

All samples MUST be labelled with FULL name, date of birth and NHS number

Processing of samples will be delayed if information is incomplete

**Send samples at room temperature
by 1st class post or courier to:**

East of England, Genomic Medicine Centre
Clinical Genetics
Genetics Laboratories, Box 143, ATC Level 6, Addenbrooke's Hospital,
Cambridge University Hospital
Hills Road, Cambridge, CB2 0QQ

Laboratory opening hours: 8.30am - 5.30pm Monday to Friday
Telephone: 01223 348866
Fax: 01223 348712
Email: geneticslaboratories@nhs.net

For further information about sample requirements and tests available see:
www.cuh.org.uk/genetics-labs

Please select one or more relevant genes below. This will help the Multi Disciplinary Team (MDT) to generate a report. The sample will be tested for all the genes on the platform using the next generation sequencing.

For further information about the diagnostic GRID test:
please visit: gridgenomics.org.uk

(Gene list - Version GRID V2.0)

I. Immunodeficiencies affecting cellular and humoral immunity	
ADA	<input type="checkbox"/>
AK2	<input type="checkbox"/>
B2M	<input type="checkbox"/>
BCL10	<input type="checkbox"/>
CARD11	<input type="checkbox"/>
CD247	<input type="checkbox"/>
CD27	<input type="checkbox"/>
CD3D	<input type="checkbox"/>
CD3E	<input type="checkbox"/>
CD3G	<input type="checkbox"/>
CD40	<input type="checkbox"/>
CD40LG	<input type="checkbox"/>
CD8A	<input type="checkbox"/>
CIITA	<input type="checkbox"/>
CORO1A	<input type="checkbox"/>
CTPS1	<input type="checkbox"/>
DCLRE1C	<input type="checkbox"/>
DOCK2	<input type="checkbox"/>
DOCK8	<input type="checkbox"/>
ICOS	<input type="checkbox"/>
IKBKB	<input type="checkbox"/>
IL21	<input type="checkbox"/>
IL21R	<input type="checkbox"/>
IL2RG	<input type="checkbox"/>
IL7R	<input type="checkbox"/>
ITK	<input type="checkbox"/>
JAK3	<input type="checkbox"/>
LCK	<input type="checkbox"/>
LIG4	<input type="checkbox"/>
LRBA	<input type="checkbox"/>
MAGT1	<input type="checkbox"/>
MALT1	<input type="checkbox"/>
MAP3K14 (NIK)*	<input type="checkbox"/>
NHEJ1 (Cernunnos/XLF)*	<input type="checkbox"/>
PRKDC	<input type="checkbox"/>
PTPRC	<input type="checkbox"/>
RAG1	<input type="checkbox"/>
RAG2	<input type="checkbox"/>
RFX5	<input type="checkbox"/>
RFXANK	<input type="checkbox"/>
RFXAP	<input type="checkbox"/>
RHOH	<input type="checkbox"/>
STK4 (MST1)*	<input type="checkbox"/>
TAP1	<input type="checkbox"/>
TAP2	<input type="checkbox"/>
TAPBP	<input type="checkbox"/>
TNFRSF4 (OX40)*	<input type="checkbox"/>
TRAC	<input type="checkbox"/>
UNC119	<input type="checkbox"/>
ZAP70	<input type="checkbox"/>

II. CID with associated or syndromic features	
ACD	<input type="checkbox"/>
ATM	<input type="checkbox"/>
BLM	<input type="checkbox"/>
CCBE1	<input type="checkbox"/>
CHD7	<input type="checkbox"/>
DCLRE1B	<input type="checkbox"/>
DKC1 (DKC)*	<input type="checkbox"/>
DNMT3B	<input type="checkbox"/>
EPG5	<input type="checkbox"/>
FOXN1	<input type="checkbox"/>
IKBKG (NEMO)*	<input type="checkbox"/>
MCM4	<input type="checkbox"/>
MTHFD1	<input type="checkbox"/>
NFKBIA (IKBA)*	<input type="checkbox"/>
NHP2 (NOLA2)*	<input type="checkbox"/>
NBN (NBS1)*	<input type="checkbox"/>
NOP10 (NOLA3)*	<input type="checkbox"/>
ORAI1	<input type="checkbox"/>
PARN	<input type="checkbox"/>
PGM3	<input type="checkbox"/>
PMS2	<input type="checkbox"/>
RNF31 (HOIP1)*	<input type="checkbox"/>
PNP (NP)*	<input type="checkbox"/>
POLE (POLE1)*	<input type="checkbox"/>
RBCK1 (HOIL1)*	<input type="checkbox"/>
RMRP	<input type="checkbox"/>
RNF168	<input type="checkbox"/>
RTEL1	<input type="checkbox"/>
SEMA3E	<input type="checkbox"/>
SLC46A1	<input type="checkbox"/>
SMARCAL1	<input type="checkbox"/>
SPINK5	<input type="checkbox"/>
SP110	<input type="checkbox"/>
STAT3	<input type="checkbox"/>
STAT5B	<input type="checkbox"/>
STIM1	<input type="checkbox"/>
TBX1	<input type="checkbox"/>
TCN2	<input type="checkbox"/>
TERC	<input type="checkbox"/>
TERT	<input type="checkbox"/>
TINF2	<input type="checkbox"/>
TTC7A	<input type="checkbox"/>
WAS	<input type="checkbox"/>
WIPF1	<input type="checkbox"/>
ZBTB24	<input type="checkbox"/>

