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:_____

Inborn Errors of Immunity (IEI)

DISEASE

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

(Only for Bone marrow failures)

Has the patient developed features of Bone marrow failure?

- ☐ No
- ☐ Yes: **Please fill in the "Bone Marrow Failure Syndromes (BMF) incl. Aplastic Anaemia (AA)" indication diagnosis form in addition to the current form**
- ☐ Unknown

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: _____

Immune profiling

(Only for Inborn errorrs of immunity)

Immune profiling done at diagnosis: ☐ No ☐ Yes ☐ Unknown

Test date: _ _ _ _ / _ _ / _ _ *(YYYY/MM/DD)* ☐ Unknown

Cell type and test results		**Units** *(for CD4 and CD8, select unit)*
CD3 T-cells: _____	☐ Not evaluated ☐ Unknown	Cells/µl
CD4 T-cells: _____	☐ Not evaluated ☐ Unknown	Cells/µl
CD8 T-cells: _____	☐ Not evaluated ☐ Unknown	Cells/µl
B-cells (i.e. CD19): _____	☐ Not evaluated ☐ Unknown	Cells/µl
NK-cells (CD16/CD56): _____	☐ Not evaluated ☐ Unknown	Cells/µl
Naive CD4 T-cells (CD4/CD45RA): _____	☐ Not evaluated ☐ Unknown	☐ % of CD4 ☐ Cells/µl
Naive CD8 T-cells (CD8/CD45RA): _____	☐ Not evaluated ☐ Unknown	☐ % of CD8 ☐ Cells/µl
IgG: _____	☐ Not evaluated ☐ Unknown	Gram/l
IgA: _____	☐ Not evaluated ☐ Unknown	Gram/l
IgM: _____	☐ Not evaluated ☐ Unkown	Gram/l

APPENDIX 1

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

10p13-p14 deletion syndrome	BRCA2	CD3D	COPG1	ERBB2IP	GOF	IL21
11q23del (Jacobsen)	BRIP1	CD3E	CORO1A	ERCC4	HAVCR2	IL21R
14q32	BTK	CD3G	CRACR2A	ERCC6L2	HAX1	IL23R
22q.11.2 deletion syndrome	C1QA	CD3Z	CSF2RA	ERZ	HCK	IL27RA
ACD	C1QB	CD40	CSF3R	EXTL3	HELLS	IL2RA
ACP5	C1QC	CD40LG	CSFR2B	FAAP24	HMOX	IL2RB
ACTB	C1R	CD46	CTC1	FADD	HYOU1	IL2RG
ADA	C1S	CD55	CTLA4	FANCA	ICOS	IL36RN
ADA2	C2	CD59	CTNNBL1	FANCB	ICOSLG	IL6R
ADAM17	C2orf69	CD70	CTPS1	FANCC	IFIH1	IL6ST
ADAR1	C3	CD79A	CTSC	FANCD2	IFNAR1	IL7R
AICDA	C4A	CD79B	CXCR2	FANCE	IFNAR2	INO80
AIRE	C4B	CD81	CXCR4	FANCF	IFNG	IRAK1
AK2	C5	CD8A	CYBA	FANCI	IFNGR1	IRAK4
ALPI	C6	CDC42	CYBB	FANCL	IFNGR2	IRF1
ALPK1	C7	CDCA7	CYBC1	FANCM	IGHM	IRF2BP2
AP1S3	C8A	CEBPE	DBF4	FAT4	IGKC	IRF3
AP3B1	C8B	CFB	DBR1	FCGR3A	IGLL1	IRF4
AP3D1	C8G	CFD	DCLRE1C	FCHO1	IKBKB	IRF7
APOL1	C9	CFH	DEF6	FCN3	IKBKG	IRF8
Apollo/SNM1B	CARD11	CFHR1	DIAPH1	FERMT1	IKZF1	IRF9
ARHGEF1	CARD14	CFHR2	DKC1	FERMT3	IKZF2	ISG15
ARPC1B	CARD9	CFHR3	DNAJC21	FLT3L	IKZF3	ITCH
ARPC5	CARMIL2	CFHR4	DNASE1L3	FNIP1	IL10	ITGB2
ATAD3A	CASP10	CFHR5	DNASE2	FOXN1	IL10RA	ITK
ATG4	CASP8	CFI	DNMT3B	FOXP3	IL10RB	ITPKB
ATM	CBLB	CFP	DOCK11	FPR1	IL12B	ITPR3
ATP6AP1	CCBE1	CFTR	DOCK2	G6PC3	IL12RB1	JAGN1
B2M	CCR2	CHD7	DOCK8	G6PD	IL12RB2	JAK1
BACH2	CD19	CHUK	DPP9	G6PT1	IL17F	JAK3
BCL10	CD20	CIB1	DUT	GATA2	IL17RA	KARS1
BCL11B	CD21	CIITA	EFL1	GFI1	IL17RC	KDM6A
BLM	CD27	CLCN7	ELANE	GIMAP6	IL18BP	KMT2A
BLNK	CD274	CLPB	ELF4	GINS1	IL-1R1	KMT2D
BRCA1	CD28	COPA	EPG5	GINS4	IL1RN	KRAS

