

## LMM Reference Sequences - GRCh37

### Cardiomyopathy

Gene	Region	Transcript
ABCC9	Exon 01 - 37	NM_005691.2
	Exon 38A	NM_005691.2
	Exon 38B	NM_020297.2
ACTC1	Exon 01 - 06	NM_005159.4
ACTN2	Exon 01 - 21	NM_001103.2
ANKRD1	Exon 01 - 09	NM_014391.2
BAG3	Exon 01 - 04	NM_004281.3
CASQ2	Exon 01 - 11	NM_001232.3
CAV3	Exon 01 - 02	NM_033337.2
CHRM2	Exon 05	NM_001006626.1
CRYAB	Exon 01 - 03	NM_001885.1
CSRP3	Exon 02 - 06	NM_003476.3
DES	Exon 01 - 09	NM_001927.3
DMD	Exon 01	NM_004006.2
	Exon 01A	NM_000109.3
	Exon 01C	NM_004009.3
	Exon 01E	NM_004011.3
	Exon 01F	NM_004012.3
	Exon 01H	NM_004014.2
	Exon 01J	NM_004016.2
	Exon 02 - 09	NM_004006.2
	Exon 09M	NM_004019.2
	Exon 10 - 35	NM_004006.2
	Exon 35O	NM_004021.2
	Exon 36 - 69	NM_004006.2
	Exon 71 - 78	NM_004006.2
DOLK	Exon 01	NM_014908.3
DSC2	Exon 01 - 14	NM_024422.3
	Exon 15A	NM_024422.3
	Exon 15B	NM_004949.3
	Exon 16	NM_024422.3
DSG2	Exon 01 - 15	NM_001943.3
DSP	Exon 01 - 24	NM_004415.2
DTNA	Exon 03 - 12	NM_032975.3
	Exon 12A	NM_001128175.1
	Exon 12B, 12C	NM_032978.6

### Cardiomyopathy

Gene	Region	Transcript
DTNA <i>cont.</i>	Exon 13 - 16	NM_032975.3
	Exon 16A	NM_032978.6
	Exon 17 - 21	NM_032975.3
EMD	Exon 01 - 06	NM_000117.2
FHL2	Exon 02 - 06	NM_201557.3
GATAD1	Exon 01 - 05	NM_021167.3
GLA	Exon 01 - 04	NM_000169.2
	Intron 04	NM_000169.2
	Exon 05 - 07	NM_000169.2
ILK	Exon 01B	NM_001014795.1
	Exon 03 - 13	NM_004517.2
JPH2	Exon 01 - 02	NM_020433.4
	Exon 02A	NM_175913.3
	Exon 03 - 05	NM_020433.4
JUP	Exon 02 - 14	NM_021991.2
LAMA4*	Exon 02 - 06	NM_002290.3
	Exon 07	NM_001105206.1
	Exon 09 - 39	NM_002290.3
<i>Not covered</i>	Exon 08	<i>NM_002290.3</i>
LAMP2	Exon 01 - 08, 09A	NM_002294.2
	Exon 09B	NM_013995.2
	Exon 09C	NM_001122606.1
LDB3	Exon 01 - 04	NM_007078.2
	Exon 05 - 06	NM_001080116.1
	Exon 07 - 08	NM_007078.2
	Exon 09	NM_001080116.1
	Exon 10 - 16	NM_007078.2
LMNA	Exon 01 - 09	NM_170707.2
	Exon 10	NM_005572.3
	Exon 11 - 12	NM_170707.2
MURC	Exon 01 - 02	NM_001018116.1
MYBPC3	Exon 01 - 34	NM_000256.3
MYH6*	Exon 03 - 36	NM_002471.3
	Exon 38 - 39	NM_002471.3
	<i>Not covered</i>	Exon 37
MYH7	Exon 03 - 40	NM_000257.2

### Cardiomyopathy

Gene	Region	Transcript
MYL2	Exon 01 - 07	NM_000432.3
MYL3	Exon 01 - 06	NM_000258.2
MYLK2	Exon 02 - 13	NM_033118.3
MYOM1	Exon 02 - 38	NM_003803.3
MYOZ2	Exon 02 - 06	NM_016599.3
MYPN	Exon 03 - 06	NM_001256267.1
	Exon 06D	NM_001256268.1
	Exon 07 - 21	NM_001256267.1
NEBL	Exon 01	NM_006393.2
	Exon 01A	NM_213569.2
	Exon 02	NM_006393.2
	Exon 02A	NM_213569.2
	Exon 03	NM_006393.2
	Exon 03A	NM_213569.2
	Exon 04	NM_006393.2
	Exon 04A	NM_213569.2
	Exon 05 - 28	NM_006393.2
NEXN	Exon 02 - 13	NM_144573.3
PDLIM3	Exon 01 - 04	NM_014476.4
	Exon 04A	NM_001114107.3
	Exon 05 - 08	NM_014476.4
PKP2	Exon 01 - 14	NM_004572.3
PLN	Exon 02	NM_002667.3
PRDM16	Exon 01 - 17	NM_022114.3
PRKAG2	Exon 01 - 16	NM_016203.3
PTPN11	Exon 01 - 15	NM_002834.3
RAF1	Exon 02 - 17	NM_002880.3
RBM20	Exon 01 - 14	NM_001134363.1
RYR2	Exon 01 - 105	NM_001035.2
SCN5A	Exon 02 - 06	NM_198056.2
	Exon 06A	NM_001099404.1
	Exon 07 - 28	NM_198056.2
SGCD	Exon 02 - 08	NM_172244.2
	Exon 09	NM_000337.5

