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Diagnostic Evaluation of Myelodysplastic Syndromes (MDS) in the Democratic Republic of Congo: A Review of Current Practices and Challenges

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Abstract

Background and Aim: Myelodysplastic syndromes (MDS) are a heterogeneous group of clonal disorders characterised by ineffective haematopoiesis and an increased risk of progression to acute myeloid leukaemia. In the Democratic Republic of Congo (DRC) healthcare institutions face major diagnostic challenges due to minimal infrastructure, limited access to testing equipment and a shortage of trained professionals. This study analyses current MDS diagnostic practices in the DRC, identifies unmet needs and proposes strategies for improving early and accurate detection. **Methods:** A comprehensive literature review was conducted using international databases (PubMed, Google Scholar and Scopus) alongside local health reports. Peer-reviewed articles, hospital-based studies and World Health Organization (WHO) guidelines were synthesised, with a focus on MDS diagnosis in sub-Saharan Africa and the DRC. Grey literature, including ministry of health reports and National Laboratory assessments, was also examined. **Results:** The findings reveal persistent diagnostic barriers in the DRC, including limited availability of bone marrow aspiration tools, under-resourced laboratories and a lack of trained

haematopathologists. MDS diagnosis largely depends on peripheral blood analysis and basic marrow examinations, leading to frequent under-diagnosis and misclassification. The absence of standardised diagnostic protocols and inconsistent reporting practices further hampers accurate disease identification. Misdiagnosis can occur when nutritional deficiencies, such as copper or vitamin B_{12} deficiency—present with cytopenias and dysplastic changes that mimic MDS. **Conclusion:** MDS diagnostic evaluation in the DRC is hindered by systemic and technical limitations including infrastructure deficits and workforce shortages. There is an urgent need for a national diagnostic guideline tailored to the DRC's healthcare context and a tiered diagnostic framework that aligns basic, district-level and tertiary-level investigations. Addressing these issues requires strengthening laboratory capacity, expanding access to diagnostic technologies and investing in specialist training through international collaborations and local educational initiatives.

Keywords

Diagnosis, Myelodysplastic Syndromes, Haematology, Democratic Republic of Congo, Health Systems

1. Introduction

Myelodysplastic syndromes (MDS) comprise a group of clonal haematopoietic disorders characterised by ineffective blood cell production, cytopenias and a variable risk of progression to acute myeloid leukaemia. Genetic and epigenetic mutations in haematopoietic stem cells underlie these disorders, causing dysplasia in one or more myeloid lineages. Clinicians face substantial challenges because the disease has diverse diagnostic features and patient outcomes. Globally MDS predominantly affects older adults; most diagnoses occur in people over 60 years of age.

Epidemiological data from Western countries show that the median age of MDS onset is about 71 years [1]. However, a systematic review of studies from the Middle East and North Africa (MENA) region found that the pooled mean age of MDS diagnosis was 58.4 years and that some country-level studies reported even younger mean ages 50 years in Saudi Arabia and 65.7 years in Morocco [2]. These findings suggest that MDS may present at a younger age in non-Western populations. Epidemiological data from sub-Saharan Africa remain scarce; therefore the age at presentation and disease patterns in this region are uncertain.

The current WHO and IPSS-R classifications require precise morphologic and cytogenetic data for accurate diagnosis and prognosis. These requirements are rarely met in low-income settings. Diagnostic systems are limited to high-income countries where advanced laboratory infrastructure and molecular testing are available. In resource-scarce health facilities, clinicians rely on clinical manifestations, blood counts, bone marrow smears and, occasionally, cytogenetic findings.

Consequently, there is considerable variation in diagnostic accuracy and reporting across regions.

2. Methods

Our review followed a structured search strategy. PubMed, Scopus and Google Scholar were searched using combinations of the terms "myelodysplastic syndromes," "diagnosis," "sub-Saharan Africa," "Democratic Republic of Congo," "haematology" and "resource-limited settings." Peer-reviewed publications, hospital-based studies, conference proceedings and relevant WHO documents were included if they addressed MDS diagnosis or diagnostic barriers in the DRC or neighbouring countries. Studies focusing exclusively on treatment were excluded.

