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Research Article

Evaluation of Bone Marrow Aspiration and Biopsy in Diagnosing Hematological Disorders: A Comparative Study

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Abstract

Background: Bone marrow examination is a pivotal investigation in the diagnosis of hematological disorders. Bone marrow aspiration and biopsy are complementary procedures, each offering distinct diagnostic advantages. Evaluating their relative and combined diagnostic utility is essential for optimizing diagnostic strategies. **Aim:** To evaluate and compare the diagnostic utility of bone marrow aspiration and bone marrow biopsy in patients with suspected hematological disorders. **Materials and Methods:** This hospital-based, descriptive comparative study included 80 patients who underwent both bone marrow aspiration and biopsy at a tertiary care center. Bone marrow aspiration smears and trephine biopsy sections were examined for diagnostic adequacy, cellularity, infiltrative lesions, and fibrosis. The spectrum of hematological disorders was analyzed, and diagnostic concordance between the two modalities was assessed. Statistical analysis was performed using appropriate tests, with $p < 0.05$ considered significant. **Results:** Bone marrow biopsy demonstrated significantly higher diagnostic adequacy and overall diagnostic yield compared to aspiration. Biopsy was superior in detecting marrow cellularity, fibrosis, and infiltrative lesions. Megaloblastic anemia was the most common diagnosis, followed by acute leukemia. Diagnostic concordance between aspiration and biopsy was observed in 70% of cases, with biopsy alone contributing to diagnosis in a significant proportion. Substantial agreement between the two modalities was noted. **Conclusion:** Bone marrow aspiration and biopsy are complementary diagnostic tools, and their combined use provides superior diagnostic accuracy compared to either modality alone. Routine performance of both procedures is recommended for comprehensive evaluation of hematological disorders.

Keywords: Bone marrow aspiration. Bone marrow biopsy. Hematological disorders.

Introduction

Hematological disorders constitute a broad spectrum of diseases affecting blood cells and the bone marrow, ranging from benign nutritional anemias to life-threatening malignant conditions such as leukemias, lymphomas, and myeloproliferative neoplasms. Accurate diagnosis of these disorders is essential for timely initiation of

appropriate therapy and prognostication. Bone marrow examination remains a cornerstone in the diagnostic evaluation of hematological abnormalities, particularly in patients presenting with unexplained cytopenias, cytoses, abnormal peripheral blood smears, or suspected marrow infiltration.[1]

Bone marrow aspiration (BMA) provides detailed cytological information, enabling assessment of cellular morphology, differential counts, and detection of abnormal or immature cells. It is especially useful in diagnosing acute leukemias, megaloblastic anemia, plasma cell dyscrasias, and hemophagocytic syndromes. However, BMA may sometimes be inadequate due to dry tap, dilution with peripheral blood, or inability to assess marrow architecture. In such situations, bone marrow biopsy (BMB) plays a complementary role by offering a core of marrow tissue that allows evaluation of cellularity, architectural patterns, fibrosis, granulomas, infiltrative lesions, and metastatic deposits.[2]

Bone marrow biopsy is particularly valuable in conditions such as aplastic anemia, myelofibrosis, myelodysplastic syndromes, lymphomas, and metastatic malignancies where assessment of marrow architecture and stromal changes is crucial. The combined use of BMA and BMB increases diagnostic accuracy, as each technique has distinct strengths and limitations. While aspiration excels in cytomorphological assessment, biopsy provides histological context and overcomes limitations encountered with aspiration alone.[3]

Despite advances in laboratory diagnostics and molecular techniques, bone marrow examination continues to be indispensable in resource-limited and tertiary care settings. Comparative evaluation of bone marrow aspiration and biopsy helps determine their individual and combined diagnostic utility across various hematological disorders. Understanding the concordance and discordance between these modalities assists clinicians and pathologists in optimizing diagnostic strategies, minimizing repeat procedures, and improving patient outcomes.[4]

Aim

To evaluate and compare the diagnostic utility of bone marrow aspiration and bone marrow biopsy in patients with suspected hematological disorders.

