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)*

INBORN ERRORS

DISEASE

Date of diagnosis: _ _ _ _ / _ _ / _ _ *(YYYY/MM/DD)*

Classification:

- ☐ Inborn errors of immunity (IEI)
- ☐ Inborn errors of metabolism
- ☐ Other inborn errors

Is the patient registered in another registry?

- ☐ No
- ☐ Yes:
  - ☐ ESID
    Patient Registry Number: _____
  - ☐ Other; specify: _____
    Patient Registry Number: _____

Inborn Errors of Immunity (IEI)

DISEASE

Specify the gene in which a disease-causing variant was observed*:*

☐ Gene from *Appendix 1*; specify:_____ ☐ Other gene; specify:_____ ☐ Unknown	**Effect of mutation:** ☐ Gain of function ☐ Loss of function **Mutation type:** ☐ Germline mutation ☐ Somatic mutation
Mutation in additional gene; specify:_____	

Classification *(IUIS)*:

- ☐ Combined Immune deficiency including SCID:
  - ☐ SCID
  - ☐ CID
  - ☐ CID with associated or syndromic features
- ☐ Predominantly antibody deficiency:
  - ☐ Hypogammaglobulinemia
  - ☐ Other antibody deficiency
- ☐ Diseases of immune dysregulation:
  - ☐ HLH and susceptibility to EBV
  - ☐ Syndromes with autoimmunity and others
- ☐ Phagocyte defects:
  - ☐ Neutropenia without anti-neutrophil antibodies
  - ☐ Functional defects
- ☐ Defects in intrinsic and innate immunity:
  - ☐ Predisposition to bacterial, fungal and parasitic infections including Mendelian Susceptibility to Mycobacterial Disease (MSMD)
  - ☐ Predisposition to viral infections
- ☐ Autoinflammatory disorders:
  - ☐ Recurrent and systemic inflammations
  - ☐ Sterile inflammation and interferonopathies
- ☐ Complement deficiencies
- ☐ Bone marrow failures*
- ☐ Phenocopies of IEI

*Please fill in the "Bone Marrow Failure Syndromes (BMF) incl. Aplastic Anaemia (AA)" indication diagnosis form in addition to the current form (optional)

Inborn Errors of Metabolism

DISEASE

Specify the gene in which a disease-causing variant was observed*:*

☐ Gene from *Appendix 2*; specify:_____

☐ Other gene; specify:_____

☐ Unknown

Effect of mutation:
☐ Gain of function
☐ Loss of function

Mutation type:
☐ Germline mutation
☐ Somatic mutation

Mutation in additional gene; specify:_____

Classification:

☐ MPS: ☐ Severe ☐ Attenuated

MPS type:
- ☐ MPSI (Hurler)
- ☐ MPSII (Hunter)
- ☐ MPSIII (Sanfilippo), type:
  - ☐ A ☐ B
- ☐ MPSIV (Morquio), type:
  - ☐ A ☐ B
- ☐ MPSVI (Maroteax Lamy)
- ☐ MPSVII (Sly)
- ☐ I-cell disease

☐ Other lysosomal storage diseases:
- ☐ Tay sachs
- ☐ Metachromatic leukodystrophy (MLD)
- ☐ Aspartylglucosaminuria (AGU)
- ☐ Gaucher
- ☐ Alfa-mannosidosis
- ☐ CLN
- ☐ Wolman
- ☐ Niemann-Pick disease
- ☐ Fucosidosis
- ☐ Sialidosis
- ☐ Fabry
- ☐ Pompe
- ☐ Other; specify: _____

DISEASE continued

Classification (continued):

- ☐ Other metabolic disorders (non LSD):
  - ☐ X-ALD
  - ☐ Glycogen storage disease Ib
  - ☐ MNGIE
  - ☐ SIFD
  - ☐ Krabbe (globoid cell leukodystrophy)
  - ☐ Other; specify: _____

Other Inborn Errors

DISEASE

Specify the gene in which a disease-causing variant was observed*:*

☐ Gene from *Appendix 3*; specify:_____ ☐ Other gene; specify:_____ ☐ Unknown	**Effect of mutation:** ☐ Gain of function ☐ Loss of function **Mutation type:** ☐ Germline mutation ☐ Somatic mutation
Mutation in additional gene; specify:_____	

Classification:

- ☐ Glanzmann thrombasthenia
- ☐ Other inherited platelet abnormalities; specify: _____
- ☐ Porphyria
- ☐ Osteopetrosis
- ☐ Other inborn error; specify: _____

APPENDIX 1

-- List of genes/abnormalities involved in Inborn Errors of Immunity (IEI) --

10p13-p14 deletion syndrome	C1QA	CD40	CTLA4	FANCD2	IFNGR2
11q23del (Jacobsen)	C1QB	CD40LG	CTNNBL1	FANCE	IGHM
14q32	C1QC	CD46	CTPS1	FANCF	IGKC
22q.11.2 deletion syndrome (DiGeorge)	C1R	CD55	CTSC	FANCI	IGLL1
ACD	C1R	CD59	CXCR2	FANCL	IKBKB
ACP5	C1S	CD70	CXCR4	FANCM	IKBKG
ACTB	C1S	CD79A	CYBA	FAT4	IKZF1
ADA	C2	CD79B	CYBB	FCGR3A	IKZF2
ADA2	C2orf69	CD81	CYBC1	FCHO1	IKZF3
ADAM17	C3	CD8A	DBF4	FCN3	IL10
ADAR1	C3	CDC42	DBR1	FERMT1	IL10RA
AICDA	C4A+C4B	CDCA7	DCLRE1C	FERMT3	IL10RB
AIRE	C5	CEBPE	DEF6	FLT3L	IL12B
AK2	C6	CFB	DIAPH1	FNIP1	IL12RB1
ALPI	C7	CFD	DKC1	FOXN1	IL12RB2
ALPK1	C8A	CFH	DNAJC21	FOXP3	IL17F
AP1S3	C8B	CFHR1	DNASE1L3	FPR1	IL17RA
AP3B1	C8G	CFHR2	DNASE2	G6PC3	IL17RC
AP3D1	C9	CFHR3	DNMT3B	G6PD	IL18BP
APOL1	CARD11	CFHR4	DOCK11	G6PT1	IL-1R1
Apollo/SNM1B	CARD14	CFHR5	DOCK2	GATA2	IL1RN
ARHGEF1	CARD9	CFI	DOCK8	GFI1	IL21
ARPC1B	CARMIL2	CFP	DPP9	GIMAP6	IL21R
ARPC5	CASP10	CFTR	DUT	GINS1	IL23R
ATAD3A	CASP8	CHD7	EFL1	GINS4	IL27RA
ATG4	CBLB	CHUK	ELANE	HAVCR2	IL2RA
ATM	CCBE1	CIB1	ELF4	HAX1	IL2RB
ATP6AP1	CCR2	CIITA	EPG5	HCK	IL2RG
B2M	CD19	CLCN7	ERBB2IP	HELLS	IL36RN
BACH2	CD20	CLPB	ERCC4	HMOX	IL6R
BCL10	CD21	COPA	ERCC6L2	HYOU1	IL6ST
BCL11B	CD27	COPG1	ERZ	ICOS	IL7R
BLM	CD274	CORO1A	EXTL3	ICOSLG	INO80
BLNK	CD28	CRACR2A	FAAP24	IFIH1	IRAK1
BRCA1	CD3D	CSF2RA	FADD	IFNAR1	IRAK4
BRCA2	CD3E	CSF3R	FANCA	IFNAR2	IRF1
BRIP1	CD3G	CSFR2B	FANCB	IFNG	IRF2BP2
BTK	CD3Z	CTC1	FANCC	IFNGR1	IRF3

APPENDIX 1

-- List of genes/abnormalities involved in Inborn Errors of Immunity (IEI) -- continued