*non official HGNC gene name

CUH Laboratory Use Only:		Other Information:
Receipt date and time:		
Volume:	No of tubes:	
Tube type:		

III. Predominantly antibody deficiencies			
AICDA	<input type="checkbox"/>	MSH6	<input type="checkbox"/>
BLNK	<input type="checkbox"/>	MS4A1 (CD20*)	<input type="checkbox"/>
BTK	<input type="checkbox"/>	MOGS	<input type="checkbox"/>
CARD11	<input type="checkbox"/>	NFKB2	<input type="checkbox"/>
CD19	<input type="checkbox"/>	PIK3CD	<input type="checkbox"/>
CD79A	<input type="checkbox"/>	PIK3R1	<input type="checkbox"/>
CD79B	<input type="checkbox"/>	SART3 (p110)*	<input type="checkbox"/>
CD81	<input type="checkbox"/>	TCF3	<input type="checkbox"/>
CR2 (CD21)*	<input type="checkbox"/>	TNFRSF13B (TACI)*	<input type="checkbox"/>
DKC1 (DKC)*	<input type="checkbox"/>	TNFRSF13C (BAFF-R)*	<input type="checkbox"/>
GATA2	<input type="checkbox"/>	TNFSF12 (TWEAK)*	<input type="checkbox"/>
IGKC	<input type="checkbox"/>	TRNT1	<input type="checkbox"/>
IGHM	<input type="checkbox"/>	TTC37	<input type="checkbox"/>
IGLL1	<input type="checkbox"/>	UNG	<input type="checkbox"/>
INO80	<input type="checkbox"/>		

IV. Diseases of immune dysregulation			
ACPS5	<input type="checkbox"/>	LYST	<input type="checkbox"/>
ADAR (ADAR1)*	<input type="checkbox"/>	NFAT5	<input type="checkbox"/>
AIRE	<input type="checkbox"/>	PRF1	<input type="checkbox"/>
AP3B1	<input type="checkbox"/>	PRKCD	<input type="checkbox"/>
BLOC1S6 (PLDN)*	<input type="checkbox"/>	RAB27A	<input type="checkbox"/>
CASP10	<input type="checkbox"/>	RNASEH2A	<input type="checkbox"/>
CASP8	<input type="checkbox"/>	RNASEH2B	<input type="checkbox"/>
CECR1 (ADA2)*	<input type="checkbox"/>	RNASEH2C	<input type="checkbox"/>
CTLA4	<input type="checkbox"/>	SAMHD1	<input type="checkbox"/>
FADD	<input type="checkbox"/>	SH2D1A	<input type="checkbox"/>
FASLG	<input type="checkbox"/>	STAT3	<input type="checkbox"/>
FOXP3	<input type="checkbox"/>	STX11	<input type="checkbox"/>
FAS (TNFRSF6)*	<input type="checkbox"/>	STXB2	<input type="checkbox"/>
IFIH1	<input type="checkbox"/>	TMEM173 (STING)*	<input type="checkbox"/>
IL10	<input type="checkbox"/>	TPP2	<input type="checkbox"/>
IL10RA	<input type="checkbox"/>	TREX1	<input type="checkbox"/>
IL10RB	<input type="checkbox"/>	UNC13D	<input type="checkbox"/>
IL2RA	<input type="checkbox"/>	XIAP	<input type="checkbox"/>
ITCH	<input type="checkbox"/>		

V. Congenital defects of phagocyte number, function, or both			
ACTB	<input type="checkbox"/>	ITGB2	<input type="checkbox"/>
CEBPE	<input type="checkbox"/>	JAGN1	<input type="checkbox"/>
CLPB	<input type="checkbox"/>	LAMTOR2 (ROBLD3)*	<input type="checkbox"/>
CSF2RA	<input type="checkbox"/>	NCF1	<input type="checkbox"/>
CSF3R	<input type="checkbox"/>	NCF2	<input type="checkbox"/>
CTSC	<input type="checkbox"/>	NCF4	<input type="checkbox"/>
CYBA	<input type="checkbox"/>	RAC2	<input type="checkbox"/>
CYBB	<input type="checkbox"/>	SBDS	<input type="checkbox"/>
ELANE	<input type="checkbox"/>	SLC35C1	<input type="checkbox"/>
FERMT3	<input type="checkbox"/>	SLC37A4 (G6PT1)*	<input type="checkbox"/>
FPR1	<input type="checkbox"/>	TAZ	<input type="checkbox"/>
G6PC3	<input type="checkbox"/>	USB1 (c16orf57)*	<input type="checkbox"/>
GATA2	<input type="checkbox"/>	VPS13B	<input type="checkbox"/>
GFI1	<input type="checkbox"/>	VPS45	<input type="checkbox"/>
HAX1	<input type="checkbox"/>	WAS	<input type="checkbox"/>

