biopsy, Bone marrow examination.

ABSTRACT:

Background: Bone marrow is involved in variety of hematological and non-hematological disorders. The hematological disorders include acute leukemia, myeloproliferative neoplasm (MPN), hemato-lymphoid neoplasm, nutritional deficiency diseases. On the other hand non-hematological disorders include infectious diseases infiltrating the bone marrow such as tuberculosis, parasitic infections and metastatic deposits

Aim:The aim of study is the diagnostic utility of bone marrow aspiration and biopsy findings in various hematological disorders

Study Design: This is retrospective study. The findings of bone marrow aspiration and biopsy is correlated in various hematological disorders after retrieving the data from record available in pathology department.

Material and Methods:This was a 3 year retrospective study done in the department of pathology, UPUMS, Saifai, Etawah from Jan 2023 to December 2025. A total of 39 cases presented with clinical haematological disorders. All the smears and sections were reviewed for morphological details and findings on peripheral blood,aspirate and biopsy and compared to each other. A detailed history, clinical findings, routine relevant laboratory investigations and radiological findings were carried out in each case

Results:The most common diagnosis based on BMA cytology was erythroid hyperplasia (normoblastic/micronormoblastic) seen in 9 cases (23.1%), followed by erythroid hyperplasia with megaloblastoid change in 7 cases (17.9%) (Fig 3 a,b) and hypocellular marrow in 5 cases (12.8%)(Fig 4). Other findings included hemodiluted marrow (10.3%), CML (7.7%), myelodysplastic syndrome and ITP (5.1% each), and a few rare entities like ALL, pure red cell aplasia, idiopathic eosinophilia, and inflammatory pathology (2.6% each). In 4 cases (10.3%), aspirate was inadequate for opinion.Concordant diagnosis was observed in 17 cases (48.6%), including conditions like erythroid hyperplasia with megaloblastoid change, marrow hypoplasia, and CML

Conclusion: The study demonstrates a high diagnostic yield of bone marrow examination in evaluating hematological disorders. While BMA is effective and easier, it may be insufficient in certain neoplastic or hypo-plastic disorders. BMB significantly enhances diagnostic accuracy, especially in cases of inadequate aspirate or marrow failure, which is supported by multiple referenced studies

Introduction

The adult haematopoietic system consists of specialized tissues and organs responsible for the production, development, and breakdown of blood cells. These include the bone marrow, thymus, spleen, and lymph nodes.The bone marrow is the primary site where myeloid, erythroid, megakaryocytic, and lymphoid cells originate and develop.[1]

Bone marrow examination is a crucial diagnostic procedure employed to assess a variety of

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hematological disorders, encompassing both neoplastic and non-neoplastic conditions. This evaluation plays a pivotal role in confirming clinically suspected diagnoses and can also uncover previously unrecognized hematological diseases. [2,3]

Bone marrow aspiration is a widely performed and minimally invasive diagnostic procedure routinely conducted in hospitals for the evaluation and management of various hematological disorders. It is considered a safe and effective method for obtaining bone marrow samples to diagnose conditions such as anemia, leukemia, and myelodysplastic syndromes. [4]

Trephine biopsy offers significant advantages by providing intact, three-dimensional bone marrow architecture, allowing for comprehensive assessment of both cellular morphology and stromal elements. This technique enables pathologists to examine the spatial arrangement of cells and the bone marrow microenvironment, facilitating the identification of infiltrative processes and abnormalities that may not be apparent in aspirate samples. Additionally, trephine biopsy is particularly valuable in cases where aspiration yields insufficient material or when a "dry tap" occurs, ensuring a more complete evaluation of the marrow. [5]

Bone marrow aspiration (BMA) and bone marrow biopsy (BMB) are fundamental diagnostic procedures routinely performed in hospitals across India to evaluate hematological disorders. While numerous studies have examined the individual roles of BMA and BMB in diagnosing various hematological conditions, fewer have directly compared the relative diagnostic value of these two techniques. This comparative analysis is crucial, as it can guide clinicians in selecting the most appropriate method based on the specific clinical scenarioand available resources.[6-8]

The present study was conducted to correlate the findings of peripheral bloodsmear with that of bone marrow aspirate cytology and trephine biopsy and to formulate an effective and rapid method for diagnosing wide spectrum of hematological diseases.

