

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:

☐ Aplastic anaemia (AA) Severity of Aplastic Anaemia (AA): ☐ Moderate ☐ Severe ☐ Very Severe ☐ Unknown	Etiology: ☐ Secondary to hepatitis ☐ Secondary to toxin/other drug ☐ Idiopathic ☐ Other; specify: _____
☐ Pure red cell aplasia (non-congenital PRCA)	
☐ PNH PNH presentation: ☐ Haemolytic ☐ Aplastic ☐ Thrombotic ☐ Other; specify: _____	
☐ Pure white cell aplasia	
☐ Amegakaryocytosis / Thrombocytopenia (non-congenital)	
☐ Other acquired cytopenic syndrome; specify: _____	

☐ Genetic*:

☐ Amegakaryocytosis / Thrombocytopenia (congenital)																							
☐ Fanconi anaemia <table><tr><td>Mutated gene: ☐ FANCA</td><td>☐ FANCM</td></tr><tr><td>☐ FANCB</td><td>☐ FANCN (PALB2)</td></tr><tr><td>☐ FANCC</td><td>☐ FANCO (RAD51C)</td></tr><tr><td>☐ FANCD1 (BRCA2)</td><td>☐ FANCP (SLX4)</td></tr><tr><td>☐ FANCD2</td><td>☐ FANCQ (XPF)</td></tr><tr><td>☐ FANCE</td><td>☐ FANCS (BRCA1)</td></tr><tr><td>☐ FANCF</td><td>☐ FANCT (UBE2T)</td></tr><tr><td>☐ FANCG</td><td>☐ FANCU (XRCC2)</td></tr><tr><td>☐ FANCI</td><td>☐ FANCV (REV7)</td></tr><tr><td>☐ FANCI (BRIP1)</td><td>☐ FANCW (RFDW3)</td></tr><tr><td>☐ FANCL</td><td>☐ Other; specify: _____</td></tr></table>		Mutated gene: ☐ FANCA	☐ FANCM	☐ FANCB	☐ FANCN (PALB2)	☐ FANCC	☐ FANCO (RAD51C)	☐ FANCD1 (BRCA2)	☐ FANCP (SLX4)	☐ FANCD2	☐ FANCQ (XPF)	☐ FANCE	☐ FANCS (BRCA1)	☐ FANCF	☐ FANCT (UBE2T)	☐ FANCG	☐ FANCU (XRCC2)	☐ FANCI	☐ FANCV (REV7)	☐ FANCI (BRIP1)	☐ FANCW (RFDW3)	☐ FANCL	☐ Other; specify: _____
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☐ FANCL	☐ Other; specify: _____																						
☐ Diamond-Blackfan anaemia (congenital PRCA)																							
☐ Shwachman-Diamond syndrome																							
☐ Dyserythropoietic anaemia																							
☐ Dyskeratosis congenita																							
☐ Congenital sideroblastic anaemia (CSA)																							
☐ Other congenital anaemia; specify: _____																							



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

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

DISEASE

(Only for Genetic BMF)

Has the patient developed features of an inborn error of immunity?

- ☐ No
- ☐ Yes:

Please fill in the "Inborn Errors" indication diagnosis form in addition to the current form (Mandatory)
- ☐ Unknown

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	☐ Absent	☐ Present	☐ Not evaluated	☐ Unknown
del(13q)	☐ Absent	☐ Present	☐ Not evaluated	☐ Unknown
Monosomy 7	☐ Absent	☐ Present	☐ Not evaluated	☐ Unknown
Trisomy 8	☐ Absent	☐ Present	☐ Not evaluated	☐ Unknown
Other; specify: _____	☐ Absent	☐ Present		

OR

Transcribe the complete karyotype: _____

Chromosomal breakage test *(for Fanconi only):*

- ☐ Negative
- ☐ Positive
- ☐ Not done or failed
- ☐ Unknown



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



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Treatment Date ____/____/____ (YYYY/MM/DD)

BONE MARROW INVESTIGATION

Bone marrow assessments:

Cellularity in the bone marrow aspirate	☐ Acellular ☐ Hypocellular ☐ Normocellular ☐ Hypercellular	☐ Focal cellularity ☐ Not evaluated ☐ Unknown
Cellularity in the bone marrow trephine	☐ Acellular ☐ Hypocellular ☐ Normocellular ☐ Hypercellular	☐ Focal cellularity ☐ Not evaluated ☐ Unknown
Fibrosis on bone marrow biopsy	☐ No ☐ Mild ☐ Moderate ☐ Severe	☐ Not evaluable ☐ Not evaluated ☐ Unknown
CD34+ cell count percentage (%)	_____ %	☐ Not evaluated ☐ Unknown
Blast count percentage (%)	_____ % If the precise blast count is not available, please indicate whether it is: ☐ ≤ 5% ☐ > 5%	☐ Not evaluated ☐ 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)
☐ Eculizumab	____/____/____ ☐ Unknown	☐ No ☐ Yes: ____/____/____ ☐ Unknown ☐ Unknown
☐ Ravalizumab	____/____/____ ☐ Unknown	☐ No ☐ Yes: ____/____/____ ☐ Unknown ☐ Unknown
☐ Pegcetacoplan	____/____/____ ☐ Unknown	☐ No ☐ Yes: ____/____/____ ☐ Unknown ☐ Unknown
☐ Other; specify*: _____	____/____/____ ☐ Unknown	☐ No ☐ Yes: ____/____/____ ☐ Unknown ☐ 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.