Section: Pathology

Original Research Article

HISTOPATHOLOGICAL VERSUS CYTOLOGICAL EVALUATION OF BONE MARROW: A STUDY ON THE CONCORDANCE BETWEEN ASPIRATION AND TREPHINE BIOPSY

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ABSTRACT

Background: Bone marrow evaluation (BME) is crucial in testing haematological and certain non-haematological conditions. The bone marrow evaluation not only confirm clinically suspected but also previously unsuspected diagnoses. The cytological preparation of bone marrow, which is obtained by aspiration, reveals cellular morphology and allows ancillary testing (flow cytometry, molecular genetics) and the histological sample, usually obtained with a Jamshidi needle, allows optimal evaluation of cellularity, fibrosis or infiltrative disease. Bone marrow aspirate and trephine biopsy specimens nowadays are considered complementary and when both are obtained, they provide a thorough examination of bone marrow.

Materials and Methods: During the study period of 1 year 111 BMEs, both bone marrow aspirations and BMB were performed for various indications.

Results: The mean age was 40.6 years of these, the Male: Female ratio was 1.3:1. In this study comprising 111 patients, the majority were in the 21–40 years age group (42 cases, 37.8%), followed by the 41–60 years group (28 cases, 25.2%), and the 2–20 years group (24 cases, 21.6%). The least representation was from the 61–80 years age group (17 cases, 15.3%). A slight male predominance was observed overall, with 62 males (55.9%) and 49 females (44.1%). Assessment of bone marrow cellularity revealed that normocellular marrow was most common (42%), followed by hypercellular (34%), hypocellular (19%), and diluted samples (16%).

Conclusion: This study reaffirms that while aspiration remains a valuable diagnostic tool, trephine biopsy is indispensable for a comprehensive assessment especially in complex, inconclusive, or architecturally dependent marrow pathologies. For optimal diagnostic accuracy and patient care, the combined use of both BMA and BMB should remain the standard practice.

Keywords: Bone Marrow Aspiration, Bone Marrow Biopsy, Leukaemia.

INTRODUCTION

Haematological disorders are now quite common in general population. The Haematological disorders may range from simple conditions like iron deficiency anemia to infiltrative carcinomas involving the bone marrow. Most of these haematological disorders require bone marrow aspiration examination for definitive diagnosis.[1]

Bone marrow evaluation is pivotal in diagnosing haematological and certain non haematological conditions.[1] Sometimes, the haematological disorders present with ambiguous symptoms. The clinicians find it difficult to reach to final diagnosis in such cases. Bone marrow aspiration and biopsy plays a vital prognostic tool and a crucial part of the follow-up in patients receiving chemotherapy or bone marrow transplants.[2] The bone marrow evaluation may either confirm clinically suspected or previously unsuspected diagnoses.[3,4]

The bone marrow examination helps with the diagnosis and management of many hematologic

diseases, which usually involves two separate specimens: a cytologic and a histologic preparation. The cytological preparation of bone marrow, which is obtained by aspiration, reveals cellular morphology and allows ancillary testing (flow cytometry, molecular genetics), and the histological sample, usually obtained with a Jamshidi needle, allows optimal evaluation of cellularity, fibrosis or infiltrative disease.[5]

Bone marrow aspirate and trephine biopsy specimens nowadays are considered complementary and when both are obtained, they provide a thorough examination of bone marrow.[6]

In haematology, examining bone marrow is essential for the differential diagnosis of various haematological disorders such as acute leukaemia, CLL, hairy cell leukaemia, myeloproliferative disorders like polycythaemia vera (PV), essential thrombocythemia (ET), primary myelofibrosis (PMF), and chronic myelogenous leukaemia (CML), plasma cell dyscrasias like multiple myeloma (MM), staging of lymphomas, and marrow infiltration by foreign cells.[7-11]

When all other clinical diagnostic tools have been done with bone marrow examination (BME) is the final endpoint to the diagnosis of many haematological and non-haematological disorders.[12]

Initially, aspiration techniques were more popular in view of the simplicity of the procedure, good representation of marrow cells' morphology and ready acceptability by the patient. However, knowledge about diagnostic limitations of aspirated marrow, coupled with introduction of simplified percutaneous needle biopsy procedures (under local anaesthesia) led to a progressive increase in the use of bone marrow trephine biopsy as an indispensable adjunct to bone marrow aspirations.[13]

MATERIALS AND METHODS

The study was a retrospective study conducted in the Department of Pathology. All case records of bone marrow examination were retrieved, and the bone marrow findings were reviewed from Jan 2024 to Dec 2024; and those diagnosed as infections were included in the study. The present study was conducted in Dept of pathology. All reports of BM were retrieved and BM findings were reviewed from Jan 2024 to Dec 2024 wherein total 118 BM aspirate and 118 BM biopsies were reviewed. In 111 cases, both bone marrow aspiration and bone marrow biopsy were performed simultaneously.

