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Original Research Article

A Retrospective Study of Pancytopenia Based on Diagnostic Reliability of Bone Marrow Aspiration and Biopsy

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Abstract:

Background: Pancytopenia is a common hematological condition characterized by reduction in all three blood cell lines and presents a diagnostic challenge due to its diverse etiologies. Bone marrow aspiration and biopsy are key diagnostic tools used to evaluate its underlying causes.

Aim: To assess the diagnostic reliability and concordance of bone marrow aspiration and biopsy in determining the etiology of pancytopenia.

Methodology: This retrospective observational study was conducted over one year at Department of Pathology, Phulo Jhano Medical College and Hospital, Dumka, Jharkhand, India, including 86 patients with pancytopenia. Clinical, hematological, and bone marrow findings were analyzed. Bone marrow aspiration and biopsy results were compared for diagnostic agreement, and statistical measures such as sensitivity, specificity, and accuracy were calculated.

Results: The majority of patients were young adults (41.9%) with slight male predominance (55.8%). Pallor was the most common clinical feature (100%). Megaloblastic anemia (32.6%) and aplastic anemia (30.2%) were the leading causes. Diagnostic concordance between aspiration and biopsy was observed in 81.4% of cases, while 7% of cases required biopsy for definitive diagnosis. Bone marrow aspiration showed high sensitivity (89.5%), specificity (85%), and overall accuracy (87.2%).

Conclusion: Bone marrow aspiration is a reliable initial diagnostic modality; however, biopsy plays a crucial complementary role, particularly in inconclusive cases, enhancing overall diagnostic accuracy in pancytopenia.

Keywords: Pancytopenia, Bone marrow aspiration, Bone marrow biopsy, Diagnostic reliability, Megaloblastic anemia, Aplastic anemia.

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Introduction

Pancytopenia is a relatively common hematological disorder that is very often seen in clinical practice and is a very difficult diagnosis to be established in clinical practice because of its various etiological spectrum. Defined as the decrease of all three formed elements of blood (red blood cells, white blood cells and platelets) below normal reference range. Pancytopenia is not a disease but merely a haematological feature that can be a result of many underlying pathological processes [1]. These processes may be simple and non- threatening and reversible, or life threatening and require prompt and accurate diagnosis for management and good patient outcomes.

Pancytopenia is a complex and multifactorial pathophysiology. It could be due to a reduction in production of blood cells, destruction or sequestration of blood cells, or both. Reduction in production may be caused by direct damage to bone marrow stem cells by toxins, drugs, infections or radiation or by the replacement of normal marrow by malignant or abnormal infiltrative processes. Or, normal hematopoiesis can be suppressed, leading to the growth and differentiation of precursor cells [2]. Other causes are poor hematopoiesis (poor production of blood cells, which die in the marrow before they are formed), blood cell destruction in the periphery, by immune mechanisms or hyper-splenism, or blood cell sequestration in the spleen or other organs. Under these conditions, antibodies or hyperactive

reticuloendothelial system (RES) can play a part in the destruction of cells or the trapping of normal cells [3].

The bone marrow morphology is of great importance in the understanding of the underlying mechanism of pancytopenia. Marrow is cellular in different diseases. The marrow is generally hypocellular in conditions where there is a primary production defect, including aplastic anemia. Compared with these, the disorders where myeloid production is defective, peripheral destruction is increased or malignancy is present tend to appear hyper or normocellular on the marrow. Thus, bone marrow cellularity is an important parameter that helps to understand the pathogenesis and can further direct the diagnostic work-up [4].

Patients with pancytopenia may have signs and symptoms of anemia, neutropenia, and/or thrombocytopenia. Symptoms of anemia include fatigue, pallor and generalized weakness, and bleeding disorders are typical symptoms of thrombocytopenia, including petechiae, ecchymosis, and mucosal hemorrhages. Leucopenia is not a common presenting symptom, but it makes patients more vulnerable to infections and can be the worst and life-threatening symptom that may appear during the course of the disease [5]. Comprehensive diagnostic evaluation is emphasized by the variable clinical presentation.

