

R91 Cytopenia panel (NOT Fanconi anaemia) (Panelapp v3)

Whole panel or single gene analysis only

Panel	Gene	Transcript
Cytopenia	ACD	NM_001082486
Cytopenia	ACKR1	NM_002036
Cytopenia	AK2	NM_001625
Cytopenia	ANKRD26	NM_014915
Cytopenia	CECR1 (ADA2)	NM_001282225
Cytopenia	CLPB	NM_030813
Cytopenia	CSF3R	NM_156039
Cytopenia	CTC1	NM_025099
Cytopenia	CXCR4	NM_003467
Cytopenia	CYCS	NM_018947
Cytopenia	DDX41	NM_016222
Cytopenia	DKC1	NM_001363
Cytopenia	DNAJC21	NM_001012339
Cytopenia	EFL1	NM_024580
Cytopenia	ELANE	NM_001972
Cytopenia	ERCC6L2	NM_020207
Cytopenia	ETV6	NM_001987
Cytopenia	FYB (FYB1)	NM_001465
Cytopenia	G6PC3	NM_138387
Cytopenia	GATA1	NM_002049
Cytopenia	GATA2	NM_032638
Cytopenia	GFI1	NM_005263
Cytopenia	HAX1	NM_006118
Cytopenia	HEATR3	NM_182922
Cytopenia	JAGN1	NM_032492
Cytopenia	KLF1	NM_006563
Cytopenia	MECOM	NM_004991
Cytopenia	MPL	NM_005373
Cytopenia	MYH9	NM_002473
Cytopenia	MYSM1	NM_001085487
Cytopenia	NBEAL2	NM_015175
Cytopenia	NHP2	NM_017838
Cytopenia	PARN	NM_002582
Cytopenia	RMRP	NR_003051
Cytopenia	RPA1	NM_002945

Next generation sequencing gene panel list – RCGPv7

Cytopenia	RPL11	NM_000975
Cytopenia	RPL15	NM_002948
Cytopenia	RPL31	NM_000993
Cytopenia	RPL35A	NM_000996
Cytopenia	RPL5	NM_000969
Cytopenia	RPL9	NM_001024921
Cytopenia	RPS10	NM_001014
Cytopenia	RPS17	NM_001021
Cytopenia	RPS19	NM_001022
Cytopenia	RPS24	NM_033022
Cytopenia	RPS26	NM_001029
Cytopenia	RPS29	NM_001032
Cytopenia	RPS7	NM_001011
Cytopenia	RTEL1	NM_001283009
Cytopenia	RUNX1	NM_001754
Cytopenia	SAMD9	NM_017654
Cytopenia	SAMD9L	NM_152703
Cytopenia	SBDS	NM_016038
Cytopenia	SRC	NM_005417
Cytopenia	SRP54	NM_003136
Cytopenia	STN1	NM_024928
Cytopenia	TAZ	NM_000116
Cytopenia	TERC	NR_001566
Cytopenia	TERT	NM_198253
Cytopenia	THPO	NM_000460
Cytopenia	TINF2	NM_001099274
Cytopenia	USB1	NM_024598
Cytopenia	VPS45	NM_007259
Cytopenia	WAS	NM_000377
Cytopenia	WIPF1	NM_001077269
Cytopenia	WRAP53	NM_018081

R92 Rare anaemia panel (Panelapp v3)

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

Next generation sequencing gene panel list – RCGPv7

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	Haemoglobinopathy	SUPT5H	NM_001111020
Rare anaemia	Congenital dyserythropoietic anaemia	C15orf41 (CDIN1)	NM_001130010
Rare anaemia	Congenital dyserythropoietic anaemia	CDAN1	NM_138477
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	Congenital dyserythropoietic anaemia	VPS4A	NM_013245
Rare anaemia	Diamond-Blackfan anaemia	CECR1 (ADA2)	NM_001282225
Rare anaemia	Diamond-Blackfan anaemia	GATA1	NM_002049
Rare anaemia	Diamond-Blackfan anaemia	HEATR3	NM_182922
Rare anaemia	Diamond-Blackfan anaemia	RPL5	NM_000969
Rare anaemia	Diamond-Blackfan anaemia	RPL9	NM_001024921
Rare anaemia	Diamond-Blackfan anaemia	RPL11	NM_000975
Rare anaemia	Diamond-Blackfan anaemia	RPL15	NM_002948

