North West GLH R15 Primary Immunodeficiency Panel Proforma

NHS Manchester University NHS Foundation Trust

NHS North West NHS Genomic Laboratory Hub

PATIENT:	
SURNAME	
FIRST NAME	
DATE OF BIRTH / SEX	
NHS NUMBER	
HOSPITAL NUMBER	
HOSPITAL LAB NUMBER	
ETHNIC ORIGIN	
SAMPLE DATE/TIME	
ADDRESS & POSTCODE	

REFERRAL INFORMATION:	
Physician name	
Physician phone	
Physician email	
Dept. name	
Dept. email	
Dept. address	
Local genetics hub/ address	
Lab phone	
Lab email	

Indication for testing:	Family history:	Suspected inborn error type(s):
❑ DIAGNOSTIC TEST ❑ CARRIER TEST ❑ PREDICTIVE TEST	❑ Consanguinity? ❑ Previous cases in family? Gene/mutation if known:	❑ Unknown ❑ Combined immunodeficiency ❑ Predominantly antibody deficiency ❑ Immune dysregulation ❑ Phagocyte defect ❑ Defect of intrinsic/innate immunity ❑ Autoinflammatory disorder ❑ Complement disorder
Pedigree:	Index patient's name & DOB: Patient's relation to index:	
	Urgent?	

Abnormal infection history? (tick box if feature present)	**Site, organism (if known)**
Recurrent &/or severe bacterial infection	
Sepsis	
Recurrent &/or severe viral infection	
Chronic mucocutaneous candidiasis	
Opportunistic infection	
Susceptibility to mycobacterial disease	
Other infection, please specify:	

Organomegaly, lympho-proliferation, neoplasia?
Hepatomegaly
Splenomegaly
Lymphadenopathy
Lymphoma Specify:
Other Specify:

Autoinflammatory/autoimmune features?		
Fever	Aphthous ulcers	SLE
Haemophagocytosis	Elevated ESR	Urticaria
Cytopenias	Amyloidosis	Vasculitis
Intracranial calcification	Arthritis	Atypical HUS
CNS symptoms	Hypothyroidism	Dermatitis
GLILD	Other, please specify:	
Other granuloma		

Impaired growth & nutrition?
Failure to thrive
Diarrhoea
Enteropathy / enterocolitis Specify age at onset (yr): Small bowel / large bowel Histology:
Microcephaly
Dysmorphic features Specify:

Please provide any results of laboratory investigations performed:

Immunoglobulin levels g/L (required)		**Specific antibody production (required)**		
			Pre-booster	**(Post-booster)**
IgG*				
IgA		Tetanus		
IgM		HiB		
IgE (IU/L)		Pneumococcus		
*Please state if on Ig replacement:		Other (specify)		

Haematology (required)	
Hb (g/L)	
WBC	
Neutrophils	
Lymphocytes	
Platelets	
Mean platelet vol (if available)	❑ machine, fL ❑ Film
Monocytes	
Blood film	

Complement (if relevant)	
C3	
C4	
CH100	
AP100	

Autoantibodies (if relevant)	
DAT (Coomb's)	
ANA	
Other autoantibody (please specify)	

HLH lab parameters (if relevant)	
Ferritin	
Triglycerides	
Fibrinogen	
sCD25	
other	

Lymphocyte subsets (cells/µl)	
CD3	
CD4	
CD8	
B cells	❑ CD19 or ❑ CD20?
NK cells	

Additional immunophenotype data (if available)	
Class-switched memory B cells %	
Neonatal TREC count	❑ normal ❑ abnormal
Naïve CD4 (specify units)	
Naïve CD8 (specify units)	
TCR$\gamma\delta$ %	
TCR$\alpha\beta$+CD4-CD8- (DNT) %	
HLA-DR+ T %	
TCR Vβ usage	❑ normal ❑ abnormal
T cell proliferation to PHA	❑ normal ❑ abnormal
Neutrophil oxidative burst	❑ normal ❑ abnormal

Any other diagnostic information (please detail, including reason urgent if stated above):