MATERIALSANDMETHODS

This was a 3 year retrospective study done in the department of pathology, UPUMS, Saifai, Etawah from Jan 2023 to December 2025. A total of 39 cases presented with clinical haematological disorders. All the smears and sections were reviewed for morphological details and findings on peripheral blood,aspirate and biopsy and compared to each other. A detailed history, clinical findings, routine relevant laboratory investigations and radiological findings were carried out in each case. Only those cases in which bone marrow examination was done by using BMA and BMB were included in the study. The standard technique was employed for obtaining the aspirate samples using the Salah's needle from posterior superior iliac spine. The trephine biopsy was performed using Jamshidi needle with the length of the biopsy core ranging from 1 to 3 cm. The biopsy was then fixed for minimum of 24 hours in 10% buffered formalin and then decalcified overnight in mixture of 8% hydrochloric acid and 10% formic acid in equal amounts. The fixation of the biopsy core was followed by automated tissue processing, paraffin embedding and sectioning. All the aspirate smears were routinely stained by Jenner Giemsa while the trephine biopsy sections were stained by routine Hematoxylin and Eosin stain. The relevant cytochemistry staining was performed as and when required. All the smears and sections were reviewed for morphological details by pathologists and the findings on peripheral smear, BMA and BMB were compared and the final correlation was done.

RESULT

The most common diagnosis based on BMA cytology was erythroid hyperplasia (normoblastic/micronormoblastic) seen in 9 cases (23.1%), followed by erythroid hyperplasia with megaloblastoid change in 7 cases (17.9%) (Fig 3 a,b) and hypocellular marrow in 5 cases (12.8%)(Fig 4). Other findings included hemodiluted marrow (10.3%), CML (7.7%), myelodysplastic syndrome and ITP (5.1% each), and a few rare entities like ALL, pure red cell aplasia, idiopathic eosinophilia, and inflammatory pathology (2.6% each). In 4 cases (10.3%), aspirate was inadequate for opinion.Histopathological examination from BMB revealed erythroid hyperplasia 8 (24.2%), followed closely by erythroid hyperplasia with megaloblastoid changes 6 (18.2%) and hypocellular marrow 3 (9.1%). Neoplastic lesions like CML 3 (9.1%), AML/MDS 3 (9.1%), and acute leukemia 1(3%) were also detected (Fig 1a,b and 2a,b). Biopsies were non-diagnostic in 6 cases (18.2%), which were either inadequate or dry tap.

Journal of Chemical Health Risks

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Concordant diagnosis was observed in 17 cases (48.6%), including conditions like erythroid hyperplasia with megaloblastoid change, marrow hypoplasia, and CML.

Out of 39 patients, 22 (56.4%) were female and 17 (43.6%) were male. Female preponderance was noted in Erythroid hyperplasia with megaloblastoid change (F:M = 8:2), MDS/AML (F only), Megakaryocytic hyperplasia (F only). The majority of hematological abnormalities occurred in the 1–5 year age group (17.9%), followed by 21–30 years (15.4%) and 16–20 years (10.3%). Erythroid hyperplasia was spread across pediatric and young adult age groups. Hypocellular marrow and ALL were mainly seen in younger patients. MDS, CML, and AML were more common in adults and elderly.

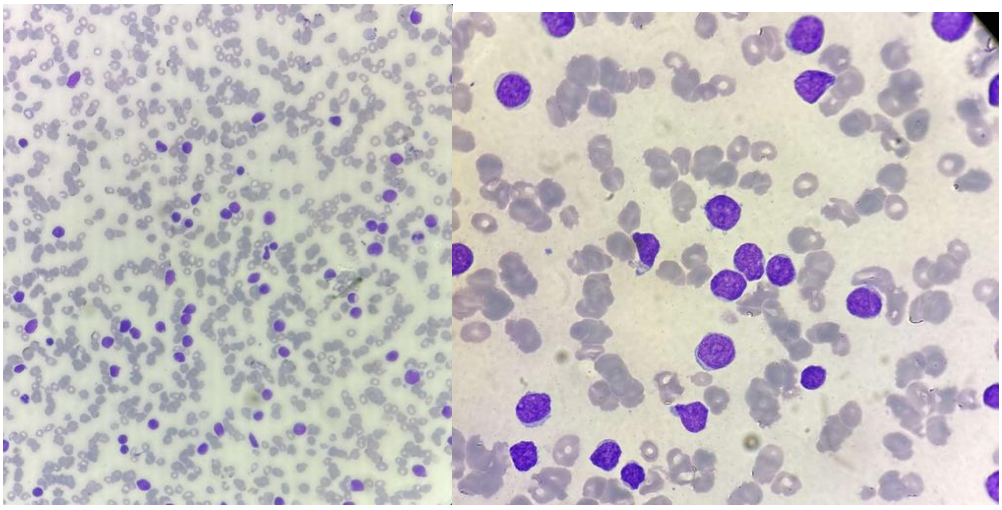

Figure 1(a) **1(b)**

Bone marrow aspirate showing blasts Acute Leukemia (Giemsa stain, 40X and 100X).