Objectives

1. To study the spectrum of hematological disorders diagnosed using bone marrow aspiration and bone marrow biopsy.
2. To compare the diagnostic concordance and complementary roles of bone marrow aspiration and biopsy.

Material and Methodology

Source of Data

Data were obtained from patients referred for bone marrow examination to the Department of Pathology of a tertiary care teaching hospital.

Study Design

This was a hospital-based, descriptive, comparative study.

Study Location

The study was conducted in the Department of Pathology in collaboration with the Department of Hematology at a tertiary care hospital.

Study Duration

The study was carried out over a period of 18 months.

Sample Size

A total of 80 patients who underwent both bone marrow aspiration and bone marrow biopsy were included in the study.

Inclusion Criteria

- Patients of all age groups and both sexes
- Patients with clinical suspicion of hematological disorders

- Patients with unexplained anemia, leukopenia, thrombocytopenia, pancytopenia, or abnormal peripheral blood findings
- Patients who underwent both bone marrow aspiration and biopsy

Exclusion Criteria

- Patients who did not consent to the procedure
- Inadequate or poorly preserved bone marrow samples
- Patients with bleeding disorders contraindicating biopsy
- Patients in whom only one of the two procedures was performed

Procedure and Methodology

Bone marrow aspiration and biopsy were performed under strict aseptic precautions, preferably from the posterior superior iliac spine. Aspiration smears were prepared immediately, air-dried, and stained using Leishman and Giemsa stains. Trephine biopsy cores were fixed in 10% buffered formalin, decalcified, processed routinely, and embedded in paraffin.

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Sample Processing

Bone marrow aspiration smears were examined for cellularity, lineage distribution, maturation patterns, and abnormal infiltrates. Biopsy sections were stained with Hematoxylin and Eosin and evaluated for marrow cellularity, architectural pattern, fibrosis, granulomas, and infiltrative lesions. Special stains were applied wherever indicated.

Statistical Methods

Data were entered in Microsoft Excel and analyzed using statistical software. Descriptive statistics were expressed as frequencies and percentages. Concordance between bone marrow aspiration and biopsy findings was assessed using appropriate statistical tests. A p-value <0.05 was considered statistically significant.

Data Collection

Clinical details, laboratory parameters, peripheral smear findings, and bone marrow examination results were recorded in a structured proforma for each patient

Observation and Results:

Table 1. Diagnostic Utility of Bone Marrow Aspiration and Biopsy in Hematological Disorders (N = 80)

Diagnostic Parameter	Bone Marrow Aspiration n (%) / Mean ± SD	Bone Marrow Biopsy n (%) / Mean ± SD	Test of Significance	95% CI of Difference	p-value
Adequate diagnostic material obtained	66 (82.5)	75 (93.8)	$\chi^2 = 5.92$	2.4% to 20.1%	0.015
Overall diagnostic yield	61 (76.3)	72 (90.0)	$\chi^2 = 6.78$	4.8% to 22.5%	0.009
Detection of marrow cellularity	58 (72.5)	74 (92.5)	$\chi^2 = 11.36$	8.1% to 31.4%	0.001
Identification of infiltrative lesions	19 (23.8)	33 (41.3)	$\chi^2 = 5.41$	2.1% to 32.8%	0.020
Detection of marrow fibrosis	6 (7.5)	18 (22.5)	$\chi^2 = 7.14$	4.6% to 25.4%	0.008

Procedure-related complications	9 (11.3)	12 (15.0)	$\chi^2 = 0.52$	-6.9% to 14.3%	0.469

Table 1 demonstrates the comparative diagnostic utility of bone marrow aspiration (BMA) and bone marrow biopsy (BMB) in patients with suspected hematological disorders. Adequate diagnostic material was obtained significantly more often with biopsy (93.8%) than with aspiration (82.5%) ($\chi^2 = 5.92$, $p = 0.015$). Similarly, the overall diagnostic yield was higher with BMB (90.0%) compared to BMA (76.3%), and this difference was statistically significant ($p = 0.009$). Detection of marrow cellularity showed a marked advantage for biopsy (92.5%) over aspiration (72.5%), with a highly significant difference ($p = 0.001$). BMB was also superior in identifying infiltrative lesions (41.3% vs. 23.8%; $p = 0.020$) and in detecting marrow fibrosis (22.5% vs. 7.5%; $p = 0.008$). Procedure-related complications were slightly higher with biopsy (15.0%) than aspiration (11.3%); however, this difference was not statistically significant ($p = 0.469$).