The site for Bone marrow aspiration (BMA) and Bone marrow biopsy (BMB) was the posterior superior iliac spine. Jamshidi bone marrow biopsy/aspiration needle with Luer lock adapter was used, 13-gauge needle for lower age group and 11-gauge needle for higher age groups.

BMA slides were stained with Leishman and Giemsa stain. The BMB was fixed in 10% neutral buffered formalin for overnight. Biopsies underwent tissue decalcification and tissue processing followed by staining with haematoxylin and eosin stain.

Inclusion criteria

The study subjects consisted of clinically suspected cases of either haematological or non-haematological disorders in the age group between 2 and 80 years. Children and adolescents were included in this study.

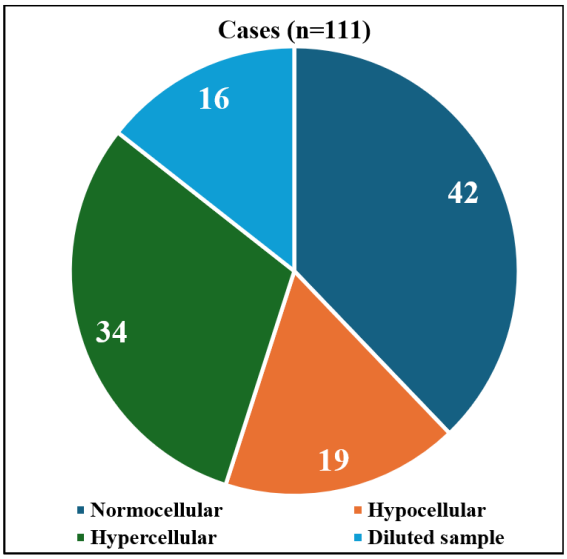
Exclusion criteria

Patients with bleeding or coagulation disorders, ages less than two years or more than 80 years, and terminally ill patients were excluded.

Patients who did not undergo bone marrow biopsy due to a deranged coagulation profile were excluded from the study.

The reports of both aspirate and biopsy for each case were reviewed and compared. The reasons for inconclusive reports were also recorded.

RESULTS



Graph 1: Pie chart showing distribution of cases according to cellularity of marrow.

Table 1: Diagnostic findings of correlation between bone marrow aspirate and bone marrow biopsy (n=111).

S.No.	Diagnosis	Complete correlation	No correlation	Total (n=111)
1	AML	08	03	11
2	CML	10	00	10
3	ALL	05	02	07
4	CLL	01	00	01
5	Megaloblastic anemia	02	00	02
6	Iron deficiency anemia	0	0	0
7	Aplastic anemia	0	0	0
8	Erythroid hyperplasia	10	1	11
9	Plasma cell dyscrasias	01	0	01
10	Hypoplastic marrow	05	01	06

11	Normal marrow	08	00	08
12	ITP	01	0	01
13	Micronormoblastic erythropoiesis	13	02	15
14	Normoblastic erythropoiesis	17	01	18
15	Macronormoblastic erythropoiesis	01	00	01
16	Megaloblastic erythropoiesis	02	00	02
17	Hypercellular marrow	01	00	01
18	Dyserythropoiesis	01	00	01
19	Inadequate for opinion	10	05	15

(AML: Acute myeloblastic leukaemia, CML: Chronic myeloid leukaemia, ALL- Acute lymphoblastic leukaemia, CLL – Chronic lymphocytic leukaemia, ITP: Immune thrombocytopenic purpura)

Table 2: Age and sex distribution of cases (n= 111)

S. No.	Age	Male	Female	Total
1	2-20 yr	13	11	24
2	21-40yrs	24	18	42
3	41-60 yrs	15	13	28
4	61-80 yrs	10	07	17

Table 3: Gender-wise distribution of bone marrow cellularity

S. No.	Variables	Cases (n=111)	Male	Female	Percentage%
1.	Normocellular	42	23	17	38
2.	Hypocellular	19	10	8	17
3.	Hypercellular	34	18	14	31
4	Diluted sample	16	9	6	14

Table 4: Comparison of the diagnostic efficacy of BMA and BMB with the previous studies.