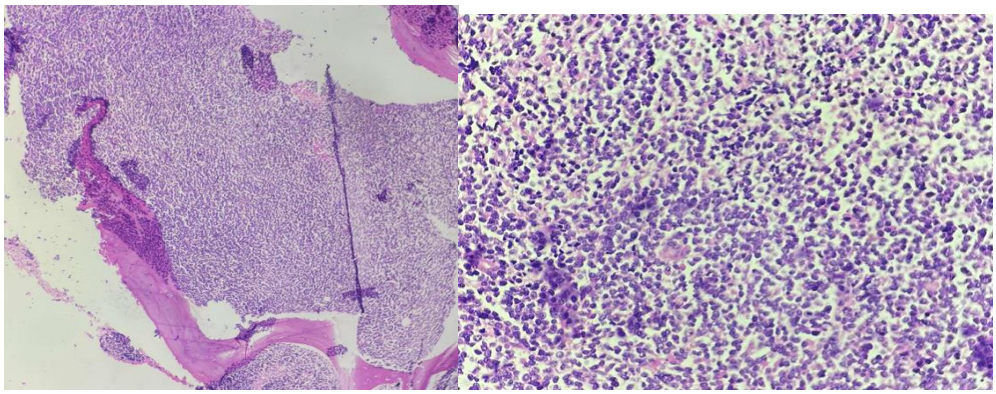

Figure 2(a) **Figure 2(b)**

Bone marrow biopsy showing hypercellular bone marrow fragments with absence fat cells (HE stain, X10.)

Bone marrow biopsy showing infiltration by blast cells (HE stain, X40.)

Journal of Chemical Health Risks

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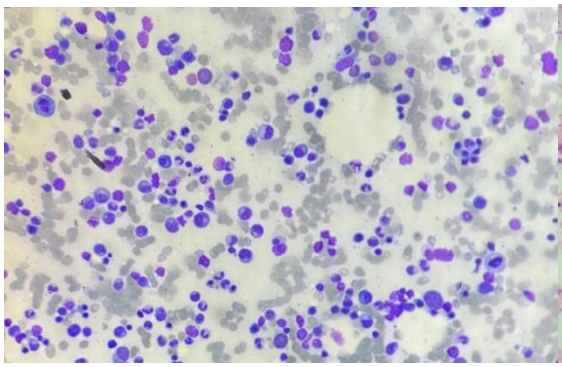

Fig 3(a), BMA and BMB showing megaloblastoid Changes (Giemsa stain, 40X)

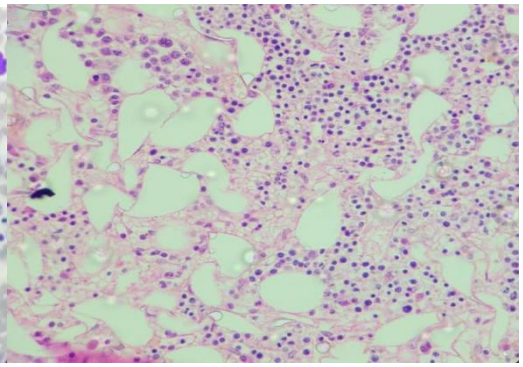

Fig 3(b) BMB showing megaloblastoid changes (HE stain, X40.)

