

## R91 Cytopenia panel (NOT Fanconi anaemia)

Can be analysed as a whole panel – all genes analysed

Or as individual sub-panels – please specify in referral

Panel	Sub-panel	Gene	Transcript
Cytopenia	Thrombocytopenia	ABCG5	NM_022436
Cytopenia	Thrombocytopenia	ABCG8	NM_022437
Cytopenia	Thrombocytopenia	ACTN1	NM_001130004
Cytopenia	Thrombocytopenia	ADAMTS13	NM_139025
Cytopenia	Thrombocytopenia	ANKRD26	NM_014915
Cytopenia	Thrombocytopenia	AP3B1	NM_003664
Cytopenia	Thrombocytopenia	CECR1 (ADA2)	NM_001282225
Cytopenia	Thrombocytopenia	CYCS	NM_018947
Cytopenia	Thrombocytopenia	ETV6	NM_001987
Cytopenia	Thrombocytopenia	FLI1	NM_002017
Cytopenia	Thrombocytopenia	FYB1	NM_001465
Cytopenia	Thrombocytopenia	GATA1	NM_002049
Cytopenia	Thrombocytopenia	GP1BA	NM_000173
Cytopenia	Thrombocytopenia	GP1BB	NM_000407
Cytopenia	Thrombocytopenia	GP9	NM_000174
Cytopenia	Thrombocytopenia	IKZF1	NM_006060
Cytopenia	Thrombocytopenia	LAT	NM_001014987
Cytopenia	Thrombocytopenia	MECOM	NM_004991
Cytopenia	Thrombocytopenia	MPL	NM_005373
Cytopenia	Thrombocytopenia	MYH9	NM_002473
Cytopenia	Thrombocytopenia	NBEAL2	NM_015175
Cytopenia	Thrombocytopenia	RUNX1	NM_001754
Cytopenia	Thrombocytopenia	SAMD9	NM_017654
Cytopenia	Thrombocytopenia	SLFN14	NM_001129820
Cytopenia	Thrombocytopenia	SRC	NM_005417
Cytopenia	Thrombocytopenia	STIM1	NM_003156
Cytopenia	Thrombocytopenia	THPO	NM_000460
Cytopenia	Thrombocytopenia	TUBB1	NM_030773
Cytopenia	Thrombocytopenia	WAS	NM_000377
Cytopenia	Thrombocytopenia	WIPF1	NM_001077269
Cytopenia	Neutropenia	ACKR1	NM_002036
Cytopenia	Neutropenia	AP3B1	NM_003664
Cytopenia	Neutropenia	CECR1 (ADA2)	NM_001282225
Cytopenia	Neutropenia	CSF3R	NM_156039
Cytopenia	Neutropenia	CXCR4	NM_003467
Cytopenia	Neutropenia	ELANE	NM_001972
Cytopenia	Neutropenia	FCGR3B	NM_001244753
Cytopenia	Neutropenia	G6PC3	NM_138387

