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

☐ Genetic*:

☐ Amegakaryocytosis / Thrombocytopenia (congenital)

☐ Fanconi anaemia

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)
☐ FANCJ (BRIP1)	☐ FANCW (RFWD3)
☐ FANCL	☐ Other; specify: _____

☐ Diamond-Blackfan anaemia (congenital PRCA)

☐ Shwachman-Diamond syndrome

☐ Dyserythropoietic anaemia

☐ Dyskeratosis congenita

☐ Congenital sideroblastic anaemia (CSA)

☐ Other congenital anaemia; specify: _____

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

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

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 (%)	_____ % ☐ Not evaluated ☐ Unknown **If the precise blast count is not available, please indicate whether it is:** ☐ ≤ 5% ☐ > 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)
☐ 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.