## LMM Reference Sequences - GRCh37

### Cardiomyopathy

Gene	Region	Transcript
TAZ	Exon 01 - 11	NM_000116.3
TCAP	Exon 01 - 02	NM_003673.3
TMEM43	Exon 01 - 12	NM_024334.2
TNNC1	Exon 01 - 06	NM_003280.2
TNNI3	Exon 01 - 08	NM_000363.4
TNNT2	Exon 02 - 04	NM_001001430.1
	Exon 04A	NM_000364.2
	Exon 05 - 16	NM_001001430.1
TPM1	Exon 01	NM_000366.5
	Exon 01A	NM_001018020.1
	Exon 02	NM_000366.5
	Exon 02A	NM_001018008.1
	Exon 03 - 05, 6A	NM_000366.5
	Exon 06B	NM_001018005.1
	Exon 07 - 09	NM_000366.5
	Exon 10	NM_001018005.1
	Exon 10A	NM_001018008.1
	Exon 10B	NM_001018020.1
TRDN	Exon 01 - 06	NM_006073.3
	Exon 06D	NM_001256022.1
	Exon 07	NM_006073.3
	Exon 08C	NM_001256021.1
	Exon 09	NM_006073.3
	Exon 09B	NM_001256020.1
	Exon 11	NM_006073.3
	Exon 12	NM_001251987.1
	Exon 13 - 20	NM_006073.3
	Exon 21	NM_001251987.1
	Exon 22 - 41	NM_006073.3
TTN*	Exon 02 - 44	NM_133378.4
	Exon 44A	NM_133432.3
	Exon 44B	NM_133437.3
	Exon 45	NM_133378.4
	Exon 45B	NM_133432.3
	Exon 46 - 152	NM_133378.4
	Exon 156 - 312	NM_133378.4
<i>Not covered</i>	<i>Exon 45A</i>	<i>NM_133379.3</i>
	<i>Exon 154 - 153</i>	<i>NM_133378.4</i>
TTR	Exon 01 - 04	NM_000371.3
VCL	Exon 01 - 22	NM_014000.2

### Congenital Heart Disease

Gene	Region	Transcript
ELN	Exon 01 - 07	NM_000501.2
	Exon 08	NM_001081754.1
	Exon 09 - 21	NM_000501.2
	Exon 21A	Chromosome 7 (hg19)
	Exon 22	NM_000501.2
	Exon 23	Chromosome 7 (hg19)
	Exon 24	NM_000501.2
	Exon 25	Chromosome 7 (hg19)
	Exon 26 - 33	NM_000501.2
GATA4	Exon 01 - 07	NM_002052.3
JAG1	Exon 01 - 26	NM_000214.2
NKX2-5	Exon 01 - 02	NM_004387.3
	Exon 02B	NM_001166176.1

### Connective Tissue

Gene	Region	Transcript
ACTA2	Exon 02 - 09	NM_001613.2
CBS	Exon 03 - 16	NM_001178009.1
	Exon 17B	NM_001178008.1
COL3A1	Exon 01 - 51	NM_000090.3
FBN1	Exon 01 - 65	NM_000138.4
FBN2	Exon 01 - 65	NM_001999.3
MED12*	Exon 22 only	NM_005120.2
MYH11	Exon 02 - 42	NM_001040113.1
	Exon 42A	NM_001040114.1
MYLK	Exon 04 - 34	NM_053025.3
SKI	Exon 01 - 07	NM_003036.3
SLC2A10	Exon 01 - 05	NM_030777.3
SMAD3	Exon 01	NM_005902.3
	Exon 01B	NM_001145103.1
	Exon 02 - 09	NM_005902.3
TGFB2	Exon 01 - 08	NM_001135599.2
TGFBR1	Exon 01 - 09	NM_004612.2
TGFBR2	Exon 01	NM_003242.5
	Exon 01A	NM_001024847.2
	Exon 02 - 07	NM_003242.5

### Ectodermal Dysplasia

Gene	Region	Transcript
EDA	Exon 01 - 08	NM_001399.4
GJB6 (CX30)	Exon 03	NM_006783.4
GJB2 (CX26)	Exon 01 - 02	NM_004004.5
DSP	Exon 01 - 24	NM_004415.2

### RASopathies/Noonan Spectrum Disorders

Gene	Region	Transcript
BRAF	Exon 01 - 18	NM_004333.4
CBL	Exon 01 - 16	NM_005188.2
HRAS	Exon 02 - 04	NM_005343.2
	Exon 05A	NM_176795.3
	Exon 05B	NM_005343.2
KRAS	Exon 02 - 03	NM_004985.3
	Exon 04 - 05	NM_033360.2
	Exon 06	NM_004985.3
MAP2K1	Exon 01 - 11	NM_002755.3
MAP2K2	Exon 01 - 11	NM_030662.3
NF1	Exon 01 - 58	NM_001042492.2
NRAS	Exon 02 - 04	NM_002524.3
PTPN11	Exon 01 - 15	NM_002834.3
RAF1	Exon 02 - 17	NM_002880.3
RIT1	Exon 01 - 06	NM_001256821.1
SHOC2*	Exon 02 only	NM_007373.3
SOS1	Exon 01 - 23	NM_005633.3
SPRED1	Exon 01 - 07	NM_152594.2

