

Next Generation Sequencing: Red Cell Panel V3 - Subpanels

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
EPB41	NM_001166005.1
EPB42	NM_000119.2
EPB49 (DMTN)	NM_001978
GYPA	NM_002099.7
GYPB	NM_002100.5
GYPE	NM_002101.4
KCNN4	NM_002250.2
MTTP	NM_001300785.1
PIEZO1 (FAM38A)	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 Enzymes	
Gene	Transcript
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
PGM1	NM_002633.2
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

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 (CDANIII)	NM_138555.3
KLF1	NM_006563.3
LPIN2	NM_014646.2
SEC23B	NM_032985.4
TAL1	NM_003189.5

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

Megaloblastic Anaemia	
Gene	Transcript
AMN	NM_030943.3
ATP4A	NM_000704.2
ATP4B	NM_000705.3
CBL	NM_005188.3
CBS	NM_000071.2

Megaloblastic Anaemia continued...	
Gene	Transcript
CD320	NM_016579.3
CUBN	NM_001081.3
DHFR	NM_000791
DUT	NM_001025248.1
FBXO7	NM_012179.3
FTCD	NM_206965.1
GAST	NM_000805.4
GIF	NM_005142.2
HACL1	NM_012260.3
HPRT1	NM_000194.2
LMBRD1	NM_018368.3
MMAA	NM_172250.2
MMACHC	NM_015506.2
MMADHC	NM_015702.2
MT-CO1	YP_003024028.1
MTR	NM_000254.2
MTRR	NM_024010.2
MUT	NM_000255.3
PSG2	NM_031246.3
SLC19A1	NM_194255.2
SLC19A2	NM_006996.2
SLC19A3	NM_025243.3
SLC46A1	NM_080669.5
SLC5A6	NM_021095.2
TCN1	NM_001062.3
TCN2	NM_000355.3
TKT	NM_001135055.2
TPK1	NM_022445.3
UMPS	NM_000373.3

Diamond-Blackfan Anaemia	
Gene	Transcript
RPL11	NM_000975.3
RPL15	NM_002948.3
RPL19	NM_000981.3
RPL26	NM_001315530.1
RPL27	NM_000988.3
RPL35A	NM_001316311.1
RPL5	NM_000969.3
RPL9	NM_000661.4
RPS10	NM_001203245.2
RPS17	NM_001021.4
RPS19	NM_001022.3
RPS24	NM_001142285.1
RPS26	NM_001029.3
RPS29	NM_001032.4
RPS7	NM_001011.3

Bone Marrow Failure	
Gene	Transcript
DKC1	NM_001363.4
GATA1	NM_002049.3
GATA2	NM_032638.4
NHP2	NM_017838.3
NOP10	NM_018648.3
NT5C3A	NM_001002010.2
SBDS	NM_016038.2
TERC	NR_001566.1
TERT	NM_198253.2
TINF2	NM_001099274.1

Sideroblastic Anaemia

Gene	Transcript
ABCB6	NM_005689.2
ABCB7	NM_004299.4
ALAS1	NM_000688.5
ALAS2	NM_000032.4
GLRX5	NM_016417.2
PUS1	NM_001002020.2
SF3B1	NM_012433.3
SLC19A2	NM_006996.2
SLC25A38	NM_017875.2
YARS2	NM_001040436.2

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

Iron Regulation

Gene	Transcript
ACVR1	NM_001105.4
BMP6	NM_001718
BMPR1A	NM_004329
BMPR1B	NM_001256793.1
CCND3	NM_001760.4
CHRD	NM_003741.3
CP	NM_000096.3
CYBRD1	NM_024843.3
FTH1	NM_002032.2
FTL	NM_000146.3
HAMP	NM_021175
HEPH	NM_138737.4
HFE	NM_000410.3
HFE2	NM_213653.3
MCOLN1	NM_020533
NOG	NM_005450.4
PCSK7	NM_004716
SLC11A2	NM_001174125.1
SLC40A1	NM_014585
SMAD4	NM_005359
SMAD6	NM_005585.4
SMAD7	NM_005904.3
STEAP1	NM_012449.2
STEAP3	NM_182915.2
TF	NM_001063.3
TFR2	NM_003227.3
TFRC	NM_003234.2
TMPRSS6	NM_001289000.1

Haemolytic Uraemic Syndrome

Gene	Transcript
CFH	NM_000186.3
CFI	NM_000204.3

Secondary Modifiers

Gene	Transcript
HP	NM_005143.3
MTHFR	NM_005957.4
UGT1A1	NM_000463.2

Single Genes

Gene	Transcript
ATP7B	NM_000053.3
PIGA	NM_002641.3
SERPINA1	NM_000295.4

Sex Chromosome Markers

Gene	Transcript
AMELX	NM_182680.1
SRY	