TPM Vol. 32, No. S3, 2025
ISSN: 1972-6325
https://www.tpmap.org/

COMPARATIVE EVALUATION OF BONE MARROW ASPIRATION AND TREPHINE BIOPSY: INSIGHTS FROM AN OBSERVATIONAL STUDY

PRABAKARAN B[2], AKSHAYA VISWANATHAN[1], NEHA BRAHMA[1], VIMAL S[1]*, DR. ISHWARYA DHEVI[3]

[1]DEPARTMENT OF BIOCHEMISTRY, SAVEETHA MEDICAL COLLEGE, SAVEETHA INSTITUTE OF MEDICAL AND TECHNICAL SCIENCES, SAVEETHA UNIVERSITY, CHENNAI, TAMIL NADU, INDIA.
[2]DEPARTMENT OF PATHOLOGY, SAVEETHA MEDICAL COLLEGE, SAVEETHA INSTITUTE OF MEDICAL AND TECHNICAL SCIENCES, SAVEETHA UNIVERSITY, CHENNAI, TAMIL NADU, INDIA.
[3]SENIOR LECTURER, DEPARTMENT OF PEDIATRIC DENTISTRY, SREE BALAJI DENTAL COLLEGE & HOSPITAL, CHENNAI, INDIA
CORRESPONDING AUTHOR *
DEPARTMENT OF BIOCHEMISTRY, SAVEETHA MEDICAL COLLEGE & HOSPITAL, SAVEETHA INSTITUTE OF MEDICAL AND TECHNICAL SCIENCES (SIMATS), THANDALAM, CHENNAI - 602105, TAMIL NADU, INDIA.

Abstract

Bone marrow aspiration (BMA) and bone marrow trephine biopsy (BMB) are crucial diagnostic procedures in hematology, providing complementary insights into marrow pathology. BMA offers detailed cytological evaluation and facilitates ancillary studies, while BMB assesses marrow architecture, stromal alterations, fibrosis, and focal infiltrates. BMA identifies pathology in cases where BMA may be limited, while BMB detects early cytological abnormalities missed by BMB. The combined use of both techniques increases diagnostic yield to over 95%. Disease-specific considerations influence procedural choice, with BMA preferred in acute leukemias and BMB in myeloproliferative neoplasms.

Keywords

Bone marrow trephine biopsy, Hematological diagnosis, Cytology, Marrow architecture, Myelofibrosis, Metastatic disease

Bone marrow examination has long been a cornerstone in the evaluation and management of hematological disorders, offering crucial insights into marrow architecture, cellularity, and disease processes (1). Among the various diagnostic techniques available, bone marrow aspiration (BMA) and bone marrow trephine biopsy (BMB) are the most widely employed and often performed together to provide a comprehensive evaluation. While both aim to investigate the hematopoietic compartment, their diagnostic strengths, limitations, and applicability differ significantly (2). BMA, first introduced as a clinical procedure in the early twentieth century, involves the aspiration of liquid marrow, usually from the posterior superior iliac spine, to obtain cellular material for cytological examination. This allows for detailed evaluation of hematopoietic cell morphology, assessment of differential counts, detection of blast cells, and evaluation of iron stores (3). Additionally, aspirated samples can be subjected to ancillary investigations such as flow cytometry, cytogenetic analysis, and molecular testing, enabling precise disease classification and prognostication. However, the technique is not without its limitations, as it can be affected by hemodilution and may fail to detect focal lesions, patchy infiltrates, or significant marrow fibrosis (4). In contrast, trephine biopsy provides a cylindrical core of bone and marrow tissue, enabling histological assessment of the marrow architecture, spatial distribution of hematopoietic elements, and evaluation of stromal and fibrotic changes (5) . BMB is particularly advantageous in detecting focal or patchy infiltrates, metastatic tumor deposits, granulomatous inflammation, and myelofibrosis, all of which may be missed on aspiration smears. It also provides reliable information on overall marrow cellularity, which can be variably represented in aspirate smears. However, trephine biopsy requires specialized equipment, careful handling, and histopathological processing, which can prolong turnaround time compared to BMA. Despite these differences, both techniques are regarded as complementary, and simultaneous performance often yields the most accurate diagnostic outcome (6).