\* A few exons have been excluded due to technical difficulties

## LMM Reference Sequences - GRCh37

### Hearing Loss

Gene	Region	Transcript
ACTG1	Exon 02 - 06	NM_001199954.1
ATP6V1B1	Exon 01 - 14	NM_001692.3
BSND	Exon 01 - 04	NM_057176.2
CACNA1D	Exon 01 - 08 Exon 08B Exon 09 - 49	NM_000720.2 NM_001128840.1 NM_000720.2
CCDC50	Exon 01 - 12	NM_178335.2
CDH23	Exon 01A Exon 02 - 10 Exon 11A Exon 12 - 13 Exon 14A Exon 15 - 25 Exon 26A Exon 27 - 31 Exon 32A Exon 33 - 70	NM_001171935.1 NM_022124.5 NM_001171932.1 NM_022124.5 NM_052836.3 NM_022124.5 NM_001171931.1 NM_022124.5 NM_001171930.1 NM_022124.5
CEACAM16	Exon 02 - 07	NM_001039213.2
CIB2	Exon 01 - 06	NM_006383.2
CLDN14	Exon 03	NM_001146077.1
CLPP	Exon 01 - 06	NM_006012.2
CLRN1*	Exon 01 Exon 01A Exon 02 - 03 Exon 03A Exon 03C <i>Not covered</i> Exon 01B	NM_174878.2 NM_052995.2 NM_174878.2 NM_052995.2 NM_001195794.1 NM_174880.1
COCH	Exon 01A Exon 03 - 12	NM_001135058.1 NM_004086.2
COL11A2	Exon 01 - 04 Exon 05A Exon 06 - 66	NM_080680.2 NM_001163771.1 NM_080680.2
DFNA5	Exon 02 - 10	NM_004403.2
DFNB31	Exon 01 - 12	NM_015404.3
DFNB59	Exon 02 - 07	NM_001042702.3
DIABLO	Exon 02 - 07	NM_019887.4

### Hearing Loss

Gene	Region	Transcript
DIAPH1	Exon 01 - 28	NM_005219.4
EDN3	Exon 01 - 03 Exon 04A Exon 04B	NM_207034.1 NM_207032.1 NM_207033.1
EDNRB	Exon 01 - 07 Exon 07B Exon 08	NM_001201397.1 NM_003991.3 NM_001201397.1
ESPN*	Exon 02 - 03, 05 - 06, 09 - 13 <i>Not covered</i> Exon 01, 04, 07-08	NM_031475.2 NM_031475.2
ESRRB	Exon 04 - 11	NM_004452.3
EYA1	Exon 01A Exon 02 - 17	NM_172060.2 NM_172058.2
EYA4	Exon 02 - 18 Exon 18A Exon 19 - 20	NM_004100.4 NM_172105.3 NM_004100.4
GIPC3	Exon 01 - 06	NM_133261.2
GJB2	Exon 01 - 02	NM_004004.5
GJB6	Exon 03	NM_006783.4
GPR98	Exon 01 - 90	NM_032119.3
GPSM2	Exon 02 - 15	NM_013296.4
GRHL2	Exon 01 - 16	NM_024915.3
GRXCR1	Exon 01 - 04	NM_001080476.2
HARS	Exon 01 - 13	NM_002109.3
HARS2	Exon 01 - 13	NM_012208.2
HGF	Exon 01 - 04 Exon 05A Exon 06 - 08 Exon 08A Exon 09 - 11, 13 - 18 <i>Not covered</i> Exon 12	NM_000601.4 NM_001010934.1 NM_000601.4 NM_001010931.1 NM_000601.4 NM_000601.4
HSD17B4	Exon 01 Exon 01A Exon 02 - 25	NM_001199291.1 NM_000414.3 NM_001199291.1
ILDRL1	Exon 01 - 08	NM_001199799.1

### Hearing Loss

Gene	Region	Transcript
KARS	Exon 01A Exon 02 - 15	NM_005548.2 NM_001130089.1
KCNE1	Exon 03	NM_000219.3
KCNQ1	Exon 01 Exon 01A Exon 02 - 16	NM_000218.2 NM_181798.1 NM_000218.2
KCNQ4*	Exon 02 - 14 <i>Not covered</i> Exon 01	NM_004700.3 NM_004700.3
LARS2	Exon 03 - 22	NM_015340.3
LHFPL5	Exon 01 - 03	NM_182548.3
LOXHD1	Exon 01 - 32 Exon 32A Exon 33 - 40 Exon 40A	NM_144612.6 NM_001145472.2 NM_144612.6 NM_001145472.2
LRTOMT*	Exon 04 Exon 04B Exon 05 Exon 05B Exon 06 - 07 <i>Not covered</i> Exon 03B, 06B Exon 06A	NM_001145308.1 NM_001145307.1 NM_001145308.1 NM_001145307.1 NM_001145308.1 NM_001145307.1 NM_145309.2
MARVELD2	Exon 02 - 07	NM_001038603.2
MIR96	Exon 01	NR_029512.1
MITF	Exon 01 Exon 01A Exon 01B Exon 01C Exon 02 - 03 Exon 03E Exon 04 - 06 Exon 06A Exon 08 - 10	NM_198159.2 NM_000248.3 NM_006722.2 NM_198177.2 NM_198159.2 NM_001184968.1 NM_198159.2 NM_000248.3 NM_198159.2
MSRB3	Exon 01A Exon 03 - 08	NM_198080.3 NM_001193460.1
MTRNR1	Exon 01	mitochondrial
MTTS1	Exon 01	mitochondrial
MYH14	Exon 02 - 43	NM_001145809.1

