

BONE MARROW FAILURE SYNDROMES (BMF) incl. APLASTIC ANAEMIA (AA)

DISEASE

Note: complete this form only if this diagnosis was the indication for the HCT/IST or if it was specifically requested. Consult the manual for further information.

Date of diagnosis: ____/____/____ (YYYY/MM/DD)

Classification:

Acquired:

<input type="checkbox"/> Aplastic anaemia (AA) <input type="checkbox"/> Moderate <input type="checkbox"/> Severe <input type="checkbox"/> Very Severe <input type="checkbox"/> Unknown	Etiology: <input type="checkbox"/> Secondary to hepatitis <input type="checkbox"/> Secondary to toxin/other drug <input type="checkbox"/> Idiopathic <input type="checkbox"/> Other; specify: _____
<input type="checkbox"/> Pure red cell aplasia (non-congenital PRCA)	
<input type="checkbox"/> PNH presentation <input type="checkbox"/> Haemolytic <input type="checkbox"/> Aplastic <input type="checkbox"/> Thrombotic <input type="checkbox"/> Other; specify: _____	
<input type="checkbox"/> Pure white cell aplasia	
<input type="checkbox"/> Amegakaryocytosis / Thrombocytopenia (non-congenital)	
<input type="checkbox"/> Other acquired cytopenic syndrome; specify: _____	

Genetic*:

<input type="checkbox"/> Amegakaryocytosis / Thrombocytopenia (congenital)		
<input type="checkbox"/> Fanconi anaemia <table style="width: 100%; border: none;"> <tr> <td style="width: 50%; vertical-align: top;"> Mutated gene: <input type="checkbox"/> FANCA <input type="checkbox"/> FANCB <input type="checkbox"/> FANCC <input type="checkbox"/> FANCD1 (BRCA2) <input type="checkbox"/> FANCD2 <input type="checkbox"/> FANCE <input type="checkbox"/> FANCF <input type="checkbox"/> FANCG <input type="checkbox"/> FANCI <input type="checkbox"/> FANCI (BRIP1) <input type="checkbox"/> FANCL </td> <td style="width: 50%; vertical-align: top;"> <input type="checkbox"/> FANCM <input type="checkbox"/> FANCN (PALB2) <input type="checkbox"/> FANCO (RAD51C) <input type="checkbox"/> FANCP (SLX4) <input type="checkbox"/> FANCQ (XPF) <input type="checkbox"/> FANCS (BRCA1) <input type="checkbox"/> FANCT (UBE2T) <input type="checkbox"/> FANCU (XRCC2) <input type="checkbox"/> FANCV (REV7) <input type="checkbox"/> FANCW (RFWD3) <input type="checkbox"/> Other; specify: _____ </td> </tr> </table>	Mutated gene: <input type="checkbox"/> FANCA <input type="checkbox"/> FANCB <input type="checkbox"/> FANCC <input type="checkbox"/> FANCD1 (BRCA2) <input type="checkbox"/> FANCD2 <input type="checkbox"/> FANCE <input type="checkbox"/> FANCF <input type="checkbox"/> FANCG <input type="checkbox"/> FANCI <input type="checkbox"/> FANCI (BRIP1) <input type="checkbox"/> FANCL	<input type="checkbox"/> FANCM <input type="checkbox"/> FANCN (PALB2) <input type="checkbox"/> FANCO (RAD51C) <input type="checkbox"/> FANCP (SLX4) <input type="checkbox"/> FANCQ (XPF) <input type="checkbox"/> FANCS (BRCA1) <input type="checkbox"/> FANCT (UBE2T) <input type="checkbox"/> FANCU (XRCC2) <input type="checkbox"/> FANCV (REV7) <input type="checkbox"/> FANCW (RFWD3) <input type="checkbox"/> Other; specify: _____
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<input type="checkbox"/> Diamond-Blackfan anaemia (congenital PRCA)		
<input type="checkbox"/> Shwachman-Diamond syndrome		
<input type="checkbox"/> Dyserythropoietic anaemia		
<input type="checkbox"/> Dyskeratosis congenita		
<input type="checkbox"/> Congenital sideroblastic anaemia (CSA)		
<input type="checkbox"/> Other congenital anaemia; specify: _____		

*Please fill in the "Inborn Errors" indication diagnosis form in addition to the current form (optional)



EBMT Centre Identification Code (CIC): _____
 Hospital Unique Patient Number (UPN): _____
 Patient Number in EBMT Registry: _____

Treatment Type HCT CT IST Other
 Treatment Date ____/____/____ (YYYY/MM/DD)

CHROMOSOME ANALYSIS

Chromosome analysis done before IST/HCT:

(Describe results of the most recent complete analysis)

- No
 Yes: **Output of analysis:** Separate abnormalities Full karyotype
 Unknown

If chromosome analysis was done:

What were the results?