The difference between normal bone homeostasis and the vicious cycle of bone metastasis. Under healthy conditions, bone homeostasis is maintained by a balance between bone formation by osteoblasts (OB) and bone resorption by osteoclasts (OC). However, in bone metastasis, metastatic cancer cells disrupt this balance by inducing osteoblasts to secrete RANKL (Receptor Activator of Nuclear Factor κB Ligand), which binds to RANK on osteoclasts, increasing their proliferation and activity. This heightened osteoclast activity accelerates bone resorption, releasing growth factors such as IGF-1 and TGF-beta from the bone matrix, which in turn promote further tumor growth, creating a self-perpetuating cycle where bone formation is outweighed by bone resorption (figure 1) (7) (8).

Conversely, aspiration detected early cytological abnormalities in acute leukemias that were not apparent in the trephine sections, underscoring its superior cellular detail. Similar studies have shown that while BMA alone achieves a diagnostic accuracy of around 75-80% and BMB alone achieves around 85%, the combined approach can increase diagnostic yield to over 95%, making the practice of performing both in tandem clinically justifiable in most cases (9). The choice between BMA and BMB, however, can be influenced by the suspected diagnosis. In acute leukemia, for example, aspiration is preferred for its morphological clarity and suitability for immunophenotyping and genetic profiling, while trephine biopsy may offer limited added value in early disease stages (10). In myeloproliferative neoplasms and myelofibrosis, on the other hand, trephine biopsy is indispensable for assessing stromal changes and grading fibrosis. Similarly, in cases of suspected metastatic carcinoma or granulomatous diseases, the architectural preservation in BMB enables more sensitive detection of focal lesions and localized granulomas. These differences make it clear that both procedures have unique niches in hematopathology, and optimal diagnostic strategies often involve tailoring the use of one or both techniques to the clinical context (11).

While the existing literature provides valuable insights, most comparative studies are observational in nature and are subject to limitations such as sampling bias, operator variability, and differences in institutional diagnostic criteria (12). Nevertheless, advances in diagnostic technology are beginning to bridge the gaps between the two methods. For instance, artificial intelligence assisted digital morphology is improving the efficiency and accuracy of cytological interpretation from aspirates, while innovations in spatial transcriptomics and multiplex imaging are enhancing the molecular and immunohistochemical utility of trephine biopsy specimens. Additionally, minimally invasive micro-biopsy devices are being explored to reduce patient discomfort while retaining the architectural benefits of a core sample, which could further refine diagnostic workflows in the future (13) (14).

In conclusion, bone marrow aspiration and trephine biopsy are indispensable and complementary diagnostic tools, each offering distinct yet overlapping contributions to the diagnosis and management of haematological and metastatic diseases. Observational data consistently emphasize that the highest diagnostic yield is achieved when both are used in combination, with aspiration providing rapid and detailed cytological information, and trephine biopsy delivering comprehensive architectural and stromal evaluation. The integration of morphological, immunophenotypic, and molecular data from both techniques, along with emerging technological innovations, holds the promise of improving diagnostic precision, reducing procedural invasiveness, and ultimately enhancing patient outcomes (Table 1).

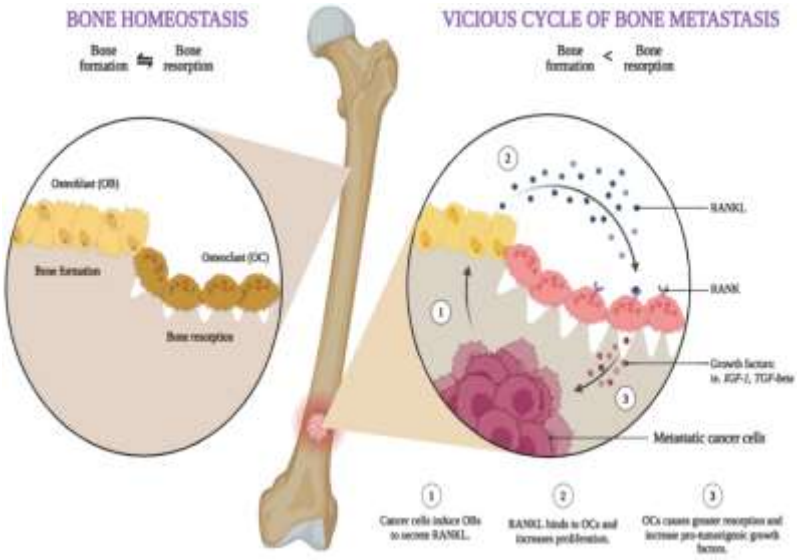

Figure 1. The black double-headed arrow in the left panel represents the balanced state of bone metabolism in normal bone homeostasis, where bone formation and bone resorption occur in equilibrium. In contrast, the black arrow with the smaller-than sign in the right panel indicates the disrupted balance during bone metastasis, where bone formation is reduced and bone resorption predominates, leading to pathological bone degradation.