The etiological factors of pancytopenia vary widely in their nature and are related to a number of factors such as geographic distribution, nutritional factors, environment, and infectious diseases [6]. For example, folate or vitamin B12 deficiency can cause megaloblastic anemia in some populations but in others, infections, autoimmune diseases and hematological malignancies can be more prevalent. The variability requires a systematic and region-specific approach to diagnosis.

The bone marrow is still a very important component in the workup of pancytopenia. Usually, it is a combination of two complementary procedures: bone marrow aspiration and trephine biopsy. Bone marrow aspiration allows for a detailed cytological evaluation, which includes the ability to assess the morphology of individual cells, to identify abnormal cells, and to detect hematological malignancies. It is, however, sometimes able to provide poor or thin samples, especially when marrow fibrosis or infiltration is present [7]. However, trephine biopsy provides a more detailed picture of marrow architecture, marrow cellularity and focal lesions or infiltrative processes. This is especially helpful in situations where aspiration results are ambiguous or inadequate [8].

Even though bone marrow aspiration and biopsy are widely used tests, the reliability of the diagnostic results can be variable depending on the disease. Biopsy may be better to diagnose focal lesions, fibrosis or granulomatous changes, but aspiration can be very informative in cases with diffuse marrow involvement. A combined approach is, therefore, often more helpful in improving the accuracy of the diagnosis and in gaining a more comprehensive picture of marrow pathology. It is crucial to assess the agreement and diagnostic value of these two methods, to optimize clinical decision-making.

Retrospective studies are useful for revealing trends in distribution of the disease and the efficacy of the diagnostic methods in the heterogenous group of causes and clinical implications of pancytopenia. These kinds of studies are useful for correlation of clinical findings with laboratory and bone marrow examination results, and for the refinement of diagnostic algorithms and care strategies.

The present study is designed to compare the diagnostic reliability of bone marrow aspiration and trephine biopsy in the diagnosis of the underlying causes of pancytopenia. To compare the effectiveness of these diagnostic methods, determine their limitations and emphasise their complementary role in clinical practice, a retrospective analysis of these data is performed. In addition, it aims to give insights into the spectrum of diseases associated with pancytopenia in the investigated population, which will improve the understanding of this complicated hematological disease and facilitate the development of more precise and efficient diagnostic methods.

Methodology

Study Design: This study was a retrospective observational study conducted to evaluate the diagnostic reliability of bone marrow aspiration and biopsy in patients presenting with pancytopenia. The study involved review and analysis of previously recorded clinical, hematological, and bone marrow examination data.

Study Area: The study was carried out in the Department of Pathology, Phulo Jhano Medical College and Hospital, Dumka, Jharkhand, India.

Study Duration: The duration of the study was one year from January 2025 to December 2025.

Sample Size: A total of 86 cases (N = 86) fulfilling the inclusion criteria were included in the study. These cases were selected from departmental records over the defined study period.

Study Population: The study population comprised patients of all age groups and both sexes who presented with pancytopenia and underwent bone marrow aspiration and/or biopsy in the Department of Pathology during the study period. These patients were evaluated for underlying etiologies of pancytopenia.

Data Collection: Data were collected retrospectively from patient case records, laboratory registers,

and bone marrow reports. Information regarding demographic details, clinical presentation, and hematological parameters such as hemoglobin levels, total leucocyte count, and platelet count was obtained. Bone marrow aspiration smears and biopsy slides were retrieved and reviewed. Aspirate smears were stained with Leishman stain, while biopsy sections were stained with Hematoxylin and Eosin. Special stains and immunohistochemistry were applied in selected cases wherever necessary to reach a definitive diagnosis.

Inclusion Criteria

- All patients with pancytopenia defined as:
  - Hemoglobin < 10 g/dL
  - Total leucocyte count < 4000/mm³
  - Platelet count < 150,000/mm³
- Patients who underwent both bone marrow aspiration and/or biopsy
- Cases available with complete hematological and bone marrow records

Exclusion Criteria

- Cases of chemotherapy-induced pancytopenia
- Incomplete or missing medical or laboratory records
- Poor quality or inadequate bone marrow samples

Procedure: All eligible cases were identified from departmental archives and their records were systematically reviewed. Peripheral blood findings were correlated with bone marrow aspiration and biopsy findings. The slides of bone marrow aspirates and biopsies were re-examined to ensure diagnostic accuracy. The findings obtained from bone marrow aspiration were compared with those from biopsy to assess concordance and diagnostic reliability. Final diagnoses were established by integrating clinical features, hematological parameters, and bone marrow findings.