## LMM Reference Sequences - GRCh37

### Hearing Loss

Gene	Region	Transcript
MYH9	Exon 02 - 41	NM_002473.4
MYO15A	Exon 02 (c.-15-c.1794,c.2946-c.3609+15), Exons 3-66	NM_016239.3
<i>Not covered</i>	Exon 02 (c.1795-c.2945)	NM_016239.3
MYO3A	Exon 03 - 35	NM_017433.4
MYO6	Exon 02 - 35	NM_004999.3
MYO7A	Exon 02 - 26	NM_000260.3
	Exon 27A	NM_001127179.2
	Exon 28 - 49	NM_000260.3
OTOA	Exon 01	NM_144672.3
	Exon 01A	NM_001161683.1
	Exon 02 - 10	NM_144672.3
	Exon 10A	NM_170664.2
	Exon 11 - 21, 28	NM_144672.3
OTOF	Exon 01	NM_194248.2
	Exon 01A	NM_194322.2
	Exon 02 - 04, 05A	NM_194248.2
	Exon 06 - 18, 20 - 46	NM_194248.2
	Exon 47A	NM_194323.2
OTOG	Exon 01 - 55	NM_001277269.1
OTOGL	Exon 01 - 58	NM_173591.3
P2RX2	Exon 01 - 09	NM_174873.1
	Exon 10B	NM_170683.2
PAX3	Exon 01 - 02	NM_001127366.2
	Exon 03A, 04A	NM_000438.5
	Exon 05	NM_001127366.2
	Exon 05B	NM_013942.4
	Exon 07	NM_001127366.2
	Exon 08C	NM_181457.3
	Exon 08E	NM_181461.3
	Exon 10D	NM_181459.3
PCDH15	Exon 02 - 03	NM_033056.3
	Exon 03A	NM_001142769.1
	Exon 04 - 13	NM_033056.3
	Exon 13A	NM_001142769.1
	Exon 14 - 33	NM_033056.3
	Exon 34B	NM_001142770.1
	Exon 35A, 36A	NM_001142769.1
	Exon 36C	NM_001142771.1
	Exon 37A	NM_001142769.1
POU3F4	Exon 01	NM_000307.3

### Hearing Loss

Gene	Region	Transcript
POU4F3	Exon 01 - 02	NM_002700.2
PRPS1	Exon 01 - 07	NM_002764.3
RDX	Exon 02 - 14	NM_002906.3
SERPINB6	Exon 02C - 07C	NM_004568.5
SIX1	Exon 01 - 02	NM_005982.3
SLC26A4	Exon 01 - 21	NM_000441.1
SMPX	Exon 02 - 04	NM_014332.1
SOX10	Exon 02 - 04	NM_006941.3
STRC	Exon 01 - 29	NM_153700.2
SYNE4	Exon 01 - 08	NM_001039876.1
TBC1D24	Exon 02 - 08	NM_001199107.1
TECTA	Exon 01 - 23	NM_005422.2
TIMM8A	Exon 01 - 02	NM_004085.3
	Exon 01A	NM_001145951.1
TMC1	Exon 05 - 24	NM_138691.2
TMIE	Exon 01 - 04	NM_147196.2
TMPRSS3	Exon 02 - 08	NM_024022.2
	Exon 09A	NM_032405.1
	Exon 10 - 13	NM_024022.2
TPRN*	Exon 02 - 04	NM_001128228.2
<i>Not covered</i>	Exon 01	NM_001128228.2
TRIOBP*	Exon 03 - 06, 08 - 17	NM_001039141.2
	Exon 17A	NM_138632.2
	Exon 18 - 23	NM_001039141.2
<i>Not covered</i>	Exon 01A	NM_138632.2
TSPEAR	Exon 01 - 12	NM_144991.2
USH1C	Exon 01 - 15	NM_153676.3
	Exon 15B	NM_005709.3
	Exon 16 - 27	NM_153676.3
USH1G	Exon 01 - 03	NM_173477.2
USH2A	Exon 02 - 20	NM_206933.2

### Hearing Loss

Gene	Region	Transcript
USH2A cont.	Exon 21A	NM_007123.5
	Exon 22 - 40	NM_206933.2
	Intron 40	NM_206933.2
	Exon 41 - 72	NM_206933.2
WFS1	Exon 02 - 08	NM_006005.3