Cytopenia	Neutropenia	GFI1	NM_005263
Cytopenia	Neutropenia	HAX1	NM_006118
Cytopenia	Neutropenia	JAGN1	NM_032492
Cytopenia	Neutropenia	RAC2	NM_002872
Cytopenia	Neutropenia	RMRP	NR_003051
Cytopenia	Neutropenia	TAZ	NM_000116
Cytopenia	Neutropenia	TCN2	NM_000355
Cytopenia	Neutropenia	USB1	NM_024598
Cytopenia	Neutropenia	VPS45	NM_007259
Cytopenia	Neutropenia	WAS	NM_000377
Cytopenia	Neutropenia	WIPF1	NM_001077269
Cytopenia	Inherited bone marrow failure	ACD	NM_001082486
Cytopenia	Inherited bone marrow failure	ANKRD26	NM_014915
Cytopenia	Inherited bone marrow failure	CECR1 (ADA2)	NM_001282225
Cytopenia	Inherited bone marrow failure	CTC1	NM_025099
Cytopenia	Inherited bone marrow failure	CXCR4	NM_003467
Cytopenia	Inherited bone marrow failure	DKC1	NM_001363
Cytopenia	Inherited bone marrow failure	DNAJC21	NM_001012339
Cytopenia	Inherited bone marrow failure	EFL1	NM_024580
Cytopenia	Inherited bone marrow failure	ERCC6L2	NM_020207
Cytopenia	Inherited bone marrow failure	G6PC3	NM_138387
Cytopenia	Inherited bone marrow failure	GATA2	NM_032638
Cytopenia	Inherited bone marrow failure	GFI1	NM_005263
Cytopenia	Inherited bone marrow failure	HAX1	NM_006118
Cytopenia	Inherited bone marrow failure	IKZF1	NM_006060
Cytopenia	Inherited bone marrow failure	KIF23	NM_138555
Cytopenia	Inherited bone marrow failure	KLF1	NM_006563
Cytopenia	Inherited bone marrow failure	LAT	NM_001014987
Cytopenia	Inherited bone marrow failure	MPL	NM_005373
Cytopenia	Inherited bone marrow failure	MYSM1	NM_001085487
Cytopenia	Inherited bone marrow failure	NBN	NM_002485
Cytopenia	Inherited bone marrow failure	NHP2	NM_017838
Cytopenia	Inherited bone marrow failure	NOP10	NM_018648
Cytopenia	Inherited bone marrow failure	PARN	NM_002582
Cytopenia	Inherited bone marrow failure	RTEL1	NM_001283009
Cytopenia	Inherited bone marrow failure	RUNX1	NM_001754
Cytopenia	Inherited bone marrow failure	SAMD9	NM_017654
Cytopenia	Inherited bone marrow failure	SAMD9L	NM_152703
Cytopenia	Inherited bone marrow failure	SBDS	NM_016038
Cytopenia	Inherited bone marrow failure	SRP72	NM_006947
Cytopenia	Inherited bone marrow failure	TCN2	NM_000355
Cytopenia	Inherited bone marrow failure	TERC	NR_001566
Cytopenia	Inherited bone marrow failure	TERT	NM_198253
Cytopenia	Inherited bone marrow failure	TINF2	NM_001099274
Cytopenia	Inherited bone marrow failure	USB1	NM_024598
Cytopenia	Inherited bone marrow failure	VPS45	NM_007259

Cytopenia	Inherited bone marrow failure	WRAP53	NM_018081
Cytopenia	Diamond-Blackfan anaemia	CECR1 (ADA2)	NM_001282225
Cytopenia	Diamond-Blackfan anaemia	GATA1	NM_002049
Cytopenia	Diamond-Blackfan anaemia	RPL5	NM_000969
Cytopenia	Diamond-Blackfan anaemia	RPL9	NM_001024921
Cytopenia	Diamond-Blackfan anaemia	RPL11	NM_000993
Cytopenia	Diamond-Blackfan anaemia	RPL15	NM_002948
Cytopenia	Diamond-Blackfan anaemia	RPL18	NM_000979
Cytopenia	Diamond-Blackfan anaemia	RPL26	NM_000987
Cytopenia	Diamond-Blackfan anaemia	RPL27	NM_000988
Cytopenia	Diamond-Blackfan anaemia	RPL31	NM_000993
Cytopenia	Diamond-Blackfan anaemia	RPL35A	NM_000996
Cytopenia	Diamond-Blackfan anaemia	RPS7	NM_001011
Cytopenia	Diamond-Blackfan anaemia	RPS10	NM_001014
Cytopenia	Diamond-Blackfan anaemia	RPS17	NM_001021
Cytopenia	Diamond-Blackfan anaemia	RPS19	NM_001022
Cytopenia	Diamond-Blackfan anaemia	RPS24	NM_033022
Cytopenia	Diamond-Blackfan anaemia	RPS26	NM_001029
Cytopenia	Diamond-Blackfan anaemia	RPS27	NM_001030
Cytopenia	Diamond-Blackfan anaemia	RPS28	NM_001031
Cytopenia	Diamond-Blackfan anaemia	RPS29	NM_001032
Cytopenia	Diamond-Blackfan anaemia	TSR2	NM_058163

