NHS
North Thames
Genomic Laboratory Hub

R15 Primary Immunodeficiency Pre-Test Proforma

To be sent with sample and request card to your local NHS Genomic Laboratory Hub

PATIENT DETAILS:		REFERRAL INFORMATION:	
Surname		Physician name	
First name		Physician phone	
Date of birth / sex	F ☐ M ☐ Other ☐	Physician email	
NHS Number		Dept. name	
Hospital Number		Dept. email	
Hospital lab number		Dept. address	
Ethnic origin		Local genetics hub/address	
Sample date / time		Lab phone	
Address & Postcode		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
Pedigree:	Index patient's name & DOB: Index patient's DOB Patient's relation to index:	☐ Phagocyte defect ☐ Defect of intrinsic/innate immunity ☐ Autoinflammatory disorder ☐ Complement disorder
Urgent? Yes/No Please select an option from the drop d		

Abnormal infection history? (detail if feature present)	Site, organism (if known)	Organomegaly, lympho-proliferation, neoplasia?	
Recurrent &/or severe bacterial infection		Hepatomegaly	
Sepsis		Splenomegaly	
Recurrent &/or severe viral infection		Lymphadenopathy	
Chronic mucocutaneous candidiasis		Lymphoma Specify:	
Opportunistic infection		Other Specify:	
Susceptibility to mycobacterial disease			
Other infection, please specify:			

Autoinflammatory/autoimmune features? (tick box if feature present)			**Impaired growth & nutrition?**	
☐ Fever	☐ Aphthous ulcers	☐ SLE	Failure to thrive	
☐Haemophagocytosis	☐ Elevated ESR	☐ Urticaria	Diarrhoea	
☐ Cytopenias	☐ Amyloidosis	☐ Vasculitis	Enteropathy / enterocolitis	
☐ Intracranial calcification	☐ Arthritis	☐ Atypical HUS	Microcephaly	
☐ CNS symptoms	☐ Hypothyroidism	☐ Dermatitis	Dysmorphic features, specify:	
☐ GLILD	Other, please specify:			
☐ Other granuloma				

Please provide any results of laboratory investigations performed:

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

Haematology (required)		**Lymphocyte subsets (cells/µl)**	
Hb (g/L)		CD3	
WBC		CD4	
Neutrophils		CD8	
Lymphocytes		B cells	☐CD19 ☐CD20
Platelets		NK cells	
Mean platelet volume (if available)			
Monocytes			
Blood film			

Complement (if relevant)		**Autoantibodies (if relevant)**	
C3		DAT (Coomb's)	
C4		ANA	
CH100		Other autoantibody (please specify)	
AP100			

HLH lab parameters (if relevant)	
Ferritin	
Fibrinogen	
sCD25	
Other	

Additional immunophenotype data (if available)	
Class-switched memory B cells %	
Neonatal TREC count	Please select an option from the drop down
Naïve CD4 (specify units)	
Naïve CD8 (specify units)	
TCRgd %	
TCRab+CD4-CD8- (DNT) %	
HLA-DR+ T %	
TCR Vb usage	Please select an option from the drop down
T cell proliferation to PHA	Please select an option from the drop down
Neutrophil oxidative burst	Please select an option fro the drop down

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