### Pulmonary

Gene	Region	Transcript
ABCA3	Exon 04 - 33	NM_001089.2
ACVRL1	Exon 01A	NM_001077401.1
	Exon 03 - 10	NM_000020.2
AP3B1	Exon 01 - 27	NM_003664.3
ASCL1	Exon 01 (c.351 - c.*15)	NM_004316.3
<i>Not covered</i>	Exon 01 (c.1 - c.350)	NM_004316.3
BDNF	Exon 01A	NM_170731.4
	Exon 01E	NM_001143809.1
	Exon 01I	NM_170734.3
	Exon 02	NM_001143810.1
	Exon 02E	NM_001143809.1
BLOC1S3	Exon 02	NM_212550.3
BLOC1S6	Exon 01 - 05	NM_012388.2
BMPR2	Exon 01 - 13	NM_001204.6
CCDC39	Exon 01 - 20	NM_181426.1
CCDC40*	Exon 01 - 20	NM_017950.2
<i>Not covered</i>	Exon 18A	NM_001243342.1
CFTR	Exon 01 - 12	NM_000492.3
	Intron 12	NM_000492.3
	Exon 13 - 22	NM_000492.3
	Intron 22	NM_000492.3
	Exon 23 - 27	NM_000492.3
CSF2RA*	Exon 04 - 11	NM_001161529.1
	Exon 11A	NM_172246.2
	Exon 12 - 13	NM_001161529.1
	Exon 13B	NM_001161531.1
<i>Not covered</i>	Exon 09A	NM_001161530.1
CSF2RB	Exon 02 - 14	NM_000395.2
DNAAF1	Exon 01 - 12	NM_178452.4
DNAAF2	Exon 01 - 03	NM_018139.2

## LMM Reference Sequences - GRCh37

### Pulmonary

Gene	Region	Transcript
DNAH11	Exon 01 - 82	NM_003777
DNAH5	Exon 01 - 79	NM_001369.2
DNAI1	Exon 01 - 20	NM_012144.2
DNAI2	Exon 02 - 13	NM_023036.4
DNAL1	Exon 01 - 03	NM_031427.3
	Exon 05 - 09	NM_001201366.1
DOCK8	Exon 01 - 48	NM_203447.3
DTNBP1	Exon 01 - 08	NM_032122.4
	Exon 09A	NM_183040.2
	Exon 10	NM_032122.4
EDN3	Exon 01 - 03	NM_207034.1
	Exon 04A	NM_207032.1
	Exon 04B	NM_207033.1
EFEMP2	Exon 02 - 11	NM_016938.4
ELMOD2	Exon 02 - 09	NM_153702.3
ELN	Exon 01 - 07	NM_000501.2
	Exon 08	NM_001081754.1
	Exon 09 - 21	NM_000501.2
	Exon 21A	Chromosome 7 (hg19)*
	Exon 22	NM_000501.2
	Exon 23	Chromosome 7 (hg19)*
	Exon 24	NM_000501.2
	Exon 25	Chromosome 7 (hg19)*
	Exon 26 - 33	NM_000501.2
ENG	Exon 01 - 13	NM_001114753.1
	Exon 14A	NM_000118.2
	Exon 15	NM_001114753.1
FBLN5	Exon 01 - 11	NM_006329.3
FBN1	Exon 01 - 65	NM_000138.4
FLCN	Exon 04 - 07	NM_144997.5
	Exon 08A	NM_144606.5
	Exon 09 - 14	NM_144997.5
FOXF1	Exon 01 - 02	NM_001451.2
GDNF	Exon 01 - 03	NM_001190468.1
HPS1	Exon 03 - 09	NM_000195.2

### Pulmonary

Gene	Region	Transcript
HPS1 cont.	Exon 10A	NM_182639.1
	Exon 11 - 20	NM_000195.2
HPS3	Exon 01 - 17	NM_032383.3
HPS4	Exon 01A	NM_152841.1
	Exon 02, 04 - 14	NM_022081.4
HPS5	Exon 02 - 23	NM_181507.1
HPS6	Exon 01	NM_024747.4
HRAS	Exon 02 - 04	NM_005343.2
	Exon 05A	NM_176795.3
	Exon 05B	NM_005343.2
LTBP4	Exon 01	NM_001042544.1
	Exon 01A	NM_001042545.1
	Exon 01B	NM_003573.2
	Exon 02	NM_001042544.1
	Exon 03B	NM_003573.2
	Exon 04 - 33	NM_001042544.1
MUC5B*	Promoter (c.-3133G>T)	NM_002458.2
	Exon 01 - 16, 18 - 24	NM_002458.2
	Exon 27 - 30, 32 - 49	NM_002458.2
	<i>Not covered</i> Exon 17, 25 - 26, 31	<i>NM_002458.2</i>
NF1	Exon 01 - 58	NM_001042492.2
NKX21	Exon 01 - 03	NM_001079668.2
NME8	Exon 03 - 17	NM_016616.4
PHOX2B	Exon 01 - 03	NM_003924.3
RET	Exon 01 - 18	NM_020975.4
	Exon 19A	NM_020630.4
	Exon 20	NM_020975.4
RPGR*	Exon 01 - 12, 14	NM_001034853.1
	Exon 16A	NM_000328.2
	Exon 17A	NM_000328.2
	Exon 18A	NM_000328.2
	Exon 19A	NM_000328.2
	<i>Not covered</i> Exon 13, 15	<i>NM_001034853.1</i>
RSPH1	Exon 01 - 09	NM_080860.2
RSPH4A	Exon 01 - 06	NM_001010892.2
RSPH9	Exon 01 - 02	NM_001193341.1