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

LAMTOR2	NCF1	PIK3CD	RAC2	SASH3	STAT4	TMC8	WAS
LAT	NCF2	PIK3CG	RAD51	SBDS	STAT5B	TMEM173	WDR1
LCK	NCF4	PIK3R1	RAD51C	SEC61A1	STAT6	TNFAIP3	WIPF1
LCP2	NCKAP1L	PLCG1	RAG1	SEMA3E	STIM1	TNFRSF11A	WRAP53
LIG1	NCSTN	PLCG2	RAG2	SERPING1	STK4	TNFRSF13B	XIAP
LIG4	NFAT1	PLEKHM1	RANBP2	SGPL1	STN1	TNFRSF13C	XRCC2
LPIN2	NFAT5	PMS2	RASGRP1	SH2B3	STX11	TNFRSF1A	XRCC9
LRBA	NFATC1	PMVK	RBCK1	SH2D1A	STXBP2	TNFRSF4	ZAP70
LSM11	NFE2L2	PNP	REL	SH3BP2	SYK	TNFRSF6	ZBTB24
LY96	NFKB1	POLA1	RELA	SH3KBP1	TAP1	TNFSF11	ZNF341
LYN	NFKB2	POLD1	RELB	SHARPIN	TAP2	TNFSF12	ZNFX1
LYST	NFKBIA	POLD2	RFWD3	SKIV2L	TAPBP	TNFSF13	
MAD2L2	NHEJ1	POLE1	RFX5	SLC19A1	TAZ	TNFSF6	
MAGT1	NLRC4	POLE2	RFXANK	SLC29A3	TBK1	TNFSF9	
MALT1	NLRP1	POLR3A	RFXAP	SLC35C1	TBX1	TOP2B	
MAN2B2	NLRP12	POLR3C	RHBDF2	SLC39A7	TBX21	TP53	
MAP1LC3B2	NLRP3	POLR3F	RHOG	SLC46A1	TCF3	TPP2	
MAP3K14	NOD2	POMP	RHOH	SLC7A7	TCIRG1	TRAC	
MAPK8	NOLA2	POU2AF1	RIPK1	SLX4	TCN2	TRAF3	
MASP2	NOLA3	PRF1	RIPK3	SMARCAL1	TERC	TRAF3IP2	
MCM10	NOS2	PRKCD	RMRP	SMARCD2	TERT	TREX1	
MCM4	NRAS	PRKDC	RNASEH2A	SNORA31	TET2	TRIM22	
MCTS1	NSMCE3	PSEN	RNASEH2B	SNX10	TFRC	TRNT1	
MECOM	OAS1	PSENEN	RNASEH2C	SOCS1	TGFB1	TTC37	
MEFV	OAS2	PSMB10	RNASEL	SP110	TGFBR1	TTC7A	
MKL1	ORAI1	PSMB4	RNF168	SPI1	TGFBR2	TYK2	
MOGS	OSTM1	PSMB8	RNF31	SPINK5	THBD	UBA1	
MSH6	OTULIN	PSMB9	RNU4ATAC	SPPL2A	TICAM1	UBE2T	
MSN	PALB2	PSMD12	RNU7-1	SRP19	TINF2	UNC13D	
MTHFD1	PARN	PSMG2	RORC	SRP54	TIRAP	UNC93B1	
MVK	PAX1	PSTPIP1	RPSA	SRP72	TLR3	UNG	
MYD88	PAX5	PTCRA	RTEL1	SRPRA	TLR4	USB1	
MYSM1	PDCD1	PTEN	SAMD9	STAT1	TLR7	USP18	
NBAS	PEPD	PTPRC	SAMD9L	STAT2	TLR8	VPS13B	
NBS1	PGM3	RAB27A	SAMHD1	STAT3	TMC6	VPS45	

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 --

ALAS2	GP1BB	MYH9
ANKRD26	GP9	NBEAL2
CA2	HOXA11	NEMO
CLCN7	ITGA2B	RANK
ETV6	ITGA2B	RBM8A
FECH	ITGB3	RUNX1
FLI1	ITGB3	SLFN14
		TCIRG1
GNE	MECOM	THPO
GP1BA	MPL	UROS