VI. Defects in intrinsic and innate immunity			
APOL1	<input type="checkbox"/>	IRF8	<input type="checkbox"/>
CARD9	<input type="checkbox"/>	ISG15	<input type="checkbox"/>
CXCR4	<input type="checkbox"/>	MYD88	<input type="checkbox"/>
CYBB	<input type="checkbox"/>	RORC	<input type="checkbox"/>
FCGR3A (CD16)*	<input type="checkbox"/>	RPSA	<input type="checkbox"/>
FCGR3B (CD16)*	<input type="checkbox"/>	STAT1	<input type="checkbox"/>
IFNGR1	<input type="checkbox"/>	STAT2	<input type="checkbox"/>
IFNGR2	<input type="checkbox"/>	TBK1	<input type="checkbox"/>
IKBK (NEMO)*	<input type="checkbox"/>	TICAM1 (TRIF)*	<input type="checkbox"/>
IL12B	<input type="checkbox"/>	TLR3	<input type="checkbox"/>
IL12RB1	<input type="checkbox"/>	TMC6 (EVER1)*	<input type="checkbox"/>
IL17F	<input type="checkbox"/>	TMC8 (EVER2)*	<input type="checkbox"/>
IL17RA	<input type="checkbox"/>	TRAF3	<input type="checkbox"/>
IL17RC	<input type="checkbox"/>	TRAF3IP2 (ACT1)*	<input type="checkbox"/>
IRAK4	<input type="checkbox"/>	TYK2	<input type="checkbox"/>
IRF7	<input type="checkbox"/>	UNC93B1	<input type="checkbox"/>

VII. Auto-inflammatory disorders			
ADAM17	<input type="checkbox"/>	NLR4	<input type="checkbox"/>
CARD14	<input type="checkbox"/>	NLRP12	<input type="checkbox"/>
COPA	<input type="checkbox"/>	NOD2	<input type="checkbox"/>
IL1RN	<input type="checkbox"/>	PLCG2	<input type="checkbox"/>
IL36RN	<input type="checkbox"/>	PSMB8	<input type="checkbox"/>
LPIN2	<input type="checkbox"/>	PSTPIP1	<input type="checkbox"/>
MEFV	<input type="checkbox"/>	SH3BP2	<input type="checkbox"/>
MVK	<input type="checkbox"/>	SLC29A3	<input type="checkbox"/>
NLRP3	<input type="checkbox"/>	TNFRSF1A	<input type="checkbox"/>

VIII. Complement deficiencies			
C1QA	<input type="checkbox"/>	CD59	<input type="checkbox"/>
C1QB	<input type="checkbox"/>	CFB	<input type="checkbox"/>
C1QC	<input type="checkbox"/>	CFD	<input type="checkbox"/>
C1R	<input type="checkbox"/>	CFH	<input type="checkbox"/>
C1S	<input type="checkbox"/>	CFHR1	<input type="checkbox"/>
C2	<input type="checkbox"/>	CFHR2	<input type="checkbox"/>
C3	<input type="checkbox"/>	CFHR3	<input type="checkbox"/>
C4A	<input type="checkbox"/>	CFHR4	<input type="checkbox"/>
C4B	<input type="checkbox"/>	CFHR5	<input type="checkbox"/>
C5	<input type="checkbox"/>	CFI	<input type="checkbox"/>
C6	<input type="checkbox"/>	CFP (PFC)*	<input type="checkbox"/>
C7	<input type="checkbox"/>	FCN3	<input type="checkbox"/>
C8A	<input type="checkbox"/>	ITGAM	<input type="checkbox"/>
C8B	<input type="checkbox"/>	MASP2	<input type="checkbox"/>
C8G	<input type="checkbox"/>	SERPING1	<input type="checkbox"/>
C9	<input type="checkbox"/>	THBD	<input type="checkbox"/>
CD46	<input type="checkbox"/>		

IX. Phenocopies of PID		Additional genes	
CSF2RA	<input type="checkbox"/>	ARPC1B	<input type="checkbox"/>
FAS (TNFRSF6)*	<input type="checkbox"/>	F12	<input type="checkbox"/>
IL17A	<input type="checkbox"/>	IKZF1 (IKAROS)*	<input type="checkbox"/>
IL22	<input type="checkbox"/>	LIG1	<input type="checkbox"/>
KRAS	<input type="checkbox"/>	MBL2	<input type="checkbox"/>
NLRP3	<input type="checkbox"/>	MPO	<input type="checkbox"/>
NRAS	<input type="checkbox"/>	MRE11	<input type="checkbox"/>
STAT3	<input type="checkbox"/>	NFKB1	<input type="checkbox"/>

CUH Laboratory Use Only:

Receipt date and time:	
Volume:	No of tubes:
Tube type:	

Other Information:

*non official HGNC gene

Clinical Synopsis

CUH Laboratory Use Only:

Receipt date and time:		Other Information:
Volume:	No of tubes:	
Tube type:		