Statistical Analysis: The collected data was compiled and entered into Microsoft Excel and subsequently analyzed using appropriate statistical software such as SPSS. Descriptive statistics, including mean and standard deviation for continuous variables and frequencies and percentages for categorical variables, were calculated. The diagnostic performance of bone marrow aspiration in comparison to biopsy was evaluated using sensitivity, specificity, positive predictive value, and negative predictive value. Agreement between the two diagnostic methods was assessed using the Kappa coefficient, and a p-value of less than 0.05 was considered statistically significant."

Result

Table 1 shows the age distribution of 86 cases, with the majority of patients belonging to the 15–35 years age group, accounting for 36 cases (41.9%). This was followed by the 36–55 years group with 24 cases (27.9%) and those above 55 years with 18 cases (20.9%). The least number of cases were observed in patients below 15 years, comprising 8 cases (9.3%). Overall, the data indicate that the study population was predominantly composed of young adults.

Table 1: Age Distribution of Cases (N = 86)

Age Group (Years)	Number of Cases	Percentage (%)
<15	8	9.3
15–35	36	41.9
36–55	24	27.9
>55	18	20.9
Total	**86**	**100**

Table 2 depicts the gender distribution of the study population comprising 86 cases. Males constituted the majority, with 48 cases (55.8%), while females accounted for 38 cases (44.2%). This indicates a slight male predominance in the study group.

Table 2: Gender Distribution (N = 86)

Gender	Number of Cases	Percentage (%)
Male	48	55.8
Female	38	44.2
Total	**86**	**100**

Table 3 outlines the clinical presentation of 86 cases, with pallor being the most common feature, observed in all patients (100%), indicating its universal presence in the study group. Weakness was the next most frequent symptom, seen in 52 cases (60.5%), followed by fever in 34 cases (39.5%). Hepatosplenomegaly was noted in 22 cases (25.6%), while bleeding tendency was present in 18 cases (20.9%). Overall, the findings suggest that general symptoms like pallor and weakness were predominant, whereas systemic and hemorrhagic manifestations were less commonly observed.

Table 3: Clinical Presentation (N = 86)		
Clinical Feature	**Number of Cases**	**Percentage (%)**
Pallor	86	100
Fever	34	39.5
Weakness	52	60.5
Bleeding tendency	18	20.9
Hepatosplenomegaly	22	25.6

Table 4 presents the spectrum of diagnoses obtained from bone marrow aspiration and biopsy in 86 cases. The most common diagnosis was megaloblastic anaemia, observed in 28 cases (32.6%), followed closely by hypoplastic/aplastic anaemia in 26 cases (30.2%), together accounting for the majority of cases. Normal marrow findings were seen in 10 cases (11.6%). Less frequent diagnoses included myelofibrosis and acute leukemia (each 4.7%), as well as myeloproliferative disorders and erythroid hyperplasia (each 3.5%). Chronic myeloid leukemia, acute lymphoblastic leukemia, and granulomatous lesions each contributed 2.3% of cases, while myelodysplastic syndrome and hemolytic anaemia were the least common, each observed in 1.2% of cases. Overall, the data indicate that nutritional and marrow failure disorders were more prevalent than malignant conditions in this study population.

Table 4: Bone Marrow Aspiration and Biopsy Diagnosis (N = 86)		
Diagnosis	**Number of Cases**	**Percentage (%)**
Megaloblastic Anaemia	28	32.6
Hypoplastic/Aplastic Anaemia	26	30.2
Normal Marrow	10	11.6
Myelofibrosis	4	4.7
Myeloproliferative Disorder	3	3.5
Acute Leukemia	4	4.7
Erythroid Hyperplasia	3	3.5
Chronic Myeloid Leukemia	2	2.3
Acute Lymphoblastic Leukemia	2	2.3
Myelodysplastic Syndrome	1	1.2
Hemolytic Anaemia	1	1.2
Granulomatous Lesion	2	2.3
Total	**86**	**100**

Table 5 shows the diagnostic correlation between bone marrow aspiration and biopsy in 86 cases. A concordant diagnosis was observed in 70 cases (81.4%), indicating a high level of agreement between the two methods. Discordant diagnoses were noted in 10 cases (11.6%), reflecting some differences in findings between aspiration and biopsy. Additionally, in 6 cases (7%), the aspiration results were inconclusive, but the biopsy provided a definitive diagnosis. Overall, the data suggest that bone marrow aspiration generally correlates well with biopsy, though biopsy remains essential in cases where aspiration is inconclusive or discrepant.