Grey literature was critically examined to contextualise published findings. Local health reports consulted included ministry of health annual reports, hospital inventory summaries and laboratory leadership assessments. The 2024 ASLM *Status Report on Laboratory Leadership in Africa* used a mixed-methods approach, desk reviews, surveys and interviews, and drew on perspectives from civil servants across sub-Saharan Africa [3]. These grey-literature sources complemented peer-reviewed evidence by highlighting on-the-ground laboratory capacity gaps and governance issues.

Data were extracted on diagnostic strategies, infrastructure barriers and proposed solutions for resource-constrained environments. When national-level data were unavailable, figures were taken from studies in comparable sub-Saharan African settings. The synthesis drew on both qualitative and quantitative evidence to identify recurring themes and gaps.

3. Overview of MDS Diagnosis

Diagnosing MDS involves multiple phases that integrate clinical evaluation, haematological testing, morphological assessment and genetic analysis. Sustained or severe bone marrow failure presents as non-specific symptoms, fatigue, pallor, bruising and recurrent infections, reflecting anaemia, thrombocytopenia or neutropenia. Clinicians begin with a complete blood count (CBC), where cytopenias, macrocytic red blood cells and reduced reticulocyte counts are typical. Peripheral blood smears allow morphological assessment and reveal abnormalities such as hypogranular neutrophils, pseudo-Pelger Huët anomalies and macrocytic erythrocytes. However, confirmation requires bone marrow aspiration with biopsy for cellularity assessment, dysplasia evaluation and blast percentage estimation. Cytogenetic and molecular testing refine subtype classification and inform prognosis.

Nutritional deficiencies can mimic these findings. Case reports describe copper deficiency and vitamin B_{12} deficiency presenting with cytopenias and dysplastic changes similar to MDS; failure to exclude these conditions can result in misdiagnosis [4] [5]. Clinicians must therefore evaluate dietary history and basic nutri-

tional markers before establishing an MDS diagnosis.

4. Current Diagnostic Practices in the DRC

Diagnostic practices vary markedly between urban and rural healthcare facilities in the DRC. Urban centres such as Kinshasa and Lubumbashi provide basic diagnostic services, including CBC testing and peripheral blood smear evaluation under microscopy, but bone marrow aspiration and biopsy are scarce and almost unavailable in rural areas. Cytogenetic and molecular testing are largely absent. Consequently, diagnoses often rely heavily on clinical judgement, and definitive testing is delayed or replaced by presumptive assessments. Semi-automated haematology analysers perform CBCs, but interpretation of smears is limited by shortages of trained laboratory staff, frequent reagent stock-outs and equipment failures. Patients may be referred to distant facilities or abandon evaluation due to cost and distance, contributing to under-diagnosis and misclassification.

5. Barriers to Accurate Diagnosis

Multiple overlapping barriers limit accurate MDS diagnosis in the DRC:

1) Technical constraints: inadequate laboratory equipment, lack of bone marrow aspiration kits, microscopes and centrifuges; unreliable power supply and poor cold-chain management; absence of cytogenetic and molecular testing capabilities.

2) Human-resource shortages: limited numbers of haematologists, haematopathologists and trained laboratory technologists; concentration of expertise in a few urban centres; lack of continuing professional education.

3) Economic barriers: high out-of-pocket costs for basic tests; unaffordable bone marrow procedures; travel costs to urban centres; limited insurance coverage.

4) Systemic issues: absence of national diagnostic guidelines or cancer registries; disorganised referral systems; inconsistent documentation and reporting.

5) **Diagnostic mimics:** nutritional deficiencies such as copper or vitamin B_{12} deficiency and chronic infections can present with cytopenias and dysplastic features, leading to misdiagnosis if not excluded [4] [5].