- Normal
 Abnormal: number of abnormalities present: _____
 Failed

Date of chromosome analysis: ____/____/____ (YYYY/MM/DD) Unknown

For abnormal results, indicate below whether the abnormalities were absent, present or not evaluated.

abn 3	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
del(13q)	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
Monosomy 7	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
Trisomy 8	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
Other; specify: _____	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	

OR

Transcribe the complete karyotype: _____

Chromosomal breakage test *(for Fanconi only)*:

- Negative
 Positive
 Not done or failed
 Unknown

MOLECULAR MARKER ANALYSIS

Molecular markers analysis done before IST/HCT:

- No
 Yes
 Unknown

Date of molecular marker analysis (if applicable): ____/____/____ (YYYY/MM/DD) Unknown

Indicate below whether the markers were absent, present or not evaluated.

ASXL1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
BCOR	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
BCORL1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
CBL	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
CSMD1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
DNMT3A	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
ETV6	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
EZH2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
FLT3	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
GNAS	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
IDH1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
IDH2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
JAK2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
KRAS	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
MPL	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
NPM1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
NRAS	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
PHF6	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
PIGA	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
PPM1D	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
PTPN11	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
RAD21	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
RUNX1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
SETBP1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
SF3B1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
SRSF2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
STAG2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
TET2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
TP53	TP53 mutation type: <input type="checkbox"/> Single hit <input type="checkbox"/> Multi hit <input type="checkbox"/> Unknown			
U2AF1	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
ZRSR2	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated	<input type="checkbox"/> Unknown
Other; specify: _____	<input type="checkbox"/> Absent	<input type="checkbox"/> Present		



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Treatment Type HCT CT IST Other
 Treatment Date ____/____/____ (YYYY/MM/DD)

BONE MARROW INVESTIGATION

Bone marrow assessments:

Cellularity in the bone marrow aspirate	<input type="checkbox"/> Acellular <input type="checkbox"/> Hypocellular <input type="checkbox"/> Normocellular <input type="checkbox"/> Hypercellular	<input type="checkbox"/> Focal cellularity <input type="checkbox"/> Not evaluated <input type="checkbox"/> Unknown
Cellularity in the bone marrow trephine	<input type="checkbox"/> Acellular <input type="checkbox"/> Hypocellular <input type="checkbox"/> Normocellular <input type="checkbox"/> Hypercellular	<input type="checkbox"/> Focal cellularity <input type="checkbox"/> Not evaluated <input type="checkbox"/> Unknown
Fibrosis on bone marrow biopsy	<input type="checkbox"/> No <input type="checkbox"/> Mild <input type="checkbox"/> Moderate <input type="checkbox"/> Severe	<input type="checkbox"/> Not evaluable <input type="checkbox"/> Not evaluated <input type="checkbox"/> Unknown
CD34+ cell count percentage (%)	_____ %	<input type="checkbox"/> Not evaluated <input type="checkbox"/> Unknown
Blast count percentage (%)	_____ %	<input type="checkbox"/> Not evaluated <input type="checkbox"/> Unknown If the precise blast count is not available, please indicate whether it is: <input type="checkbox"/> ≤ 5% <input type="checkbox"/> > 5%

PNH TESTS

only applicable for Aplastic Anaemia and/or PNH at time of diagnosis

PNH test done?

- No
 Yes: **Date of PNH test:** ____/____/____ (YYYY/MM/DD) Unknown
 Unknown

PNH diagnostics by flow cytometry:

- Clone absent
 Clone present: Size of PNH clone in percentage (%): _____
 Unknown

Flow cytometry assessment done on:

- Granulocytes
 RBC
 Both
 Other; specify: _____

If clone present:

Clinical manifestation of PNH:

- No
 Yes: Date of clinical manifestation of PNH: ____/____/____ (YYYY/MM/DD) Unknown

Anti-complement treatment given?

- No
 Yes, complete the table:

Drug	Start date (YYYY/MM/DD)	Treatment stopped/date (YYYY/MM/DD)
<input type="checkbox"/> Eculizumab	____/____/____ <input type="checkbox"/> Unknown	<input type="checkbox"/> No <input type="checkbox"/> Yes: ____/____/____ <input type="checkbox"/> Unknown <input type="checkbox"/> Unknown
<input type="checkbox"/> Ravalizumab	____/____/____ <input type="checkbox"/> Unknown	<input type="checkbox"/> No <input type="checkbox"/> Yes: ____/____/____ <input type="checkbox"/> Unknown <input type="checkbox"/> Unknown
<input type="checkbox"/> Pegcetacoplan	____/____/____ <input type="checkbox"/> Unknown	<input type="checkbox"/> No <input type="checkbox"/> Yes: ____/____/____ <input type="checkbox"/> Unknown <input type="checkbox"/> Unknown
<input type="checkbox"/> Other; specify*: _____	____/____/____ <input type="checkbox"/> Unknown	<input type="checkbox"/> No <input type="checkbox"/> Yes: ____/____/____ <input type="checkbox"/> Unknown <input type="checkbox"/> Unknown

*Please consult the **LIST OF CHEMOTHERAPY DRUGS/AGENTS AND REGIMENS** on the EBMT website for drugs/regimens names

If there were more drugs given during one line of treatment add more copies of this page.