

## 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	<b>Etiology:</b>  <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;"> <b>Mutated gene:</b> <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 (RFDW3)  <input type="checkbox"/> Other; specify: _____           </td> </tr> </table>	<b>Mutated gene:</b> <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 (RFDW3) <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.

<b>abn 3</b>	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
<b>del(13q)</b>	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
<b>Monosomy 7</b>	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
<b>Trisomy 8</b>	<input type="checkbox"/> Absent	<input type="checkbox"/> Present	<input type="checkbox"/> Not evaluated
<b>Other; specify: _____</b>	<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	<b>TP53 mutation type:</b> <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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### 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 (%)	_____ %  <b>If the precise blast count is not available, please indicate whether it is:</b> <input type="checkbox"/> ≤ 5% <input type="checkbox"/> > 5%	<input type="checkbox"/> Not evaluated <input type="checkbox"/> Unknown

### 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.*