### Pulmonary

Gene	Region	Transcript
RSPH9 cont.	Exon 03A	NM_152732.4
	Exon 04 - 06	NM_001193341.1
SCNN1A	Exon 01	NM_001159576.1
	Exon 01A	NM_001159575.1
	Exon 02 - 12	NM_001159576.1
SCNN1B	Exon 02 - 13	NM_000336.2
SCNN1G	Exon 02 - 13	NM_001039.3
SERPINA1	Exon 04 - 07	NM_001127701.1
SFTPA1	Exon 02B	NM_001164645.1
	Exon 03 - 06	NM_001093770.2
SFTPA2	Exon 03 - 06	NM_001098668.2
SFTPB	Exon 02 - 11	NM_000542.3
SFTPC	Exon 01 - 04	NM_001172357.1
	Exon 05A	NM_003018.3
SFTPD	Exon 02 - 08	NM_003019.4
SMAD9	Exon 02 - 07	NM_001127217.2
STAT3	Exon 02 - 24	NM_139276.2
TERC	Exon 01	NR_001566.1
TERT	Exon 01 - 16	NM_198253.2
TSC1	Exon 03 - 23	NM_000368.4
TSC2	Exon 02 - 42	NM_000548.3

\* A few exons have been excluded due to technical difficulties

## LMM Reference Sequences - GRCh37

### Myopathy

Gene	Region	Transcript
ACTA1	Exon 02 - 07	NM_001100.3
ANO5	Exon 01 - 22	NM_213599.2
BAG3	Exon 01 - 04	NM_004281.3
BIN1	Exon 01 - 10	NM_139343.2
	Exon 10D	NM_139346.2
	Exon 11 - 19	NM_139343.2
CAPN3	Exon 01 - 24	NM_000070.2
CAV3	Exon 01 - 02	NM_033337.2
CFL2*	Exon 01A	NM_021914.7
	Exon 02 - 04	NM_138638.4
<i>Not covered</i>	Exon 01	NM_138638.4
CRYAB	Exon 01 - 03	NM_001885.1
DAG1	Exon 05C - 06C	NM_001177634.2
DES	Exon 01 - 09	NM_001927.3
DMD	Exon 01	NM_004006.2
	Exon 01A	NM_000109.3
	Exon 01C	NM_004009.3
	Exon 01E	NM_004011.3
	Exon 01F	NM_004012.3
	Exon 01H	NM_004014.2
	Exon 01J	NM_004016.2
	Exon 02 - 09	NM_004006.2
	Exon 09M	NM_004019.2
	Exon 10 - 35	NM_004006.2
	Exon 35O	NM_004021.2
	Exon 36 - 69, 71 - 78	NM_004006.2
DNAJB6	Exon 02 - 07	NM_058246.3
	Exon 08A	NM_005494.2
	Exon 09 - 10	NM_058246.3
DNM2	Exon 01 - 10	NM_001005360.2
	Exon 10B	NM_001005361.2
	Exon 11 - 21	NM_001005360.2
DYSF	Exon 01	NM_001130987.1
	Exon 01H	NM_001130981.1
	Exon 02 - 18	NM_001130987.1
	Exon 18I	NM_001130982.1
	Exon 19 - 56	NM_001130987.1
EMD	Exon 01 - 06	NM_000117.2

### Myopathy

Gene	Region	Transcript
FHL1	Exon 01C	NM_001159699.1
	Exon 01E	NM_001159701.1
	Exon 02E	NM_001159701.1
	Exon 04 - 06	NM_001159702.2
	Exon 06E	NM_001159701.1
	Exon 07	NM_001159702.2
FKRP	Exon 04	NM_001039885.2
FKTN	Exon 03 - 11	NM_001079802.1
	Exon 12B	NM_001198963.1
FLNC	Exon 01 - 48	NM_001458.4
ISPD	Exon 01 - 10	NM_001101426.3
KLHL40	Exon 01 - 06	NM_152393.2
LDB3	Exon 01 - 04	NM_007078.2
	Exon 05 - 06	NM_001080116.1
	Exon 07 - 08	NM_007078.2
	Exon 09	NM_001080116.1
	Exon 10 - 16	NM_007078.2
LMNA	Exon 01 - 09	NM_170707.2
	Exon 10	NM_005572.3
	Exon 11 - 12	NM_170707.2
MTM1	Exon 02 - 15	NM_000252.2
MYH7	Exon 03 - 40	NM_000257.2
MYOT	Exon 02 - 10	NM_006790.2
NEB*	Exon 03 - 89, 106 - 183	NM_001271208.1
<i>Not covered</i>	Exon 90 - 105 (repeat region)	NM_001271208.1
PLEC*	Exon 01	NM_201380.2
	Exon 01B	NM_201379.1
	Exon 01C	NM_201381.1
	Exon 01D	NM_201382.2
	Exon 01E	NM_201383.1
	Exon 01F	NM_201384.1
	Exon 02	NM_201380.2
	Exon 02A	NM_000445.3
	Exon 03 - 32	NM_201380.2
<i>Not covered</i>	Exon 01G	NM_201378.2
POMGNT1	Exon 02 - 22	NM_017739.3
POMT1	Exon 02 - 20	NM_007171.3

