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Myelodysplastic syndrome: An update narrative review of literature

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Abstract
Myelodysplastic syndromes (MDS) are a group of clonal malignant hematopoietic stem cell disorders characterized by dysplastic morphology, cytopenia and a predisposition to acute myeloid leukemia. These dysplastic changes are a result of chromosomal abnormalities and somatic mutations. MDS is the most common myeloid neoplasm of the older adults with median age at diagnosis being 72 years and an average incidence rate of 4.9 per 100,000 people per year. MDS is diagnosed and classified according to the WHO classification system, which utilizes peripheral blood and bone marrow findings. Other essential investigations include flow cytometry, genetic profile and chromosomal analysis. Various prognostic scoring system have been developed which help guide the treatment. Treatment of complications associated with MDS also forms an essential component of the management of this disease.

Keywords: Myelodysplastic syndromes, history, epidemiology, etiology, classification, symptoms, diagnosis, prognosis, treatment

Introduction
Myelodysplastic syndrome (MDS) are group of haematological disorders characterized by reduced numbers of mature blood cells and increased numbers of immature blood cells (blasts) in the peripheral circulation (Shim, 2025, Zhuo *et al*, 2026) [1]. It is characterised by cytopenias (anaemia, neutropenia, and/or thrombocytopenia); significant bone marrow dysplasia and/or bone marrow (classically <20%); cytogenetic/genetic abnormalities (e.g., 5q deletion, SF3B1 mutation, TP53 mutation); and a predilection to progress to leukemias (Sekeres and Taylor, 2022, Gantana *et al*, 2025, Debi *et al*, 2025) [3, 4, 5]. The MDS can arise de novo or occur secondary to antecedent haematological malignancy or following exposure to chemotherapy or radiotherapy (Silva *et al*, 2025) [32].

History
First described in 1900 by von Leube as a 'leukanemia', on the basis of an alleged co-existence of pernicious anemia and leukemia, MDS were named and described in a variety of ways until 1976, when the French-American-British (FAB) classification named them 'dysmyelopoietic syndromes' and categorized them separately from AML. In 1982, the FAB group refined the proposal, changed the designation to 'myelodysplastic syndromes' (Zini, 2017) [6].

Epidemiology
The precise incidence of MDS is not well-defined due to variable clinical presentations and evolving diagnostic criteria (Zhahid *et al*, 2017, Guo *et al*, 2025). According to the latest data from the Surveillance, Epidemiology, and End Results Programme (SEER) registry, the annual incidence of MDS is 4.9 per 100 000 people. In patients aged 65–69 years, it is 13.9 per 100 000 people, with at least 20 cases per 100 000 people aged > 70 years, while in people over 85 years it is 64 per 100 000 people. Myelodysplastic syndromes with excess blasts accounting for about 40% of all MDS cases (Cogle, 2015; Yahyoui *et al*,2016; Zeidan *et al*, 2019; Shrestha *et al*,2021; Gantana *et al*, 2025, Xie *et al*, 2025) [4, 9, 11, 12, 13].

Etiology
Some of the causes identified are:

- **Age:** The risk of being affected by MDS increases with age, and MDS is more seen in older people. Most people with MDS are above sixty years of age.
- **Genetic Mutations:** Some genetic mutations could result in an individual having a high chance of developing MDS. Mutation that has occurred may affect the proper functioning of blood cells and the bone marrow. Some people are born with a propensity for developing MDS due to inheriting specific genes, while others develop these changes in their genetics during aging (Erlacher *et al*, 2023) [14]
- **Exposure to Certain Chemicals and Radiation:** In particular, some studies have associated a higher chance of developing MDS with prolonged exposure to chemicals like benzene or ionizing radiation.
- **Previous Cancer Treatment:** Some cancers, like chemotherapy and radiation therapy, increase the chance of getting MDS. Secondary or therapy-induced MDS occurs as a consequence of repeated radiation exposure against the bone marrow by the treatments.
- **Inherited Disorders:** Such conditions include Fanconi anemia and bone marrow failure syndrome, among others, that may predispose one to MDS. These may arise from inherent errors in blood cells' normal functions caused by genetic mutations.

- **Smoking:** There is some indication that cigarette smoke can serve as one of the causative factors of MDS. The development of aplastic anemia can be a result of chemicals that are found in tobacco smoke, like benzene.

- **Viral Infections:** Human immunodeficiency virus (HIV) and EBV are considered among some of the viral infections that could trigger this disease. Nonetheless, the linkage between viral infectious disease and MDS remains elusive and complicated.

- **Autoimmune Diseases:** This is because some autoimmune diseases like rheumatoid arthritis and lupus are risk factors for MDS development. Autoimmune conditions can also interfere with the immune system's normal response, thus affecting the bone marrow (Greenberg *et al*, 2017) [15].

Classifications

MDS subtype identification is guided by the WHO framework, which integrates morphological and genetic findings to distinguish among subtypes. Diagnostic evaluation using the Revised International Prognostic Scoring System (IPSS-R) considers cytopenias, bone marrow blast percentage, and cytogenetic abnormalities (Garcia-Manero 2023) [16]. Clinical decision-making incorporates this risk stratification system to guide urgency of intervention and choice of treatment strategy (Sutton *et al*. 2020) [17].

- The classification of MDS by the WHO includes:
- MDS with single lineage dysplasia
- MDS with ring sideroblasts (MDS-RS)
- MDS with multi-lineage dysplasia (MDS-MLD)
- MDS with excess blasts (MDS-EB-1), with 5% to 9% blasts in the bone marrow
- MDS with excess blasts (MDS-EB-2), with 10% to 19% blasts in the bone marrow
- MDS with isolated deletion (5q)
- MDS, unclassifiable (MDS-U) (Locatelli and Strahm, 2018; Pantea *et al*, 2026) [18,19].

