

## Next Generation Sequencing: Red Cell Panel V4 - Subpanels

Red cell membranopathy	
Gene	Transcript
ABCG5	NM_022436.2
ABCG8	NM_022437.2
ADD1	NM_014189.3
ADD2	NM_001617.3
AK1	NM_000476.2
ANK1	NM_001142446.1
APOB	NM_000384.2
DMTN	NM_001978
EPB41	NM_001166005.1
EPB42	NM_000119.2
KCNN4	NM_002250.2
MTTP	NM_001300785.1
PIEZO1	NM_001142864.2
RhAG	NM_000324.2
SLC2A1	NM_006516.2
SLC4A1	NM_000342.3
SPTA1	NM_003126.2
SPTB	NM_001024858.2
STOM	NM_004099.5
TMOD1	NM_003275.3
TPM3	NM_153649.3
XK	NM_021083.2

Red Cell Enzyme	
Gene	Transcript
AK1	NM_000476.2
ALDOA	NM_000034.3
BPGM	NM_199186.2
CYB5A	NM_148923.3
CYB5R1	NM_016243.2
CYB5R2	NM_001302826.1
CYB5R3	NM_001129819.2
CYB5R4	NM_016230.3
CYB5RL	NM_001031672.2
ENO1	NM_001428.3
G6PD	NM_000402.4
GAPDH	NM_002046.5
GCLC	NM_001498.3
GPI	NM_001289789.1
GPX1	NM_000581.2
GSR	NM_000637.3
GSS	NM_000178.2
HK1	NM_033496.2
HK2	NM_000189.4
NT5C3A	NM_001002010.2
PFKM	NM_001166686.1
PGAM1	NM_002629.2
PGD	NM_002631.3
PGK1	NM_000291.3
PGM1	NM_001172818.1
PKLR	NM_000298.5
TPI1	NM_001159287.1

Haemoglobinopathies	
Gene	Transcript
AHSP	NM_016633.2
Alpha globin HS40	Genomic region
ATRX	NM_000489.4
Beta globin LCR HS1-5	Genomic region
HBA1	NM_000558.4
HBA2	NM_000517.4
HBB	NM_000518.4
HBD	NM_000519.3
HBE1	NM_005330.3
HBG1	NM_000559.2
HBG2	NM_000184.2
HBM	NM_001003938.3
HBQ1	NM_005331.4
HBZ	NM_005332.2

## Next Generation Sequencing: Red Cell Panel V4 - Subpanels

Congenital Dyserythropoietic Anaemia	
Gene	Transcript
CDAN1	NM_138477.2
C15ORF41	NM_001130010.2
COX4I2	NM_032609.2
GATA1	NM_002049.3
GATA2	NM_032638.4
KIF23	NM_138555.3
KLF1	NM_006563.3
LPIN2	NM_014646.2
SEC23B	NM_032985.4
TAL1	NM_003189.5

Megaloblastic Anaemia	
Gene	Transcript
AMN	NM_030943.3
CUBN	NM_001081.3
DHFR	NM_000791
FTCD	NM_206965.1
GIF	NM_005142.2
MT-CO1	YP_003024028.1
MTR	NM_000254.2
MTRR	NM_024010.2
SLC19A2	NM_006996.2
UMPS	NM_000373.3

Congenital Erythrocytosis	
Gene	Transcript
BHLHE41	NM_030762.2
BPGM	NM_199186.2
EGLN1	NM_022051.2
EGLN2	NM_080732.3
EGLN3	NM_022073.3
EPAS1	NM_001430.4
EPO	NM_000799.2
EPOR	NM_000121.3
GFI1B	NM_004188.6
HBA1	NM_000558.4
HBA2	NM_000517.4
HBB	NM_000518.4
HIF1A	NM_001243084.1
HIF1AN	NM_017902.2
HIF3A	NM_152795.3
JAK2	NM_004972.3
KDM6A	NM_001291415.1
OS9	NM_006812.3
SH2B3	NM_005475.2
VHL	NM_000551.3
ZNF197	NM_006991.3

Diamond-Blackfan Anaemia	
Gene	Transcript
CECR1	NM_001282225.1
GATA1	NM_002049.3
RPL5	NM_000969.3
RPL9	NM_000661.4
RPS10	NM_001203245.2
RPL11	NM_000975.3
RPL15	NM_002948.3
RPL19	NM_000981.3
RPL26	NM_001315530.1
RPL27	NM_000988.3
RPL35A	NM_001316311.1
RPS7	NM_001011.3
RPS19	NM_001022.3
RPS24	NM_001142285.1
RPS26	NM_001029.3
RPS29	NM_001032.4