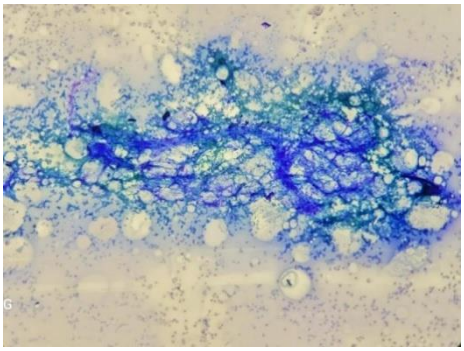

Fig 4 BMA showing only fat cells absence of hematopoietic cells(Marrow hypoplasia)

(Giemsa 10x)

Discussion:

The present study aligns closely with several previous investigations cited in the reference section, affirming the diagnostic value of combined bone marrow aspiration (BMA) and trephine biopsy (BMB) in evaluating hematological disorders. Chandra et al. [2] and Nanda et al. [3] highlighted the significance of BMB in cases where BMA is inadequate or hemodiluted, a pattern echoed in our study where BMB provided more definitive diagnoses in multiple discordant cases. The predominance of erythroid hyperplasia in our findings is consistent with those of Kibria et al. [4], who documented similar trends in patients with anemia and nutritional deficiencies. Dacie and Lewis [5] emphasized the structural advantages of BMB for assessing marrow architecture, which our study confirms by identifying conditions such as MDS and CML more accurately on biopsy. Gupta et al. [6] and Moid and DePalma [7] also support the superior role of BMB in identifying dysplastic and infiltrative lesions, which matched our higher detection rate of MDS/AML on BMB compared to BMA.

Sabharwal et al. [8] reinforced the utility of BMB for diagnosing benign conditions, a finding that aligns with our observations of ITP, eosinophilia, and red cell aplasia. Our correlation statistics48.6% concordance, 20% partial concordance, and 14.3% discordanceare similar to those reported by Ch Toi et al. [9], who found 64% complete and 28% partial correlation, reaffirming the diagnostic complimentary of BMA and BMB. The importance of biopsy in identifying subtle marrow changes, as stressed by Varma et al. [10] and Pasquale and Chikkappa [11], was supported in our study, especially in hypoplastic and

Journal of Chemical Health Risks

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JCHR (2026) 16(1), 285-292 | ISSN:2251-6727

myelodysplastic conditions. Finally, studies by Aboul-Nasr et al. [13] and Barekman and Cotelingam [14] on the comparative efficacy of smears and imprints validate the morphological limitations of aspiration alone, as seen in our inadequate BMA cases. Overall, our findings reinforce the literature consensus that simultaneous use of BMA and BMB significantly improves diagnostic accuracy across a wide range of hematological disorders

Conclusion

The study demonstrates a high diagnostic yield of bone marrow examination in evaluating hematological disorders. While BMA is effective and easier, it may be insufficient in certain neoplastic or hypo-plastic disorders. BMB significantly enhances diagnostic accuracy, especially in cases of inadequate aspirate or marrow failure, which is supported by multiple referenced studies.

Table 1: Diagnosis on bone marrow aspiration (BMA) cytology

S.No	Diagnosis (BMA Finding)	Count	Percentage (%)
1	Erythroid hyperplasia (normoblastic/micronormoblastic)	9	23.1%
2	Erythroid hyperplasia with megaloblastoid change	7	17.9%
3	Hypocellular marrow/marrow hypoplasia	5	12.8%
4	Hemodiluted	4	10.3%
5	CML / CML-CP / CML-Blast	3	7.7%
6	Megakaryocytic hyperplasia (incl. ITP)	2	5.1%
7	Myelodysplastic syndrome (MDS / AML)	2	5.1%
8	Mild megaloblastoid change (nutritional anemia)	2	5.1%
9	ALL (Acute Lymphoblastic Leukemia)	1	2.6%
10	Pure red cell aplasia	1	2.6%
11	Lymphoproliferative disorder	1	2.6%
12	Inflammatory pathology	1	2.6%
13	Idiopathic eosinophilia	1	2.6%
14	No opinion / Inadequate	4	10.3%

Table 2: Histopathological diagnosis of lesions based on trephine biopsy

S.No	Diagnosis (BMB Finding)	Count	Percentage (%)
1	Erythroid hyperplasia (normoblastic/micronormoblastic)	8	24.2%
2	Erythroid hyperplasia with megaloblastoid change	6	18.2%
3	Hypocellular marrow / Marrow hypoplasia	3	9.1%
4	CML / CML-CP / CML-Blast	3	9.1%
5	Myelodysplastic syndrome (MDS / AML)	3	9.1%
6	Acute Leukemia (myeloid)	1	3.0%
7	ALL (Acute Lymphoblastic Leukemia)	1	3.0%
8	Immune Thrombocytopenic Purpura (Megakaryocytic hyperplasia)	1	3.0%
9	Dysmegakaryopoiesis + Erythroid hyperplasia	1	3.0%
10	Normocellular marrow with eosinophilia	1	3.0%
11	Inadequate for evaluation / No opinion	6	18.2%