## R92 Rare anaemia panel

Can be analysed as a whole panel – all genes analysed

Or as individual sub-panels – please specify in referral

Panel	Sub-panel	Gene	Transcript
Rare anaemia	Membranopathy	ANK1	NM_000037
Rare anaemia	Membranopathy	EPB41	NM_001166005
Rare anaemia	Membranopathy	EPB42	NM_000119
Rare anaemia	Membranopathy	KCNN4	NM_002250
Rare anaemia	Membranopathy	PIEZO1	NM_001142864
Rare anaemia	Membranopathy	RHAG	NM_000324
Rare anaemia	Membranopathy	SLC2A1	NM_006516
Rare anaemia	Membranopathy	SLC4A1	NM_000342
Rare anaemia	Membranopathy	SPTA1	NM_003126
Rare anaemia	Membranopathy	SPTB	NM_001024858
Rare anaemia	Membranopathy	XK	NM_021083
Rare anaemia	Enzymopathy	AK1	NM_000476
Rare anaemia	Enzymopathy	ALDOA	NM_000034
Rare anaemia	Enzymopathy	CYB5R3	NM_000398
Rare anaemia	Enzymopathy	G6PD	NM_000402

Rare anaemia	Enzymopathy	GCLC	NM_001498
Rare anaemia	Enzymopathy	GPI	NM_000175
Rare anaemia	Enzymopathy	GSR	NM_000637
Rare anaemia	Enzymopathy	GSS	NM_000178
Rare anaemia	Enzymopathy	HK1	NM_033496
Rare anaemia	Enzymopathy	NT5C3A	NM_016489
Rare anaemia	Enzymopathy	PFKM	NM_000289
Rare anaemia	Enzymopathy	PGK1	NM_000291
Rare anaemia	Enzymopathy	PKLR	NM_000298
Rare anaemia	Enzymopathy	TPI1	NM_000365
Rare anaemia	Haemoglobinopathy	HBA1	NM_000558
Rare anaemia	Haemoglobinopathy	HBA2	NM_000517
Rare anaemia	Haemoglobinopathy	HBB	NM_000518
Rare anaemia	Haemoglobinopathy	HBD	NM_000519
Rare anaemia	Haemoglobinopathy	HBG1	NM_000559
Rare anaemia	Haemoglobinopathy	HBG2	NM_000184
Rare anaemia	Congenital dyserythropoietic anaemia	C15orf41 (CDIN1)	NM_001130010
Rare anaemia	Congenital dyserythropoietic anaemia	CDAN1	NM_138477
Rare anaemia	Congenital dyserythropoietic anaemia	COX4I2	NM_032609
Rare anaemia	Congenital dyserythropoietic anaemia	GATA1	NM_002049
Rare anaemia	Congenital dyserythropoietic anaemia	KIF23	NM_138555
Rare anaemia	Congenital dyserythropoietic anaemia	KLF1	NM_006563
Rare anaemia	Congenital dyserythropoietic anaemia	LPIN2	NM_014646
Rare anaemia	Congenital dyserythropoietic anaemia	SEC23B	NM_006363
Rare anaemia	Diamond-Blackfan anaemia	CECR1 (ADA2)	NM_001282225
Rare anaemia	Diamond-Blackfan anaemia	GATA1	NM_002049
Rare anaemia	Diamond-Blackfan anaemia	RPL5	NM_000969
Rare anaemia	Diamond-Blackfan anaemia	RPL9	NM_001024921
Rare anaemia	Diamond-Blackfan anaemia	RPL11	NM_000993
Rare anaemia	Diamond-Blackfan anaemia	RPL15	NM_002948
Rare anaemia	Diamond-Blackfan anaemia	RPL18	NM_000979
Rare anaemia	Diamond-Blackfan anaemia	RPL26	NM_000987
Rare anaemia	Diamond-Blackfan anaemia	RPL27	NM_000988
Rare anaemia	Diamond-Blackfan anaemia	RPL31	NM_000988
Rare anaemia	Diamond-Blackfan anaemia	RPL35A	NM_000996
Rare anaemia	Diamond-Blackfan anaemia	RPS7	NM_001011
Rare anaemia	Diamond-Blackfan anaemia	RPS10	NM_001014
Rare anaemia	Diamond-Blackfan anaemia	RPS17	NM_001021
Rare anaemia	Diamond-Blackfan anaemia	RPS19	NM_001022
Rare anaemia	Diamond-Blackfan anaemia	RPS24	NM_033022
Rare anaemia	Diamond-Blackfan anaemia	RPS26	NM_001029
Rare anaemia	Diamond-Blackfan anaemia	RPS27	NM_001030
Rare anaemia	Diamond-Blackfan anaemia	RPS28	NM_001031
Rare anaemia	Diamond-Blackfan anaemia	RPS29	NM_001032
Rare anaemia	Diamond-Blackfan anaemia	TSR2	NM_058163