## Next Generation Sequencing: Red Cell Panel V4 - Subpanels

Inherited Bone Marrow Failure	
Gene	Transcript
ACD	NM_001082486.1
CTC1	NM_025099.5
DKC1	NM_001363.4
GATA2	NM_032638.4
HOXA11	NM_005523.5
LIG4	NM_002312.3
MASTL	NM_001320757.1
MPL	NM_005373.2
NHP2	NM_017838.3
NOP10	NM_018648.3
NT5C3A	NM_001002010.2
PALB2	NM_024675.3
PARN	NM_002582.3
RAD51C	NM_058216.2
RMRP	NR_003051.3
RTEL1	NM_001283009.1
RUNX1	NM_001754.4
SBDS	NM_016038.2
SMARCAL1	NM_014140.3
SRP72	NM_006947.3
TERC	NR_001566.1
TERT	NM_198253.2
TINF2	NM_001099274.1
WRAP53	NM_018081.2
XRCC2	NM_005431.1

Inherited Thrombocytopenia	
Gene	Transcript
ANKRD26	NM_014915.2
CTC1	NM_025099.5
GP1BA	NM_000173.6
HOXA11	NM_005523.5
MASTL	NM_001320757.1
MPL	NM_005373.2
MYH9	NM_002473.5
NBEAL2	NM_015175.2
RBM8A	NM_005105.4
RMRP	NR_003051.3
RUNX1	NM_001754.4
SMARCAL1	NM_014140.3
TINF2	NM_001099274.1
TUBB1	NM_030773.3
WAS	NM_000377.2

Porphyria	
Gene	Transcript
ALAD	NM_000031.5
ALAS2	NM_000032.4
CPOX	NM_000097.5
FECH	NM_001012515.2
GATA1	NM_002049.3
HMBS	NM_000190.3
PPOX	NM_001122764.1
UROD	NM_000374.4
UROS	NM_000375.2

Inherited Neutropenia	
Gene	Transcript
ACKR1	NM_002036.3
CECR1	NM_001282225.1
CXCR4	NM_001348056.1
ELANE	NM_001972.3
G6PC3	NM_138387.3
GFI1	NM_005263.3
HAX1	NM_006118.3
SLC37A4	NM_001164277
SMARCAL1	NM_014140.3
TAZ	NM_000116.4
USB1	NM_024598.3
WAS	NM_000377.2

Sideroblastic Anaemia	
Gene	Transcript
ABCB6	NM_005689.2
ABCB7	NM_004299.4
ALAS1	NM_000688.5
ALAS2	NM_000032.4
GLRX5	NM_016417.2
MT-CO1	YP_003024028.1
PUS1	NM_001002020.2
SF3B1	NM_012433.3
SLC19A2	NM_006996.2
SLC25A38	NM_017875.2
YARS2	NM_001040436.2

## Next Generation Sequencing: Red Cell Panel V4 - Subpanels

Iron Regulation	
Gene	Transcript
BMP4	NM_001202.5
BMP6	NM_001718
CP	NM_000096.3
FTH1	NM_002032.2
FTL	NM_000146.3
HAMP	NM_021175
HFE	NM_000410.3
HFE2	NM_213653.3
SLC11A2	NM_001174125.1
SLC40A1	NM_014585
SMAD4	NM_005359
SMAD6	NM_005585.4
SMAD7	NM_005904.3
TF	NM_001063.3
TFR2	NM_003227.3
TFRC	NM_003234.2
TMPRSS6	NM_001289000.1

Lymphedema	
Gene	Transcript
CCBE1	NM_133459.3
FAT4	NM_001291303.1
GATA2	NM_0032638.4
PIEZO1	NM_001142864.3

Single Genes	
Gene	Transcript
HP	NM_005143.3
SERPINA1	NM_000295.4
UGT1A1	NM_000463.2

Sex Chromosome Markers	
Gene	Transcript
AMELX	NM_182680.1
SRY	

HLH	
Gene	Transcript
PRF1	NM_001083116.1
STX11	NM_003764.3
STXBP2	NM_001272034.1
UNC13D	NM_199242.2