6. Implications of Diagnostic Limitations

The limited availability of diagnostic resources in the DRC creates clinical, social and economic challenges. Patients often receive inappropriate treatments for nutritional anaemia or chronic infections, while underlying clonal disorders remain undetected. Late identification of high-risk MDS subtypes reduces survival and worsens quality of life. Under-reporting hampers the assessment of disease burden and resource allocation. Households face financial strain from repeated consultations and inappropriate therapies; many abandon care or turn to traditional healers. Psychosocial stress, including anxiety and isolation, compounds the burden in a setting with low disease awareness and inadequate support systems.

7. Opportunities and Strategic Recommendations

7.1. Strengthening Workforce and Infrastructure

Capacity-building programmes for laboratory technicians, haematologists and pathologists should be prioritised. Partnerships with universities, professional associations and regional training centres can strengthen local expertise and improve retention. Resource-limited settings require cost-effective diagnostic solutions. Point-of-care (PoC) haematology analysers, smartphone-based microscopy and cloud-based image analysis have shown promise. Telepathology and remote consultations can also bridge diagnostic gaps when physical equipment is unavailable.

7.2. Addressing Barriers to PoC Diagnostics

Recent reviews emphasise that technology alone is insufficient to improve diagnostics. Successful adoption of PoC analysers requires regulatory approval, supply-chain reliability, workforce training and awareness among healthcare workers [6]. A scoping review of supply-chain management found that most low-income-country studies reported stock-outs of PoC tests for HIV and syphilis due to supply-chain failures [7]. These experiences underscore the need to integrate PoC devices into existing logistics systems and to plan for maintenance and supervision.

7.3. Developing a Tiered Diagnostic Framework

We propose a tiered diagnostic framework tailored to the DRC. At the primary-care level, healthcare workers should perform basic evaluations, clinical assessment, CBC and peripheral smear, supported by PoC analysers. At the district-hospital level, facilities should offer bone marrow aspiration and improved microscopy. Tertiary centres should provide cytogenetic and molecular testing with referral pathways for complex cases. To design this framework systematically, a standardised assessment tool is needed. The Service Availability and Readiness Assessment (SARA) instrument divides facility readiness into four domains, staff and guidelines, equipment, diagnostic capacity and medicines/commodities, and uses tracer items such as blood-pressure apparatus, haemoglobin tests and iron tablets [8]. Adapting SARA to haematology would allow policymakers to evaluate laboratory readiness and identify gaps in each domain.

7.4. Establishing National Guidelines and Data Systems

National diagnostic guidelines for MDS should be developed and aligned with WHO classifications and the realities of DRC facilities. These guidelines should outline minimum diagnostic criteria at each healthcare level and standardise documentation and reporting. A national cancer registry capturing MDS cases will enable surveillance and resource planning. Collaboration with international partners can facilitate technology transfer and funding for essential diagnostics.

8. Conclusion

MDS diagnosis in the Democratic Republic of Congo is constrained by infrastructure deficits, workforce shortages, financial barriers and systemic weaknesses. Current diagnostic practices rely on limited tests and clinical judgement, leading to misclassification and under-reporting. Epidemiological data suggest that MDS may present at younger ages in non-Western populations [2], but the absence of local data highlights the urgent need for surveillance. Nutritional deficiencies and chronic infections that mimic MDS must be excluded to prevent misdiagnosis [4] [5]. Integrated strategies are required: workforce development, cost-effective PoC technologies with adequate supply-chain support, a tiered diagnostic framework based on SARA, and national guidelines and data systems. Addressing these gaps will improve individual patient care and strengthen the DRC's health system.

9. Ethical Considerations

Ethical approval for this review was granted by the Institutional Ethics Committee of the Higher Institute of Medical Techniques of Uvira (approval number ISTM/UVIRA/CEM/004/2025). The study used published and publicly available data, and no patient identifiers were collected.

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Authors' Contributions

All authors contributed equally to the conception and writing of this manuscript and approved the final version.

Conflicts of Interest

The authors declare no conflicts of interest.

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