Myelodysplastic syndromes (MDS)

**A Guide to the completion of the EBMT Diagnosis form:**

**DRAFT\_MDS\_v0.5**

DAY Month YEAR

**EBMT Registry**

*Helpdesk Unit*

EBMT Science Department

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# Introduction

Myelodysplastic syndrome (MDS) is a heterogeneous group of clonal haematopoietic stem cell disorders characterised by ineffective, dysplastic haematopoiesis, peripheral cytopenia and a variable rate of progression to acute myelogenous leukaemia (AML).

This form must be completed for all patients whose primary disease for which the reported treatment is being given is MDS. When the MDS has transformed to AML before the treatment, please complete both the MDS diagnosis form and the Acute Leukaemias diagnosis form.

If MDS originated from Fanconi Anaemia or Aplastic Anaemia, also complete Bone Marrow Failure Syndromes (BMF) incl. Aplastic Anaemia (AA) diagnosis or Non-indication diagnosis form in addition to the current form. If the patient received treatment for the Fanconi Anaemia or Aplastic Anaemia diagnosis, please complete the Bone Marrow Failure Syndromes (BMF) incl. Aplastic Anaemia (AA) diagnosis form. If no treatment was given, please complete the Non-indication diagnosis form.

In addition, the MDS diagnosis form can be completed if it was requested for specific studies.

No data items should be left blank unless specifically stated in the definition.

# Disease

## 1. Date of diagnosis

Report the date of the first pathological diagnosis of the disease. Add the date when the sample was collected for examination or (in its absence) the date indicated by a physician within the patient's medical record.

## 2. MDS transformed into Acute Leukaemia and treatment was done for Acute Leukaemia?

MDS can progress through different phases (subclassifications) from the time of diagnosis to transplantation. One of these phases can be AML.

If the patient is being transplanted for AML that has transformed from MDS, select **Yes** and complete the Acute Leukaemias diagnosis form in addition to the current form. Otherwise, check the **No** option.

## 3. MDS Classification at diagnosis (WHO 2016) (1)

| **Subclassification** | **Dysplastic lineages** | **Cytopenias[[1]](#footnote-1)** | **Ring sideroblasts as % of marrow erythroid elements** | **BM and PB blasts** | **Cytogenetics by conventional karyotype analysis** |
| --- | --- | --- | --- | --- | --- |
| MDS with single lineage dysplasia (MDS-SLD) | 1 | 1 or 2 | <15%/<5%[[2]](#footnote-2) | BM <5%, PB <1%, no Auer rods | Any, unless fulfils all criteria for MDS with isolated del(5q) |
| MDS with ring sideroblasts (MDS-RS) | 1 | 1 | ≥15% | BM <5%, PB ≤1% | Any, unless fulfils all criteria for MDS with isolated del(5q) |
| Myelodysplastic syndrome with isolated del(5q) chromosomal abnormality | 1-3 | 1-2 | None or any | BM <5%, PB <1%, no Auer rods | del(5q) alone or with 1 additional abnormality except −7 or del(7q) |
| MDS with multilineage dysplasia (MDS-MLD) | 2 or 3 | 1-3 | <15%/<5%3 | BM <5%, PB <1%, no Auer rods | Any, unless fulfils all criteria for MDS with isolated del(5q) |
| MDS-RS with single lineage dysplasia (MDS-RS-SLD) | 1 | 1 or 2 | ≥15%/≥5%3 | BM <5%, PB <1%, no Auer rods | Any, unless fulfils all criteria for MDS with isolated del(5q) |
| MDS-RS with multilineage dysplasia (MDS-RS-MLD) | 2 or 3 | 1-3 | ≥15%/≥5%3 | BM <5%, PB <1%, no Auer rods | Any, unless fulfils all criteria for MDS with isolated del(5q) |
| MDS with excess blasts (EB)-1 | 0-3 | 1-3 | None or any | BM 5%-9% or PB 2%-4%, no Auer rods | Any |
| MDS with excess blasts (EB)-2 | 0-3 | 1-3 | None or any | BM 10%-19% or PB 5%-19% or Auer rods | Any |
| Refractory cytopenia of childhood | 1-3 | 1-3 | None | BM <5%, PB <2% | Any |

## 4. Therapy-related MDS

Indicate if MDS developed in response to medical treatment (therapeutic agents or radiation). If the diagnosis of MDS is therapy-related, answer **Yes**. Otherwise, check **No**. If it is unknown whether or not the diagnosis of MDS was therapy-related, check **Unknown**.

# Chromosome analysis

5. Chromosome analysis done before treatment (all methods including FISH):

In this section describe the results of all chromosome analyses (performed at/after diagnosis but before the treatment).

**Not done or failed** - the chromosome analysis has not been done or failed;

**Yes, abnormal results** - the chromosome analysis has been performed and at least one of the results has been found to be abnormal. In addition, indicate the total number of different abnormalities present in all analyses with abnormal results **(number of abnormalities present)**.

**Yes, normal results** - the chromosome analysis has been performed and all the results have been found normal;

**Unknown** - it is unknown whether the chromosome analysis has been done or not.

### 5.1. Date of chromosome analysis (if tested)

Indicate the date of the chromosome analysis. If the results were normal, add the date of the first test with normal results.

### 5.2. Chromosome analysis details:

See the cytogenetics form or ask the cytogenetics team and consult your physician.

If chromosome analysis was performed, indicate for each abnormality in the table whether it was **Absent** or **Present**. If a chromosome abnormality was not evaluated, report **Not** **evaluated**.

If a chromosome abnormality was checked, but not listed as an option in the table, select **Other** and specify the abnormality, marking whether it was **Absent** or **Present**.

### 5.3. Transcribe the complete karyotype

if it is not possible to report the chromosome analysis results as per the abnormalities table. Preferably the table above with abnormalities should be completed. If the result of the chromosome analysis is too complex, the complete karyotype should be described here. Describe all abnormalities according to the ISCN karyotype nomenclature. This notation includes the total number of chromosomes, the sex chromosomes, and any extra or missing autosomal chromosomes. For example, **47, XY, +18** indicates that the patient has 47 chromosomes, is a male, and has an extra autosomal chromosome 18.

# Molecular marker analysis

## 6. Molecular markers analysis done before treatment

Indicate if molecular marker analysis was done or not before the treatment. Check **Unknown** if it is not known whether it was performed.

### 6.1. Date of molecular marker analysis (if tested)

Indicate the date of the molecular marker analysis.

### 6.2. Molecular marker analysis details

If molecular marker analysis was performed, indicate for each marker in the table whether it was **Absent** or **Present**. If a molecular marker was not evaluated, report **Not evaluated**.

If a molecular marker was evaluated, but not listed as an option in the table, select **Other** and specify the marker, indicating whether it was **Absent** or **Present**.

# Bibliography

1. Campo E, Harris NL, Pileri SA, Jaffe ES, Stein H, Thiele J. WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. IARC Who Classification of Tum; 2017. 586 p.

1. Cytopenias are defined as: hemoglobin, <10 g/dL; platelet count, <100 × 109/L; and absolute neutrophil count, <1.8 × 109/L. Rarely, MDS may present with mild anaemia or thrombocytopenia above these levels. PB monocytes must be <1 × 109/L [↑](#footnote-ref-1)
2. If SF3B1 mutation is present. [↑](#footnote-ref-2)