Next generation sequencing gene panel list – RCGPv7

Rare anaemia	Diamond-Blackfan anaemia	RPL31	NM_000993
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	RPS29	NM_001032
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	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)	CD59	NM_203330
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)	TF	NM_001063
Rare anaemia	Rare anaemia (whole panel only)	TMPRSS6	NM_001289000

R96 Iron metabolism disorders (Panelapp v2)

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

Next generation sequencing gene panel list – RCGPv7

Iron metabolism disorders	BMP6	NM_001718
Iron metabolism disorders	CP	NM_000096
Iron metabolism disorders	CYBRD1	NM_024843
Iron metabolism disorders	FTL	NM_000146
Iron metabolism disorders	GBA (GBA1)	NM_000157
Iron metabolism disorders	GLRX5	NM_016417
Iron metabolism disorders	HAMP	NM_021175
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	TF	NM_001063
Iron metabolism disorders	TFR2	NM_003227
Iron metabolism disorders	TMPRSS6	NM_001289000

R168 Porphyria (Panelapp v1.4)

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	HMBS	NM_000190
Porphyria	PPOX	NM_001122764
Porphyria	UROD	NM_000374
Porphyria	UROS	NM_000375
Porphyria	GATA1	NM_002049

R347 Inherited predisposition to acute myeloid leukaemia (AML) (Panelapp v3)

Whole panel or single gene analysis only

Panel	Gene	Transcript
Inherited predisposition to AML	ANKRD26	NM_014915
Inherited predisposition to AML	CEBPA	NM_004364
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	MBD4	NM_003925

Inherited predisposition to AML	RUNX1	NM_001754
Inherited predisposition to AML	SAMD9L	NM_152703
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) (Panelapp v1)

Whole panel analysis only

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

R405 Hereditary erythrocytosis (Panelapp v2)

Whole panel or single gene analysis only

Panel	Gene	Transcript
Hereditary erythrocytosis	BPGM	NM_001724
Hereditary erythrocytosis	EGLN1	NM_022051
Hereditary erythrocytosis	EPAS1	NM_001430
Hereditary erythrocytosis	EPO	NM_000799
Hereditary erythrocytosis	EPOR	NM_000121
Hereditary erythrocytosis	HBA1	NM_000558
Hereditary erythrocytosis	HBA2	NM_000517
Hereditary erythrocytosis	HBB	NM_000518
Hereditary erythrocytosis	SLC30A10	NM_018713
Hereditary erythrocytosis	VHL	NM_000551

R406 Hereditary thrombocythaemia

Whole panel analysis only

Panel	Gene	Transcript
Hereditary thrombocythaemia	JAK2	NM_004972
Hereditary thrombocythaemia	MPL	NM_005373
Hereditary thrombocythaemia	THPO	NM_000460

R169 Acute intermittent porphyria (Panelapp v1)

Panel	Gene	Transcript
Porphyria	HMBS	NM_000190

R170 Variegate porphyria (Panelapp v1)

Panel	Gene	Transcript
Porphyria	PPOX	NM_001122764

R313 Neutropenia consistent with ELANE mutations (Panelapp v1)

Panel	Gene	Transcript
Neutropenia	ELANE	NM_001972

R323 Sitosterolaemia

Panel	Gene	Transcript
Sitosterolaemia	ABCG5	NM_022436
Sitosterolaemia	ABCG8	NM_022437

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