	Diagnostic efficacy	
Name of the study	**Bone marrow aspiration (%)**	**Bone marrow biopsy (%)**
Aljadayeh et al. [14]	72.6	98.8
Gilotra et al. [15]	72.4	83.9
Khan et al. [16]	73.8	99
Chandra and Chandra [17]	77.5	99.2

BMA: Bone marrow aspirate, BMB: Bone marrow biopsies

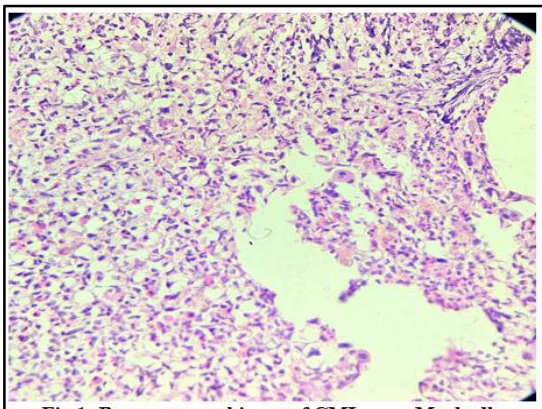


Fig 1: Bone marrow biopsy of CML case. Markedly hypercellular marrow. Predominance of cells of myeloid lineage seen.

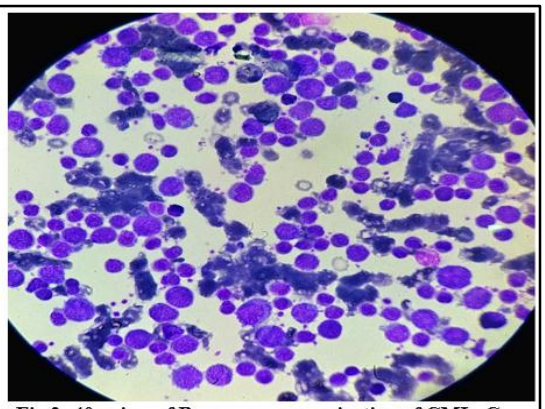


Fig 2: 40x view of Bone marrow aspiration of CML. Case showing high cellularity and cells of myeloid cells and basophil.

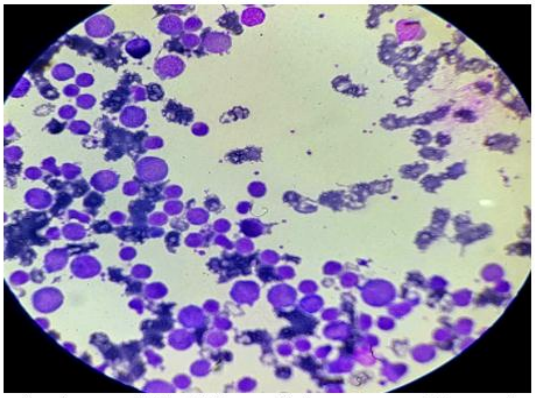


Fig 3: 100x view of CML case showing myeloblast. Hypercellular marrow with granulocytic proliferation. High M:E ratio. Blasts <10% (chronic phase)

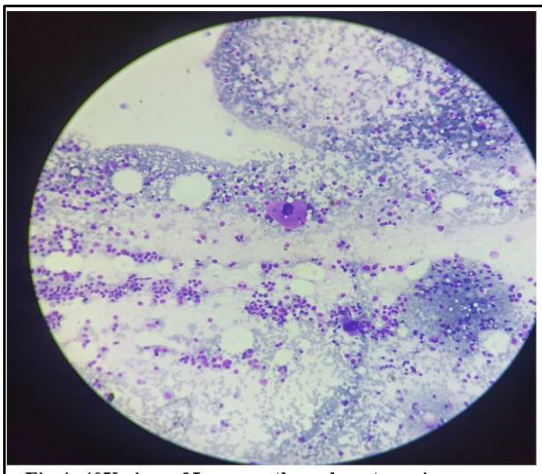


Fig 4: 40X view of Immune thrombocytopenic purpura (ITP), showing megakaryocytes.