Signs/Symptoms

The symptoms associated with myelodysplastic syndrome could vary wildly from one patient to another, and in some instances, a patient diagnosed with this condition may have insignificant symptoms or none at all at the onset (Hochman and Dezern, 2022, Balimo *et al*, 2025) [20, 21]. Nevertheless, some common symptoms can surface as an individual age, and the disorder continues. However, it should be noted that those symptoms could point to some other haematological conditions. Here are some of the typical myelodysplastic syndrome symptoms:

- **Fatigue:** One of the most commonly reported features of MDS is persistent and unexplained fatigue. Sometimes, people may be so exhausted despite having enough sleep.

- **Recurrent Infections:** MDS may reduce the natural formation of white blood cells essential for the body's defense mechanisms. Consequently, people with MDS might quickly end up contracting recurring infectious diseases on several occasions.

- **Easy Bruising and Bleeding:** This may cause easy bruising since it results from ineffective platelet production, which is majorly involved in blood coagulation.

- **Shortness of Breath:** Anemia, with fewer red blood cells, reduces the ability of blood to transport oxygen, causing shortness of breath.

- **Pale Skin:** A low number of red blood cells leads to pale-looking skin due to anemia.

- **Unexplained Weight Loss:** Unexplained weight loss can also be seen among some people with MDS and could be related to changes in blood cell production.

- **Fever:** Some studies have shown that MDS can sometimes lead to a chronic low-grade fever related to an ongoing inflammation.

- **Increased Heart Rate:** Anemia may increase heart rate because the heart tries to compensate for decreased blood that carries oxygen.

- **Changes in Skin Color:** At times, skin pigmentation, for example, resulting in a yellow or sallow skin color, may be caused by MDS.

- **Bone Pain:** Discomfort or bone pain may be experienced due to MDS, which typically entails dysfunctional production of blood cells within the bone marrow.

Diagnosis

The MDS are diagnosed by evaluating clinical signs and performing a complete blood count (CBC), where cytopenias, macrocytic red blood cells, and reduced reticulocyte counts are typically (Balimo, 2025) [21]. Peripheral blood smears allow for morphological assessment of cells, revealing abnormalities such as pogranular neutrophils, pseudo-Pelger-Hu¨et anomalies, and macrocytic erythrocytes exhibiting anisopoikilocytosis (Parisi and Bledsoe 2024) [22]. However, accurate diagnosis of MDS requires more than peripheral findings alone. Bone marrow aspiration with biopsy remains the gold standard for confirmation. This method enables the evaluation of cellularity, dysplasia in haematopoietic stem cells, and blast percentages—critical for diagnosis and treatment planning (Fenaux *et al*. 2020) [23]. Molecular testing enhances diagnostic precision by detecting gene mutations such as SF3B1, TET2, and ASXL1. While these advanced diagnostics are routinely employed in high-income settings, they are largely unavailable in low-resource environments (van Zyl *et al*. 2021, Yusuf and Ibrahim, 2024) [24, 25].

Prognosis

The prognosis of patients with MDS varies greatly and is predicted using validated scoring systems such as the International Prognostic Scoring System (IPSS).11 Factors such as the percentage of blasts, karyotype and number of cytopenias influence the prognosis. Advancements in molecular methods, such as next-generation sequencing (NGS), have enabled the integration of molecular data into risk stratification systems, hat is IPSS-M, thereby enhancing

the prognostic capabilities of traditional scoring systems. Those with low-risk MDS have a median survival of three to ten years, while individuals with high-risk disease have a median survival of less than three years (Naidoo and Parasnath, 2025, Lewis *et al*, 2024; Al-Haidose *et al*, 2023) [26, 27, 28].

Treatment Approaches

Myelodysplastic syndrome treatment will involve an individualized, multidisciplinary strategy depending on the type and level of MDS. The spectrum of myelodysplastic syndrome treatments includes observation (for asymptomatic patients) to highly aggressive procedures such as chemotherapy, stem cell transplants, and palliative therapies (Fozza *et al*, 2022, Ugwu *et al*, 2021) [29, 30]. Myelodysplastic syndrome medication is meant to enhance normal haematopoiesis and relieve their symptoms. With ongoing advancement in research, more efficient treatment options are available for myelodysplastic syndrome. Supportive care is central to management and includes: red blood cell (RBC) transfusions with iron chelation support (for anaemia); platelet transfusions (for thrombocytopenia and bleeding); and anti-infective therapy (e.g., antibiotics). Treatment is guided by risk assessment, disease type/characteristics, symptoms/severity of cytopenias, and erythropoietin levels. Treatment includes drug therapy (e.g., lenalidomide, erythropoiesis-stimulating agents, hypomethylating agents, luspatercept, imetelstat, immunosuppressive therapy [antithymocyte globulin], ivosidenib) (Shahnor *et al*, 2025, Silvie *et al*, 2025, Sen *et al*, 2021) [33].

Conclusion

Although there has been significant improvement in the diagnosis of myelodysplastic syndrome in the last decades in low- and – middle income, several improvements are yet to be made. There is currently no comprehensive data on the incidence of MDS in so many poor resource countries. This is due to the challenges of limited expertise, poor infrastructure and funding. To address this, some actions need to be taken which may involve investing more resources to train more skilled personnel, acquire more equipment and maintain them better. It is equally important to improve on the coverage of health insurance scheme and sensitize policy makers on the need for these services and more funding for medical diagnosis.

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