### Myopathy

Gene	Region	Transcript
POMT2	Exon 01 - 21	NM_013382.5
RYR1	Exon 01 - 106	NM_000540.2
SEPN1*	Exon 02 - 13	NM_020451.2
<i>Not covered</i>	Exon 01	NM_020451.2
SGCA	Exon 01 - 09	NM_000023.2
SGCB	Exon 01 - 06	NM_000232.4
SGCD	Exon 02 - 08	NM_172244.2
	Exon 09	NM_000337.5
SGCG	Exon 02 - 08	NM_000231.2
TCAP	Exon 01 - 02	NM_003673.3
TNNT1*	Exon 02 - 04, 06 - 14	NM_003283.4
<i>Not covered</i>	Exon 05	NM_003283.4
TPM2	Exon 01 - 06	NM_213674.1
	Exon 06A	NM_003289.3
	Exon 07 - 09	NM_213674.1
	Exon 09A	NM_003289.3
TPM3	Exon 01	NM_152263.2
	Exon 01B	NM_001043351.1
	Exon 02 - 05	NM_152263.2
	Exon 05A	NM_153649.3
	Exon 06 - 08	NM_152263.2
	Exon 08B	NM_001043351.1
	Exon 08D	NM_001043353.1
	Exon 09 - 10	NM_152263.2
TRIM32	Exon 02	NM_012210.3
TTN*	Exon 02 - 44	NM_133378.4
	Exon 44A	NM_133432.3
	Exon 44B	NM_133437.3
	Exon 45	NM_133378.4
	Exon 45A	NM_133379.3
	Exon 45B	NM_133432.3
	Exon 46 - 152	NM_133378.4
	Exon 156 - 312	NM_133378.4
<i>Not covered</i>	Exon 154 - 153	NM_133378.4

\* A few exons have been excluded due to technical difficulties

## LMM Reference Sequences - GRCh37

### Renal Disease

Gene	Region	Transcript
ACE*	Exon 01-12, 14-25	NM_000789.3
	Exon 01A	NM_152830.2
<i>Not covered</i>	Exon 13	NM_000789.3
ACTN4*	Exon 02-21	NM_004924.4
<i>Not covered</i>	Exon 01	NM_004924.4
ADAMTS13*	Exon 03-29	NM_139025.4
<i>Not covered</i>	Exon 02	NM_139025.4
AGXT	Exon 01-11	NM_000030.2
ALMS1*	Exon 02-23	NM_015120.4
<i>Not covered</i>	Exon 01	NM_015120.4
ANKS6	Exon 01-15	NM_173551.3
AP2S1	Exon 02-05	NM_004069.3
APOL1	Exon 03-06	NM_003661.3
	Exon 03C	NM_145343.2
APRT	Exon 02-05	NM_000485.2
AQP2	Exon 01-04	NM_000486.5
ATP6V0A4*	Exon 03, 05-14, 16-22	NM_020632.2
<i>Not covered</i>	Exon 04, 15	NM_020632.2
ATP6V1B1	Exon 01-14	NM_001692.3
AVP*	Exon 01, 03	NM_000490.4
<i>Not covered</i>	Exon 02	NM_000490.4
AVPR2	Exon 01, 03	NM_000054.4
AVPR2	Exon 02A	NM_001146151.1
BMP4	Exon 03-04	NM_001202.3
BSND	Exon 01-04	NM_057176.2
C1QA	Exon 02-03	NM_015991.2
C1QC	Exon 02-03	NM_172369.3
C3	Exon 01-41	NM_000064.2
CASR	Exon 02-07	NM_001178065.1
CD46	Exon 01-13	NM_002389.4
CD46	Exon 10G	NM_172350.2

### Renal Disease

Gene	Region	Transcript
CDC73	Exon 01-17	NM_024529.4
CEP290*	Exon 02-04, 06-15, 18-31, 33	NM_025114.3
<i>Not covered</i>	Exon 05,16,17,32	NM_025114.3
CFB	Exon 01-18	NM_001710.5
CFH	Exon 01-22	NM_000186.3
CFHR1	Exon 01-06	NM_002113.2
CFI	Exon 01-13	NM_000204.3
CLCN5	Exon 03-15	NM_001127898.2
	Exon 04C	NM_001272102.1
CLCNKB*	Exon 01A	NM_001165945.2
	Exon 02-12, 14-20	NM_000085.4
<i>Not covered</i>	Exon 13-14	NM_000085.4
CLDN16	Exon 01-05	NM_006580.3
CLDN19	Exon 01-05	NM_148960.2
	Exon 03B	NM_001185117.1
COL4A3*	Exon 01-35, 37-50, 52	NM_000091.4
<i>Not covered</i>	Exon 36, 51	NM_000091.4
COL4A4*	Exon 02-30, 32-48	NM_000092.4
<i>Not covered</i>	Exon 31	NM_000092.4
COL4A5	Exon 01-53	NM_033380.2
CTNS	Exon 03-13	NM_001031681.2
	Exon 12A	NM_004937.2
CUL3*	Exon 01-02, 04-16	NM_001257198.1
	Exon 01A	NM_003590.4
<i>Not covered</i>	Exon 03	NM_001257198.1
CYP11B2	Exon 01-09	NM_000498.3
CYP24A1	Exon 01-11	NM_000782.4
DGKE	Exon 02-12	NM_003647.2
ENPP1*	Exon 01-11, 13-25	NM_006208.2
<i>Not covered</i>	Exon 012	NM_006208.2
EYA1	Exon 01A	NM_172060.2
	Exon 02-17	NM_172058.2