Rare anaemia	Sideroblastic anaemia	ABCB7	NM_004299
Rare anaemia	Sideroblastic anaemia	ALAS2	NM_000032
Rare anaemia	Sideroblastic anaemia	GLRX5	NM_016417
Rare anaemia	Sideroblastic anaemia	HSPA9	NM_004134
Rare anaemia	Sideroblastic anaemia	NDUFB11	NM_019056
Rare anaemia	Sideroblastic anaemia	PUS1	NM_025215
Rare anaemia	Sideroblastic anaemia	SLC25A38	NM_017875
Rare anaemia	Sideroblastic anaemia	TRNT1	NM_182916
Rare anaemia	Sideroblastic anaemia	LARS2	NM_015340
Rare anaemia	Sideroblastic anaemia	YARS2	NM_001040436
Rare anaemia	Megaloblastic anaemia	AMN	NM_030943
Rare anaemia	Megaloblastic anaemia	CUBN	NM_001081
Rare anaemia	Megaloblastic anaemia	DHFR	NM_000791
Rare anaemia	Megaloblastic anaemia	GIF (CBLIF)	NM_005142
Rare anaemia	Megaloblastic anaemia	MTR	NM_000254
Rare anaemia	Megaloblastic anaemia	MTRR	NM_002454
Rare anaemia	Megaloblastic anaemia	SLC19A2	NM_006996
Rare anaemia	Megaloblastic anaemia	TCN2	NM_000355
Rare anaemia	Megaloblastic anaemia	UMPS	NM_000373
Rare anaemia	Sitosterolaemia	ABCG5	NM_022436
Rare anaemia	Sitosterolaemia	ABCG8	NM_022437
Rare anaemia	Rare anaemia (whole panel only)	ATRX	NM_000489
Rare anaemia	Rare anaemia (whole panel only)	CD59	NM_203330
Rare anaemia	Rare anaemia (whole panel only)	MAD2L2	NM_006341
Rare anaemia	Rare anaemia (whole panel only)	NHP2	NM_017838
Rare anaemia	Rare anaemia (whole panel only)	SBDS	NM_016038
Rare anaemia	Rare anaemia (whole panel only)	SLC11A2	NM_000617
Rare anaemia	Rare anaemia (whole panel only)	STEAP3	NM_182915
Rare anaemia	Rare anaemia (whole panel only)	TF	NM_001063
Rare anaemia	Rare anaemia (whole panel only)	THPO	NM_000460
Rare anaemia	Rare anaemia (whole panel only)	TMPRSS6	NM_001289000

## R96 Iron metabolism disorders

Whole panel or single gene analysis only

Panel	Gene	Transcript
Iron metabolism disorders	ABCB7	NM_004299
Iron metabolism disorders	ALAS2	NM_000032
Iron metabolism disorders	ATP7B	NM_000053
Iron metabolism disorders	BMP6	NM_001718
Iron metabolism disorders	CP	NM_000096
Iron metabolism disorders	CYBRD1	NM_024843
Iron metabolism disorders	FECH	NM_001012515
Iron metabolism disorders	FTH1	NM_002032