Table 5: Diagnostic Correlation Between Aspiration and Biopsy (N = 86)		
Diagnostic Outcome	**Number of Cases**	**Percentage (%)**
Concordant Diagnosis	70	81.4
Discordant Diagnosis	10	11.6
Inconclusive Aspiration (Biopsy Diagnostic)	6	7
Total	**86**	**100**

Table 6 illustrates the diagnostic validity of bone marrow aspiration, demonstrating high overall performance. The sensitivity was 89.5%, indicating a strong ability to correctly identify positive cases, while the specificity of 85% reflects a good capacity to correctly exclude negative cases. The positive predictive value (91.2%) suggests that a high proportion of positive test results were truly positive, whereas the negative predictive value (81%) indicates a relatively reliable identification of true negatives. The overall diagnostic accuracy was 87.2%, highlighting that bone marrow aspiration is a dependable and effective diagnostic tool in the evaluated cases.

Table 6: Diagnostic Validity of Bone Marrow Aspiration	
Parameter	**Value (%)**
Sensitivity	89.5
Specificity	85
Positive Predictive Value (PPV)	91.2
Negative Predictive Value (NPV)	81
Diagnostic Accuracy	87.2

Table 7 presents the distribution of bone marrow cellularity patterns among 86 cases. The majority of cases were hypercellular, accounting for 42 cases (48.8%), indicating increased marrow activity in nearly half of the subjects. Hypocellular marrow was observed in 28 cases (32.6%), reflecting reduced cellularity in about one-third of the cases. Normocellular marrow constituted the smallest proportion, with 16 cases (18.6%), suggesting normal marrow cellularity in fewer individuals. Overall, the findings show that abnormal marrow cellularity (both hypercellular and hypocellular) was more prevalent than normal cellularity in the study population.

Table 7: Bone Marrow Cellularity Pattern (N = 86)		
Cellularity Type	**Number of Cases**	**Percentage (%)**
Hypercellular	42	48.8
Hypocellular	28	32.6
Normocellular	16	18.6
Total	**86**	**100**

Discussion

The present survey of pancytopenia, with special reference to the diagnostic usefulness and reliability of bone marrow aspiration and the biopsy, confirms much of what has already been reported in the literature, and adds a few regional differences. In our study age distribution revealed that the young adult age group (15-35 years) had the maximum number of cases (41.9%) as observed by Jha et al., (2008) and Pathak et al., (2012) [9,10] also mentioned that pancytopenia occurs more frequently among young adults. This resemblance implies that younger populations in developing areas may be more vulnerable as a result of nutritional deficiencies and infectious etiologies. Some western studies, however, have found it to be more prevalent in older age groups, which may be due to the variation in disease pattern including malignancies and chronic marrow disorders [11]. Therefore, this geographic and socioeconomic influence on the age-related variation in pancytopenia seems to exist."

The gender distribution in our study has a slight male predominance of 55.8%, similar to that reported by Pathak et al., (2012) and Jha et al., (2008) of 1.5:1 and 1.3:1 respectively [9,10]. This could be due to an increase in exposure to environmental risk factors for males or may be due to differential access to care-seeking. However, in contrast, Tariq et al., (2010) [12] noted a predominance of females, which suggests that the gender distribution might differ from population to population. Both of these conflicting results highlight the fact that the disease pancytopenia is relatively equally distributed amongst the sexes and that the differences are probably due to the availability of health care services in the various sociocultural and regional settings rather than a real disparity of sexes.

Among the clinical features our study was most consistent with pallor (100%), followed by weakness (60.5%) and fever (39.5%). The results were found to be consistent with the previous studies conducted by Tilak & Jain (1999) [13] and Shah et al., (2017) [14] with symptoms of anemia being predominant. A small number of patients also had hepatosplenomegaly and bleeding problems, which also correspond with previous reports and are linked to the underlying causes (marrow infiltration or platelet deficiency). Although the etiology for each case of pancytopenia varies, the clinical features are relatively uniform among different studies.