Journal of Chemical Health Risks

www.jchr.org

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Table 3 :Gender-wise Distribution of Diagnoses (Combined BMA + BMB)

S.No	Diagnosis Category	Male (M)	Female (F)	Total
1	Normoblastic/Mild normoblastic erythroid hyperplasia	1	6	7
2	Erythroid hyperplasia with megaloblastoid change	2	8	10
3	Hypocellular marrow / Marrow hypoplasia	2	4	6
4	Hemodiluted	3	2	5
5	CML / CML-CP / CML-Blast	2	3	5
6	Myelodysplastic syndrome (MDS / AML)	0	4	4
7	Megakaryocytic hyperplasia (ITP)	0	2	2
8	ALL (Acute Lymphoblastic Leukemia)	1	1	2
9	Inflammatory pathology	1	0	1
10	Idiopathic eosinophilia	1	0	1
11	Pure red cell aplasia	1	0	1
12	Lymphoproliferative disorder	1	0	1
13	Erythroid hyperplasia with dyserythropoiesis	0	1	1
14	Dysmegakaryopoiesis + erythroid hyperplasia	0	1	1
15	Acute leukemia (myeloid)	0	1	1
16	Normocellular marrow with eosinophilia	1	0	1
17	Inadequate / No opinion	4	3	7

Table 4. Frequency of Different Diagnoses Across Age Groups

Diagnosis	<1 yr	1–5 yrs	6–10 yrs	11–15 yrs	16–20 yrs	21–30 yrs	31–40 yrs	41–50 yrs	51–60 yrs	61–70 yrs	71–80 yrs	Total	% of Total
Erythroid hyperplasia (normoblastic)	1	3	–	–	–	1	–	–	–	–	–	5	15.2%
Erythroid hyperplasia with megaloblastoid	–	–	–	1	–	4	–	–	–	2	–	7	21.2%
Marrow hypoplasia / Hypocellular marrow	–	3	–	–	2	–	–	–	–	–	–	5	15.2%
Megakaryocytic hyperplasia (ITP)	–	1	1	–	–	–	–	–	–	–	–	2	6.1%
ALL (Acute Lymphoblastic Leukemia)	–	2	–	–	–	–	–	–	–	–	–	2	6.1%
MDS / AML	–	–	–	1	–	2	–	–	1	–	–	4	12.1%
CML / CML-CP / CML-Blast	–	–	–	–	–	–	–	–	2	1	–	3	9.1%

Journal of Chemical Health Risks

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JCHR (2026) 16(1), 285-292 | ISSN:2251-6727

Nutritional anemia (megaloblastoid)	–	–	1	–	–	–	–	–	–	–	–	1	3.0%
Red cell aplasia	1	–	–	–	–	–	–	–	–	–	–	1	3.0%
Idiopathic eosinophilia / eosinophilia	1	–	–	–	1	–	–	–	–	–	–	2	6.1%
Inflammatory pathology	–	–	–	–	1	–	–	–	–	–	–	1	3.0%
Lymphoproliferative disorder	–	1	–	–	–	–	–	–	–	–	–	1	3.0%
Dysmegakaryopoiesis + erythroid hyperplasia	–	–	–	–	–	–	1	–	–	–	–	1	3.0%
Acute Leukemia (myeloid)	–	–	–	–	–	–	–	–	1	–	–	1	3.0%
Megaloblastic hyperplasia	–	–	–	–	–	–	–	–	–	–	1	1	3.0%

Table 5: Correlation between BMA and BMB Findings

Correlation Type	**Description**	**No. of Cases**	**Examples (Findings)**
Concordant	Same diagnosis on both BMA and BMB	17	- CML-CP (BMA & BMB) - Marrow hypoplasia - Erythroid hyperplasia with megaloblastoid change
Partially Concordant	Same general pathology type, but variation in degree or cellularity	7	- Normoblastic vs. mild normoblastic erythroid hyperplasia - Megaloblastoid change with dysplasia
Discordant	Different or more definitive diagnosis on BMB than BMA	5	- Hemodiluted BMA → AML on BMB - Inadequate BMA → MDS on BMB
Inadequate/No opinion	Either BMA or BMB (or both) reported as inadequate or non-diagnostic	6	- Inadequate BMA/BMB - No opinion due to poor cellularity
Total Correlated Cases	Cases where both BMA and BMB were done and could be compared	**35**	

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