Iron metabolism disorders	FTL	NM_000146
Iron metabolism disorders	GBA	NM_000157
Iron metabolism disorders	GLRX5	NM_016417
Iron metabolism disorders	HAMP	NM_021175
Iron metabolism disorders	HEPH	NM_138737
Iron metabolism disorders	HFE	NM_000410
Iron metabolism disorders	HFE2 (HJV)	NM_213653
Iron metabolism disorders	SLC11A2	NM_000617
Iron metabolism disorders	SLC25A38	NM_017875
Iron metabolism disorders	SLC40A1	NM_014585
Iron metabolism disorders	STEAP3	NM_182915
Iron metabolism disorders	TF	NM_001063
Iron metabolism disorders	TFR2	NM_003227
Iron metabolism disorders	TMPRSS6	NM_001289000

## R168 Porphyria

Whole panel or single gene analysis only

Panel	Gene	Transcript
Porphyria	ALAD	NM_000031
Porphyria	ALAS2	NM_000032
Porphyria	CPOX	NM_000097
Porphyria	FECH	NM_001012515
Porphyria	GATA1	NM_002049
Porphyria	HMBS	NM_000190
Porphyria	PPOX	NM_001122764
Porphyria	UROD	NM_000374
Porphyria	UROS	NM_000375

## R347 Inherited predisposition to acute myeloid leukaemia (AML)

Whole panel or single gene analysis only

Panel	Gene	Transcript
Inherited predisposition to AML	ACD	NM_001082486
Inherited predisposition to AML	ANKRD26	NM_014915
Inherited predisposition to AML	CEBPA	NM_004364
Inherited predisposition to AML	CHEK2	NM_007194
Inherited predisposition to AML	DDX41	NM_016222
Inherited predisposition to AML	ETV6	NM_001987
Inherited predisposition to AML	GATA2	NM_032638
Inherited predisposition to AML	RTEL1	NM_001283009
Inherited predisposition to AML	RUNX1	NM_001754
Inherited predisposition to AML	SAMD9	NM_017654

Inherited predisposition to AML	SRP72	NM_006947
Inherited predisposition to AML	TERC	NR_001566
Inherited predisposition to AML	TERT	NM_198253
Inherited predisposition to AML	TP53	NM_000546

### R366 Inherited predisposition to acute lymphocytic leukaemia (ALL)

Whole panel analysis only

Panel	Gene	Transcript
Inherited predisposition to ALL	ETV6	NM_001987
Inherited predisposition to ALL	PAX5	NM_016734

### R405 Hereditary erythrocytosis

Whole panel or single gene analysis only

Panel	Gene	Transcript
Hereditary erythrocytosis	BPGM	NM_001724
Hereditary erythrocytosis	EGLN1	NM_022051
Hereditary erythrocytosis	EGLN2	NM_080732
Hereditary erythrocytosis	EGLN3	NM_022073
Hereditary erythrocytosis	EPAS1	NM_001430
Hereditary erythrocytosis	EPO	NM_000799
Hereditary erythrocytosis	EPOR	NM_000121
Hereditary erythrocytosis	HIF1A	NM_001530
Hereditary erythrocytosis	HBA1	NM_000558
Hereditary erythrocytosis	HBA2	NM_000517
Hereditary erythrocytosis	HBB	NM_000518
Hereditary erythrocytosis	PKLR	NM_000298
Hereditary erythrocytosis	VHL	NM_000551

### R406 Hereditary thrombocythaemia

Whole panel analysis only

Panel	Gene	Transcript
Hereditary thrombocythaemia	CALR	NM_004343
Hereditary thrombocythaemia	JAK2	NM_004972
Hereditary thrombocythaemia	MPL	NM_005373
Hereditary thrombocythaemia	SH2B3	NM_005475
Hereditary thrombocythaemia	THPO	NM_000460

## Haemophagocytic lymphohistiocytosis (HLH)

Whole panel analysis only

Panel	Gene	Transcript
HLH	PRF1	NM_001083116
HLH	STX11	NM_003764
HLH	STXBP2	NM_006949
HLH	UNC13D	NM_199242

## Single gene analysis

Panel	Gene	Transcript
Single gene only	SERPINA1	NM_000295
Single gene only	UGT1A1	NM_000463