

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: www.gridgenomics.org.uk or contact Ilenia Simeoni PhD info@gridgenomics.org.uk

(Gene list - Version GRID V2.0)

I. Immunodeficiencies affecting cellular and humoral immunity		II. CID with associated or syndromic features	
ADA	<input type="checkbox"/>	ITK	<input type="checkbox"/>
AK2	<input type="checkbox"/>	JAK3	<input type="checkbox"/>
B2M	<input type="checkbox"/>	LCK	<input type="checkbox"/>
BCL10	<input type="checkbox"/>	LIG4	<input type="checkbox"/>
CARD11	<input type="checkbox"/>	LRBA	<input type="checkbox"/>
CD247	<input type="checkbox"/>	MAGT1	<input type="checkbox"/>
CD27	<input type="checkbox"/>	MALT1	<input type="checkbox"/>
CD3D	<input type="checkbox"/>	MAP3K14 (NIK)*	<input type="checkbox"/>
CD3E	<input type="checkbox"/>	NHEJ1 (Cernunnos/XLF)*	<input type="checkbox"/>
CD3G	<input type="checkbox"/>	PRKDC	<input type="checkbox"/>
CD40	<input type="checkbox"/>	PTPRC	<input type="checkbox"/>
CD40LG	<input type="checkbox"/>	RAG1	<input type="checkbox"/>
CD8A	<input type="checkbox"/>	RAG2	<input type="checkbox"/>
CIITA	<input type="checkbox"/>	RFX5	<input type="checkbox"/>
CORO1A	<input type="checkbox"/>	RFXANK	<input type="checkbox"/>
CTPS1	<input type="checkbox"/>	RFXAP	<input type="checkbox"/>
DCLRE1C	<input type="checkbox"/>	RHOH	<input type="checkbox"/>
DOCK2	<input type="checkbox"/>	STK4 (MST1)*	<input type="checkbox"/>
DOCK8	<input type="checkbox"/>	TAP1	<input type="checkbox"/>
ICOS	<input type="checkbox"/>	TAP2	<input type="checkbox"/>
IKBKB	<input type="checkbox"/>	TAPBP	<input type="checkbox"/>
IL21	<input type="checkbox"/>	TNFRSF4 (OX40)*	<input type="checkbox"/>
IL21R	<input type="checkbox"/>	TRAC	<input type="checkbox"/>
IL2RG	<input type="checkbox"/>	UNC119	<input type="checkbox"/>
IL7R	<input type="checkbox"/>	ZAP70	<input type="checkbox"/>
ACD	<input type="checkbox"/>	POLE (POLE1)*	<input type="checkbox"/>
ATM	<input type="checkbox"/>	RBCK1 (HOIL1)*	<input type="checkbox"/>
BLM	<input type="checkbox"/>	RMRP	<input type="checkbox"/>
CCBE1	<input type="checkbox"/>	RNF168	<input type="checkbox"/>
CHD7	<input type="checkbox"/>	RTEL1	<input type="checkbox"/>
DCLRE1B	<input type="checkbox"/>	SEMA3E	<input type="checkbox"/>
DKC1 (DKC)*	<input type="checkbox"/>	SLC46A1	<input type="checkbox"/>
DNMT3B	<input type="checkbox"/>	SMARCAL1	<input type="checkbox"/>
EPG5	<input type="checkbox"/>	SPINK5	<input type="checkbox"/>
FOXN1	<input type="checkbox"/>	SP110	<input type="checkbox"/>
IKBKG (NEMO)*	<input type="checkbox"/>	STAT3	<input type="checkbox"/>
MCM4	<input type="checkbox"/>	STAT5B	<input type="checkbox"/>
MTHFD1	<input type="checkbox"/>	STIM1	<input type="checkbox"/>
NFKBIA (IKBA)*	<input type="checkbox"/>	TBX1	<input type="checkbox"/>
NHP2 (NOLA2)*	<input type="checkbox"/>	TCN2	<input type="checkbox"/>
NBN (NBS1)*	<input type="checkbox"/>	TERC	<input type="checkbox"/>
NOP10 (NOLA3)*	<input type="checkbox"/>	TERT	<input type="checkbox"/>
ORAI1	<input type="checkbox"/>	TINF2	<input type="checkbox"/>
PARN	<input type="checkbox"/>	TTC7A	<input type="checkbox"/>
PGM3	<input type="checkbox"/>	WAS	<input type="checkbox"/>
PMS2	<input type="checkbox"/>	WIPF1	<input type="checkbox"/>
RNF31 (HOIP1)*	<input type="checkbox"/>	ZBTB24	<input type="checkbox"/>
PNP (NP)*	<input type="checkbox"/>		

*non official HGNC gene name

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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"/>

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Clinical Synopsis

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