Etiological distribution, our findings showed that megaloblastic anemia was the most prevalent (32.6%) followed by aplastic/hypoplastic anemia (30.2%). The results obtained are very similar to the findings by Tilak & Jain (1999) and Shah et al., (2017) [13,14] in which megaloblastic anemia was found to be the most common cause. In the same way, Tariq et al., (2010) [12] reported megaloblastic anemia as the most common cause. This uniformity among South Asian studies shows how prevalent are nutritional deficiencies in those areas, especially B12 and folate deficiencies.

The results of this study however are in contrast with the results of studies from western populations in which the leading cause of pancytopenia was the neoplastic disorders (Keisu & Ost (1990) [11]). Furthermore, aplastic anemia and myelodysplastic syndromes have been found to be more common causes by the International Agranulocytosis and Aplastic

Anemia Study Group [15]. The variations can be explained by differences in nutritional status, environmental exposures, and healthcare infrastructure. In developing countries nutritional anemia is still a major contributor while in developed countries, malignancies and marrow failure syndromes are predominant.

In our study, the highest percentage of cases were found to be Aplastic anemia (30.2%), which is similar to the data of Jha et al., (2008) and Pathak et al., (2012) who reported it as a major or leading cause [9,10]. Relatively high incidence in our study might be related to environmental exposures to pesticides, drugs, or toxic chemicals, that are more prevalent in rural and semi-urban areas. Geographic variations were present here as well since the rate of aplastic anemia was higher in our study than reported in the western literature (10–25%) [12].

Acute leukemia in our study was a smaller proportion (6.1%) similar to Tariq et al., (2010) [12] that was about 10% compared to the findings of Jha et al., (2008) [9] that was 19.59%. This variation could be a result of different referral patterns, sample size and diagnostic facilities. Furthermore, rare conditions like myelodysplastic syndrome and erythroid hyperplasia were seldom seen in our study, as was described in previous studies, but their frequency has been reported to be higher in some international studies [15].

One of the important parts of our study is an assessment of the diagnostic correlation between bone marrow aspiration and biopsy. In our study, the concordance rate was 81.4%, which agreed with the results of other studies that reported the reliability of bone marrow aspiration as a first line investigation [9,10]. The discordant and inconclusive cases (18.6% combined) further highlight the need for bone marrow biopsy, especially in cases of myelofibrosis and aplastic anemia that may have a dry tap. This is similar to standard hematological practice and previous studies identifying the complementary nature of both procedures.

Moreover, the diagnostic validity parameters obtained in our study are high sensitivity (89.5%), high specificity (85%) and high overall accuracy (87.2%), highlighting the effectiveness of bone marrow aspiration. The results are consistent with previous studies which showed that aspiration was useful for diagnosis of most cases of pancytopenia and recommended biopsy to confirm the diagnosis in selected cases [9].

In Summary, the results of our study agree with the previous studies that have taken place in similar geographical areas except for the prevalence of megaloblastic anemia and aplastic anemia. However, there are distinct differences between this, and studies done in the west which emphasize the role of regional, nutritional and environmental factors in pancytopenia etiology. The study also stresses the importance of bone marrow aspiration and biopsy in making a correct diagnosis for proper clinical management.

Conclusion

This retrospective study highlights that pancytopenia affects a wide age range with a predominance in younger adults and a slight male preponderance, with pallor being the most consistent clinical presentation followed by generalized weakness and fever. The underlying etiology was varied, with nutritional deficiencies and bone marrow failure states coming into the fore as the most common causes, whereas malignant and infiltrative disorders were less common. Bone marrow was an important diagnostic process as the findings by aspiration and biopsy had a high degree of agreement but were useful in cases which yielded inconclusive or conflicting results in aspiration. The overall diagnostic performance of bone marrow aspiration was satisfactory, but the complementary role of trephine biopsy enhanced diagnostic reliability, particularly in structurally altered marrow conditions. Variations in marrow cellularity further reflected the heterogeneity of underlying pathologies. Thus, combined use of bone marrow aspiration and biopsy remains crucial for accurate diagnosis and effective evaluation of pancytopenia.

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