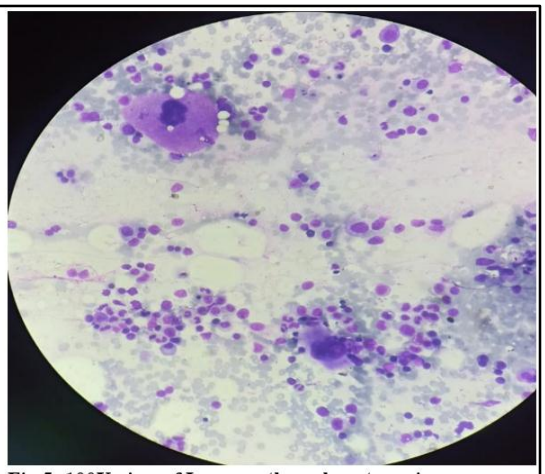


Fig 5: 100X view of Immune thrombocytopenic purpura (ITP), showing large lobulated megakaryocytes.

DISCUSSION

Bone marrow aspiration and biopsy are indispensable and often complementary procedures in haematology. While aspiration offers detailed cytological analysis, biopsy enables evaluation of the structural integrity and cellular distribution of the marrow. Each technique has its own diagnostic strengths and limitations. The choice of procedure should be guided by clinical indications, suspected pathology, and the likelihood of obtaining diagnostic material. In most settings, performing both procedures concurrently ensures the highest diagnostic yield and minimizes the need for repeat interventions. A high degree of diagnostic concordance was observed in conditions such as normoblastic erythropoiesis, micronormoblastic erythropoiesis, erythroid hyperplasia and chronic myeloid leukaemia, supporting the adequacy of cytology in many routine marrow evaluations. Complete correlation was found in cases of chronic myeloid leukaemia. Correlation of bone marrow aspiration and biopsy was found in 78.05 % of cases and non –correlated cases were 21.9%. However, discordance was noted in cases of acute leukaemia, megaloblastic anemia and those labelled "inadequate for opinion" on aspiration, emphasizing the limitations of aspiration alone in cases with poor yield or architectural distortion. [Table 1]

During the study period of 1 year 111 BMEs, both bone marrow aspirations and BMB were performed for various indications. The mean age was 40.6 years of these, the Male: Female ratio was 1.3:1.

In this study comprising 111 patients, the majority were in the 21–40 years age group (42 cases, 37.8%), followed by the 41–60 years group (28 cases, 25.2%), and the 2–20 years group (24 cases, 21.6%). The least representation was from the 61–80 years age group (17 cases, 15.3%). A slight male predominance was observed overall, with 62 males (55.9%) and 49 females (44.1%). [Table 2]

Assessment of bone marrow cellularity revealed that normocellular marrow was most common (42%), followed by hypercellular (34%), hypocellular (19%), and diluted samples (16%).

The presence of diluted or hypocellular samples particularly affected the reliability of aspiration, reinforcing the diagnostic superiority of trephine biopsy in such scenarios. [Table 3]

Our study is consistent with the studies of Aljadayeh et al,[14] Gilotra et al,[15] Khan et al,[16] Chandra and Chandra.[17] In their study they found that BMB is diagnostically more efficient than BMA. The diagnostic efficacy in our study is 86.5%.

BMA and BMB are crucial diagnostic tools for identifying various haematological and non-haematological disorders. These procedures are also valuable for follow-up in patients undergoing chemotherapy and other medical treatments, including bone marrow transplantation.[9,10] It is well-established that BMA and BMB are complementary procedures. Nowadays, it is customary to collect both specimens simultaneously, preferably from the same location.[11] Inadequate for opinion resulted from dry tap due to fibrosis, haemodilution, inadequate bone marrow sampling and some technical errors.

CONCLUSION

In conclusion, bone marrow aspiration (BMA) remains an indispensable initial diagnostic modality for the evaluation of haematological and certain non-haematological disorders. It provides rapid and detailed cytomorphological assessment, facilitating early diagnosis and therapeutic planning. However, its diagnostic yield may be limited in conditions such as myelofibrosis, focal marrow infiltration, granulomatous lesions, metastatic deposits, and in cases of haemodilution or dry tap.

Bone marrow biopsy (BMB), particularly trephine biopsy, overcomes these limitations by enabling comprehensive assessment of marrow architecture, overall cellularity, stromal alterations, fibrosis grading, and focal pathological involvement. It plays a decisive role in diagnostically challenging cases where aspiration findings are inadequate,

inconclusive, or discordant with clinical and laboratory parameters.

The findings of the present study reaffirm that BMA and BMB are complementary procedures rather than alternatives. The combined interpretation of cytological and histological features significantly enhances diagnostic accuracy, improves clinico-pathological correlation, and strengthens prognostic assessment. Therefore, the integrated use of both aspiration and biopsy should continue to be regarded as the gold standard for thorough bone marrow evaluation in routine practice as well as in complex haematological disorders.

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