### Renal Disease

Gene	Region	Transcript
FAH	Exon 01-14	NM_000137.2
FAM20A	Exon 01A-02A	NM_017565.3
	Exon 04-12	NM_001243746.1
FAM20C	Exon 01-10	NM_020223.3
FGF23	Exon 01-03	NM_020638.2
FRAS1	Exon 01-74	NM_025074.6
	Exon 42A	NM_001166133.1
GALNT3	Exon 02-11	NM_004482.3
GATA3	Exon 02-06	NM_002051.2
GCM2	Exon 01-05	NM_004752.3
GLA	Exon 01-07	NM_000169.2
	Intron 04	NM_000169.2
GNA11	Exon 01-07	NM_002067.2
GNAS	Exon 01-13	NM_080425.2
	Exon 01B	NM_001077488.2
	Exon 01D	NM_016592.2
	Exon 04B	NM_001077488.2
GRHPR	Exon 01-09	NM_012203.1
HNF1B	Exon 01-09	NM_000458.2
HOGA1	Exon 01-07	NM_138413.3
HPRT1	Exon 01-09	NM_000194.2
HSD11B2	Exon 01-05	NM_000196.3
INF2*	Exon 02-07, 09-22	NM_022489.3
	Exon 05B	NM_032714.2
	Exon 22A	NM_001031714.3
<i>Not covered</i>	Exon 08	NM_022489.3
INVS*	Exon 02-10, 12-18	NM_183245.2
	Exon 14A	NM_014425.3
<i>Not covered</i>	Exon 11	NM_183245.2
IQCB1	Exon 03-15	NM_001023570.2
ITGB4*	Exon 02-25, 27-40	NM_000213.3
	Exon 33A	NM_001005619.1
<i>Not covered</i>	Exon 26	NM_000213.3

## LMM Reference Sequences - GRCh37

### Renal Disease

Gene	Region	Transcript
KCNJ1	Exon 01C-02C	NM_000220.4
	Exon 02A	NM_153765.2
KLHL3	Exon 01-15	NM_017415.2
LAMB2	Exon 01-32	NM_002292.3
LMX1B	Exon 01-06, 08	NM_001174146.1
	Exon 07A	NM_001174147.1
MUC1*	Exon 01, 04-08	NM_001204286.1
	<i>Not covered</i> Exon 02-03	<i>NM_001204286.1</i>
MYH9	Exon 02-41	NM_002473.4
MYO1E*	Exon 01-02, 04-17, 19-28	NM_004998.3
	<i>Not covered</i> Exon 03, 18	<i>NM_004998.3</i>
NPHP1	Exon 01-20	NM_000272.3
NPHP3*	Exon 02-07, 09-27	NM_153240.4
	<i>Not covered</i> Exon 01, 08	<i>NM_153240.4</i>
NPHP4	Exon 02-30	NM_015102.3
NPHS1*	Exon 01-22, 24-25, 28	NM_004646.3
	<i>Not covered</i> Exon 23,26,27,29	<i>NM_004646.3</i>
NPHS2	Exon 01-08	NM_014625.2
NR3C2	Exon 02-09	NM_000901.4
OCRL*	Exon 02-24	NM_000276.3
	<i>Not covered</i> Exon 01	<i>NM_000276.3</i>
PAX2	Exon 01-11	NM_003990.3
	Exon 09B	NM_003988.3
PHEX	Exon 01-22	NM_000444.4
	Intron 22	NM_000444.4
PKD2	Exon 01-15	NM_000297.3
PKHD1	Exon 02-67	NM_138694.3
	Exon 61A	NM_170724.2
PLCE1*	Exon 01A	NM_001165979.1
	Exon 02-04, 06-32	NM_016341.3
	<i>Not covered</i> Exon 05	<i>NM_016341.3</i>
PTH	Exon 02-03	NM_000315.2

### Renal Disease

Gene	Region	Transcript
REN	Exon 01-10	NM_000537.3
SCNN1A	Exon 01-12	NM_001159576.1
	Exon 01A	NM_001159575.1
SCNN1B	Exon 02-13	NM_000336.2
SLC12A1	Exon 02-27	NM_000338.2
	Exon 05A	NM_001184832.1
SLC12A3*	Exon 01-21, 23-26	NM_000339.2
	<i>Not covered</i> Exon 22	<i>NM_000339.2</i>
SLC22A12	Exon 01-10	NM_144585.3
SLC2A2	Exon 01-11	NM_000340.1
SLC34A3*	Exon 02B	NM_001177317.1
	Exon 03-05, 07, 10-13	NM_080877.2
	<i>Not covered</i> Exon 06,08,09	<i>NM_080877.2</i>
SLC4A1	Exon 02-20	NM_000342.3
TRPC6	Exon 01-13	NM_004621.5
TSC1	Exon 03-23	NM_000368.4
TSC2	Exon 02-42	NM_000548.3
UMOD	Exon 02-12	NM_001278614.1
VDR	Exon 01A-03A	NM_001017536.1
	Exon 05-11	NM_001017535.1
WNK4	Exon 01-19	NM_032387.4
WT1*	Exon 01B	NM_001198551.1
	Exon 02-10	NM_024426.4
	<i>Not covered</i> Exon 01	<i>NM_024426.4</i>

\* A few exons have been excluded due to technical difficulties