IRF4	MCM10	OAS2	PSMB8*	RORC	STAT1	TMC6	WRAP53
IRF4	MCM4	ORAI1	PSMB9	RPSA	STAT2	TMC8	XIAP
IRF7	MCTS1	OSTM1	PSMD12	RTEL1	STAT3	TMEM173	XRCC2
IRF8	MECOM	OTULIN	PSMG2	SAMD9	STAT3	TNFAIP3	XRCC9
IRF9	MEFV	PALB2	PSTPIP1	SAMD9L	STAT4	TNFRSF11A	ZAP70
ISG15	MKL1	PARN	PTCRA	SAMHD1	STAT5B	TNFRSF13B	ZBTB24
ITCH	MOGS	PAX1	PTEN	SASH3	STAT6	TNFRSF13C	ZNF341
ITGB2	MSH6	PAX5	PTPRC	SBDS	STIM1	TNFRSF1A	ZNFX1
ITK	MSN	PDCD1	RAB27A	SEC61A1	STK4	TNFRSF4	
ITPKB	MTHFD1	PEPD	RAC2	SEMA3E	STN1	TNFRSF6	
ITPR3	MVK	PGM3	RAD51	SERPING1	STX11	TNFSF11	
JAGN1	MYD88	PIK3CD	RAD51C	SGPL1	STXBP2	TNFSF12	
JAK1	MYSM1	PIK3CD GOF	RAG1	SH2B3	SYK	TNFSF13	
JAK3	NBAS	PIK3CG	RAG2	SH2D1A	TAP1	TNFSF6	
KARS1	NBS1	PIK3R1	RANBP2	SH3BP2	TAP2	TNFSF9	
KDM6A	NCF1	PLCG1	RASGRP1	SH3KBP1	TAPBP	TOP2B	
KMT2A	NCF2	PLCG2	RBCK1	SHARPIN	TAZ	TP53	
KMT2D	NCF4	PLEKHM1	REL	SKIV2L	TBK1	TPP2	
LAMTOR2	NCKAP1L	PMS2	RELA	SLC19A1	TBX1	TRAC	
LAT	NCSTN	PMVK	RELB	SLC29A3	TBX21	TRAF3	
LCK	NFAT1	PNP	RFWD3	SLC35C1	TCF3	TRAF3IP2	
LCP2	NFAT5	POLA1	RFX5	SLC39A7	TCIRG1	TREX1	
LIG1	NFATC1	POLD1	RFXANK	SLC46A1	TCN2	TRIM22	
LIG4	NFE2L2	POLD2	RFXAP	SLC7A7	TERC	TRNT1	
LPIN2	NFKB1	POLE1	RHBDF2	SLX4	TERT	TTC37	
LRBA	NFKB2	POLE2	RHOG	SMARCAL1	TET2	TTC7A	
LSM11	NFKBIA	POLR3A	RHOH	SMARCD2	TFRC	TYK2	
LY96	NHEJ1	POLR3C	RIPK1	SNORA31	TGFB1	UBE2T	
LYN	NLRC4	POLR3F	RIPK3	SNX10	TGFBR1	UNC13D	
LYST	NLRP1	POMP	RMRP	SOCS1	TGFBR2	UNC93B1	
MAD2L2	NLRP12	POU2AF1	RNASEH2A	SP110	THBD	UNG	
MAGT1	NLRP3	PRF1	RNASEH2B	SPI1	TICAM1	USB1	
MALT1	NOD2	PRKCD	RNASEH2C	SPINK5	TINF2	USP18	
MAN2B2	NOLA2	PRKDC	RNASEL	SPPL2A	TIRAP	VPS13B	
MAP1LC3B2	NOLA3	PSEN	RNF168	SRP19	TLR3	VPS45	
MAP3K14	NOS2	PSENEN	RNF31	SRP54	TLR4	WAS	
MAPK8	NSMCE3	PSMB10	RNU4ATAC	SRP72	TLR7	WDR1	
MASP2	OAS1	PSMB4	RNU7-1	SRPRA	TLR8	WIPF1	

APPENDIX 2
-- List of genes involved in Inborn Errors of Metabolism --

ABCD1	GUSB	PPT1
AGA	HEXA	RRM2B
ARSA	IDS	SGSH
ARSB	IDUA	SLC37A4
FUCA1	LIPA	SMPD1
GAA	MAN2B1	TRNT1
GALNS	MANBA	TYMP
GBA	NAGLU	
GLA	NEU1	
GLB1	NPC1	
GNPTAB	NPC2	
GNPTG	POLG	

APPENDIX 3
-- List of genes involved in Other Inborn Errors --

11q23del (Jacobsen)	GP1BB	OSTM1
ALAS2	GP9	PLEKHM1
ANKRD26	HOXA11	RANK
CA2	ITGA2B	RBM8A
CICN7	ITGA2B	RUNX1
DIAPH1	ITGB3	SLFN14
ETV6	ITGB3	SNX10
FECH	MECOM	TCIRG1
FERMT3	MPL	THPO
FLI1	MYH9	UROS
GNE	NBEAL2	WAS
GP1BA	NEMO	