

# Supraregional Service for Molecular Diagnosis of Primary Immune Deficiencies

Departments of Immunology and Clinical Molecular Genetics  
Camelia Botnar Laboratories

**\* COMPLETE BOTH SIDES OF THIS FORM IN FULL \***

SAMPLES CAN NOT BE ANALYSED IF UNSUITABLE /OLD/ INSUFFICIENT

**Patient:**

Name

Address (including postcode)

Date of Birth:

Sex

Patient NHS number

**For GOS Use:**

Date received at GOS	
GOS ref No	
Sample to Genetics	
Immunology lab No	

Date & Time of Sample		
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**Referring Physician:**

Name

Phone

Address

Fax

Email

Health authority/Trust

**Analysis requested**

- |                                      |                          |  |
|--------------------------------------|--------------------------|--|
| BTK                                  | <input type="checkbox"/> | 5 ml EDTA (patient and control)                        |
| CD40                                 | <input type="checkbox"/> | 2 ml EDTA (patient and control)                        |
| CD40 Ligand                          | <input type="checkbox"/> | 10 ml EDTA +2 ml Lithium Heparin (patient and control) |
| CGD Proteins                         | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| Common gamma chain                   | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| gp91                                 | <input type="checkbox"/> | 5 ml EDTA (patient and control)                        |
| HLH proteins                         | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| (SAP/Perforin/Granule release assay) |                          |  |
| IL-7Ra                               | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| JAK3                                 | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| Perforin                             | <input type="checkbox"/> | 5 ml EDTA (patient and control)                        |
| SAP                                  | <input type="checkbox"/> | 5 ml EDTA (patient and control)                        |
| Stat5 phosphorylation                | <input type="checkbox"/> | 5 ml EDTA (patient and control)                        |
| WASP                                 | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| ZAP-70                               | <input type="checkbox"/> | 10 ml EDTA (patient and control)                       |
| Other (please specify) _____         |                          | Check with laboratory                                  |

**Clinical Details**

Family History

(include pedigree and names of known affected family members if available and any consanguinity)

Ethnicity

Age at presentation

Major infections

Other complications/comments

Immunoglobulin levels g/l

(please indicate if levels performed on or off replacement immunoglobulin therapy)

IgG	
IgA	
IgM	

Full blood count

Hb	
WBC	
Neutrophils	
Lymphocytes	
Platelets	
Mean platelet volume (machine/film)	

Lymphocyte subpopulations

CD3	
CD4	
CD8	
B cells*	
NK cells	

\*please indicate if CD19 or 20

Specific antibody production

	Pre-booster	Post-Booster
Tetanus		
Hib		
Pneumococcus		
Other (specify)		

For HLH complete the following

Cytopenias		fibrinogen	
Fever		rash	
splenomegaly/lympadenopathy		CNS symptoms	
ferritin		Haemophagocytosis (site)	
triglycerides		Other	

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