Genetic Immunology Flagship
Optimal Clinical Data
Version 1, 24.9.19

Australian Genomics

GI Subtype					
☐ Autoimmune ☐ Autoinflammatory ☐ CVID & CID			☐ HLH ☐ SCID		
Autoimmune Disease					
Age of onset:			Psychosis	Yes ☐	No ☐
Malar rash	Yes ☐	No ☐	Seizures	Yes ☐	No ☐
Photosensitivity	Yes ☐	No ☐	Proteinuria	Yes ☐	No ☐
Alopecia	Yes ☐	No ☐	Glomerulonephritis (on biopsy)	Yes ☐	No ☐
Aphthous ulcers	Yes ☐	No ☐	Lymphocyte count:	_______________	(x10^9/L)
Arthritis	Yes ☐	No ☐	White cell count:	_______________	(x10^9/L)
Serositis	Yes ☐	No ☐	Platelet count:	_______________	(x10^9/L)
Antinuclear antibodies: _______________ (titre)					
dsDNA antibodies	Yes ☐	No ☐	CNS vasculitis	Yes ☐	No ☐
Sm antibodies	Yes ☐	No ☐	Autoimmune gastritis/pernicious anaemia	Yes ☐	No ☐
Cardiolipin antibodies	Yes ☐	No ☐	Autoimmune thyroid disease	Yes ☐	No ☐
B2 glycoprotein 1 antibodies	Yes ☐	No ☐	Autoimmune parathyroid disease	Yes ☐	No ☐
Lupus anticoagulant	Yes ☐	No ☐	Autoimmune adrenalitis	Yes ☐	No ☐
Low C3	Yes ☐	No ☐	Hypophysitis	Yes ☐	No ☐
Low C4	Yes ☐	No ☐	Gonadal failure	Yes ☐	No ☐
Pneumonitis	Yes ☐	No ☐	Inflammatory bowel disease	Yes ☐	No ☐
Autoimmune liver disease	Yes ☐	No ☐			
Treatment:					
Steroids (equivalent to >1mg/kg prednisone)	Yes ☐	No ☐	Rituximab	Yes ☐	No ☐
Azathioprine	Yes ☐	No ☐	Abatacept	Yes ☐	No ☐
Mycophenolate	Yes ☐	No ☐	Other Mab	Yes ☐	No ☐
Cyclophosphamide	Yes ☐	No ☐			
Autoinflammatory Disease					
Recurrent fevers:	(Number)		Serositis:	☐ Pleuris ☐ Pericarditis ☐ Peritonitis	
Pharyngitis	Yes ☐	No ☐	Amyloidosis:	☐ Kidney ☐ Skin ☐ Other	
Aphthous ulcers	Yes ☐	No ☐	Erysipelas-like erythema	Yes ☐	No ☐
Neutrophil count: _____________	(x10^9/L)		C-reactive protein (while afebrile)	____________	(mg/L)
Urticaria	Yes ☐	No ☐	Musculoskeletal symptoms	Yes ☐	No ☐
Cold-induced manifestations	Yes ☐	No ☐	Aseptic meningitis	Yes ☐	No ☐
Stress-induced manifestations	Yes ☐	No ☐	Splenomegaly	Yes ☐	No ☐
Sensorineural hearing loss	Yes ☐	No ☐	Response to colchicine	Yes ☐	No ☐
Common Variable Immunodeficiency (CVID)/Combined Immune Deficiency (CID)					
Bacterial infection:	☐ Pneumonia ☐ Meningitis ☐ Sinusitis		Autoimmune cytopenia:	☐ Anaemia ☐ Thrombocytopenia	

Opportunistic infections: ☐ PJP ☐ Mucocutaneous candidiasis ☐ Low virulence mycobacteria ☐ Disseminated viral infection	Other autoimmunity/inflammation: ☐ Pneumonitis ☐ Colitis ☐ Coeliac disease ☐ Thyroiditis ☐ Adrenalitis ☐ Hypophysitis ☐ Arthritis
Granulomas: Yes ☐ No ☐	
IgG: ________________ (g/L)	
IgM: ________________ (g/L)	
IgA: ________________ (g/L)	
Lymphocyte count: ________________ (x 10^9/L)	Neutrophil count: ________________ (x 10^9/L)
CD19+ B cell count: ________________ (x 10^9/L)	CD3+ T cell count: ________________ (x 10^9/L)
Immunoglobulin replacement Yes ☐ No ☐	

Hemophagocytic Lymphohistiocytosis (HLH)

Fever: ____________ °C	
Splenomegaly: Yes ☐ No ☐	
Haemoglobin: _____________ (g/L)	
Haemophagocytosis:	
Bone marrow: Yes ☐ No ☐ ND ☐	Lymph node: Yes ☐ No ☐ ND ☐
Spleen: Yes ☐ No ☐ ND ☐	Liver: Yes ☐ No ☐ ND ☐
Platelets: _____________ (x 10^9/L)	Neutrophil count: _____________ (x 10^9/L)
Triglycerides (fasting) - Please check yes to enter data if applicable. Yes ☐ ND ☐ Triglycerides (fasting): _____________ (mmol/L)	Fibrinogen - Please check yes to enter data if applicable. Yes ☐ ND ☐ Fibrinogen: _____________ (mg/dL)
Ferritin - Please check yes to enter data if applicable. Yes ☐ ND ☐ Fibrinogen: _____________ (ng/mL)	Serum CD25 - Please check yes to enter data if applicable. Yes ☐ ND ☐ Serum CD25: _____________ (U/ mL)
Absent NK cell cytotoxicity: Yes ☐ No ☐ ND ☐	High suspicion of CNS involvement: Yes ☐ No ☐

Herpesvirus infection:			Associated clinical syndrome:		
HSV	☐ Yes	☐ No	Autoimmune disease	☐ Yes	☐ No
EBV	☐ Yes	☐ No	Malignancy	☐ Yes	☐ No
VZV	☐ Yes	☐ No	Metabolic disease	☐ Yes	☐ No
HHV6	☐ Yes	☐ No	Leishmania infection	☐ Yes	☐ No
HHV8	☐ Yes	☐ No			

Treatment:		
Corticosteroids	☐ Yes	☐ No
Etoposide	☐ Yes	☐ No
Cyclosporin	☐ Yes	☐ No
Antithymocyte globulin	☐ Yes	☐ No

Severe Combined Immunodeficiency (SCID)

CD3+ T cells: _____________ (x10^6/L)	CD3+ CD45RO+ T cells: ___________ (%CD3+ cells)
Maternal T cells: Yes ☐ No ☐	
T cell proliferation Mitogen: _____________ (%)	T cell proliferation Antigen: _____________ (%)
Neutrophil count: (x10^6/L)	

Erythroderma: Yes ☐ No ☐
Sensorineural deafness: Yes ☐ No ☐