Parameter	Bone Marrow Aspiration (BMA)	Bone Marrow Trephine Biopsy (BMB)
Sample type	Liquid marrow aspirate for cytological evaluation (1,2,3)	Cylindrical core of bone and marrow tissue for histological assessment (5,6)
Primary diagnostic strength	Detailed hematopoietic cell morphology, detection of blasts, iron stores assessment, and ancillary studies such as flow cytometry, cytogenetics, and molecular testing (1,3,4)	Marrow architecture, cellularity, stromal and fibrotic changes, focal infiltrates, metastatic deposits, granulomas (5,6,11)
Advantages	Rapid results, suitable for immunophenotyping and genetic profiling, higher sensitivity for early cytological abnormalities (e.g., acute leukemias) (3,10)	Superior for detecting patchy lesions, myelofibrosis, architectural changes, and focal infiltrates (5,6,11)
Limitations	May be affected by hemodilution, can miss focal or patchy lesions and significant fibrosis (4,6)	Requires more time for processing, patient discomfort may be higher, less suitable for rapid immunophenotyping (5,6)
Preferred clinical contexts	Acute leukemias, cases requiring rapid cytology and molecular profiling (3,10)	Myeloproliferative neoplasms, myelofibrosis, suspected metastatic carcinoma, granulomatous diseases (5,6,11)
Diagnostic accuracy (alone)	~75–80% (6,9)	~85% (6,9)
Combined diagnostic yield	>95% when both performed together (6,9)	>95% when both performed together (6,9)

Table 1. This table compares Bone Marrow Aspiration (BMA) and Bone Marrow Trephine Biopsy (BMB) in terms of diagnostic features, advantages, limitations, and clinical indications. BMA offers rapid cytology, early blast detection, and molecular study suitability, whereas BMB is essential for architectural assessment and detecting focal infiltrates. Combined use improves diagnostic accuracy to over 95%, underscoring their complementary roles in hematopathology.

REFERENCES

1. Dogan A, Demircioglu S. Diagnostic importance of bone marrow aspiration evaluation: A single-center study. Pak J Med Sci. 2022;38(4Part-II):811-5.
2. Kaur M, Singh Rana AP, Kapoor S, Puri A. Diagnostic value of bone marrow aspiration and biopsy in routine hematology practice. J Clin Diagn Res. 2014;8(8):Fc13-6.
3. Bhaskar N. Bone Marrow Aspiration and Biopsy in Critical Pediatric Patients: A Pathologist's Perspective. Cureus. 2021;13(8):e17423.
4. Gabali A. Flow cytometry, molecular analysis, and other special techniques (in Serous Fluid Cytopathology). Cytojournal. 2022;19:18.
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.
6. Gilotra M, Gupta M, Singh S, Sen R. Comparison of bone marrow aspiration cytology with bone marrow trephine biopsy histopathology: An observational study. J Lab Physicians. 2017;9(3):182-9.

7. Riquelme MA, Cardenas ER, Jiang JX. Osteocytes and Bone Metastasis. Front Endocrinol (Lausanne). 2020;11:567844.
8. Sharma. K P. Mandibular (Gnathic) osteosarcoma: Narrative review emphasizing clinical, radiological, and histopathological correlation. Oral Oncology Reports. 2024;10:100479.
9. .
10. Ansari Z, Chaurasia A, Neha, Kalani A, Bachheti RK, Gupta PC. Comprehensive insights into leishmaniasis: From etiopathogenesis to a novel therapeutic approach. Microb Pathog. 2025;204:107535.
11. Zahr AA, Salama ME, Carreau N, Tremblay D, Verstovsek S, Mesa R, et al. Bone marrow fibrosis in myelofibrosis: pathogenesis, prognosis and targeted strategies. Haematologica. 2016;101(6):660-71.
12. Song JW, Chung KC. Observational studies: cohort and case-control studies. Plast Reconstr Surg. 2010;126(6):2234-42.
13. Giansanti D. Advancements in Digital Cytopathology Since COVID-19: Insights from a Narrative Review of Review Articles. Healthcare (Basel). 2025;13(6).
14. Muthu S, Visawanathan V, Chellamuthu G, Thabrez M. Clinical Effectiveness of Various Treatments for Cartilage Defects Compared to Microfracture: a Network Meta-analysis of Randomized Controlled Trials. Journal of Cartilage & Joint Preservation. 2023;4:100163.