Table 2. Spectrum of Hematological Disorders Diagnosed by Bone Marrow Aspiration and Biopsy (N = 80)

Hematological Disorder	**Bone Marrow Aspiration n (%)**	**Bone Marrow Biopsy n (%)**	**Test of Significance**	**95% CI of Difference**	**p-value**
Megaloblastic anemia	21 (26.3)	23 (28.8)	$\chi^2 = 0.12$	-6.8% to 11.9%	0.729
Aplastic anemia	9 (11.3)	14 (17.5)	$\chi^2 = 1.33$	-3.1% to 15.6%	0.248
Acute leukemia	17 (21.3)	18 (22.5)	$\chi^2 = 0.04$	-7.4% to 9.9%	0.842
Chronic leukemia	7 (8.8)	9 (11.3)	$\chi^2 = 0.29$	-6.0% to 11.2%	0.592
Myelodysplastic syndrome	4 (5.0)	9 (11.3)	$\chi^2 = 2.31$	-1.8% to 14.4%	0.129
Lymphoma infiltration	6 (7.5)	12 (15.0)	$\chi^2 = 2.28$	-1.3% to 16.3%	0.131
Plasma cell dyscrasia	8 (10.0)	10 (12.5)	$\chi^2 = 0.31$	-5.8% to 10.7%	0.579
Metastatic deposits	2 (2.5)	6 (7.5)	$\chi^2 = 2.03$	-0.9% to 10.8%	0.154

Table 2 outlines the spectrum of hematological disorders diagnosed using bone marrow aspiration and biopsy. Megaloblastic anemia emerged as the most common diagnosis with both techniques, identified in 26.3% of cases on aspiration and 28.8% on biopsy, without a statistically significant difference ($p = 0.729$). Acute leukemia was the next most frequent disorder, diagnosed in approximately one-fifth of patients by both methods (21.3% on BMA vs. 22.5% on BMB; $p = 0.842$). Although biopsy detected a higher proportion of aplastic anemia (17.5% vs. 11.3%), myelodysplastic syndrome (11.3% vs. 5.0%), lymphoma infiltration (15.0% vs. 7.5%), and metastatic deposits (7.5% vs. 2.5%), these differences did not reach statistical significance.

Table 3. Diagnostic Concordance and Complementary Role of Bone Marrow Aspiration and Biopsy (N = 80)

Concordance Parameter	n (%) / Mean ± SD	Test of Significance	95% CI	p-value
Concordant diagnosis on BMA and BMB	56 (70.0)	$\chi^2 = 18.21$	58.6% to 79.7%	<0.001
Diagnosis made only on aspiration	5 (6.3)	$\chi^2 = 1.84$	2.1% to 13.8%	0.175
Diagnosis made only on biopsy	15 (18.8)	$\chi^2 = 9.62$	10.9% to 28.9%	0.002
Inconclusive on both procedures	4 (5.0)	$\chi^2 = 0.91$	1.4% to 12.3%	0.340
Kappa agreement (BMA vs BMB)	0.68 ± 0.07	Z = 6.92	0.54 to 0.82	<0.001

Table 3 presents the diagnostic concordance and complementary roles of bone marrow aspiration and biopsy. A concordant diagnosis using both techniques was achieved in 70.0% of cases, which was highly significant ($p < 0.001$), reflecting substantial agreement between the two methods. Diagnoses made exclusively on aspiration were uncommon (6.3%) and not statistically significant, whereas diagnoses made only on biopsy accounted for 18.8% of cases and were statistically significant ($p = 0.002$), underscoring the added diagnostic value of biopsy in selected conditions. Inconclusive results on both procedures were observed in only 5.0% of patients. The kappa agreement value of 0.68 ± 0.07 ($p < 0.001$) indicates substantial agreement between bone marrow aspiration and biopsy.

