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 an additional immunophenotype form

**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	C1QB	CD8A	DCLRE1C (ARTEMIS)	FPR1
11q23del (Jacobsen)	C1QC	CDC42	DEF6	G6PC3
14q32	C1R	CEBPE	DIAPH1	G6PD
22q.11.2 deletion syndrome	C1S	CFB	DKC1	G6PT1
ACD	C2	CFD	DNAJC21	GATA2
ACP5	C2orf69	CFH	DNASE1L3	GFI1
ACT1	C3	CFHR1	DNASE2	GINS1
ACTB	C4A	CFHR2	DNMT3B	HAVCR2
ADA	C4B	CFHR3	DOCK2	HAX1
ADA2	C6	CFHR4	DOCK8	HCK
ADAM17	C7	CFHR5	EFL1	HELLS
ADAR1	C8A	CFTR	ELANE	HMOX
AICDA	C8B	CHD7	ELF4	HYOU1
AIRE	C8G	CIB1	EPG5	ICOS
AK2	C9	CIITA	ERBB21P	ICOSL
ALP1	CARD11	CLCN7	ERCC4	IFIH1
AP1S3	CARD14	CLPB	ERCC6L2	IFNAR1
AP3B1	CARD9	CNFX1	EXTL3	IFNAR2
AP3D1	CASP10	COH1	FAAP24	IFNG
APOL1	CASP8	COPA	FADD	IFNGR1
ARHGEF	CCBE1	COPG1	FANCA	IFNGR2
ARPC1B	CD132	CORO1A	FANCB	IGHM
ATAD3A	CD19	CRACR2A	FANCC	IGKC
ATG4A	CD20	CSF2RB	FANCD2	IGLL1
ATM	CD21	CSF3R	FANCE	IKBKB
ATP6AP1	CD27	CTC1	FANCF	IKBKG (NEMO)
B2M	CD28	CTLA4	FANCI	IKKA (CHUK)
BACH2	CD3D	CTNNBL1	FANCL	IKZF1
BCL10	CD3E	CTPS1	FANCM	IKZF2
BCL11B	CD3Z	CTSC	FAT4	IKZF3
BLM	CD40L	CXCR2	FCGR3A	IL10
BLNK	CD40LG	CXCR4	FCH01	IL10RA
BRCA1	CD46	CYBA	FCN3	IL10RB
BRCA2 (FANCD1)	CD55	CYBB	FERMT1	IL12B
BRIP1	CD59	CYBC1	FERMT3 (LAD3)	IL12RB1
BTK	CD79A	DBR1	FNIP1	IL12RB2
C16ORF57	CD79B	DCAB1	FOXN1	IL17F
C1QA	CD81	DCLRE1B (APOLLO)	FOXP3	IL17RA

APPENDIX 1

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

IL17RC	LIG1	NFKBIB (IKBB)	POU2AF1	RNU4ATAC
IL18BP	LIG4	NHEJ1	PRF1	RORC
IL1RN	LPIN2	NLRC4	PRKCD	RPSA
IL21	LRBA	NLRP1	PRKDC	RTEL1
IL21R	LSM11	NLRP12	PSEN	SAMD9
IL23R	LYST	NLRP3	PSENEN	SAMD9L
IL2RA	MAD2L2	NOD2	PSMB8	SAMHD1
IL2RB	MAGT1	NOLA2 (NHP2)	PSMB9	SASH3
IL2RG	MALT1	NOLA3 (NOP10)	PSTPIP1	SBDS
IL36RN	MAN282	NOS2	PTEN	SEC61A1
IL6R	MAP1LC3B2	NRAS	PTPRC	SEMA3E
IL6ST	MAP3K14	NSMCE3	RAB27A	SERPING1
IL7R	MAPK8	OAS1	RAC2	SH2D1A
IMCDCA7	MASP2	ORAI1	RAD51	SH3BP2
INO80	MCM4	OSTM1	RAD51C	SH3KBP1 (CIN85)
IRAK1	MECOM	OTULIN	RAG1	SKIV2L
IRAK4	MEFV	PALB2	RAG2	SLC29A3
IRF2BP2	MKL1	PARN	RANBP2	SLC35C1 (LAD2)
IRF3	MOGS (GCS1)	PAX1	RASGRP1	SLC39A7
IRF4	MSH6	PDCD1	RBCK1	SLC46A1
IRF7	MSN	PEPD	REL	SLC7A7
IRF8	MTHFD1	PFC	RELA	SLP76
IRF9	MVK	PGM3	RELB	SLX4
ISG15	MYD88	PIK3CD	RFWD3	SMARCAL1
ITCH	MYSM1	PIK3CG	RFX5	SMARCD2
ITGB2 (LAD1)	NBAS	PIK3R1	RFXANK	SNORA31
ITK	NBS1	PLCG2	RFXAP	SNX10
ITPKB	NCF1	PLEKHM1	RHOG	SOCS1
JAGN1	NCF2	PLOD2	RHOH	SP110
JAK1	NCF4	PMS2	RIPK1	SPI1
JAK3	NCKAP1L	PNP	RLTPR (CARMIL2)	SPINK5
KDM6A	NCM10	POLA1	RMRP	SPPL2A
KMT2A (MLL)	NCSTN	POLD1	RNASEH2A	SRP54
KMT2D (MLL2)	NFAT5	POLE1	RNASEH2B	SRP72
KRAS	NFE2L2	POLE2	RNASEH2C	STAT1
LAMTOR2	NFKB1	POLR3A	RNF168	STAT2
LAT	NFKB2	POLR3C	RNF31	STAT3
LCK	NFKBIA (IKBA)	POLR3F	RNO7	STAT5B

APPENDIX 1
-- List of genes/abnormalities involved in Inborn Errors of Immunity (IEI) -- continued

STIM1	TNFRSF4
STK4	TNFRSF6
STN1	TNFRSF9
STX11	TNFSF11
STXBP2	TNFSF13 (APRIL)
SYK	TNFSF6
TAP1	TNFSF7 (CD70)
TAP2	TOP2B
TAPBP	TP53
TAZ	TPP1
TBK1	TPP2
TBX1	TRAC
TBX21	TRAF3
TCF3	TREX1
TCIRG1	TRIM22
TCN2	TRNT1
TERC	TTC37
TERT	TTC7A
TET2	TWEAK (TNFSF12)
TFRC	TYK2
TGFB1	UBA1
TGFBR1	UBE2T
TGFBR2	UNC13D (Munc13.4)
THBD	UNC93B1
TICAM1 (TRF)	UNG
TINF2	USP18
TIRAP	VPS45
TLR3	WAS
TLR7	WDR1
TLR8	WIPF1
TMC6	WRAP53
TMC8	XIAP
TMEM173	XRCC2
TNFAIP3	XRCC9
TNFRSF11A	ZAP70
TNFRSF13B (TACI)	ZBTB24
TNFRSF13C	ZNF341
TNFRSF1A	ZNKFX1

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

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

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

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