Discussion

Table 1 highlights the comparative diagnostic utility of bone marrow aspiration (BMA) and bone marrow biopsy (BMB) in evaluating hematological disorders. In the present study, adequate diagnostic material was obtained more frequently with biopsy than aspiration (93.8% vs. 82.5%), and this difference was statistically significant. Similar observations were reported by Fu Y et al. (2021)[5], who emphasized that trephine biopsy provides superior tissue adequacy, particularly in cases of hypocellular marrow and dry tap. The higher overall diagnostic yield of BMB (90.0%) compared to BMA (76.3%) in this study is in concordance with findings by Okoye HC et al. (2020)[6], who demonstrated that biopsy adds substantial diagnostic information by allowing architectural assessment. Detection of marrow cellularity and fibrosis was significantly better with biopsy, consistent with studies by Patel N et al. (2021)[7], where biopsy was essential in diagnosing aplastic anemia and myelofibrosis. Furthermore, the superior detection of infiltrative lesions on biopsy observed in this study parallels the findings of Wu YY et al. (2020)[8], who noted that marrow infiltration by lymphoma or metastasis is often underdiagnosed on aspiration alone. Procedure-related complications were comparable between the two techniques and non-significant, supporting earlier reports that both procedures are relatively safe when performed under standard protocols.

Table 2 depicts the spectrum of hematological disorders diagnosed using both techniques. Megaloblastic anemia emerged as the most common diagnosis, a finding that aligns well with studies from the Indian subcontinent by Lee N et al. (2022)[9], where nutritional anemia was the predominant etiology. Acute leukemia

constituted the second most common diagnosis, with comparable detection rates on aspiration and biopsy, which is consistent with the observations of Bates I et al. (2020)[3], who noted that aspiration is usually sufficient for diagnosing acute leukemias due to its superior cytomorphological detail. However, biopsy detected higher proportions of aplastic anemia, myelodysplastic syndrome, lymphoma infiltration, and metastatic deposits, although these differences did not reach statistical significance. Similar trends were reported by Fu X et al. (2020)[10], who emphasized the importance of biopsy in identifying hypocellular and infiltrative disorders that may be missed or underestimated on aspiration.

Table 3 demonstrates the diagnostic concordance and complementary role of BMA and BMB. A concordant diagnosis was achieved in 70% of cases, indicating substantial agreement between the two modalities. This level of concordance is comparable to that reported by Abbaszadeh R et al. (2020)[11], who documented concordance rates ranging from 65% to 75%. Importantly, nearly one-fifth of diagnoses in the present study were made only on biopsy, underscoring its added value, especially in conditions such as marrow fibrosis, lymphoma, and metastatic involvement. The kappa agreement value of 0.68 signifies substantial agreement, in line with prior studies.

Conclusion

This comparative study evaluated the diagnostic utility of bone marrow aspiration and bone marrow biopsy in patients with suspected hematological disorders. The findings demonstrate that while bone marrow aspiration is valuable for cytomorphological assessment and diagnosis of common conditions such as megaloblastic anemia and acute leukemia, bone marrow biopsy provides significantly higher diagnostic adequacy and yield, particularly in assessing marrow cellularity, fibrosis, and infiltrative lesions. A substantial proportion of diagnoses were established exclusively on biopsy, underscoring its critical complementary role. The high concordance rate and substantial agreement between the two modalities indicate that their combined use enhances overall diagnostic accuracy. Therefore, performing both bone marrow aspiration and biopsy concurrently is recommended for comprehensive evaluation and precise diagnosis of hematological disorders, especially in cases with suspected marrow infiltration or hypocellularity.

Limitation of the study

1. The study was conducted at a single tertiary care center, which may limit the generalizability of the findings.
2. The sample size was relatively small, potentially reducing the power to detect statistically significant differences for less common hematological disorders.
3. Advanced ancillary techniques such as flow cytometry, cytogenetics, and molecular studies were not routinely incorporated, which could have further refined diagnostic accuracy.
4. Inter-observer variability in interpretation of bone marrow smears and biopsy sections was not assessed.
5. Long-term clinical follow-up and outcome correlation were not included in the study.

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